

**Untersuchungen im Bereich:**

Medizinische Laboratoriumsdiagnostik

**Untersuchungsgebiete:**

Klinische Chemie

Immunologie

Humangenetik (Zytogenetik, Molekulare Humangenetik)

Mikrobiologie

Virologie

Transfusionsmedizin

**Untersuchungsgebiet: Klinische Chemie**

**Untersuchungsart:**

**Chromatographie (Flüssigkeitschromatographie LC-MS/MS)\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
2C-B	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
2C-I	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
3-Hydroxybromazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
5-Hydroxyindolessigsäure	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0111-00.3
6-Acetylcodein	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
6-MAM (6-Monoacetylmorphin)	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
7-Aminoclonazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
7-Aminoflunitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
7-Aminonitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
9-OH-Risperidon/Paliperidon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
10-OH-Carbazepin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Alpha-Hydroxyalprazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Alpha-Hydroxymidazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Alpha-Hydroxytriazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Alprazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Aminosäuren	EDTA-Plasma	LC-MS/MS	BIO-ING-SPCH-SAA-0065-01.1
Amiodaron	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0105-00
Amisulprid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Amitriptylin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00
Amphetamin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Amphetamine-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Aripiprazol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Benzodiazepine-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Benzoylcegonin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
BDB	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Booster-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Brivaracetam	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Bromazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Buprenorphin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Brotizolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Butylon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Cannabinoide-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Cathinon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Chlordiazepoxid	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Chlorprothixen	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Ciclosporin	ETDA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0079-00.2
Citalopram	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Clobazam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Clomipramin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0108-00
Clonazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Clozapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Cocaethylen	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Cocain	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Cocain-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Codein	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Dehydroaripiprazol	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Demoxepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Desalkylflurazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Desethylamiodaron	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0105-00
Desipramin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00
Desmethylcitalopram	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Norclomipramin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0108-00
Desmethylclozapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Nordoxepin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Desmethylflunitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Desmethylmirtazapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Desmethylsertralın	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
O-Desmethylvenlafaxin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Diazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Dihydrocodein	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Doxepin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00
Duloxetin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
EDDP	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Escitalopram	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Estazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Ethosuximid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Ethylglucuronid	Urin, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0061-01
Ethylsulfat	Urin, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0061-01
Everolimus	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0079-00.2
Fentanyl	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Felbamat	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Flecainid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0105-00
Fluconazol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0109-00
Flunitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Fluoxetin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Flupentixol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Flurazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Fluvoxamin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Gabapentin	Serum, EDTA-Plasma, Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02 BIO-ING-CHROM-SAA-0096-00.3
Haloperidol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Homocystein	NaF-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0114-00
Hydrocodon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Hydromorphon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Imipramin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00
Itraconazol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0109-00
Ketamin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Lacosamid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Lamotrigin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Levetiracetam	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Levomepromazin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Lorazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Lormetazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
LSD	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Maprotilin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0108-00
MBDB	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
MDA	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
MDEA	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
MDMA	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
MDPV	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Meconin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Medazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Meperidin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Mephedron	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Mescaline	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Methadon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Metanephrin / Normetanephrin / 3-Methoxythyramin	EDTA-Plasma, Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0002-00.5 BIO-ING-CHROM-SAA-0115-00.1
Methamphetamin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Methaqualon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Methylmalonsäure	Serum, Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0089-00.2
Methylon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Midazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Mirtazapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Morphin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Mycophenolsäure	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0110-00
Naltrexon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Naloxon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
N-Desmethylnesuximid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
N-Desmethyloanzapin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Nitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norbuprenorphin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norclobazam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norcocain	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norcodein	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Nordiazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norfentanyl	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Desmethylfluoxetin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Norketamin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Normeperidin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Norquetiapin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Nortapentadol	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Nortilidin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Nortriptylin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00
O-Desmethyltramadol	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
O-Desmethyl-Venlafaxin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Olanzapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Opiate-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Opioid-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Oxazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Oxcarbazepin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Oxycodon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Oxymorphon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Papaverin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Paroxetin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
PCP	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Perampanel	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Perazin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Pipamperon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
PMA	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Posaconazol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0109-00
Prazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Pregabalin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Pregabalin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Primidon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Promethazin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Promethazin	Urin	LC-MS/MS	ING-CHROM-SAA-0096-00.3
Propafenon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0105-00
Propoxyphen	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Psychedelika-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Quetiapin	Serum, EDTA-Plasma, Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00 BIO-ING-CHROM-SAA-0096-00.3
Risperidon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0095-00
Ritalinsäure	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Rufinamid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Serotonin	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0117-00
Sertralin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Sirolimus	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0079-00.2
Sufentanil	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Sultiam	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Synthetische Cannabinoide	Urin, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0087-00.3
Tabakalkaloide	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0016-00.3
Tacrolimus	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0079-00.2
Tapentadol	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Temazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
THC-COOH	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Thebain	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Tilidin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Topiramate	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Toxikologische Suchanalyse	Urin, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0087-00.3
Tramadol	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Triazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Trimipramin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0108-00
Trizyklische Antidepressiva (TCA Suchanalyse)	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00 BIO-ING-CHROM-SAA-0108-00
Trizyklische Antidepressiva (TCA Suchanalyse)	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0087-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Venlafaxin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Vigabatrin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
25-OH-Vitamin D (gesamt)	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0116-00
Voriconazol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0109-00
Zaleplon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Z-Hypnotika-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Ziprasidon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-00
Zolpidem	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Zonisamid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Zopiclon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3

**Untersuchungsart:  
Chromatographie (Gaschromatographie (GC))\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Organische Säuren	Urin	GC-MS	BIO-ING-SPCH-SAA-0020-00.3
Ultralangkettige Fettsäuren	Serum	GC-MS	BIO-ING-SPCH-SAA-0032-00.4

**Untersuchungsart:  
Chromatographie (Hochleistungsflüssigkeitschromatographie (HPLC))\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Vitamin B 1	EDTA-Blut	HPLC-FD	BIO-ING-CHROM-SAA-0069-02.1
Vitamin B 2	EDTA-Blut	HPLC-FD	BIO-ING-CHROM-SAA-0010-01.2
Vitamin B 6	EDTA-Plasma, Serum	HPLC-FD	BIO-ING-CHROM-SAA-0099-00.1
Vitamin K1	Serum	HPLC-FD	BIO-ING-CHROM-SAA-0088-01.3
Aminosäuren	Urin, Liquor	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0007-01
Beta-Carotin	Serum, EDTA-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0007-01.1
CDT (Carbohydrate-Deficient-Transferrin)	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0090-01.1
Coenzym Q10	Serum, EDTA-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0026-01
HbA	EDTA-Blut	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0029-01.1
HbA2	EDTA-Blut	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0029-01.1
HbF	EDTA-Blut	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0029-01.1
HbX	EDTA-Blut	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0029-01.1
Hippursäure/Methylhippursäure	Urin	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0098-00.1
Vitamin A	Serum, EDTA-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0009-02.2

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Vitamin C	Lithium-Heparin-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0077-01.1
Vitamin E	Serum, EDTA-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0009-02.2

**Untersuchungsart:  
Durchflusszytometrie (inkl. Partikeleigenschaftsbestimmungen)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Großes Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.2
Kleines Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.2
Retikulozyten	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.2
Thrombozyten	EDTA-Blut, Spezialröhrchen Thromboexact	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.2
Zellzahl	Körperflüssigkeiten wie Ascites, Dialysat, Liquor, Punktat, Synovia	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0002-00.5

**Untersuchungsart:  
Elektrochemische Untersuchungen\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Calcium (ionisiert)	Serum	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0005-01.1
Carboxyhämoglobin (COHb)	Heparinblut, EDTA-Blut, Citrat-Blut, Kapillarblut	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0005-01.1
Chlorid	Serum, Heparin-Plasma	ionenselektive Elektroden	BIO-ING-AUTO-SAA-0020-00.1
Chlorid	Urin	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0021-01.3
Kalium	Urin	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0021-01.3
Kalium	Serum, Heparin-Plasma	ionenselektive Elektroden	BIO-ING-AUTO-SAA-0036-00.2
Methämoglobin	Heparinblut, EDTA-Blut, Citrat-Blut, Kapillarblut	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0005-01.1
Natrium	Urin	ionenselektive Elektroden	BIO-ING-SPCH-SAA-0021-01.3
Natrium	Serum, Heparin-Plasma	ionenselektive Elektroden	BIO-ING-AUTO-SAA-0040-00.1



**Untersuchungsart:  
Elektrophorese\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Eiweisselektrophorese	Serum	Kapillarelektrophorese	BIO-ING-IMMU-SAA-0050-03.2
Alkalische Phosphatase-Isoenzyme	Serum	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0024-00.1
CK-Isoenzyme	Serum	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0030-00.2
HB-Differenzierung im alkalischen Milieu	EDTA-Blut	Agarose Elektrophorese	BIO-ING-SPCH-SAA-0060-00.2
HB-Differenzierung im sauren Milieu	EDTA-Blut	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0060-00.2
LDH-Isoenzyme	Serum	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0004-00.3
Mucopolysaccharide	Urin	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0014-00.4

**Untersuchungsart:  
Koagulometrie\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
APC-Resistenz	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0010-01
Fibrinogen	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0023-02.1
Lupus Antikoagulans (Lupus Inhibitoren)	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0018-01
Quick	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0023-02.1
Quick	Citratplasma	mechanisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0003-01.5
INR	Citratplasma	Berechnung	BIO-ING-HÄMA-SAA-0023-02.1
INR	Citratplasma	Berechnung	BIO-ING-HÄMA-SAA-0003-01.5
Thromboplastinzeit (aPTT)	Citratplasma	mechanisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0003-01.5
Thrombinzeit	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0023-02.1
Thromboplastinzeit (aPTT)	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0023-02.1

**Untersuchungsart:  
Ligandenassays\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
17-Hydroxy(OH)-Progesteron	Serum	RIA	BIO-ING-RIA-SAA-0010-00.7
Adiponectin	Serum	ELISA	BIO-ING-SPCH-SAA-0063-01
Adrenocorticotropes Hormon (ACTH)	EDTA-Plasma	CLIA	BIO-ING-AUTO-SAA-0152-00
Aldosteron	Serum, EDTA-Plasma	CLIA	BIO-ING-AUTO-SAA-0135-00
Alpha-Fetoprotein (AFP)	Serum	ECLIA	BIO-ING-AUTO-SAA-0043-00.1
Amphetamine	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Androstaniol Glucuronid	Serum	RIA	BIO-ING-RIA-SAA-0003-00.6
Androstendion	Serum	ECLIA	BIO-ING-AUTO-SAA-0212-00.1
Anti-Müller-Hormon	Serum, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0146-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Benzodiazepine	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Benzoylcegonin	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Beta-2-Mikroglobulin	Serum, Urin, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0139-00
Beta-Amyloid 1-40	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.4
Beta-Amyloid 1-42	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.4
Beta-CrossLaps (CTX)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0066-00
Beta-hCG, freies	Serum	ECLIA	BIO-ING-AUTO-SAA-0182-00.2
Biotin	Serum	ELISA	BIO-ING-SPCH-SAA-0063-01
Buprenorphin	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
CA 50	Serum	RIA	BIO-ING-RIA-SAA-0009-00.6
Calcitonin	Serum, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0144-00.1
Cannabinoide	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Carbamazepin	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0190-00
Ciclosporin A	EDTA-Blut	ECLIA	BIO-ING-AUTO-SAA-0184-00
Cortisol	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0049-00
Cortisol	Urin	ECLIA	BIO-ING-AUTO-SAA-0161-00
C-Peptid	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0050-00
Dehydroepiandrosteron-Sulfat (DHEA-S)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0052-00
Diaminoxidase Aktivität	Serum	REA	BIO-ING-RIA-SAA-0008-00.3
Diaminoxidase Konzentration	Serum	ELISA	BIO-ING-RIA-SAA-0032-00
Digitoxin	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0130-00.1
Digoxin	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0129-00.1
Drogenscreening	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
EDDP	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Erythropoetin (EPO)	Serum, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0150-00
Ethylglucuronid	Urin	ELISA	BIO-ING-CHROM-SAA-0057-02.3
Ferritin	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0054-00
Follikelstimulierendes Hormon (FSH)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0055-00.1
Folsäure in Erythrozyten	EDTA-Blut, Heparinblut	ECLIA	BIO-ING-AUTO-SAA-0138-00
Gastrin	Serum	CLIA	BIO-ING-AUTO-SAA-0151-00
Gentamicin	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0194-00
Hämoglobin (immunologisch)	Faeces	ELISA	BIO-ING-SPCH-SAA-0063-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Hämoglobin-Haptogl.-Komplex (immunol.)	Faeces	ELISA	BIO-ING-SPCH-SAA-0063-01
hCG/ $\beta$ -hCG	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0056-00
Inhibin A (Dimer)	Serum	ELISA	BIO-ING-SPCH-SAA-0050-00.2
Inhibin B	Serum	ELISA	BIO-ING-SPCH-SAA-0055-00
Insulin	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0058-00
Interleukin-2-Rezeptor, löslicher	Serum	CLIA	BIO-ING-AUTO-SAA-0149-00
Leptin	Serum	RIA	BIO-ING-RIA-SAA-0028-00.3
Luteinisierendes Hormon (LH)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0059-00.1
M2-PK	Faeces	ELISA	BIO-ING-SPCH-SAA-0044-01
Methotrexat	Serum	EIA	BIO-ING-SPCH-SAA-0021-01.3
Neopterin	Serum, EDTA-Plasma	ELISA	BIO-ING-SPCH-SAA-0063-01
NT-pro BNP	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0060-00.2
Opiate	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Ostase	Serum	CLIA	BIO-ING-AUTO-SAA-0137-00
Osteocalcin	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0216-00
Östradiol	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0053-00
Östriol, freies	Serum	RIA	BIO-ING-RIA-SAA-0023-00.6
Östron	Serum	RIA	BIO-ING-RIA-SAA-0017-00.6
Pankreas-Elastase	Serum	ELISA	BIO-ING-SPCH-SAA-0027-00.1
PAPP-A	Serum	ECLIA	BIO-ING-AUTO-SAA-0183-00.2
Parathormon, intakt (PTH)	EDTA-Plasma, Serum	ECLIA	BIO-ING-AUTO-SAA-0064-00
Parathormon-related Protein	EDTA-Plasma	RIA	BIO-ING-RIA-SAA-0004-00.9
Phencyclidin	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.6
Phenobarbital	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0196-00
Phenytoin (Diphenylhydantoin)	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0192-00
Phospho-Tau	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.4
PIGF	Serum	ECLIA	BIO-ING-AUTO-SAA-0200-00
Procalcitonin (PCT)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0204-00.1
Progesteron	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0061-00
Proinsulin, intaktes	Serum, EDTA-Plasma	ELISA	BIO-ING-SPCH-SAA-0063-01
Prokollagen-III-Peptid	Serum	RIA	BIO-ING-RIA-SAA-0013-00.6
Prolactin	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0062-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Protein S-100	Serum	ECLIA	BIO-ING-AUTO-SAA-0201-00.1
Renin	EDTA-Plasma	CLIA	BIO-ING-AUTO-SAA-0136-00
Sexual-Hormon-bindendes Globulin (SHBG)	Serum, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0065-00.1
sFlt-1	Serum	ECLIA	BIO-ING-AUTO-SAA-0199-00
Squamous cell carcinoma (SCC)-Ag	Serum	ECLIA	BIO-ING-AUTO-SAA-0195-00
Tacrolimus	EDTA-Blut	ECLIA	BIO-ING-AUTO-SAA-0185-00
Tau-Protein	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.4
Testosteron	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0067-00.1
Testosteron, freies	Serum	ELISA	BIO-ING-SPCH-SAA-0063-01
Theophyllin	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0209-00
Thrombin/Antithrombin-III-Komplex	Citratplasma	ELISA	BIO-ING-IMMU-SAA-0048-01.2
Thymidinkinase (TK)	Serum, EDTA-Plasma	CLIA	BIO-ING-AUTO-SAA-0140-00
Thyreoglobulin-Ak	Serum, EDTA-Plasma	ECLIA	BIO-ING-AUTO-SAA-0068-00.1
Thyroxin, freies (FT4)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0086-00.3
Tissue Polypeptide Antigen (TPA)	Serum	CLIA	BIO-ING-AUTO-SAA-0147-00
Tissue polypeptide specific antigen (TPS)	Serum	RIA	BIO-ING-RIA-SAA-0024-00.7
Trijodthyronin, freies (FT3)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0085-00.2
Troponin T	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0132-00.3
TSH basal	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0081-00.2
TSH Rezeptor Ak (TRAK)	Serum	ECLIA	BIO-ING-AUTO-SAA-0082-00.1
Valproinsäure	Serum, EDTA-, Heparin-Plasma	EIA	BIO-ING-AUTO-SAA-0191-00
Vancomycin	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0193-00.1
Vitamin D1.25	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0141-00
Wachstumshormon (HGH)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0219-00

**Untersuchungsart:  
Mikroskopie\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Differentialblutbild	EDTA-Blut, Ausstriche	Hellfeldmikroskopie mit Anfärbung	BIO-ING-HÄMA-SAA-0030-01
Leukozyten (Kammerzählung)	Punktat, Liquor	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-HÄMA-SAA-0031-00.7
Synovialanalyse	Punktat	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-HÄMA-SAA-0031-00.7
Urinsediment	Urin	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-HÄMA-SAA-0029-01.3

**Untersuchungsart:  
Osmometrie\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Osmolalität	Serum, Urin	Kryoskopie	BIO-ING-RIA-SAA-0007-00.9

**Untersuchungsart:  
Spektrometrie (UV-/VIS-Photometrie)\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Alkalische Phosphatase	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0016-00.1
Alpha-Amylase	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0015-01.1
Alpha-Amylase, Pankreas	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0010-00.1
Ammoniak	EDTA-Plasma	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Angiotensin-Converting-Enzyme (ACE)	Serum	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Anti-Faktor Xa Aktivität (Rivaroxaban)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0020-02
Anti-Faktor Xa Aktivität (Apixaban)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0020-02
Anti-Faktor Xa (LMW-Heparin)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0020-02
Antithrombin	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0010-01
Beta-Hydroxybuttersäure	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Bilirubin, direkt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0018-00.2
Bilirubin, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0017-00.2
Blutkörperchensenkungs-geschwindigkeit	EDTA-Blut	Photometrie	BIO-ING-HÄMA-SAA-0026-00.3
Bromid	Serum	Photometrie	BIO-ING-SPCH-SAA-0022-01.1
C1-Esterase-Inhibitor (Aktivität)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0015-01
Calcium	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0019-00
Calcium	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Cholesterin, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0021-00.2
Cholinesterase (Pseudocholinesterase)	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0022-00.1
CK	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0023-00.3
CK-MB	Serum, EDTA-Plasma	Photometrie	BIO-ING-AUTO-SAA-0003-00.2
Creatinin	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Creatinin, enzymatisch	Serum, EDTA-, Heparin-Plasma, Urin	Photometrie	BIO-ING-AUTO-SAA-0024-00.1
Eisen	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0025-00
Eiweiß, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0027-00.1
Ethanol	Serum, Urin, EDTA-Plasma	Photometrie	BIO-ING-CHROM-SAA-0021-00.5
Fettsäuren, frei	Serum	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Fructosamin	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0026-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Gallensäuren, gesamt	Serum	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Gamma-Hydroxybuttersäure	Urin	Photometrie	BIO-ING-CHROM-SAA-0023-00.5
GGT	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0028-00
GLDH	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0029-00
Glucose	NaF-Citrat-Plasma, Serum, EDTA-, Heparin-Plasma, Hämolytat, Urin	Photometrie	BIO-ING-AUTO-SAA-0030-00 BIO-ING-AUTO-SAA-0007-00
Glucose	Liquor, Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
GOT	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0031-00.1
GPT	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0032-00.1
Hämoglobin, frei	Heparin-Plasma, Serum	Photometrie	BIO-ING-SPCH-SAA-0003-01.1
Harnsäure	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0033-00.1
Harnsäure	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Harnstoff	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0034-00
Harnstoff	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Harnstoff-N	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
HBDH	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0035-00
HDL-Cholesterin	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0008-01.1
Kupfer	Serum, Heparinplasma	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Lactat	NaF-Plasma, EDTA-Plasma, Liquor	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
LDH	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0037-00
LDL-Cholesterin	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0009-01
Lipase	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0038-00.1
Lipoprotein (a)	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0116-00.2
Lithium	Serum, EDTA-, Heparinplasma	Photometrie	BIO-ING-AUTO-SAA-0213-00
Magnesium	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0039-00
Magnesium	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
N-Acetyl-Glucosaminidase	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Oxalat	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Phosphat, anorganisch	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0041-00
Phosphat, anorganisch	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Protein C-Aktivität	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0010-01
Saure Phosphatase, gesamt	Serum	Photometrie	BIO-ING-AUTO-SAA-0012-00
Tobramycin	Serum, EDTA-, Citrat-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0115-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Triglyceride	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0042-00.1
Urinstatus	Urin	Photometrie	BIO-ING-HÄMA-SAA-0029-01.3
Zink	Serum, Heparin-Plasma	Photometrie	BIO-ING-SPCH-SAA-0021-01.3

**Untersuchungsart:**

**Spektrometrie (Nephelometrie)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Albumin	Serum, EDTA-, Heparin-Plasma, Liquor, Urin	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2 BIO-ING-IMMU-SAA-0060-05
Alpha 1-Mikroglobulin	Urin	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Alpha-1-Antitrypsin	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Apolipoprotein A-1	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Apolipoprotein B	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Coeruloplasmin	Serum, Heparin-Plasma	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Cystatin C	Serum, Heparin-Plasma	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Haptoglobin	Serum, Heparin-, EDTA-Plasma	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Myoglobin	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Proteinurie-Diagnostik	Urin	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Serum-Amyloid-A (SAA)	Serum, Heparin-Plasma	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2

**Untersuchungsart:**

**Spektrometrie (Turbidimetrie)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
CRP	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0004-00.1
CRP high sensitive	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0203-00
D-Dimer	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0039-00.1
Eiweiß	Urin, Liquor	Turbidimetrie	BIO-ING-SPCH-SAA-0021-01.3
freies Protein S-Antigen	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0010-01
HbA1C	EDTA-Blut	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0022-00.4
Streptokokken, Streptolysin-O- Antikörper (ASL)	Serum	Turbidimetrie	BIO-ING-AUTO-SAA-0014-00.1
Transferrin	Serum, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0175-00
Transferrin Rezeptor, lösl. (sTfR)	Serum, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0013-01
VWF:Ag (Faktor VIII-assoziiertes Ag)	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0017-02
VWF Ac (Aktivität)	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0017-02

**Untersuchungsart:**

**Spektrometrie (Atomabsorptionsspektrometrie (AAS))\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Quecksilber	EDTA-Blut, Heparinblut, Urin	AAS	BIO-ING-SPCH-SAA-0049-00.4

**Untersuchungsart:**

**Spektrometrie (Induktiv gekoppelte Plasma-Massenspektrometrie (ICP-MS))\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Antimon	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00
Arsen	Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0085-00
Barium	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00
Blei	Urin, EDTA-Blut, Heparinblut	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0085-00
Bromid	Serum	ICP-MS	BIO-ING-SPCH-SAA-0089-00
Cadmium	Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0085-00
Chrom	EDTA-, Heparin-Plasma, Urin	ICP-MS	BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Jod	Urin, Serum	ICP-MS	BIO-ING-SPCH-SAA-0086-00
Kobalt	Serum, Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Kupfer	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00
Mangan	EDTA-, Heparin-Plasma, Urin, Serum, Heparinblut, EDTA-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Molybdän	Urin, EDTA-, Heparin-Plasma	ICP-MS	BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Nickel	Urin, EDTA-Blut, Heparinblut Heparin-, EDTA-Plasma	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Selen	Urin, EDTA-Blut, Heparinblut, EDTA-Plasma, Heparinplasma, Serum	ICP-MS	BIO-ING-SPCH-SAA-0064-01 BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Thallium	Serum, Urin	ICP-MS	BIO-ING-SPCH-SAA-0081-00 BIO-ING-SPCH-SAA-0085-00
Vanadium	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00
Zink	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00
Zinn	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00

**Untersuchungsgebiet: Immunologie**

**Untersuchungsart:**

**Durchflusszytometrie\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Lymphozytendifferenzierung	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung	BIO-ING-HÄMA-SAA-0033-00.2
Zelluläre Allergiediagnostik (FlowCAST)	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung	BIO-ING-HÄMA-SAA-0032-00.1 BIO-ING-HÄMA-VAW-0004-01.1
Lymphozytendifferenzierung und Aktivierungsmarker	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1



Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bestimmung des Anteils TCR- $\gamma\delta$ -positiver T-Zellen im peripheren Blut	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1
Bestimmung der Leichtketten-Expression auf B-Zellen oder Plasmazellen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1
Immunphänotypisierung bei Non-Hodgkin-Lymphomen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1
Immunphänotypisierung bei chronisch-myeloischen Erkrankungen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1
Immunphänotypisierung bei MDS und akuten Leukämien	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1
Immunphänotypisierung bei PNH	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.1

**Untersuchungsart:  
Elektrophorese\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Paraprotein, monoklonale Gammopathie	Serum, Urin	Immunfixationselektrophorese	BIO-ING-IMMU-SAA-0066-04
Oligoklonale Banden	Serum, Liquor	Isoelektrische Fokussierung	BIO-ING-IMMU-SAA-0054-02.5
SDS-Gelelektrophorese	Urin	Zonenelektrophorese (SDS)	BIO-ING-IMMU-SAA-0055-05

**Untersuchungsart:  
Ligandenassays\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Acetylcholin-Rezeptor-Ak	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0002-00.9
Cycl. citrulliniertes Peptid-Ak (CCP-Ak)	Serum	ECLIA	BIO-ING-AUTO-SAA-0047-00.2
Eosinophiles cationisches Protein (ECP)	Serum	CLIA	BIO-ING-AUTO-SAA-0145-00
C1q (C1C-C1q)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0045-02.2
Beta-2-Glycoprotein-Ak	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0034-01.1
Cardiolipin-Ak	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0031-02.2
Calcium-Kanal-Protein-Ak (P/Q-Typ)	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0012-00.8
Doppelstrang-DNA (dsDNA)-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0032-01.1
Einzelstrang-DNA (ssDNA)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0043-03.2
Gangliosid-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0052-03
Glutamat-Decarboxylase (GAD65)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0033-03
Histone-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0039-02.2
IA2-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0051-02.2
Immunglobulin E	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0057-00.1
Intrinsic-Faktor-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0042-02.4
Insulin-Ak gegen Humaninsulin (IgG)	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0019-01.4

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Leber-Nieren-Mikrosomen (LKM 1)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0062-04
MCV-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0053-02.1
Mikrosomale Schilddrüsen-Ak (MAK, TPO-Ak)	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0069-00
Mitochondriale-Ak, Subtyp 1 (IgG)	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0031-02.2
Mitochondriale-Ak, Subtyp 1 (IgM)	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0031-02.2
Mitochondriale-Ak, Subtyp 2	Serum	ELISA	BIO-ING-IMMU-SAA-0040-02.3
Muskelspezif. Rezeptor-Tyrosinkinase-Ak	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0001-00.5
Myeloperoxidase (MPO)-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0037-01.3
Parietalzellen-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0010-04.1
PF4-Heparin-Ak	Serum	Lateral-Flow-Assay	BIO-ING-IMMU-SAA-0047-03.2
PMSCL-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0035-01.3
Proteinase 3-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0038-02.2
Saccharomyces cerevisiae-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0041-02.4
SLA/LP-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0036-01.3
Allergenspezifische IgE Ak	Serum	EAST (Enzym-Allergo-Sorbent-Test)	BIO-ING-SERO-SAA-0059-05
Mitochondriale-Ak, Subtyp 4	Serum	Immunoblot	BIO-ING-IMMU-SAA-0014-01.4
Mitochondriale-Ak, Subtyp 9	Serum	Immunoblot	BIO-ING-IMMU-SAA-0014-01.4
Zentromer-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0075-00.1
Zinktransporter-8-Ak (ZnT8)	Serum	ELISA	BIO-ING-IMMU-SAA-0073-00.2

**Untersuchungsart:  
Mikroskopie\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
ANCA (Granulozytenzytoplasma-Ak)	Serum	IFT	BIO-ING-IMMU-SAA-0016-04.2
Antinukleäre-Ak (inkl. Subtypen)	Serum	IFT	BIO-ING-IMMU-SAA-0011-02.3
Aquaporin 4-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0003-02.4
Basalmembran-Ak (tubulär)	Serum	IFT	BIO-ING-IMMU-SAA-0015-01.6
Doppelstrang-DNA (dsDNA)-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0078-00.2
DPPX-Ak (Anti-Dipeptidyl aminopeptidase-like protein 6)	Serum	IFT	BIO-ING-IMMU-SAA-0072-03
Endomysium-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0019-02
Glatte Muskulatur-Ak (ASMA)	Serum	IFT	BIO-ING-IMMU-SAA-0005-03
Glutamat-Rezeptor (TypNMDA)-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0006-04
VGKC-Komplex (Typ CASPR2/Lgi1)-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0067-02.4
Herzmuskel-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0063-00.6

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Inselzell-Ak (ICA)	Serum	IFT	BIO-ING-IMMU-SAA-0017-03
Leber-Nieren-Mikrosomen (LKM)-2-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0005.03
Mitochondriale-Ak (AMA)	Serum	IFT	BIO-ING-IMMU-SAA-0005-03
MOG-Ak (Myelin-Oligodendrozyten-Glykoprotein)	Serum	IFT	BIO-ING-IMMU-SAA-0071-00.3
Nebennierenrinden-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0001-04
Neuronale Auto-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0013-02
Phospholipase-A <sub>2</sub> -Rezeptor (PLA <sub>2</sub> R)-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0007-04.1
Skelettmuskel-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0021-01.4
Spermatozoen-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0009-03.2
Thrombospondin type-1 domain-containing protein 7A-Ak (THSD7A)	Serum	IFT	BIO-ING-IMMU-SAA-0007-04.1

**Untersuchungsart:**

**Spektrometrie (UV-/VIS-Photometrie)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Komplement-Gesamtaktivität (CH50)	Serum	Photometrie	BIO-ING-SPCH-SAA-0021-01.3

**Untersuchungsart:**

**Spektrometrie (Nephelometrie)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
C1-Esterase-Inhibitor (Proteinbestimmung)	Serum, Citrat-Plasma	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
C3-Komplement-Komponente	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
C4-Komplement-Komponente	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Immunglobulin A	Serum, Liquor	Nephelometrie	BIO-ING-IMMU-SAA-0060-5
Immunglobulin D	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Immunglobulin G	Serum, Liquor	Nephelometrie	BIO-ING-IMMU-SAA-0060-05 BIO-ING-IMMU-SAA-0079-02.2
Immunglobulin G-Subklassen	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Immunglobulin M	Serum, Liquor	Nephelometrie	BIO-ING-IMMU-SAA-0060-05

**Untersuchungsart:**

**Spektrometrie (Turbidimetrie)\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Immunglobulin A (IgA)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0118-00.1
Immunglobulin G (IgG)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0119-00
Immunglobulin M (IgM)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0120-00.1
Rheumafaktor (RF)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0011-00.1

## Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

### Untersuchungsart:

### Molekularbiologische Untersuchungen (Amplifikationsverfahren)\*\*

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Achondroplasie (FGFR3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Alpha-Galaktosidase-A-Mangel (GLA) / Morbus Fabry	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Alpha-Thalassämie (HBA1, HBA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Amyloidpolyneuropathie, familiär (TTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Beta-Thalassämie (HBB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Ehlers-Danlos-Syndrom (TNXB) - Exon 32 bis 44	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Familiäres Mittelmeerfieber (MEFV, Exons 2, 3, 10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Geschlechtsdifferenzierung (SRY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Hereditäre Neuropathie mit Neigung zu Drucklähmungen (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Hereditäre Persistenz des fetalen Hämoglobin (HPFH)-Syndrom (HBG1, HBG2 -> nur Promotorbereiche)	EDTA-Blut, DNA, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Hypercholesterinämie (ApoB)	EDTA-Blut, DNA, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Hypochondropasie (FGFR3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Lebersche hereditäre Optikusneuropathie (LHON)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
MTHFR-Mangel (A1298C)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Neoplasie, multiple endokrine Typ 2A (RET, Hotspots)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pankreatitis, hereditär (SPINK1, PRSS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Schwerhörigkeit, nicht-syndromale sensorineurale (GJB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
SHOX(-Defizienz)/Leri-Weill-Syndrom	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Von Hippel Lindau Syndrom (VHL)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Alpha-1-Antitrypsin-Mangel (SERPINA1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1
Irinotecan-Therapie (UGT1A1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1
Morbus Meulengracht (UGT1A1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1
MTHFR-Mangel (MTHFR, C677T)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1
Plasminogen-Aktivator-Inhibitor-1-Mangel (PAI1,4G/5G)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Steatohepatitis, nicht-alkoholische (PNPLA3)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2 BIO-ING-HUM-SAA-0167-00.1
Adrenogenitales Syndrom (CYP21A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0082-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0071-00.1 BIO-ING-HUM-SAA-0063-02.2
3M-Syndrom (CCDC8, CUL7, OBSL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Achromatopsie (ACHM) (ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Acrocallosales Syndrom (GLI3, KIF7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adams-Oliver-Syndrom (ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adenomatöse Polyposis (APC, MUTYH, NTHL1, POLD1, POLE)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Adipositas (ALMS1, APC2, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CARTPT, CCDC28B, CEP164, CEP19, CEP290, CUL4B, DYRK1B, GNAS, IFT172, IFT27, IFT74, KDM6A, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, NFIX, NROB2, NSD1, NTRK2, PCSK1, PHF6, POMC, PPARG, PTEN, PYY, RAB23, SDCCAG8, SH2B1, SIM1, TMEM67, TRIM32, TTC8, UCP3, VPS13B, WDPCP)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adulte Leukenzephalopathie (ABCD1, ARSA, ATN1, CSF1R, CYP27A1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GALC, GBE1, GFAP, GLA, GLB1, HEXA, HMGCL, HTRA1, LMNB1, NOTCH3, POLR3B, SPG11, TUBB4A, ZFYVE26)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ahornsirupkrankheit (BCKDHA, BCKDHB, DBT, DLD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Aicardi-Goutières Syndrom (ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Akutes Leberversagen (ALDOB, BCS1L, DGUOK, DLD, FARS2, GALT, GBA, GFM1, HMGCL, IARS1, LARS1, LIPA, MPV17, MRPS16, NBAS, NPC1, NPC2, POLG, SCO1, SMPD1, SUCLG1, TRMU, TSFM, TWNK, TYMP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alagille-Syndrom (JAG1, NOTCH2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Albinismus (GPR143, LRMDA, MC1R, MITF, OCA2, SLC24A5, SLC45A2, TYR, TYRP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Alport-Syndrom (COL4A3, COL4A4, COL4A5, MYH9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alström-Syndrom (ALMS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alzheimer-Demenz (APOE, APP, PSEN1, PSEN2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Amyloidose (APOA1, APOA2, APP, B2M, CST3, FGA, GSN, ITM2B, LYZ, PRNP, TTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Amyotrophische Lateralsklerose (ALS2, ANG, ANXA11, C9orf72, CHCHD10, CHGB, CHMP2B, DAO, DCTN1, FIG4, FUS, HNRNPA1, MATR3, NEFH, NEK1, OPTN, PFN1, SETX, SIGMAR1, SOD1, SQSTM1, TARDBP, TUBA4A, UBQLN2, VAPB, VCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Anämie, angeborene dyserythroetische (C15orf41, CDAN1, GATA1, KIF23, KLF1, SEC23B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Angelman Syndrom (ADSL, ATRX, CDKL5, EHMT1, FOXG1, HERC2, KANSL1, MBD5, MECP2, MEF2C, SLC9A6, TCF4, UBE3A, WAC, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Aortopathie / Aortenaneurysma (ACTA2, ADAMTS10, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, FLNA, FOXE3, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFB1, TGFB2, THSD4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Aplastische Anämie (ASXL1, BCOR, BCORL1, DNMT3A, PIGA, PIGT)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Arrhythmogene rechtsventrikuläre Dysplasie/Kardiomyopathie (CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Arthrogrypose (ACTA1, ADGRG6, ALG3, BIN1, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, COL6A1, COL6A2, COL6A3, CRLF1, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKTN, GBA, GBE1, GLDN, GLE1, IBA57, LGI4, MEGF10, MPZ, MTM1, MYBPC1, MYH2, MYH3, MYH8, NALCN, NEB, PIEZO2, PIP5K1C, PLOD2, PMM2, RAPSN, RIPK4, SCARF2, SELENON, SYNE1, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN54, VIPAS39, VPS33B, VRK1, ZC4H2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Arthrogrypose- Nierenfunktionsstörung-Cholestase- Syndrom (VIPAS39, VPS33B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ashkenazi Panel (ASPA, BLM, CFTR, ELP1, FANCC, GBA, HEXA, MCOLN1, SMPD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Ataxie (ABCB7, ABHD12, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP1A3, ATP2B3, BEAN1, CA8, CACNA1A, CAPN1, COQ8A, CP, CWF19L1, CYP27A1, DARS2, DNM1, ELOVL4, FGF14, FLVCR1, FXN, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MARS2, MRE11, NKX6-2, PDYN, PHYH, PMPCA, PNKP, PNPLA6, POLG, PRKCG, RARS2, RNF216, SACS, SCN2A, SETX, SIL1, SLC17A5, SLC1A3, SNX14, SPG7, SPTBN2, STUB1, SYNE1, TGM6, TMEM240, TTBK2, TTC19, TTPA, TWNK, VAMP1, VLDLR, VPS13D, WDR81)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ataxie, autosomal-dominant (AFG3L2, FGF14, ITPR1, KCNC3, PDYN, PRKCG, SPTBN2, TMEM240)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ataxie, autosomal-rezessiv (ANO10, APTX, ATM, FXN, SETX, SIL1, SNX14, TTPA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Auditorische Neuropathie (AIFM1, DIAPH3, OTOF, PJVK)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Autosomal-dominante Polyzystische Nierenerkrankung (ADPKD) (ALG9, DNAJB11, GANAB, PKD1, PKD2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Autosomal-dominante nächtliche Frontallappenepilepsie (CHRNA2, CHRNA4, CHRN2, DEPDC5, KCNT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Autosomal-rezessive Polyzystische Nierenerkrankung (ARPKD) (DZIP1L, PKD1, PKHD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bardet-Biedl-Syndrom (ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, IFT172, IFT27, IFT74, LZTFL1, MKKS, MKS1, SCAPER, SDCCAG8, TMEM218, TRIM32, TTC8, WDPCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bartter-/Gitelman-Syndrom (AP2S1, BSND, CASR, CFTR, CLCNKA, CLCNKB, CLDN10, CTNS, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3, SLC26A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Basalganglien-Kalzifizierung (MYORG, PDGFB, PDGFRB, SLC20A2, XPR1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Beckwith-Wiedemann-Syndrom (CDKN1C)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Benigne Neugeborenenkrämpfe (KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Benigne rezurrenente intrahepatische Cholestase (ABCB11, ATP8B1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Bikuspide Aortenklappe (ACTA2, FBN1, GATA5, NKX2-5, NOTCH1, ROBO4, SMAD6, TGFB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Birt-Hogg-Dube-Syndrom (FLCN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bloom-Syndrom (BLM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Branchio-oto-renales-Syndrom (EYA1, SIX1, SIX5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brugada-Syndrom (ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SEMA3A, SLMAP, TRPM4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brustkrebs (BRCA1, BRCA2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Burkitt-Lymphom (ARID1A, CCND3, GNA13, ID3, MYC, RHOA, SMARCA4, TCF3, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
C1Q-Defizienz (C1QA, C1QB, C1QC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Charcot-Marie-Tooth-Krankheit (AARS1, ABHD12, AIFM1, ATL1, ATP1A1, ATP7A, BICD2, BSCL2, CNTNAP1, COA7, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, KIF5A, LITAF, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPZ, MTMR2, MYH14, NDRG1, NEFH, NEFL, PLEKHG5, PMP2, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCO2, SETX, SH3TC2, SIGMAR1, SLC12A6, SORD, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, WARS1, YARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
CHARGE-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chediak-Higashi-Syndrom (LYST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cholestatische Hepatopathie (ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP7B, ATP8B1, CFTR, CYP7B1, DCDC2, HSD3B7, MYO5B, NR1H4, SERPINA1, TJP2, UTP4, VIPAS39, VPS33B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chondrodysplasia punctata (AGPS, ANOS1, ARSL, EBP, GNPAT, LBR, MGP, NSDHL, PEX5, PEX7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chronisch progressive externe Ophthalmoplegie (ACADS, DGUOK, DNA2, MGME1, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SPG7, TK2, TOP3A, TWNK, TYMP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Chronische Neutrophilenleukämie (ASXL1, CALR, CBL, CSF3R, ELANE, NRAS, SETBP1, SRSF2, TET2, U2AF1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chronisch-entzündlichen Darmerkrankungen (CED) (ADA, ADA2, ADAM17, AICDA, AP3B1, AP3D1, ATG16L1, BACH2, CASP10, CASP8, CD27, CD70, CTLA4, CYBA, CYBB, DKC1, EGFR, FADD, FAS, FASLG, G6PD, IL10RA, IL10RB, IL21, IL21R, IL2RA, ITK, KRAS, LRBA, LYST, MAGT1, MEFV, MVK, NCF1, NCF2, NCF4, NFAT5, NLR4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PIK3R1, PLCG2, PRF1, PRKCD, PSMB8, PSTPIP1, RBCK1, RNF31, RTEL1, SH2D1A, SLC29A3, STAT1, STAT3, STX11, TNFAIP3, TNFRSF11A, TNFRSF1A, TTC7A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chylomikronämie (APOA5, APOC2, GPIHBP1, LMF1, LPL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cobalamin (Vitamin B12)-Mangel (ABCD4, ACSF3, AMN, CBLIF, CBS, CD320, CUBN, HCF1, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MTHFD1, MTR, MTRR, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cockayne-Syndrom (ERCC6, ERCC8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Coffin-Siris-Syndrom (ARID1A, ARID1B, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SOX11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Cornelia de Lange-Syndrom (ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Costello-Syndrom (BRAF, HRAS, KRAS, MAP2K1, NRAS, PTPN11, RAF1, SOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cowden-Syndrom (AKT1, PIK3CA, PTEN, SDHB, SDHD, SEC23B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cystinurie (SLC3A1, SLC7A9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Demenz (APOE, APP, CHCHD10, CHMP2B, CSF1R, FUS, GLA, GRN, MAPT, NOTCH3, PRNP, PSEN1, PSEN2, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, UBQLN2, VCP)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dent-Krankheit (CLCN5, OCRL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diabetes insipidus (AQP2, AVP, AVPR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Diabetes mellitus, neonataler (ABCC8, AGPAT2, BSLC2, CISD2, EIF2AK3, EIF2S3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, KCNJ11, KLF11, LRBA, MNX1, MT-TL1, NEUROD1, NEUROG3, NKX2-2, PAX6, PDX1, PLAGL1, PTF1A, RFX6, SLC19A2, SLC29A3, SLC2A2, STAT3, WFS1, ZFP57)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diabetes und Autoimmunität (AIRE, CTLA4, FOXP3, IL2RA, ITCH, JAK1, LRBA, SIRT1, STAT1, STAT3, STAT5A, STAT5B, TNFAIP3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diamond-Blackfan-Syndrom (GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
DICER1-assoziiertes Tumorsyndrom (DICER1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diffus großzelliges B-Zell-Lymphom (ARID1A, BCL2, BCL6, CARD11, CREBBP, EZH2, FOXO1, GNA13, KMT2D, MEF2B, MYD88, PCLO, PIM1, TET2, TNFRSF14, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Distale Myopathie (ACTA1, ACTN2, ADSS1, ANO5, CAV3, COL12A1, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, LRP12, MATR3, MYH14, MYH7, MYOT, NEB, PABPN1, RYR1, SLC5A7, SQSTM1, TCAP, TIA1, TPM3, TTN, VCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Duane Retraktionssyndrom (CHN1, DCC, HOXA1, MAFB, ROBO3, SALL4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Duchenne Muskeldystrophie (DMD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dyskeratosis congenita (ACD, AK2, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysostose mit vorwiegend vertebralem und kostalem Beteiligung (BMPER, COG1, DLL3, FGFR3, GDF3, GDF6, HES7, LFNG, MEOX1, MESP2, MNX1, NKX3-2, RIPPLY2, SNRNP, TBX6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysostosen (ABCC9, ALX1, ALX3, ALX4, BMPER, CDC45, CDC6, CDT1, CILK1, DHODH, DLL3, EDN1, EDNRA, EFN1, EFTUD2, EIF4A3, EVC, EVC2, GDF3, GDF6, GMNN, GNAI3, HAAO, HES7, KAT6B, KYNU, LFNG, LMX1B, MEOX1, MESP2, MNX1, MYO18B, NKX3-2, OFD1, ORC1, ORC4, ORC6, PLCB4, POLR1A, POLR1C, POLR1D, RIPPLY2, SF3B4, SH3PXD2B, SNRNP, TBX4, TBX6, TCOF1, ZSWIM6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysplasien der langen Knochen und/oder der Wirbelsäule (ACAN, ACP5, AIFM1, B3GALT6, B3GAT3, BGN, BPNT2, CANT1, CCN6, CFAP410, CHST3, COL10A1, COL11A1, COL11A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, DDR2, DDRGK1, DNAJC21, DYM, EFL1, EIF2AK3, EXTL3, FLNB, FN1, GPX4, HSPA9, HSPG2, IARS2, INPPL1, KIF22, LONP1, LTBP3, MAP3K7, MATN3, MMP13, MMP9, NANS, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, PTH1R, RAB33B, RMRP, RSPRY1, RUNX2, SBDS, SLC26A2, SLC35D1, SLC39A13, SMARCAL1, SRP54, TMEM165, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, XYLT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysplasien mit multiplen Gelenkdislokationen (B3GALT6, B3GAT3, BPNT2, CANT1, CHST3, CSGALNACT1, EXOC6B, FAM20B, FKBP14, FLNB, GZF1, KIF22, PLOD1, SLC10A7, SLC26A2, XYLT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Dysplasien mit Verkürzung der langen Knochen (akro-meso-rhizomele Dysplasien) (ADAMTS10, ADAMTS17, ADAMTSL2, BMPR1B, COL11A1, COL11A2, DVL1, DVL3, EXT1, FBN1, FGFR2, FGFR3, FLNA, FZD2, GDF5, GPC6, GSC, IFT122, IFT140, IFT43, IHH, LIFR, LRP4, LTBP2, LTBP3, MAB21L2, MIR140, NPR2, NXN, PDE4D, PRKAR1A, ROR2, SHOX, SLC26A2, SLC05A1, SMAD4, SOX9, SULF1, TRPS1, WDR35, WNT5A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dystonie (ADCY5, ANO3, ATP1A3, COL6A3, DNAJC12, FTL, GCH1, GNAL, GNAO1, HPCA, KMT2B, MECP, PLA2G6, PRKRA, RELN, SGCE, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ehlers-Danlos-Syndrom (ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ektodermale Dysplasie (ANTXR1, APCDD1, BANF1, BCS1L, CDH3, CDSN, CTSC, CYP26C1, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, ERCC2, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, GTF2H5, HOXC13, HR, IFT122, IFT43, IKBK, KCTD1, KREMEN1, KRT14, KRT25, KRT71, KRT74, KRT81, KRT83, KRT85, KRT86, LIPH, LPAR6, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKBIA, OFD1, PAX9, PIGL, PKP1, PORCN, PRKD1, SHOC2, SNRPE, SOX18, ST14, TP63, TRPS1, TSPEAR, TWIST2, UBR1, WDR19, WDR35, WNT10A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Elliptozytose, hereditäre (AMMECR1, EPB41, SPTA1, SPTB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Ellis-van-Creveld-Syndrom (DYNC2LI1, EVC, EVC2, WDR35)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Endokriner Kleinwuchs (ACAN, FGFR3, GH1, GHR, GHRHR, GHSR, HESX1, IGF1, IGF1R, IGFALS, IHH, LHX3, LHX4, NPR2, OTX2, POU1F1, PROP1, SHOX, SOX3, STAT5B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Epidermolysis bullosa (ATP2C1, CD151, CDSN, COL17A1, COL7A1, CSTA, DSG1, DSG2, DSG3, DSG4, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, TGM5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Epilepsie, Ataxie, Sensorineurale Taubheit und Tubulopathie Syndrom (KCNJ10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Epilepsie, fokale (CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1, LGI1, NPRL2, NPRL3, RELN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Epilepsie, metabolische (ABAT, ABCD1, ACY1, ADK, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, AUH, BCKDK, BTD, CAD, CLN3, CLN5, CLN6, CLN8, CNNM2, COQ4, COQ8A, CPT1A, CPT2, CTSD, CTSF, D2HGDH, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GALC, GAMT, GATM, GBA, GCDH, GLDC, GLUL, GPHN, GRN, HEXA, HEXB, HLCS, HPD, ITPA, L2HGDH, LIAS, MDH2, MFSB8, MLYCD, MMACHC, MOCS1, MOCS2, MTFMT, MTHFR, NPC1, NPC2, PC, PCBD1, PDHA1, PGK1, PHGDH, PLPBP, PNPO, POLG, PPT1, PSAT1, PSPH, PTS, QDPR, SLC19A3, SLC25A1, SLC25A15, SLC2A1, SLC46A1, SLC6A8, SLC6A9, SUOX, TPP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Epileptische Enzephalopathie (AARS1, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, ARHGEF9, ARX, ATP6V1A, ATRX, BRAT1, BTD, CACNA1A, CACNA1E, CACNA2D2, CACNB4, CDK19, CDKL5, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN2, CLCN4, CNNM2, CNPY3, CNTNAP2, CPA6, CYFIP2, DENND5A, DEPDC5, DNM1, DOCK7, EEF1A2, EHMT1, FOXG1, FRRS1L, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GLDC, GNAO1, GPHN, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HCN2, HNRNPU, IQSEC2, ITPA, KCNA2, KCNB1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, LGI1, MBD5, MDH2, MECP2, MEF2C, MOCS1, MOCS2, NEXMIF, NRXN1, PCDH19, PHF6, PIGA, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PURA, RHOTB2, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SEMA6B, SETD1A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SPTAN1, STAMBIP, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TSC1, TSC2, UBA5, UBE3A, UNC80, WDR45, WWOX, YWHAG, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Episodische Ataxie (CACNA1A, CACNB4, KCNA1, PNKD, PRRT2, SCN2A, SLC1A3, SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Erblicher Brust-und Eierstockkrebs (BRCA1, BRCA2, CHEK2, PALB2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Erblicher Brust-und Eierstockkrebs (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MRE11, NBN, PALB2, RAD51D, STK11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Erythrozytose, familiäre 1 (EPOR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Exsudative Vitreoretinopathie (inkl. Wagner- und Norrie-Syndrom) (CAPN5, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, RCBTB1, RS1, TSPAN12, TUBGCP4, VCAN, ZNF408)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Extremitätenfehlbildung (ARHGAP31, BHLHA9, BMP2, BMPR1B, CCNQ, CDH3, CHSY1, CKAP2L, DHODH, DLL4, DLX5, DOCK6, EOGT, ESCO2, FBLN1, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, FMN1, GDF5, GDF6, GJA1, GLI3, GNAS, GREM1, GSC, HDAC4, HDAC8, HOXA11, HOXA13, HOXD13, IHH, KCNJ2, KIF7, LMBR1, LRP4, MECOM, MYCN, NIPBL, NOG, NOTCH1, PAX3, PDE3A, PIGV, PITX1, PTHLH, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RSPO2, SALL1, SALL4, SHH, SMC1A, SMC3, TBX15, TBX3, TBX5, TGDS, THPO, TP63, WNT10B, WNT3, WNT6, WNT7A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Familiäres Silver-Russell-Syndrom (HMG2, IGF2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
FAMMM Syndrom (BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fanconi-Anämie (BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, PALB2, SLX4, UBE2T, XRCC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Fokale segmentale Glomerulosklerose (ACTN4, ANLN, APOL1, ARHGAP24, ARHGDI1, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAT1, FN1, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, LRP2, MAGI2, MT-TL1, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5, YRDC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Follikuläres Lymphom (BCL2, CREBBP, EZH2, KMT2D, MEF2B, TNFRSF14, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fruktoseintoleranz (ALDOB)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Galaktosämie (GALE, GALK1, GALT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gastrointestinale Stromatumoren (KIT, NF1, PDGFRA, SDHA, SDHB, SDHC, SDHD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gefleckte-Retina Syndrom (CHM, EFEMP1, PLA2G5, PRPH2, RDH5, RHO, RLBP1, RPE65, RS1, VPS13B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFS+) (GABRG2, SCN1A, SCN1B, SCN2A, STX1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glanzmann-Thrombasthenia (ITGA2B, ITGB3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glaukom (B3GLCT, CYP1B1, FOXC1, FOXE3, LMX1B, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SIX6, TBK1, TEK, WDR36)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gliedergürteldystrophie (ALG2, ANO5, BVES, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DMD, DNAJB6, DOK7, DPM3, DYSF, FKRP, FKTN, FLNC, GAA, GMPPB, HNRNPDL, LAMA2, LIMS2, LMNA, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RYR1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gliom (BRCA2, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glukokortikoid-Mangel (AIRE, CYP11A1, MC1R, MC2R, MCM4, MRAP, NNT, NR0B1, NR3C1, POMC, STAR, TXNRD2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glukoneogenese-Störung (CA5A, FBP1, G6PC, PC, PCK1, PCK2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Glykogenspeicherkrankheit (AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glykosylierungs-Defekt Typ 1 (ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, ATP6V1A, ATP6V1E1, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, GFPT1, GMPPA, MAGT1, MPDU1, MPI, NUS1, PGM1, PGM3, PMM2, RFT1, SRD5A3, SSR3, SSR4, STT3A, STT3B, TUSC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glykosylierungs-Defekt Typ 2 (ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, FCSK, FUT8, GANAB, MAN1B1, MAN2B2, MGAT2, MOGS, PRKCSH, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SLC9A7, ST3GAL3, TMEM165, TMEM199, VMA21)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glykosylierungs-Defekt, kongenitaler (ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, GMPPA, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PGM3, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, ST3GAL3, STT3A, STT3B, TMEM165, TMEM199, TUSC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gorlin-Goltz-Syndrom (PTCH1, PTCH2, SUFU)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Griscelli-Syndrom (MLPH, MYO5A, RAB27A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Haarzelleukämie (BRAF, CDKN1B, KLF2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hamartomatöse Polyposis (BMPR1A, PTEN, SMAD4, STK11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämochromatose (BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TFR2, TMPRSS6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämophagozytische Lymphohistiozytose (PRF1, SH2D1A, STX11, STXBP2, UNC13D, XIAP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hemiplegische Migräne, familiäre (ATP1A2, CACNA1A, NOTCH3, SCN1A)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hepatozelluläres Karzinom (ABCB11, AGL, APC, CDKN2A, G6PC, HMBS, PPOX, PRKAR1A, SPRTN)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre hämorrhagische Teleangiektasie (ACVRL1, ENG, GDF2, SMAD4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre motorisch-sensorische Neuropathien (axonal) (AARS1, GARS1, GDAP1, GJB1, HINT1, HSPB1, KIF1B, MFN2, MME, MPZ, NEFH, NEFL, RAB7A, TRPV4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hereditäre motorisch-sensorische Neuropathien (demyelinisierend) (EGR2, FGD4, FIG4, GDAP1, GJB1, LITAF, MPZ, MTMR2, NEFL, PMP22, PRX, SBF2, SH3TC2, TTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre Lymphödeme (ADAMTS3, CCBE1, CELSR1, EPHB4, FAT4, FLT4, FOXC2, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, VEGFC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre motorisch-sensorische Neuropathie (AARS1, ABHD12, AIFM1, ATL1, ATP1A1, ATP7A, BICD2, BSCL2, CNTNAP1, COA7, DCTN1, DNAJB2, DNMT2, DNMT1, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, KIF5A, LITAF, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPZ, MTMR2, MYH14, NDRG1, NEFH, NEFL, PLEKHG5, PMP2, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCO2, SETX, SH3TC2, SIGMAR1, SLC12A6, SORD, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, WARS1, YARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre sensorische und autonome Neuropathie (ATL1, ATL3, DNMT1, ELP1, FLVCR1, KIF1A, NGF, NTRK1, PRDM12, RAB7A, RETREG1, SCN10A, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäres nicht-polypöses kolorektales Karzinom (EPCAM, MLH1, MSH2, MSH6, PMS2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hermansky-Pudlak-Syndrom (AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Heterotaxie (ACVR2B, ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP52, CFAP53, CFC1, CRELD1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXH1, FOXJ1, GAS2L2, GAS8, GDF1, HYDIN, LMLN2, LRRC56, LRRC6, MMP21, MNS1, NFATC1, NKX2-5, NME8, NODAL, NPHP4, OFD1, PKD1L1, RSPH1, RSPH3, RSPH4A, RSPH9, SMAD2, SPAG1, TTC25, ZIC3, ZMYND10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hodgkin Lymphome, hereditär (DICER1, POT1, PTPN11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Holoprosenzephalie (CDON, CENPF, DHCR7, DISP1, FGF8, FGFR1, GLI2, KMT2D, PTCH1, RAD21, SHH, SIX3, SMC1A, SMC3, STAG2, STIL, TGIF1, ZIC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hornhautdystrophie (AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCN, FOXE3, GSN, KRT12, KRT3, LOXHD1, MIR184, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1, ZEB1, ZNF469)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hörverlust bei Erwachsenen (COCH, COL11A2, EYA4, KCNQ4, MIR96, SLC17A8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperammonämie (ACAD9, ACADVL, ACAT1, ALDH18A1, ARG1, ASL, ASS1, AUH, CA5A, CPS1, GLUD1, GLUL, HADH, HADHA, HADHB, HLCS, HMGCL, IVD, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMADHC, MMUT, MRPS22, NAGS, OAT, OTC, OXCT1, PC, PCCA, PCCB, SLC25A13, SLC25A15, SLC7A7, TAZ, TMEM70)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hypercholesterinämie (APOB, CETP, LDLR, LDLRAP1, LIPA, PCSK9, STAP1)	EDTA-Blut, DNA, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperekplexie (HKPX) (GLRA1, GLRB, SLC6A5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyper-IgE-Syndrom (DOCK8, DSG1, DSP, PGM3, SPINK5, STAT3, TYK2, ZNF341)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperinsulinismus, kongenitaler (ABCC8, ALG3, CACNA1C, CACNA1D, FOXA2, GCK, GLUD1, HADH, HK1, HNF1A, HNF4A, HRAS, INSR, KCNJ11, KCNQ1, KDM6A, KMT2D, MPI, NSD1, PGM1, PMM2, SKIL, SLC16A1, TRMT10A, UCP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperkalzämie (AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, MEN1, PTH1R, SLC12A1, SLC34A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperoxalurie (AGXT, GRHPR, HOGA1, SLC26A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperparathyreoidismus (AP2S1, CASR, CDC73, CDKN1B, CYP24A1, GCM2, HNF1B, MEN1, RET, SLC12A1, TRPV6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hypertriglyzeridämie (APOA5, APOC2, APOC3, APOE, GPD1, GPIHBP1, LIPA, LMF1, LPL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypertrophe Kardiomyopathie (ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, FHL1, FLNC, FXN, GAA, GLA, HRAS, JPH2, KLF10, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RASA2, RIT1, RRAS, RYR2, SLC25A4, SOS1, SOS2, TAZ, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypoglykämie, syndromale (AKT1, AKT2, AKT3, ALDH7A1, ALG12, ALG3, CACNA1C, CD36, CDKN1C, GPC3, GYS2, HRAS, IARS1, KCNQ1, KDM6A, KMT2D, MEF2C, MPI, NSD1, PCSK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHOX2B, PIK3CA, PIK3R2, PMM1, PMM2, PTEN, PYGL, PYGM, RNF125, SLC2A2, TALDO1, TRMT10A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypogonadotroper Hypogonadismus (ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GALT, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SLC29A3, SPRY4, TAC3, TACR3, TCF12, WDR11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypokaliämische periodische Paralyse (CACNA1S, SCN4A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypomagnesiämie (ATP1A1, BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, EGFR, FAM111A, FXYD2, HNF1B, KCNA1, KCNJ10, MT-TI, PCBD1, SARS2, SLC12A3, TRPM6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hypoparathyreoidismus (AIRE, CASR, CDC73, CHD7, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PTH, PTH1R, SOX3, STX16, TBCE, TRPM6, TRPV6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypopituitarismus (ARNT2, CDON, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RBM28, SOX2, SOX3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ichthyose und verwandte Keratinisierungsstörungen (AAGAB, ABCA12, ABHD5, ALDH3A2, ALMS1, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, ATP2C1, CARD14, CASP14, CDSN, CERS3, CLDN1, CSTA, CTSC, CYP4F22, DSG1, DSP, EBP, ELOVL4, ENPP1, ERCC2, ERCC3, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2H5, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LIPN, LORICRIN, MBTPS2, MPLKIP, NIPAL4, NSDHL, OSMR, PEX7, PHYH, PNPLA1, POMP, RHBDF2, SDR9C7, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, ST14, STS, SUMF1, TAT, TGM1, TGM5, TRPV3, VPS33B, WNT10A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Imlerslund-Gräsbeck-Syndrom (AMN, CUBN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Immundefekt, kombinierter schwerer (ADA, AK2, CD247, CD3D, CD3E, CORO1A, DCLRE1C, DOCK8, IL2RG, IL7R, JAK3, PRKDC, PTPRC, RAG1, RAG2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Insulin-Resistenz (AGPAT2, AKT1, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, FBN1, INSR, LIPE, LMNA, LMNB2, MT-TL1, NSMCE2, PCNT, PIK3R1, PLIN1, POC1A, POLD1, PPARG, PPP1R3A, SH2B1, TBC1D4, TCF7L2, WRN, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Intrahepatische Cholestase bei Schwangerschaft (ABCB11, ABCB4, ABCC2, ATP8B1, NR1H4, TJP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Jervell-Lange-Nielsen-Syndrom (KCNE1, KCNQ1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Jeune-Syndrom (CEP120, CFAP410, CILK1, DPH1, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, INTU, KIAA0586, KIAA0753, LBR, NEK1, TCTEX1D2, TRAF3IP1, TTC21B, WDR19, WDR34, WDR35, WDR60)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Joubert-Syndrom (AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CEP55, CPLANE1, CSPP1, FAM149B1, GLI3, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, POC1B, RPRGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kabuki-Syndrom (HNRNPK, KDM6A, KMT2A, KMT2D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kallmann-Syndrom (ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SPRY4, TAC3, TACR3, TCF12, WDR11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardio-fazio-kutanes Syndrom (BRAF, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, SHOC2, SOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kardiomyopathie, dilatative (ABCC9, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EMD, EYA4, FHL1, FKTN, GLA, HFE, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEXN, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, SCN5A, SDHA, SGCD, TAZ, TBX20, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie-Panel erweitert (ABCC9, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CALR3, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKTN, FLNC, FXN, GAA, GLA, HFE, HRAS, ILK, JPH2, JUP, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NKX2-5, NNT, NRAS, NSD1, OBSCN, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RASA2, RBM20, RIT1, RPS6KA3, RRAS, RYR2, SCN5A, SDHA, SGCD, SLC25A4, SOS1, SOS2, TAZ, TBX20, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Katarakt (ABCA3, ABHD12, AGK, AKR1E2, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, EYA1, FAM126A, FAR1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GJA3, GJA8, GNPAT, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, NHS, P3H2, PAX6, PEX7, PITX3, PXDN, RAB3GAP1, RNLS, SIL1, SIPA1L3, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, UNC45B, VIM, VSX2, WRN, XYLT2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ketogenese-Störung (ACADM, ACADVL, HADHA, HADHB, HMGCCL, HMGC52)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Ketolyse-Störung (ACAD9, ACAT1, ACSF3, AUH, BAAT, BCAT1, BCAT2, BCKDHA, BCKDHB, BCS1L, COQ9, DBT, DLD, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, G6PC, GLYCTK, GSS, GYS2, HADHB, HIBADH, HIBCH, HMGCL, HSD17B10, IVD, MCCC1, MCCC2, MLYCD, MMAA, MMAB, MMADHC, MMUT, OXCT1, PCCA, PCCB, PYGL, SLC16A1, SLC37A4, SUCLA2, SUCLG1, TAZ)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kleidokraniale Dysplasie (ALX4, FIG4, MSX2, RUNX2, VAC14)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Knochenmarkdysfunktions-Syndrom (AP3B1, BRCA2, BRIP1, CSF3R, CXCR4, DKC1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, MPL, NHP2, NOP10, PALB2, RAB27A, RAC2, RBM8A, RMRP, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS7, RTEL1, SBDS, SLC37A4, SLX4, SRP72, TAZ, TERC, TERT, TINF2, USB1, VPS13B, WAS, WIPF1, WRAP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolorektales Karzinom (APC, BMPR1A, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TGFBR2, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kongenitale Anomalien der Nieren und ableitenden Harnwege (ACE, AGT, AGTR1, ANOS1, BICC1, BMP4, CDC5L, CHD1L, CHRNA3, CRKL, DSTYK, EYA1, FANCB, FGF20, FOXP1, FRAS1, FREM1, FREM2, GATA3, GDNF, GLIS3, GREB1L, GREM1, GRIP1, HNF1B, ITGA8, KIF14, LHX1, LIFR, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SALL4, SIX1, SIX2, SIX5, SLIT2, SOX17, SRGAP1, TBC1D1, TBX18, TBX6, TRAP1, UMOD, UPK3A, WNT4, WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kongenitale Fibrose der äußeren Augenmuskeln (COL25A1, KIF21A, PHOX2A, TUBB3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kongenitale Muskeldystrophie (ACTA1, ANO5, B3GALNT2, B4GAT1, BIN1, CAPN3, CAVIN1, CCDC78, CFL2, CHKB, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNM2, DYSF, FHL1, FKRP, FKTN, GBE1, GMPPB, INPP5K, KBTBD13, KLHL40, KLHL41, LAMA2, LARGE1, LMNA, MEGF10, MTM1, NEB, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, RYR1, SELENON, TCAP, TNNT1, TPM2, TPM3, TRIM32, TRIP4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kongenitales Myasthenes Syndrom (AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, MYO9A, RAPSIN, SLC18A3, SLC5A7, VAMP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kraniosynostosen (ALPL, ALX4, BMP4, CCBE1, CDC45, COLEC11, CYP26B1, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT43, IHH, IL11RA, IRX5, KRAS, LRP5, MASP1, MEGF8, MSX2, MYH3, P4HB, POR, RAB23, RECQL4, SCARF2, SEC24D, SKI, SMAD6, SPECC1L, STAT3, TCF12, TGFBR1, TGFBR2, TWIST1, WDR19, WDR35, ZIC1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kutanes T-Zell-Lymphom (BRD9, CHD3, CREBBP, KMT2D, PLCG1, SMARCA4, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lebersche kongenitale Amaurose (LCA) (ACBD5, ADAMTS18, AIPL1, ALMS1, BBS2, BBS4, CABP4, CCT2, CEP290, CRB1, CRX, DHX38, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MPDZ, MYO7A, NMNAT1, OTX2, POC1B, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Lebersche hereditäre Optikusneuropathie (MT-ND1, MT-ND4, MT-ND6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, akute myeloische (ASXL1, CCND3, CEBPA, DNMT3A, FLT3, GATA2, IDH1, IDH2, KIT, KMT2A, KRAS, NPM1, NRAS, RUNX1, TET2, TP53, WT1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, atypische chronische myeloische (ASXL1, CALR, CBL, CSF3R, ETNK1, EZH2, JAK2, KRAS, MPL, NRAS, RUNX1, SETBP1, SRSF2, TET2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, B-Zell akute lymphatische (CREBBP, CRLF2, FLT3, IKZF1, NT5C2, PRDM9, SETD2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, chronische myeloische (ABL1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, chronische myelomonozytäre (ASXL1, BCOR, CBL, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KRAS, NF1, NPM1, NRAS, PHF6, RUNX1, SETBP1, SF3B1, SRSF2, TET2, U2AF1, ZRSR2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, juvenile myelomonozytische (ARHGAP26, CBL, KRAS, NRAS, PTEN, PTPN11)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, Ph-ähnliche akute lymphatische (ARID2, ASXL1, BLNK, CHD2, CRLF2, DGKH, EPOR, IKZF1, IL2RB, ITPKB, JAK1, JAK2, KRAS, LRP1B, LYN, NRAS, NTRK3, PDGFRA, PTK2B, PTPN11, TP53, TYK2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, T-Zell akute lymphatische (CDKN2A, DNMT3A, FBXW7, FLT3, IDH1, IDH2, IL7R, JAK1, JAK3, KRAS, NOTCH1, NRAS, PHF6, PTEN, RUNX1, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Leukämie, chronische lymphatische (ATM, BCL2, BIRC3, BRAF, BTK, DDX3X, EGR2, FBXW7, KRAS, MED12, MYD88, NFKBIE, NOTCH1, NRAS, PLCG2, POT1, SAMHD1, SETD2, SF3B1, TP53, XPO1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukoenzephalopathie (AARS1, ABCD1, ACOX1, ADAR, AIMP1, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, CSF1R, CTC1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FAM126A, GALC, GFAP, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, L2HGDH, MLC1, NAXE, NKX6- 2, NOTCH3, PLAA, PLP1, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SLC16A2, SNORD118, SOX10, SUMF1, TMEM106B, TREX1, TUBB4A, UFM1, VPS11, ZNHIT3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Liddle-Syndrom (SCNN1A, SCNN1B, SCNN1G)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Linksventrikuläre Noncompaction Kardiomyopathie (ACTC1, ACTN2, DTNA, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, NNT, NSD1, PRDM16, RPS6KA3, SCN5A, TAZ, TNNI3, TNNT2, TPM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Linsluxation (ADAMTSL4, FBN1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lissenzephalie (ADGRG1, ARX, CCND2, CRADD, CRPPA, DCX, DYNC1H1, FKR1, FKTN, KATNB1, KIF2A, LAMB1, LAMC3, LARGE1, MACF1, PAFAH1B1, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RELN, RTTN, RXYLT1, TMTC3, TUBA1A, TUBA8, TUBB2B, WDR62)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Long-QT-Syndrom (AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRPM4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lungenerkrankung, diffuse (ABCA3, ACVRL1, AP3B1, BMPR2, CAV1, CFTR, CSF2RA, CSF2RB, DKC1, EIF2AK4, ENG, FAM111B, FLNA, FOXF1, HPS1, HPS4, ITGA3, KCNK3, MARS1, NKX2-1, PARN, RTEL1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMAD9, SMPD1, STAT3, STRA6, TBX4, TERC, TERT, TINF2, TSC1, TSC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphozyten-Leukämie, große granuläre (STAT3, STAT5B)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lysosomale Speichererkrankungen (AGA, ARSA, ARSB, ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CSTB, CTNS, CTSB, CTSD, CTSF, CTSK, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GRN, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Magenkarzinom (APC, CDH1, CTNNA1, EPCAM, MAP3K6, MLH1, MSH2, MSH6, PMS2, SMAD4, STK11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mainzer-Saldino-Syndrom (DYNC2H1, IFT140, IFT172, WDR35)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Makrosomie (ABCC9, AIP, AKT1, CDKN1C, CHD8, DIS3L2, DNMT3A, EED, EZH2, GPC3, MC2R, NFIX, NPR2, NSD1, PDGFRB, PIK3CA, PTEN, SOST, SUZ12, ZBTB20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Makrozephalie (ABCC9, AKT1, AKT2, AKT3, AMER1, APC2, ASPA, ASXL2, BRAF, BRWD3, CCDC88C, CCND2, CDKN1C, CHD3, CHD4, CHD8, CRADD, CUL4B, DHCR24, DNMT3A, EED, EZH2, FIBP, GCDH, GFAP, GLI3, GPC3, GRIA3, H1-4, HEPACAM, HERC1, HRAS, HUWE1, IGF2, KIF7, KPTN, KRAS, L1CAM, MAP2K2, MED12, MLC1, MPDZ, MTOR, NFIB, NFIX, NRAS, NSD1, OFD1, PAK1, PHF6, PIGA, PIGN, PIGT, PIK3CA, PIK3R2, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RIN2, RNF125, RNF135, SEC24D, SETD2, SNX10, SOS1, STRADA, SUZ12, SYN1, TBC1D7, TCF20, UPF3B, ZBTB20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Maligne Hyperthermie (CACNA1S, RYR1, STAC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Männliche Infertilität (AMH, AMHR2, ANOS1, AR, ARMC2, AURKC, BNC2, CATSPER1, CCDC39, CFAP251, CFAP43, CFAP44, CFAP47, CFAP65, CFAP69, CFAP70, CFTR, CYP11B1, CYP17A1, CYP21A2, DMRT1, DNAH1, DNAH17, DNAH2, DNAH5, DNAH6, DNAH8, DNAH9, DNAJB13, DPY19L2, FANCM, FGF8, FGFR1, FSHB, FSHR, FSIP2, GNRH1, GNRHR, HSD17B3, HSD3B2, INSL3, KISS1, KISS1R, KLHL10, LHB, LHCGR, LRR6, M1AP, MAMLD1, NANOS1, NR0B1, NR5A1, NSMF, PLCZ1, PMFBP1, PPP2R3C, PROK2, PROKR2, QRICH2, RSP01, SEMA3A, SEPTIN12, SLC26A8, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPEF2, SRD5A2, SRY, SUN5, SYCP2, SYCP3, TACR3, TEX11, TEX14, TEX15, TRIM37, TTC21A, TTC29, USP26, WDR11, XRCC2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mantelzell-Lymphom (ATM, CCND1, CDKN2A, CHD2, KMT2D, NOTCH1, NOTCH2, TET2, TP53, UBR5)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Marfan-Syndrom (FBN1, TGFB1, TGFB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Marfan-Syndrom und verwandte Krankheiten (ACTA2, ADAMTSL4, COL2A1, COL4A1, DLG4, FBN1, FBN2, LTBP2, MED12, MYH11, MYLK, NKAP, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, UPF3B, ZDHC9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Marginalzonen-Lymphom (KLF2, NOTCH1, NOTCH2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mastozytose, systemische (ASXL1, CBL, EZH2, IDH1, IDH2, JAK2, KIT, KRAS, NRAS, RUNX1, SRSF2, TET2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
McCune-Albright-Syndrom (GNAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Meckel-Gruber-Syndrom (AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CEP55, CPLANE1, CSPP1, GLI3, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, POC1B, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Medulloblastom (APC, BRCA2, MSH2, MSH6, PALB2, PMS2, PTCH1, SUFU, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mentale Retardierung - männlich (ARX, ATRX, CUL4B, GPC3, HUWE1, IL1RAPL1, IQSEC2, KDM5C, LAS1L, MAOA, MECP2, MED12, OPHN1, PHF6, PQBP1, PTEN, RPS6KA3, SLC16A2, SLC2A1, SLC6A8, SLC9A6, STXBP1, TCF4, ZC4H2, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mentale Retardierung - weiblich (CDKL5, CLCN4, EHMT1, FOXG1, GDI1, HNRNP2, IL1RAPL1, MECP2, MEF2C, PCDH19, PTEN, SHANK3, SLC2A1, SLC6A8, SLC9A6, STXBP1, TCF4, THOC2, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Metaphysäre Dysplasie (COL10A1, MMP13, MMP9, POP1, PTH1R, RMRP, RUNX2, SBDS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mikroangiopathie (APP, COL4A1, COL4A2, CTSA, FOXC1, GLA, HTRA1, NOTCH3, SNORD118, TREX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mikrophthalmie/Anophthalmie (ABCB6, ALDH1A3, ATOH7, BCOR, BMP4, C12orf57, CHD7, FOXE3, FREM1, GDF3, GDF6, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, OTX2, PAX6, PORCN, PRSS56, RARB, RAX, RBP4, SALL2, SHH, SIX6, SMOG1, SOX2, STRA6, TENM3, TMEM98, VAX1, VSX2, YAP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mikrozephalie (ADARB1, ADGRG1, AGMO, ANKLE2, ANKRD11, AP4M1, ARCN1, ARFGF2, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATR, ATRIP, ATRX, BUB1B, CAR51, CASK, CDC6, CDK5RAP2, CDK6, CDKL5, CDON, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP164, CEP63, CHMP1A, CIT, CLN5, CLN6, CLN8, COG6, CREBBP, CRIPT, CTSD, CTSF, DCX, DDX11, DHCR7, DIAPH1, DNAJC5, DNM1L, DONSON, DYM, DYNC1H1, DYNC1I2, EFTUD2, EP300, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, EXOSC3, FKRP, FKTN, FLNA, FOXP1, GLI2, GRN, HDAC8, IER3IP1, IGF1, IGF1R, KAT6A, KATNB1, KCTD7, KIF11, KIF14, KIF2A, KIF5C, KNL1, LARGE1, LIG4, MCPH1, MECP2, MED17, MEF2C, MFSD2A, MFSD8, MIR17HG, MSMO1, MTHFS, MYCN, NAA10, NDE1, NIN, NIPBL, NR2E1, ORC1, ORC4, ORC6, PAFAH1B1, PARS2, PCNT, PHC1, PHF6, PHGDH, PIEZO2, PLAA, PLK4, PNKP, POMGNT1, POMT1, POMT2, PPP1R15B, PPT1, PQBP1, PRUNE1, PSAT1, PTCH1, PUS7, RAB18, RAD21, RARS2, RBBP8, RELN, RNU4ATAC, RTTN, SASS6, SHH, SIX3, SLC1A4, SLC25A19, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMPD4, SOX11, STAMBP, STIL, TCF4, TGIF1, TMX2, TPP1, TRAI, TRAPPC9, TRIO, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, UBR1, VARS1, VRK1, WDR4, WDR62, XIRP1, XRCC4, ZEB2, ZIC2, ZNF335)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mismatch-Repair-Defizienz, konstitutionelle (MLH1, MSH2, MSH6, PMS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mitochondriale DNA (MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mito-Nuklear (AARS2, ABAT, ABCB7, ACAD9, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, AK2, ALDH18A1, APTX, ATAD3A, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12orf65, C19orf12, CARS2, CHCHD10, CHKB, CLPB, CLPP, COA3, COA5, COA8, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6B1, COX7A1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DBT, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNAJC3, DNMM1L, EARS2, ECHS1, ELAC2, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, FXN, GARS1, GDAP1, GFER, GFM1, GFM2, GLRX5, GLUD1, GTPBP3, HAAO, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HMGCL, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH3A, ISCA2, ISCU, KARS1	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fortsetzung nächste Seite			

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mito-Nuklear Fortsetzung KIF5A, KYNU, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM4, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MICU2, MIPEP, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MTFMT, MTO1, MTPAP, NADK2, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NMT1, NSUN3, NUBPL, OPA1, OPA3, PANK2, PARS2, PC, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PLA2G6, PMPCA, PNPLA2, PNPLA8, PNPT1, POLG, POLG2, PPA2, PPARG, PTRH2, PUS1, QRSL1, RARS2, RMND1, RMRP, RNASEH1,  RRM2B, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC33A1, SLC39A8, SLC6A8, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TK2, TMEM126A, TMEM70, TPK1, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCRB, UQCRC2, UQCRFS1, UQCRCQ, VARS2, WARS2, WFS1, YARS2, YME1L1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mitochondriopathien (mitochondriale und nukleäre Gene) (AARS2, ABAT, ABCB7, ACAD9, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, AK2, ALDH18A1, APTX, ATAD3A, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12orf65, C19orf12, CARS2, CHCHD10, CHKB, CLPB, CLPP, COA3, COA5, COA8, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6B1, COX7A1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DBT, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, ELAC2, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, FXN, GARS1, GDAP1, GFER, GFM1, GFM2, GLRX5, GLUD1, GTPBP3, HAAO, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HMGCL, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH3A, ISCA2, ISCU, KARS1, KIF5A, KYNU, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM4, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MICU2, MIPEP, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTFMT, MTO1, MTPAP, NADK2, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13. Fortsetzung nächste Seite	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
<p>Mitochondriopathien (mitochondriale und nukleäre Gene) (Fortsetzung NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NMT1, NSUN3, NUBPL, OPA1, OPA3, PANK2, PARS2, PC, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PLA2G6, PMPCA, PNPLA2, PNPLA8, PNPT1, POLG, POLG2, PPA2, PPARG, PTRH2, PUS1, QRSL1, RARS2, RMND1, RMRP, RNASEH1, RRM2B, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC33A1,</p> <p>SLC39A8, SLC6A8, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TK2, TMEM126A, TMEM70, TPK1, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCRB, UQCRC2, UQCRFS1, UQCRCQ, VARS2, WARS2, WFS1, YARS2, YME1L1)</p>	<p>EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA</p>	<p>Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline</p>	<p>BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5</p>
<p>MNGIE-Syndrom (MGME1, POLG, RRM2B, TYMP)</p>	<p>EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA</p>	<p>Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline</p>	<p>BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5</p>
<p>MODY-Diabetes (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)</p>	<p>EDTA-Blut, DNA; DNA</p>	<p>Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline</p>	<p>BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5</p>

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Molybdän-Cofaktor-Defizienz (GPHN, MOCS1, MOCS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Monogener Diabetes (ABCC8, AGPAT2, AIRE, ALMS1, APPL1, BLK, BSCL2, CAV1, CAVIN1, CEL, CIDEA, CISD2, CTLA4, CYC1, DCAF17, DNAJC3, DUT, DYRK1B, EIF2AK3, EIF2S3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, ITCH, KCNJ11, KLF11, LMNA, LMNB2, LRBA, MNX1, MT-CO1, MT- CO3, MT-ND1, MT-ND4, MT-ND6, MT-RNR1, MT-RNR2, MT-TE, MT-TI, MT-TK, MT-TL1, MT-TR, MT-TS1, NEUROD1, NEUROG3, NKX2-2, PAX4, PAX6, PCBD1, PDX1, PLAGL1, PPARG, PPP1R15B, PPP1R3A, PTF1A, RFX6, SIRT1, SLC19A2, SLC29A3, SLC2A2, STAT1, STAT3, STAT5A, STAT5B, TRMT10A, TWNK, UCP2, WFS1, WRN, ZFP57, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Morbus Waldenström (ARID1A, CXCR4, MYD88)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Morbus Wilson (ATP7B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mukopolysaccharidose (ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mukoviszidose (CFTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Müller-Gang-Persistenzsyndrom (AMH, AMHR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Multiple Endokrine Neoplasie (CDKN1B, MEN1, RET)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Multiple epiphysäre Dysplasie (COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, PTH1R, SLC26A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Multiples Myelom (BRAF, DIS3, KRAS, NRAS, TENT5C, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myelodysplastisches Syndrom (ASXL1, BCOR, CBL, DNMT3A, ETV6, EZH2, FLT3, GATA2, IDH1, IDH2, KRAS, NRAS, RUNX1, SF3B1, SRSF2, STAG2, TET2, TP53, U2AF1, ZRSR2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myofibromatose, infantile (NOTCH3, PDGFRB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myopathie, metabolische (ABHD5, ACAD9, ACADS, ACADVL, AGK, AGL, ALDOA, AMACR, AMPD1, ATP2A1, CPT2, DGUOK, ENO3, ETFA, ETFB, ETFDH, FBXL4, FHL1, FLAD1, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMA2, LDHA, LPIN1, MGME1, MPV17, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, PNPLA8, POLG, POLG2, PUS1, PYGM, RBCK1, RRM2B, SLC16A1, SLC22A5, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, TYMP, YARS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Myopie (ARR3, COL11A1, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPSF1, LRPAP1, NYX, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nachtblindheit, kongenitale stationäre (CABP4, CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RHO, SAG, SLC24A1, TRPM1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neoplasie, myeloproliferative (ASXL1, CALR, CBL, DNMT3A, EZH2, IDH1, IDH2, JAK2, MPL, RUNX1, SF3B1, SH2B3, SRSF2, TET2, TP53, U2AF1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephroblastom (ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CTR9, DICER1, DIS3L2, GPC3, GPC4, NLRP2, PALB2, PIK3CA, REST, TP53, TRIM37, WT1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephronophthuse (NPHP) (AGXT, ANKS6, CEP164, CEP290, CEP83, DCDC2, FAN1, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TRAF3IP1, TTC21B, WDR19, XPNPEP3, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephrotisches-Syndrom (ACTN4, ANLN, APOL1, ARHGAP24, ARHGDI1, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAT1, FN1, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, LRP2, MAGI2, MT-TL1, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5, YRDC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Netzhautablösung (CAPN5, CHM, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, EFEMP1, FBN1, FZD4, KCNJ13, LRP5, NDP, P3H2, PLA2G5, RDH5, RLBP1, RS1, TSPAN12, TUBGCP4, VCAN, VPS13B, ZNF408)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neuroblastom (ALK, BARD1, BLM, CHEK2, FANCD2, KIF1B, PALB2, PHOX2B, PINK1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurodegeneration mit Eisenspeicherung im Gehirn (ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, GTPBP2, PANK2, PLA2G6, WDR45)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurofibromatose und Schwannomatose (LZTR1, NF1, NF2, SMARCB1, SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neuronale Ceroid-Lipofuszinose (ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSDB8, PPT1, TPP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neutropenie, schwere kongenitale (CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neutropenie, somatische Mutationen (CSF3R, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Nicht-syndromische Schwerhörigkeit (ACTG1, ADCY1, AIFM1, ATP2B2, BSND, CABP2, CCDC50, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, CLPP, COCH, COL11A2, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MIR96, MPZL2, MSRB3, MT-RNR1, MT-TS2, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDZD7, PJVK, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, S1PR2, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, WFS1, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nierenkrebs (BAP1, EPCAM, FH, FLCN, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCB1, TP53, TRIP13, TSC1, TSC2, VHL, WT1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nierensteinerkrankung (ABCC6, ADCY10, AGXT, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CYP24A1, CYP27B1, FAH, FAM20A, FOXI1, G6PC, GCM2, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, HSD11B2, INSR, KCNJ1, MOCOS, OCRL, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC7A9, SLC9A3R1, VDR, VIPAS39, VPS33B, WDR72, WNK1, WNK4, XDH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
NK-/T-Zell-Lymphom (ARID1A, ASXL3, BCOR, DDX3X, EP300, FAS, JAK3, KMT2D, MGA, STAT3, STAT5B, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Non-Langerhans-Zell-Histiozytosen (CBL, KRAS, NF1, NF2, NRAS, PTPN11, TP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Noonan-Syndrom (A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Noonan-Syndrom mit multiplen Lentiginen (BRAF, MAP2K1, NRAS, PTPN11, RAF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Norrie-Syndrom (NDP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nystagmus (FRMD7, GPR143)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Oligosaccharidose (AGA, CTSA, FUCA1, MAN2B1, MANBA, NAGA, NEU1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Optikusatrophie (inkl. LHON- Hotspots) (ACO2, AFG3L2, ANTXR1, ATP1A3, C12orf65, CISD2, DNAJC30, DNM1L, FA2H, MCAT, MFN2, MT-ND1, MT- ND4, MT-ND6, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Organische Azidämie (ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTB, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC13A5, SLC25A1, SLC25A19, SUCLA2, SUCLG1, TAZ, TMEM70)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Oro-fazio-digitales-Syndrom (C2CD3, CPLANE1, DDX59, FAM149B1, IFT172, IFT57, INTU, KIAA0753, NEK1, OFD1, SCLT1, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Osteolysegruppe (LMNA, MAFB, MMP14, MMP2, NOTCH2, TNFRSF11A, TREM2, TYROBP, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Osteosarkom (APC, BLM, RB1, RECQL4, TP53, WRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Otopalatodigitales Syndrom (FLNA, MAP3K7, SH3PXD2B, TAB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pachyonychia Congenita (KRT16, KRT17, KRT6A, KRT6B, KRT6C)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Palmoplantare Keratose (AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB4, GJB6, JUP, KANK2, KRT1, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LORICRIN, MBTPS2, PKP1, SERPINB7, SLURP1, SMARCAD1, TRPV3, WNT10A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pankreaskarzinom (ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, PALB2, PRSS1, STK11)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pankreatitis (CASR, CFTR, CLDN2, CPA1, CTSC, SBDS, SPINK1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Paragangliom-Phäochromozytom- Syndrom (FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Parkinson Krankheit (ATP13A2, CHCHD2, DNAJC13, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, LRRK2, PARK7, PDE8B, PINK1, PODXL, PRKN, SLC18A2, SLC6A3, SNCA, SYNJ1, TAF1, VPS13C, VPS35)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Paroxysmale Dyskinesie (ADCY5, KCNMA1, PNKD, PRRT2, SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Patellare Dysostosen (CDC6, CDT1, KAT6B, LMX1B, ORC1, ORC4, ORC6, PITX1, TBX4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pena-Shokeir-Syndrom Typ I (ADCY6, ADGRG6, CHRNA1, CHRND, CHRNA2, CNTN1, CNTNAP1, DNM2, DOK7, ERBB3, GBE1, GLE1, KIAA1109, KLHL40, MUSK, MYBPC1, NUP88, PIP5K1C, RAPSIN, RYR1, ZBTB42)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pendred-Syndrom (FOX11, KCNJ10, SLC26A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Periodische Fiebersyndrome (ADA, ADA2, ADAM17, AP1S3, BACH2, CARD14, COPA, ELANE, FOXD3, HAX1, HSPA1L, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL36RN, LPIN2, LRBA, MEFV, MVK, NFAT5, NLRC4, NLRP1, NLRP12, NLRP3, NLRP7, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RAB27A, RBCK1, RNF31, SH3BP2, SLC29A3, STING1, TNFAIP3, TNFRSF11A, TNFRSF1A, TTC7A, WDR1, XIAP, ZBTB24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Peroxisomen-Biogenese-Störungen (PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Perrault-Syndrom (CLPP, HARS2, HSD17B4, LARS2, TWNK)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Phenylketonurie und Hyperphenylalaninämie (DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Phosphatdiabetes (ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyzystische Lebererkrankung (PCLD) (ALG8, ALG9, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pontozerebelläre Hypoplasie (AMPD2, CASK, EXOSC3, RARS2, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN54, VPS53, VRK1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Porphyrie (ALAD, ALAS2, CPOX, FECH, GATA1, HMBS, PPOX, UROD, UROS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Potentiell letale Skeletterkrankungen (AGPS, ALPL, ARSL, BMPER, CANT1, CEP120, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DHCR7, DLL3, DYNC2H1, EBP, EVC, EVC2, FAM111A, FAM20C, FGFR2, FGFR3, FLNA, FLNB, GLI3, GNPAT, IFT140, IFT172, IFT80, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX7, PPIB, PTH1R, RNU4ATAC, SLC26A2, SLC35D1, SOX9, TRIP11, TTC21B, WDR19, WDR34, WDR60, WNT7A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Primäre ciliäre Dyskinesie (ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, LRRC6, MCIDAS, NME8, OFD1, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, SPEF2, TTC25, ZMYND10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Primärer Aldosteronismus (ARMC5, CACNA1D, CACNA1H, CLCN2, CTNNB1, CYP11B1, CYP11B2, HSD11B2, KCNJ5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Progressive familiäre intrahepatische Cholestase (ABCB11, ABCB4, ABCC2, ABCD3, ACOX2, AKR1D1, ATP8B1, DCDC2, HSD3B7, KIF12, LSR, MPV17, MYO5B, NR1H4, PPM1F, TJP2, USP53, VIPAS39, VPS33B, WDR83OS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Progressive myoklonische Epilepsie (ASAH1, CSTB, EFHC1, EPM2A, GOSR2, KCNC1, KCTD7, NEU1, NHLRC1, POLG, PRICKLE1, SCARB2, TBC1D24, TPP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Prolaktinom (AIP, DICER1, MEN1, SDHB)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Prostatakarzinom (ATM, BRCA1, BRCA2, CHEK2, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Proteus-Syndrom (AKT1, PIK3CA, PTEN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Proximale Tubulopathie (ABCC6, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BCS1L, CLCN5, COQ9, COX10, CTNS, CUBN, CYP27B1, CYP2R1, DMP1, EHHADH, ENPP1, FAH, FGF23, G6PC, GATM, GLA, GRHPR, HNF4A, HOGA1, HPRT1, LRP2, MOCOS, MRPS22, NDUFAF6, OCRL, PHEX, RRM2B, SLC20A1, SLC20A2, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A4, SLC7A9, SLC9A3R1, VDR, XDH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pseudohypoaldosteronismus (CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pseudohypoparathyreoidismus (GNAS, STX16)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pulmonale Hypertonie (ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, KCNK3, SMAD4, SMAD9, TBX4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pyoderma gangrenosum - Akne - Hidradenitis suppurativa (FGFR2, GJB2, NCSTN, PSEN1, PSENEN, PSTPIP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Renale Glukosurie (SLC5A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Renale tubuläre Dysgenese (ACE, AGT, AGTR1, REN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Renale tubuläre Azidose (ATP6V0A4, ATP6V1B1, CA2, FOX11, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Renales Fanconi-Syndrom (ATP6V0A4, ATP6V1B1, ATP7B, CLCN5, CTNS, EHHADH, FAH, G6PC, GATM, HNF4A, NDUFAF6, OCRL, SLC34A1, SLC4A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa, autosomal-dominant (ARL3, BEST1, CA4, CRX, CTNNA1, FSCN2, GUCA1B, HK1, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RPE65, SEMA4A, SNRNP200, TEAD1, TOPORS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Retinitis pigmentosa, autosomal-rezessiv (ABCA4, ABHD12, ADGRA3, ADIPOR1, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, ASRGL1, BBS2, BBS4, BEST1, C8orf37, CC2D2A, CDH16, CEP290, CERKL, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, DNAJC17, DYNC2H1, EMC1, EYS, FAM161A, FLVCR1, HGSNAT, IDH3A, IDH3B, IFT140, IFT172, IMPG1, IMPG2, KIAA1549, KIZ, LAMA1, LRAT, MAK, MERTK, MFSD8, NR2E3, NRL, PCARE, PDE6A, PDE6B, PDE6G, PLA2G5, PNPLA6, POMGNT1, PRCD, PROM1, PRPF31, PRPH2, RBP3, RBP4, RCBTB1, RDH11, RDH12, RDH5, REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RPE65, RPGRIP1, SAG, SCAPER, SLC7A14, SPATA7, TRNT1, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, WDR19, ZNF408, ZNF513)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa, syndromal (ABHD12, CC2D2A, CEP164, CEP19, COL9A1, ERCC6, EVC, EVC2, GNPTG, IFT140, INVS, IQCB1, KIF11, MFSD8, NPHP3, OAT, PANK2, PHYH, PLK4, POC1A, POC5, RB1, RDH11, TUBGCP6, WDR19, XPNPEP3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa, X-gekoppelt (CHM, OFD1, PGK1, RP2, RPGR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinoblastom (RB1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Rhabdomyosarkom (DICER1, NF1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Rubinstein-Taybi-Syndrom (CREBBP, EP300, SRCAP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
SANDD-Syndrom (CACNA1D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schilddrüsenerkrankung (CDCA8, DUOX1, DUOX2, DUOX2A, FOXE1, GLIS3, GNAS, IGSF1, IYD, KAT6B, LHX3, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBCE, TG, THRA, THRB, TPO, TRH, TSHB, TSHR, TUBB1, UBR1, ZFAT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schilddrüsenkarzinom (APC, CDKN1B, DICER1, FOXE1, HABP2, MEN1, MINPP1, NKX2-1, PRKAR1A, PTEN, RET, SDHB, SDHD, SRGAP1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit (ACTG1, ACTN4, ADCY1, ADGRV1, AIFM1, AMACR, ANLN, APOL1, ARHGAP24, ARHGAP24, ATP2B2, AVIL, BSND, CABP2, CACNA1D, CCDC50, CD2AP, CDC14A, CDH23, CEACAM16, CEP250, CHD7, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COQ2, COQ6, COQ8B, CRB2, CRYM, CUBN, DCDC2, DGKE, DIABLO, DIAPH1, DIAPH3, DLC1, DSPP, EDN3, EDNRB, ELMOD3, EMP2, EPS8, EPS8L2, ESPN, ESRB, EYA1, EYA4, FAT1, FN1, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HSD17B4, ILDR1, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LAGE3, LAMB2, LARS2, LHFPL5, LMX1A, LMX1B, LOXHD1, LOXL3, LRP2, LRTOMT, MAGI2, MARVELD2, MIR96, MITF, MPZL2, MSRB3, MT- RNR1, MT-TL1, MT-TS2, MYH14	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fortsetzung nächste Seite			

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Schwerhörigkeit (Fortsetzung MYH9, MYO15A, MYO1E, MYO3A, MYO6, MYO7A, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSBPL2, OSGEP, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX2, PAX3, PCDH15, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PJVK, PLCE1, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRO, PTPRQ, RDX, S1PR2, SCARB2, SERPINB6, SGPL1, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMARCAL1, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TBC1D8B, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TNS2, TP53RK, TPRKB, TPRN, TRIOBP, TRPC6, TRRAP, TSPEAR, TTC21B, TWNK, USH1C, USH1G, USH2A, WDR73, WFS1, WHRN, WT1, XPO5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit, autosomal- dominante nicht-syndromale (ACTG1, CCDC50, CEACAM16, COCH, COL11A2, CRYM, DIABLO, DIAPH1, DIAPH3, ESPN, EYA4, GJB2, GJB3, GJB6, GRHL2, GSDME, KCNQ4, KITLG, LMX1A, MIR96, MYH14, MYH9, MYO6, MYO7A, OSBPL2, P2RX2, PLS1, POU4F3, PTPRQ, SIX1, SLC17A8, TBC1D24, TECTA, TJP2, TMC1, TNC, TRRAP, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit, autosomal- rezessive nicht syndromale (ADCY1, ATP2B2, BSND, CABP2, CDC14A, CDH23, CIB2, CLDN14, CLIC5, CLPP, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PNPT1, PTPRQ, RDX, S1PR2, SERPINB6, SLC26A4, SLC26A5, SLITRK6, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, TSPEAR, USH1C, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Schwerhörigkeit, mitochondriale (MT-RNR1, MT-TS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Senior-Loken-Syndrom (CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1, WDR19)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Sensenbrenner-Syndrom (DPH1, DYNC2H1, IFT122, IFT43, WDR19, WDR35)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Septo-optische Dysplasie - Spektrum (ALG13, ARNT2, FGF8, FGFR1, FLNA, HESX1, OTX2, PAX6, PROKR2, SOX2, SOX3, STAG2, TCF7L1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Single Nucleotide Polymorphisms for Gender Determination (AMELX, AMELY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Skelettaler Kleinwuchs (ACAN, BMP2, BMPR1B, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, FBN1, FGFR3, GDF5, GNAS, IHH, MATN3, NPR2, PAPS2, PRKAR1A, PTH1R, PTHLH, ROR2, SHOX, SLC26A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Skelettdysplasie mit abnormer Mineralisierung (ALPL, ANKH, AP2S1, CASR, CDC73, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAM20C, FGF23, GCM2, GNA11, HRAS, NRAS, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, TRPV6, VDR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Skelettdysplasie mit erhöhter Knochendichte (AMER1, ANKH, CA2, CLCN7, COL1A1, CSF1R, CTSK, DHCR24, DLX3, FAM20C, FERMT3, FGF23, GALNT3, GJA1, HPGD, IKBKG, KL, LEMD3, LRP4, LRP5, LRRK1, MAP2K1, MTAP, OSTM1, PLEKHM1, POLR3B, PTDSS1, PTH1R, SFRP4, SLC29A3, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Skelettdysplasien mit reduzierter Knochendichte (ALPL, ANOS, ATP6V0A2, B3GALT6, B4GALT7, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, DDX58, FKBP10, GORAB, IFIH1, IFITM5, LRP5, MBTPS2, NBAS, NOTCH2, P3H1, P4HB, PLOD2, PLS3, POLR3A, PPIB, PYCR1, SEC24D, SERPINF1, SERPINH1, SGMS2, SP7, SPARC, TENT5A, TMEM38B, TNFRSF11B, WNT1, XYLT2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Sotos-Syndrom (EZH2, NFIX, NSD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Spastische Paraplegie, hereditäre (ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GFAP, GJC2, HSPD1, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NT5C2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TH, VAMP1, VPS37A, WASHC5, ZFYVE26, ZFYVE27)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Sphärozytose, hereditäre (ANK1, EPB42, SLC4A1, SPTA1, SPTB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Spinale Muskelatrophie (SMA), distale Form (AARS1, ASAH1, ATP7A, BICD2, BSCL2, DCTN1, DNAJB2, DYNC1H1, EXOSC3, GARS1, HSPB1, HSPB8, IGHMBP2, PLEKHG5, SIGMAR1, SMN1, TRIP4, TRPV4, UBA1, VRK1, WARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stargardt-Krankheit (ABCA4, CTNNA1, ELOVL4, IMPG1, IMPG2, PROM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stickler-Syndrom (COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Störung der Fettsäureoxidation (ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADHA, HADHB, HSD17B10, MLYCD, SLC22A5, SLC25A20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Störungen der Geschlechtsentwicklung XX (AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ATRX, BMP15, BNC1, CBX2, CHD7, CLPP, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHH, DIAPH2, DMRT1, DMRT2, DUSP6, EIF4ENIF1, ERCC6, ESR1, FANCM, FEZF1, FGF17, FGF8, FGFR1, FIGLA, FLRT3, FOXL2, FSHB, FSHR, GALT, GATA4, GDF9, GNRH1, GNRHR, HARS2, HFM1, HHAT, HNF1B, HOXA13, HOXA7, HS6ST1, HSD17B3, HSD17B4, HSD3B2, IL17RD, KISS1, KISS1R, KLHL4, LARS2, LHB, LHCGR, LHX1, MAMLD1, MAP3K1, MCM8, MCM9, MSH5, NOBOX, NROB1, NR5A1, NSMF, PAX2, POF1B, POLG, POR, PROK2, PROKR2, PSMC3IP, RSPO1, SAMD9, SEMA3A, SF1, SOX3, SOX9, SPRY4, SRD5A2, SRY, STAG3, STAR, SYCE1, TAC3, TACR3, TBX6, WDR11, WNT4, WNT9B, WT1, ZFPM2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Störungen der Geschlechtsentwicklung XY (AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, BMP15, CBX2, CHD7, CLPP, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DIAPH2, DMRT1, DMRT2, DUSP6, EIF4ENIF1, ESR1, FEZF1, FGF17, FGF8, FGFR1, FIGLA, FLRT3, FOXL2, FSHB, FSHR, GALT, GATA4, GDF9, GNRH1, GNRHR, HARS2, HFM1, HHAT, HNF1B, HOXA13, HOXA7, HS6ST1, HSD17B3, HSD17B4, HSD3B2, IL17RD, KISS1, KISS1R, KLHL4, LARS2, LHB, LHCGR, LHX1, MAMLD1, MAP3K1, MCM9, NOBOX, NROB1, NR5A1, NSMF, PAX2, POF1B, POR, PROK2, PROKR2, PSMC3IP, RPL10, RSPO1, SAMD9, SEMA3A, SF1, SGPL1, SOX3, SOX9, SPRY4, SRD5A2, SRY, STAG3, STAR, SYCE1, TAC3, TACR3, TBX6, TOE1, WDR11, WNT4, WNT9B, WT1, ZFPM2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Störungen der Knochenentwicklung (inkl. Exostosen) (ACVR1, EXT1, EXT2, FGFR1, GNAS, IDH1, IDH2, MET, NF1, PTPN11, SH3BP2, TREM2, TYROBP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Surfactant-Protein-Mangel (ABCA3, CSF2RA, CSF2RB, NKX2-1, SFTPB, SFTPC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Syndrom, autoimmun-lymphoproliferatives (AIRE, CASP10, CASP8, FADD, FAS, FASLG, FOXP3, IL2RA, ITCH, PRKCD, STAT3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Syndrom der unkämbaren Haare (PADI3, TCHH, TGM3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Syndromaler Insulinmangel (CISD2, DCAF17, DNAJC3, DUT, EIF2AK3, EIF2S3, FOXP3, GATA4, GATA6, GLIS3, IER3IP1, MT-TL1, PAX6, PPP1R15B, RFX6, SLC19A2, SLC29A3, STAT3, TRMT10A, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Syndromaler Kleinwuchs (ACAN, ANKRD11, ASXL1, ATR, ATRX, BLM, BRAF, CCDC8, CDKN1C, CDT1, CEP152, CHD7, CREBBP, CUL7, DHCR7, DVL1, DVL3, EP300, ERCC6, ERCC8, EVC, EVC2, FAM111A, FANCA, FANCC, FANCG, FGD1, FGFR3, GLI2, GLI3, HDAC8, HRAS, IARS1, IGF1R, KDM6A, KMT2D, KRAS, LARP7, LIG4, LMNA, MAP2K1, MAP2K2, NBN, NIPBL, OBSL1, ORC1, ORC4, ORC6, PCNT, PIK3R1, POC1A, PTPN11, RAD21, RAF1, RNU4ATAC, ROR2, RPS6KA3, SEMA3E, SMARCAL1, SMC1A, SMC3, SOS1, SRCAP, TBCE, TRIM37, WNT5A, XRCC4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Syndromische Schwerhörigkeit (ACTN4, ADGRV1, AMACR, ANLN, APOL1, ARHGAP24, ARHGDI, AVIL, CACNA1D, CD2AP, CDH23, CEP250, CHD7, CIB2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EDN3, EDNRB, EMP2, EYA1, FAT1, FN1, FOXI1, HARS1, HARS2, HSD17B4, INF2, ITGA3, ITGB4, ITS1N1, KANK1, KANK2, KANK4, KCNE1, KCNJ10, KCNQ1, KIT, LAGE3, LAMB2, LARS2, LMX1B, LOXL3, LRP2, MAGI2, MITF, MT-RNR1, MT-TL1, MT-TS2, MYH9, MYO1E, MYO7A, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PAX3, PCDH15, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PLCE1, POLR1C, POLR1D, PTPRO, SCARB2, SGPL1, SIX1, SIX5, SLC26A4, SMARCAL1, SNAI2, SOX10, TBC1D8B, TCOF1, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, TWNK, USH1C, USH1G, USH2A, WDR73, WHRN, WT1, XPO5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Tachykardie, katecholaminerge polymorphe ventrikuläre (ANK2, CALM1, CALM2, CALM3, CASQ2, GNAI2, KCNJ2, RYR2, SCN5A, TECRL, TRDN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thrombozytopenie (ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CYCS, DIAPH1, ETV6, FCGR2C, FLI1, FLNA, FYB1, GALE, GATA1, GF11B, GNE, GP1BA, GP1BB, GP9, HOXA11, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, ORAI1, PRKACG, RBM8A, RUNX1, SLFN14, SRC, STIM1, THBD, THPO, TPM4, TUBB1, VWF, WAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thrombozytose, familiäre (JAK2, MPL, THPO)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Treacher-Collins-Syndrom (POLR1C, POLR1D, TCOF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tuberöse Sklerose Komplex (TSC) (TSC1, TSC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tubulointerstitielle Nierenerkrankung, autosomal-dominant (DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tumorerkrankung mit unbekanntem Primärtumor (ARID1A, BRAF, KMT2D, KRAS, MYC, PTEN, SMARCA4, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Tyrosinämie (FAH, HPD, TAT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
T-Zell-Lymphom, peripheres (DNMT3A, FAS, IDH2, JAK1, PLCG1, PRDM1, PRF1, RHOA, STAT3, STIM2, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Usher Syndrom Typ 1 (CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Usher-Syndrom (ADGRV1, CDH23, CEP250, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Usher-Syndrom Typ 2 (ADGRV1, USH2A, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
VACTERL-Assoziation (CPS1, FANCB, FANCI, FGF8, FOXF1, HAAO, HOXD13, KYNU, LPP, PALB2, PCSK5, TRAP1, ZIC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Vaskulopathie (ACVRL1, COL4A1, EFEMP2, ENG, FLNA, GDF2, NOTCH1, PKD1, SLC2A10, SMAD4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Vorderkammerdysgenese (ASPH, B3GLCT, BMP4, COL4A1, CPAMD8, CYP1B1, ELP4, EYA1, FOXC1, FOXC2, FOXE3, JAG1, LAMB2, MIR184, PAX6, PITX2, PITX3, PXDN, SLC38A8, VSX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Waardenburg-Syndrom (EDN3, EDNRB, KIT, KITLG, MITF, PAX3, SNAI2, SOX10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Walker-Warburg-Syndrom (B3GALNT2, B4GAT1, CRPPA, FKR, FKTN, GMPBB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Warburg-Mikro-Syndrom (RAB18, RAB3GAP1, RAB3GAP2, TBC1D20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Weibliche Infertilität (AR, BMP15, BTG4, BUB1B, C11orf80, CBX2, CFTR, CLPP, CPEB1, CYP11A1, CYP21A2, DHH, EIF2B1, EIF2B2, EIF2B4, EIF2B5, EIF4ENIF1, ESR1, ESR2, F2, F5, FGF8, FGFR1, FIGLA, FOXL2, FSHB, FSHR, GNRH1, GNRHR, HARS2, HFM1, HSD17B3, HSD17B4, INHA, KHDC3L, KISS1, KISS1R, LARS2, LHB, LHCGR, LHX8, MAP3K1, MCM8, MCM9, MEI1, MRPS22, MTHFR, NANOS3, NLRP2, NLRP5, NLRP7, NOBOX, NROB1, NR5A1, NUP107, PADI6, PANX1, PATL2, PROC, PROKR2, PROS1, PSMC3IP, REC114, SEMA3A, SERPINC1, SERPINE1, SMC1B, SOHLH1, SPIDR, SRD5A2, SRY, STAG3, SYCE3, SYCP3, TAC3, TACR3, THBD, TRIP13, TUBB8, WDR11, WEE2, WNT4, ZP1, ZP2, ZP3)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wilms-Tumor-Aniridia-Syndrom (ELP4, PAX6, TRIM44, WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Xanthinurie (MOCOS, XDH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Xeroderma pigmentosum (DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, RECQL4, SLC6A19, XPA, XPC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
X-gekoppelte, rezessive nicht syndromale Schwerhörigkeit (XLNSHL) (AIFM1, POU3F4, PRPS1, SMPX, TIMM8A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Zapfen-Stäbchen-Dystrophie (ABCA4, ACBD5, ADAM9, AIPL1, BEST1, C1QTNF5, C8orf37, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP78, CERKL, CFAP410, CLN5, CNGA3, CNGB3, CNNM4, CRX, DRAM2, ELOVL4, FBLN5, GUCA1A, GUCA1B, GUCY2D, HMCN1, IFT81, ITM2B, KCNV2, MERTK, MFSD8, MT- ATP6, MT-TL1, PCYT1A, PDE6C, PITPNM3, PLK4, POC1B, PRDM13, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RP1L1, RPGR, RPGRIP1, SEMA4A, TEAD1, TIMP3, TTLL5, TUBGCP4, TUBGCP6, UNC119, WASF3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Zerebrale cavernöse Fehlbildungen (CCM2, KRIT1, PDCD10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Zystische Nierenerkrankung (ALG9, ANKS6, BICC1, COL4A1, COL4A4, DICER1, DNAJB11, DZIP1L, GANAB, HNF1B, INVS, KAT6B, LRP5, NOTCH2, NPHP3, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, TMEM67, UMOD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adrenogenitales-Syndrom (ARMC5, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, MC2R, PDE8B, POR, STAR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0071-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Whole Exome Sequencing (WES)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0097-00 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Whole Exome Sequencing (WES), Trio-Analyse	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0097-00 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Zapfendystrophie, X-gekoppelt ORF15 -Region des RPGR-Gens (NGS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-SAA-0099-01 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-SAA-0096-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0159-01
1p36 Deletionssyndrom	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
ABCD-Syndrom (EDNRB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Achondrogenesie Typ 2 (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
acrocallosales Syndrom (GLI3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adipositas (LEPR, MC4R, POMC, LEP, MC2R, SIM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
ADPKD (PKD1, PKD2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alagille Syndrom (JAG1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Albinismus, okulokutaner (OCA2, TYR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Albinismus, okulärer (GPR143, PITX2, FOXC1, TWIST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alpha-Galaktosidase-A-Mangel (GLA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alpha-Thalassämie (HBA1, HBA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alport-Syndrom (COL4A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03.1 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alport-Syndrom (COL4A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alport-Syndrom (COL4A5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Androgeninsensitivitäts-Syndrom (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Angelman Syndrom (SNRPN, UBE3A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Aniridie / WAGR (PAX6, WT1, SOX2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
ARPKD (PKHD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ataxia Teleangiectasia (ATM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bartter-Syndrom (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Bartter-Syndrom, klassisches (CLCNKB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Beckwith-Wiedemann-Syndrom (H19, KCNQ1OT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Beta-Thalassämie (HBB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
BOR-Syndrom (EYA1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (BARD1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (BRIP1/CHEK1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (CHEK2)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (BRCA1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (BRCA2)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (PALB2, RAD50, RAD51D)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (ATM)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (STK11)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
CADASIL (NOTCH3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Caffey-Krankheit (COL1A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Charcot-Marie-Tooth-Krankheit Typ 1A (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
CHARGE-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chorioideremie (CHM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cohen-Syndrom (VPS13B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cowden-Syndrom Typ 1 (PTEN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cystinose (CTNS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cystinurie (SLC3A1, SLC7A9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diabetes mellitus, permanenter neonataler (INS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Diabetes mellitus, transienter neonataler (6q24, UPD6, ZFP57, PLAGL1, KCNJ11, INS, ZC2HC1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
DiGeorge-Syndrom (TBX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dopa-responsive Dystonie (TH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysostose, kleidokraniale (RUNX2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Dysplasie, kampomele (SOX9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dystonie, Dopa-responsive (GCH1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
EGFR-Mutationsanalyse (EGFR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ehlers-Danlos-Syndrom Typ 6 (PLOD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ehlers-Danlos-Syndrom, hypermobiler Typ (TNXB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ehlers-Danlos-Syndrom, vaskulärer Typ (COL3A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
ektodermale Dysplasie, hypohidrotische (EDARADD, EDAR, EDA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ellis-van-Crevelde-Syndrom (EVC, EVC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
familiäres Mittelmeerfieber (MEFV)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fanconi-Anämie (FANCA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Platelet-Syndrom mit Prädisposition für akute myeloische Leukämie (FDP/AML) (RUNX1, GATA2, TERC, TERT, CEBPA)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Feingold-Syndrom Typ 1 und 2 (MYCN, MIR17HG)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fruktose-1,6-Diphosphatase- Mangel und Fruktose-Intoleranz (ALDOB, FBP1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Galaktosämie (GALT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Geschlechtsdifferenzierung (SRY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gitelman-Syndrom (SLC12A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gliedergürteldystrophie (LGMD) (SGCA, SGCB, SGCD, SGCG, FKRP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gliedergürteldystrophie (DYSF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
GLUT1-Defekt (SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glykogenspeicherkrankheit Typ 2 (GAA)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gonadendysgenese (NR5A1, NR0B1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Greig-Syndrom (GLI3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
HDR-Syndrom (GATA3, GATA4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
hereditäre Neuropathie mit Neigung zu Drucklähmungen (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Holt-Oram-Syndrom (TBX5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyper-IgE-Syndrom (DOCK8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hypercholesterinämie (LDLR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperinsulinismus, kongenitaler (ABCC8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperkalzämie, familiäre hypokalziurische (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperoxaluria, primäre (AGXT, GRHPR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypochondrogenese (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypokalzämie (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypophosphatasie (ALPL/TNSALP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypophosphatämie / hypophosphatämische Rachitis (FGF23, PHEX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypophysenhormon-Defizienz, kombinierte (LHX3, LHX4, GH1, HESX1, POU1F1, PROP1, GHRHR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämochromatose (HFE, TFR2, HFE2, HAMP, SLC40A1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämolytisch-urämisches Syndrom, atypisches (MCP/CD46, CFI)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämolytisch-urämisches Syndrom, atypisches (CFH und CFH-verbundene Gene (CFHR1-CFHR5))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ichthyose, X-chromosomale (STS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
IGF1-Mangel (IGF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kabuki-Syndrom (KDM6A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kabuki-Syndrom (MLL2/KMT2D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kallmann-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kallmann-Syndrom (FGFR1,PROK2, PROKR2, GNRHR, KISS1R, GNRH1, NELF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kallmann-Syndrom (KAL1/ANOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie (MYBPC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie, arrhythmogene ventrikuläre (PKP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kniest-Dysplasie (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (MLH1, MSH2)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (MSH6, EPCAM)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (PMS2)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kraniosynostosen (FGFR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lafora-Krankheit (EPM2A, EPM2B/NHLRC1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Laron Syndrom (GHR, IGF1, JAK2, STAT5B), Laron-ähnliches Syndrom	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leber congenitale Amaurose (GUCY2D, RDH12, RPGRIP1, CEP290 (nur Intron 26))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leber congenitale Amaurose (CRX, AIPL1, CRB1, LCA5, RPE65)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Legius-Syndrom (SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Li-Fraumeni-Syndrom (TP53)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Loeys-Dietz Syndrom (TGFB1, TGFB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Long-QT-Syndrom & Brugada- Syndrom (SCN5A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Long-QT-Syndrom (KCNQ1, KCNH2, KCNE1, KCNE2, KCNJ2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphohistiozytose, hämophagozytische (PRF1, STX11, UNC13D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Magenkarzinom, hereditäres diffuses (CDH1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Marfan-Syndrom (FBN1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mayer-Rokitansky-Küster-Hauser-Syndrom (WNT4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Medium-chain-Acyl-CoA-Dehydrogenase-Mangel (ACADM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mental retardation-hypotonic facies syndrome, X-linked (ATRX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mikrodeletionssyndrom (TSC2, PKD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mikrozephalie (STIL, ASPM, MCPH1, CDK5RAP2, CENPJ, WDR62)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
MODY (HNF1A, GCK, HNF4A, HNF1B)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
MODY (PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Morbus Gaucher (GBA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Morbus Wilson (ATP7B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mukoviszidose (CFTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Multilocus Methylierungsdefekte (PLAGL1, GRB10, MEST, H19, KCNQ1OT1, DLK1, MEG3, RTL1, SNRPN, PEG3, GNAS, MEG8, GNASXL, NESP55)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Muskeldystrophie Duchenne/Becker (DMD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Muskeldystrophie, kongenitale (LAMA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
MYH9-zugehörige Erkrankungen (MYH9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myotonia congenita Typ Becker/Thomsen (CLCN1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nagel-Patella-Syndrom (LMX1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neoplasie, multiple endokrine, Typ 1 (MEN1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephronophthise (NPHP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurodegeneration mit Eisenspeicherung im Gehirn (PANK2 + PLA2G6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurofibromatose Typ 1 (NF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurofibromatose Typ 2 (NF2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nijmegen-Chromosomenbruch- Syndrom (NBN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Norrie-Syndrom (NDP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Optikusatrophy (OPA1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ornithintranscarbamylase-Mangel (OTC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Osteogenesis imperfecta, autosomal-dominant (COL1A1,COL1A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Parkinson-Krankheit (PARK7, ATP13A2, PINK1, UCHL1, SNCA, PARK2, CAV1/2, LRRK2, GCH1)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pelizaeus-Merzbacher-Krankheit (PLP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pendred-Syndrom (SLC26A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Peutz-Jeghers-Syndrom (STK11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Phelan-McDermid-Syndrom (SHANK3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Phenylketonurie (PAH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pheochromocytoma-Paraganglioma-Syndrom (SDHB, SDHC, SDHD, SDHAF1, SDHAF2)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyposis coli, familiäre adenomatöse (APC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyposis, MUTYH-assoziierte (MUTYH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Prader-Willi-Syndrom (SNRPN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Progressive familiäre intrahepatische Cholestase (ABCB4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Propionazidämie (PCCA, PCCB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pseudohypoparathyreoidismus 1a und 1b (GNAS, STX16)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
RAS Gene (BRAF, HRAS, KRAS, NRAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
RCAD, renales Zysten- und Diabetes-Syndrom (HNF1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa (PRPF31, RP1, IMPDH1, RHO)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa (RP2, RPGR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa (ABCA4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis pigmentosa (RP25/EYS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinoblastom (RB1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Rett-Syndrom (MECP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Rett-Syndrom, atypisches (FOXP1, CDKL5, NTNG1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Robinow-Syndrom, autosomal-rezessiv (ROR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Rubinstein-Taybi Syndrom (CREBBP, EP300)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schilddrüsenerkrankung (TPO, PAX8, FOXE1, NKX2-1, TSHR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Schwannomatose (SMARCB1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwere myoklonische Epilepsie des Kindesalters (Dravet) (SCN1A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit, nicht-syndromale sensorineurale (GJB2, GJB3, GJB6, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit, nicht-syndromale sensorineurale (STRC, CATSPER2, OTOA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
SHOX-Defizienz (SHOX)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Silver-Russell-Syndrom (H19, KCNQ1, KCNQ1OT1, IGF2, CDKN1C)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Silver-Russell-Syndrom (UPD7mat) (GRB10, MEST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Simpson-Golabi-Behmel-Syndrom (GPC3, GPC4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Smith-Lemli-Opitz-Syndrom (DHCR7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Sotos-Syndrom (NSD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Spastische Paraplegie (ATL1, SPAST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Spinale Muskelatrophie (SMN1, SMN2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Spondyloepiphysäre Dysplasie, kongenitaler Typ (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stickler-Syndrom Typ 1 (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stickler-Syndrom Typ 2 (COL11A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Surfactant-Protein-Defizienz (ABCA3, SFTPC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tay-Sachs-Syndrom (HEXA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Teleangiektasie, hämorrhagische hereditäre (ENG, ACVRL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Temple-Syndrom (UPD14mat) (DLK1, MEG3, RTL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Townes-Brocks-Syndrom (SALL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Treacher-Collins-Syndrom (TCOF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tuberöse Sklerose (TSC1, TSC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tumor-Prädispositionssyndrom (BAP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Uniparentale Disomie 7 (GRB10, MEST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Uniparentale Disomie 14 (DLK1, MEG3, RTL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Usher Syndrom (PCDH15)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Usher Syndrom (USH2A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Von-Hippel-Lindau-Syndrom (VHL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Von-Willebrand-Syndrom (VWF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Waardenburg Syndrom (PAX3, MITF, SOX10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Waardenburg-Shah Syndrom (EDNRB, NRTN)	EDTA-Blut, DNA; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wachstumshormonmangel (GH1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Weaver-Syndrom (NSD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wilms-Tumor (WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Zapfen-Stäbchen-Dystrophie (PRPH2, BEST1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adrenogenitales Syndrom (CYP17A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0071-00.1
Adrenogenitales Syndrom (CYP21A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0146-03 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0071-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Angiotensin 1 konvertierendes Enzym (ACE) Insertions-/Deletions-Polymorphismus	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Atrophie, dentato-rubro-pallidoluysische (ATN1)	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Dihydropyrimidin-Dehydrogenase-Mangel (DPYD)	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Kennedy Syndrom (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Leukämie (Fusionsgen-Panel) (AML1 ETO, BCR-ABL, CALM-AF10, CBF-MYH11, DEK-CAN, MLL-AF6, MLL-AF9, MLL-ELL, MLL-PTD (part. tandem dupl.), NPM1-MLF1, PML-RARA)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0072-00.1 BIO-ING-HUM-SAA-0159-01 BIO-ING-HUM-SAA-0063-02.2
Leukämie, akute promyelozytische (PML-RARA, qualitativ)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0072-00.1 BIO-ING-HUM-SAA-0159-01 BIO-ING-HUM-SAA-0063-02.2
Mukoviszidose (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Ondine-Syndrom (PHOX2B, Repeat, Stufe 1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Pankreatitis, hereditär (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Partielle Chromosom Y-Deletion, AZF-Faktor (AZF, Devyser-Kit)	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Spinocerebelläre Ataxie (ATXN2/SCA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (ATXN3/SCA3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (ATXN7/SCA7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (CACNA1A/SCA6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (SCA1, Fragment)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (SCA1, Restriktion)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (SCA8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (SCA12)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (TBP/SCA17)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Spinocerebelläre Ataxie (SCA 1,2,3,6,7,17)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2



Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Sterilität, CBAVD (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Testung auf verschobene X-Inaktivierung (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Trisomie-Schnelltest	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	QF-PCR, Mikro /Mini Satellitenanalyse (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0063-02.2
Leukämie (BCR-ABL1, qualitativ)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0072-00.1 BIO-ING-HUM-SAA-0159-01 BIO-ING-HUM-SAA-0017-00 BIO-ING-HUM-SAA-0063-02.2
Uniparentale Disomie der Chr. 2, 4, 5, 6, 7, 8, 9, 10, 11, 11(BWS), 13, 14, 15, 16, 17, 18, 20, 22, X	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0064-02 BIO-ING-HUM-SAA-0063-02.2
Fragiles X-Syndrom (FMR1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0173-02 BIO-ING-HUM-SAA-0063-02.2
Leukämie (BCR-ABL1, Expression)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0028-01 BIO-ING-HUM-FB-0072-00.1 BIO-ING-HUM-SAA-0159-01
Leukämie, akute promyelozytische (PML-RARA, Expression)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0039-01 BIO-ING-HUM-FB-0072-00.1 BIO-ING-HUM-SAA-0159-01
Neoplasie, myeloproliferative (JAK2, Exon 14, V617F)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Real-time PCR	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0035-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Faktor V-Genmutation	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0093-00
Hämochromatose Genanalyse	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0022-02
HLA-B27	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0028-00.5
Prothrombin (Faktor II)-Genmutation	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0093-00
HLA-Typisierung	EDTA-Blut; DNA	SSO-PCR	BIO-ING-MOLE-SAA-0001-00.6
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	Schmelzpunktanalyse	BIO-ING-MOLE-SAA-0078-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	Sequenzierung	BIO-ING-MOLE-SAA-0087-00.2

### Untersuchungsgebiet: Humangenetik (Zytogenetik)

#### Untersuchungsart:

#### Chromosomenanalyse \*\*

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
angeborener Chromosomensatz	Blut, Nabelschnurblut, Fibroblasten, Amnionzellen, Chorionzotten, Abortgewebe	Chromosomenbänderungs-analyse	BIO-ING-HUM-SAA-0084-01.3 BIO-ING-HUM-SAA-0085-00.3 BIO-ING-HUM-FB-0001.-00.2 BIO-ING-HUM-VAW-0002-00.3 BIO-ING-HUM-SAA-0002-00.4 BIO-ING-HUM-SAA-0044-00.4 BIO-ING-HUM-SAA-0049-00 BIO-ING-HUM-SAA-0047-01.3 BIO-ING-HUM-VAW-0019-00.3 BIO-ING-HUM-SAA-0046-00.2 BIO-ING-HUM-SAA-0048-01.1 BIO-ING-HUM-VAW-0017-00.2 BIO-ING-HUM-VAW-0018-00.3
erworbener Chromosomensatz	Blut und/oder Knochenmark	Chromosomenbänderungs-analyse	BIO-ING-HUM-SAA-0083-00.5 BIO-ING-HUM-SAA-0001-00.3 BIO-ING-HUM-FB-0001.00.2 BIO-ING-HUM-VAW-0038-01.4
angeborener Chromosomensatz	Native und/oder kultivierte Zellen aus Blut, Fibroblasten, Amnionzellen, Chorionzotten	Fluoreszenz-in-situ-Hybridisierung (FISH)	BIO-ING-HUM-VAW-0054-01.5 BIO-ING-HUM-VAW-0034-00.6 BIO-ING-HUM-VAW-0070-00.4 BIO-ING-HUM-SAA-0151-00.2 BIO-ING-HUM-SAA-0152-00.4
erworbener Chromosomensatz	Native und/oder kultivierte Zellen aus Blut und/oder Knochenmark	Fluoreszenz-in-situ-Hybridisierung (FISH)	BIO-ING-HUM-VAW-0054-01.5 BIO-ING-HUM-VAW-0034-00.6 BIO-ING-HUM-VAW-0070-00.4 BIO-ING-HUM-SAA-0152-00.4
Molekularzytogenetische Analyse zur Detektion von Imbalancen (Zugewinnen und Verlusten von genomischen Material) im gesamten Genom	Blut, Fruchtwasser, Chorionzotten, Nabelschnurblut; DNA	Vergleichende Genom-Hybridisierung an Oligonukleotid-Arrays	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0106-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0102-00.2 BIO-ING-HUM-VAW-0031-00.1 BIO-ING-HUM-VAW-0030-00.1 BIO-ING-HUM-VAW-0032-00.1 BIO-ING-HUM-SAA-0101-00.2 BIO-ING-HUM-SAA-0183-01
Nicht invasive Pränataldiagnostik (fetale Aneuploidien für Chromosomen 13,18,21,X und Y)	peripheres Vollblut; DNA	Next Generation Sequencing (NGS), VeriSeq NIPT	BIO-ING-HUM-SAA-0187-00

## Untersuchungsgebiet: Mikrobiologie

### Untersuchungsart: Agglutinationsteste\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Haemophilus influenzae-, Escherichia coli-, Neisseria meningitidis-, Streptococcus pneumoniae-, B-Streptokokken- Antigen	Liquor, Blutkulturen, Serum, Urin, Pleurapunktat	Partikelagglutinationstest (Latex)	BIO-ING-MIKR-SAA-0057-00.5
Staphylococcus aureus, Anti- Staphylolysin, Antikörper	Serum	Partikelagglutinationstest (Latex)	BIO-ING-SERO-SAA-0043-00.3

### Untersuchungsart: Chromatographie (Immunchromatographie (IC))\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Carbapenemasen	Kulturmaterial	Immunchromatographie	BIO-ING-MIKR-SAA-0093-00.2
Cryptococcus neoformans, Antigen	Serum, Liquor	Immunchromatographie	BIO-ING-MIKR-SAA-0065-00.3

### Untersuchungsart: Spektrometrie (Nephelometrie)\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Streptokokken, DNase-B-Antikörper (ADNA)	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2

**Untersuchungsart:**

**Empfindlichkeitstestungen von Bakterien und Pilzen\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Schnellwachsende aerobe und anaerobe Bakterien sowie anspruchsvolle Bakterien	Reinkulturen	Agardiffusionstest, Bouillon-dilutionsverfahren als minimale Hemmkonzentration (MHK)Break-Point, teilmechanisiert	BIO-ING-MIKR-VAW-0018-04, BIO-ING-MIKR-VAW-0022-01.2, BIO-ING-MIKR-SAA-0061-01, BIO-ING-MIKR-SAA-0040-00.4, BIO-ING-MIKR-SAA-0052-00.3, BIO-ING-MIKR-SAA-0072-00, BIO-ING-MIKR-SAA-0031-01, BIO-ING-MIKR-SAA-0048-01, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.2, BIO-ING-MIKR-LIS-0001-00.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.1, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.2, BIO-ING-MIKR-SAA-0069-00.2, BIO-ING-MIKR-SAA-0077-00.1, BIO-ING-MIKR-SAA-0042-00.1, BIO-ING-MIKR-SAA-0076-01, BIO-ING-MIKR-SAA-0041-01, BIO-ING-MIKR-SAA-0049-00.1, BIO-ING-MIKR-SAA-0070-00.1, BIO-ING-MIKR-SAA-0044-00.3, BIO-ING-MIKR-SAA-0078-01.1, BIO-ING-MIKR-SAA-0066-00.1, BIO-ING-MIKR-SAA-0045-01.2, BIO-ING-MIKR-SAA-0046-00, BIO-ING-MIKR-SAA-0033-00.1, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-02, BIO-ING-MIKR-SAA-0049-00.1, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00
Mycobacterium-tuberculosis-Komplex	Kulturmaterial aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	modifizierte Proportionsmethode (Flüssigmedien)	BIO-ING-MIKR-SAA-0096-00
Candida albicans, non-albicans	Kulturmaterial angezüchtet aus Untersuchungsmaterial wie oben unter Kulturverfahren Hefen	Bouillon-dilutionsverfahren als minimale Hemmkonzentration (MHK)Break-Point, teilmechanisiert	BIO-ING-MIKR-VAW-0056-01.1 BIO-ING-MIKR-SAA-0098-00

**Untersuchungsart:**

**Keimdifferenzierung/-identifizierung/-typisierung\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Schnellwachsende aerobe und anaerobe Bakterien sowie anspruchsvolle Bakterien sowie Pilze	Reinkulturen	biochemisch-aufwändig (Bunte Reihe, Mehrkammerverfahren), einfach, orientierend, massenspektrometrische Erregerdifferenzierung (MALDI-TOF-MS)	BIO-ING-MIKR-SAA-0053-00.2, BIO-ING-MIKR-SAA-0058-00.1, BIO-ING-MIKR-SAA-0054-00.1, BIO-ING-MIKR-SAA-0056-00, BIO-ING-MIKR-SAA-0059-00.2, BIO-ING-MIKR-VAW-0029-01, BIO-ING-MIKR-VAW-0022-01.2, BIO-ING-MIKR-SAA-0051-00, BIO-ING-MIKR-SAA-0060-00.3, BIO-ING-MIKR-SAA-0048-01, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.2, BIO-ING-MIKR-SAA-0038-01.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.1, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.2, BIO-ING-MIKR-SAA-0069-00, BIO-ING-MIKR-SAA-0077-00.1, BIO-ING-MIKR-SAA-0042-00.1, BIO-ING-MIKR-SAA-0076-01, BIO-ING-MIKR-SAA-0041-01, BIO-ING-MIKR-SAA-0049-00.1, BIO-ING-MIKR-SAA-0070-00.1, BIO-ING-MIKR-SAA-0044-00.3, BIO-ING-MIKR-SAA-0078-01.1, BIO-ING-MIKR-SAA-0066-00.1, BIO-ING-MIKR-SAA-0045-01.2, BIO-ING-MIKR-SAA-0046-00, BIO-ING-MIKR-SAA-0033-00.1, BIO-ING-MIKR-SAA-0067-00.2, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-02.1, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00
Mykobakterien	Reinkulturen	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit	BIO-ING-MIKR-SAA-0006-01 BIO-ING-MIKR-SAA-0007-01
Mykobakterien	Kulturmateriale aus Flüssig- und Festkulturen (Primärkulturen und Isolate) angezüchtet aus Materialien wie Sputum, Bronchialsekret, Bronchial-Lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit	BIO-ING-MIKR-SAA-0010-00.1 BIO-ING-MIKR-FB-0082-00
Hefen und Hefeähnliche Pilze	Kulturmateriale angezüchtet aus Untersuchungsmaterialien wie bei Kulturverfahren (Primärkulturen und Isolate)	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferenzierung (MALDI-TOF-MS)	BIO-ING-MIKR-SAA-0085-00 BIO-ING-MIKR-VAW-0056-00 BIO-ING-MIKR-FB-0062-00
Dermatophyten und andere Erreger von Dermatomykosen	Kulturmateriale angezüchtet aus Untersuchungsmaterialien wie bei Kulturverfahren (Primärkulturen und Isolate)	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferenzierung (MALDI-TOF-MS)	BIO-ING-MIKR-SAA-0016-00, BIO-ING-MIKR-VAW-0010-00 BIO-ING-MIKR-SAA-0092-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Schimmelpilze	Kulturmaterial angezüchtet aus Untersuchungsmaterialien wie bei Kulturverfahren (Primärkulturen und Isolate)	biochemisch-aufwändig (Bunte Reihe, Mehrkammerverfahren), einfach, orientierend, massenspektrometrische Erregerdifferenzierung (MALDI-TOF-MS)	BIO-ING-MIKR-VAW-0010-00
Schimmelpilze wie Zygomyceten, z.B. Absidia, Mucor, Rhizomucor etc.; Hyalohyphomyceten, z.B. Aspergillus, Penicillium etc.; Phaeohyphomyceten, z.B. Exophiala, Cladophialophora, Alternaria	Kulturmaterial angezüchtet aus Untersuchungsmaterialien wie bei Kulturverfahren (Primärkulturen und Isolate)	biochemisch einfach, orientierend	BIO-ING-MIKR-VAW-0010-00
Schimmelpilze z.B. wie Zygomyceten	Kulturmaterial angezüchtet aus Untersuchungsmaterialien wie bei Kulturverfahren (Primärkulturen und Isolate)	biochemisch einfach, orientierend	BIO-ING-MIKR-SAA-0017-00

**Untersuchungsart:**

**Kulturelle Untersuchungen\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bakterien und Pilze	Alle geeigneten Materialien, z.B. Blut, ggf. nach Aufbereitung (z.B. Serum, Plasma), Urin, Stuhl, extravasale Körperflüssigkeiten (z.B. Sputum, Liquor, Aszites, Punktionsflüssigkeiten, Ergüsse, Lavagen), Magensaft, Galle, insbesondere von sterilen Materialien	Anreicherungsverfahren, Blutkulturverfahren, teilmechanisiert und vollmechanisiert, Keimzahlbestimmung, Kontakt-/Abklatschverfahren, spezifisch (selektiv), unspezifisch (nicht selektiv)	BIO-ING-MIKR-FB-0123-00, BIO-ING-MIKR-VAW-0049-00.1, BIO-ING-MIKR-VAW-0052-00.1, BIO-ING-MIKR-VAW-0053-00.1, BIO-ING-MIKR-VAW-0054-00.1, BIO-ING-MIKR-VAW-0051-00, BIO-ING-MIKR-VAW-0017-00.2, BIO-ING-MIKR-SAA-0031-01, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.2, BIO-ING-MIKR-SAA-0038-01.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.1, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.2, BIO-ING-MIKR-SAA-0069-00.2, BIO-ING-MIKR-SAA-0077-00.1, BIO-ING-MIKR-SAA-0042-00.1, BIO-ING-MIKR-SAA-0076-01, BIO-ING-MIKR-SAA-0041-01, BIO-ING-MIKR-SAA-0049-00.1, BIO-ING-MIKR-SAA-0070-00.1, BIO-ING-MIKR-SAA-0044-00.3, BIO-ING-MIKR-SAA-0066-00.1, BIO-ING-MIKR-SAA-0045-01.2, BIO-ING-MIKR-SAA-0046-00, BIO-ING-MIKR-SAA-0033-00.1, BIO-ING-MIKR-SAA-0067-00.2, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0048-01, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-02.1, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00, BIO-ING-MIKR-SAA-0036-00.2, BIO-ING-MIKR-SAA-0043-00.3, BIO-ING-MIKR-SAA-0034-00.5, BIO-ING-MIKR-SAA-0050-00.2

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Helicobacter pylori	Magenbiopsien	Kultur, selektiv	BIO-ING-MIKR-SAA-0078-01.1
Mykobakterien (M. tuberculosis-Komplex, nicht-tuberkulöse Mykobakterien, NTM)	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Anreicherungsverfahren, bei verschiedenen Temperaturen, spezifisch (selektiv), unspezifisch (nicht selektiv)	BIO-ING-MIKR-SAA-0086-00.2
Hefen und hefeähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	in aerober Atmosphäre, spezifisch (selektiv), unspezifisch (nicht selektiv)	BIO-ING-MIKR-VAW-0056-00
Dermatophyten und andere Erreger von Dermatomykosen	Hautgeschabsel, Haare, Nagelmateriale, Abstriche Hautläsionen, Pus, Biopsien	in aerober Atmosphäre, spezifisch (selektiv),	BIO-ING-MIKR-SAA-0016-00 BIO-ING-MIKR-VAW-0010-00 BIO-ING-MIKR-SAA-0092-00
Schimmelpilze (z.B. Zygomyceten, Hyalohyphomyceten, Phaeohyphomyceten)	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	in aerober Atmosphäre, spezifisch (selektiv),	BIO-ING-MIKR-VAW-0010-00

#### Untersuchungsart:

#### Ligandenassays\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Borrelia, IgG oder IgM	Serum	CLIA	BIO-ING-AUTO-SAA-0121-00
Campylobacter, Antigen	Stuhl	CLIA	BIO-ING-MIKR-SAA-0002-00.1
Clostridium difficile, GLDH-Antigen	Stuhl	CLIA	BIO-ING-MIKR-SAA-0003-01.1
Clostridium difficile, Toxin A und B, Antigen	Stuhl	CLIA	BIO-ING-MIKR-SAA-0004-01
Treponema pallidum, Gesamt-Antikörper	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0202-00
Mycoplasma pneumoniae, IgG oder IgM	Serum	CLIA	BIO-ING-AUTO-SAA-0126-00
Toxoplasma gondii, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0207-00
Aspergillus, Antigen	Serum	ELISA	BIO-ING-SERO-SAA-0039-01
Bordetella pertussis, Toxin, IgG oder IgA	Serum	EIA	BIO-ING-SERO-SAA-0048-02
Borrelia, IgG oder IgM, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Echinococcus multilocularis, Antikörper	Serum	ELISA	BIO-ING-SERO-SAA-0008-01
Helicobacter pylori, Antigen	Stuhl	CLIA	BIO-ING-MIKR-SAA-0028-01
Helicobacter pylori, IgA oder IgG	Serum	ELISA	BIO-ING-SERO-SAA-0016-01.1
Plasmodium ssp.-Antikörper (Malaria)	Serum	ELISA	BIO-ING-IMMU-SAA-0069-01
Mycobacterium tuberculosis, Interferon-Gamma-Nachweis nach Lymphozytenstimulation	Heparin-Blut in spezialbeschichteten Röhrchen	CLIA	BIO-ING-AUTO-SAA-0188-00.1
Legionella pneumophila Serotyp 1, Antigen	Urin	CLIA	BIO-ING-MIKR-SAA-0029-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Mycobacterium-tuberculosis-Komplex, MPT64-Antigen	Kulturmaterial aus Flüssigkulturen	ELISA	BIO-ING-MIKR-SAA-0010-00.1
Mycoplasma pneumoniae, IgA	Serum	ELISA	BIO-ING-SERO-SAA-0068-03
Streptococcus pneumoniae, Antikörper	Serum	ELISA	BIO-ING-SERO-SAA-0069-01
Salmonella-IgA Ak	Serum	ELISA	BIO-ING-SERO-SAA-0071-02
Salmonella- Ak (polyvalent)	Serum	ELISA	BIO-ING-SERO-SAA-0071-02
Treponema pallidum, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Borrelien, IgG oder IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0049-01.1
Helicobacter, IgA oder IgG	Serum	Immunoblot	BIO-ING-SERO-SAA-0015-02
Treponema pallidum, IgG oder IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0029-02
Yersinien, IgA oder IgG	Serum	Immunoblot	BIO-ING-SERO-SAA-0034-02

#### Untersuchungsart:

##### Mikroskopie\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bakterien und Pilze	Alle geeigneten, z.B. Blut, ggf. nach Aufbereitung (z. B. Serum, Plasma), Urin, Stuhl, extravasale Körperflüssigkeiten (z.B. Sputum, Liquor, Aszites, Punktionsflüssigkeiten, Ergüsse, Lavagen), Säfte (Magensaft, Galle)	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen ohne Anfärbung Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen	BIO-ING-MIKR-SAA-0060-00.3
Bartonella henselae, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0045-01
Bartonella quintana, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0045-01
Cestoden, z.B. Taenia, Diphylobothrium, Hymenolepis, Dipylidium	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0019-00
Cryptococcus neoformans	Liquor	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0012-00
Cryptosporidien	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0021-00



Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Dermatophyten	Hautgeschabsel, Haare, Nagelmaterial, Abstriche Hautläsionen, Pus, Biopsien	Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen	BIO-ING-MIKR-SAA-0018-01
Dermatophyten, z.B. Trichophyton, Microsporum, Epidermophyton und andere Erreger von Dermatomykosen, z.B. Scopulariopsis	Kulturmaterial aus Kulturen aus Hautgeschabsel, Haare, Nagelmaterial, Abstriche Hautläsionen, Pus, Biopsien	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-FB-0246-00.1 BIO-ING-MIKR-FB-0247-00.1 BIO-ING-MIKR-LIS-0003-00
Enterobius vermicularis	Klebestreifenpräparate	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen, ohne Anfärbung	BIO-ING-MIKR-LIS-0004-00.1
Hefen und Hefe- ähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen	BIO-ING-MIKR-SAA-0018-01
Hefen und Hefe-ähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0060-00.3
Hefen und Hefe-ähnliche Pilze (z.B. Candida albicans, Candida non- albicans, Klyveromyces, Pichia, Saccharomyces spp., Geotrichum, Trichosporon)	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-MIKR-LIS-0004-00.1
Legionella, non-pneumophila- Spezies, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0067-02.1
Legionella pneumophila, Serogruppen 1-6, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0067-02.1
Legionella pneumophila, Serogruppen 7-14, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0067-02.1
Malaria-Erregernachweis	EDTA-Blut	Hellfeldmikroskopie mit Anfärbung	BIO-ING-HÄMA-SAA-0027-00.3
Mykobakterien	Sputum, Bronchialsekret, Bronchial- lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-MIKR-SAA-0007-01
Mykobakterien	Sputum, Bronchialsekret, Bronchial- lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0006-01
Nematoden, z.B. Enterobius vermicularis, Trichuris, Ascaris, Hakenwürmer, Strongyloides	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0019-00
Protozoen	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0019-00
Protozoen (z.B. Giardia lamblia, Entamoeba histolytica/dispar, Entamoeba spp., sonstige Amoeben, Blastocystis hominis)	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-FB-0336-00
Rickettsia typhi, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0027-01.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Rickettsia rickettsii, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0027-01.1
Schimmelpilze	Bronchioalveoläre Lavage, Sputum, Liquor, sterile Körperflüssigkeiten, Ohrabstrich	Fluoreszenzmikroskopie	BIO-ING-MIKR-SAA-0018-01
Schimmelpilze wie Zygomyceten, z.B. Absidia, Mucor, Rhizomucor etc.; Hyalohyphomyceten, z.B. Aspergillus, Penicillium etc.; Phaeohyphomyceten, z.B. Exophiala, Cladophialophora, Alternaria	Kulturmateriel aus Kulturen des Untersuchungsmaterials (Bronchioalveoläre Lavage, Sputum, Liquor, sterile Körperflüssigkeiten, Ohrabstrich)	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-VAW-0010-00
Sporozoen/Coccidien (z.B. Cryptosporidien, Cyclospora, Isospora belli, Sarcocystis)	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0020-00
Trematoden, z.B. Schistosoma	Urin	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-MIKR-SAA-0091-00
Trematoden, z.B. Schistosoma, Clonorchis, Ophistorchis, Fasciola, Paragonimus	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0019-00
Würmer, Wurmteile, Ektoparasiten	Stuhl, ausgeschiedene Parasiten	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-MIKR-FB-0336-00

#### Untersuchungsart:

#### Molekularbiologische Untersuchungen (Amplifikationsverfahren)\*\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bordetellen, DNA	Abstrich, bronchoalveoläre Lavage	Real-time PCR	BIO-ING-MOLE-SAA-0056-00.4
Chlamydia pneumoniae, DNA	Abstrich, Sputum, Sekret	Real-time PCR	BIO-ING-MOLE-SAA-0006-00.5
Chlamydia trachomatis, DNA	Abstrich, Urin, Ejakulat, Punktat	Real-time PCR	BIO-ING-MOLE-SAA-0052-00.4
Chlamydia trachomatis L-Serovar, DNA	Abstrich, Urin	Real-time PCR	BIO-ING-MOLE-SAA-0007-01.3
Mycobacterium-tuberculosis- Komplex, DNA	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Polymerasekettenreaktion	BIO-ING-MIKR-SAA-0009-01
Mycobacterium-tuberculosis- Komplex, DNA	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl, Kulturmateriel	Empfindlichkeitsprüfung als molekularbiologische Nachweise	BIO-ING-MIKR-SAA-0095-00
Mycobacterium-tuberculosis- Komplex, DNA	Kulturmateriel aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	PCR mit nachfolgender fest-flüssig Hybridisierung (Festphasen- gebundene Fangsonden und Enzymimmunoassay basierende Detektion der Hybridisierungsereignisse)	BIO-ING-MIKR-SAA-0010-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Mykobakterien, nicht-tuberkulös (NTM), DNA	Kulturmaterial aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	PCR mit nachfolgender fest-flüssig Hybridisierung (Festphasen-gebundene Fangsonden und Enzymimmunoassay basierende Detektion der Hybridisierungsereignisse)	BIO-ING-MIKR-SAA-0010-00.1
Mykobakterien, nicht tuberkulös (NTM), DNA	Kulturmaterial aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	DNA-Sequenzierung, Kapillarelektrophorese	BIO-ING-MOLE-SAA-0002-00.6
Mycoplasma genitalium, DNA	Abstrich, Urin	Real-time PCR	BIO-ING-MOLE-SAA-0052-00.4
Mycoplasma genitalium: Genotypische Makrolid-Resistenz	Urin, Abstrich	Sequenzierung	BIO-ING-MOLE-SAA-0065-00.5
Mycoplasma genitalium: Genotypische Fluochinolon-Resistenz	Urin, Abstrich	Sequenzierung	BIO-ING-MOLE-SAA-0065-00.5
Mycoplasma pneumoniae, DNA	Abstrich, bronchoalveoläre Lavage, Sputum, Sekret	Real-time PCR	BIO-ING-MOLE-SAA-0039-00.6
Neisseria gonorrhoeae, DNA	Urin, Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0052-00.4
Pneumocystis jirovecii, DNA	BAL, Sputum, Trachealsekret, Abstriche	Real-time PCR	BIO-ING-MOLE-SAA-0060-00.4
Streptococcus pneumoniae, Legionella pneumophila, Haemophilus influenzae, Bordetellen, Mycoplasma pneumoniae, Chlamydia pneumoniae, DNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0003-00.6
Staphylococcus aureus, Methicillin-resistent (MRSA), DNA	Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0037-00.3
PVL-Staphylococcus aureus, Methicillin-resistent (MRSA), DNA	Bakterielle Reinkulturen	Real-time PCR	BIO-ING-MOLE-SAA-0089-00.1
Chlamydia trachomatis (CT), Neisseria gonorrhoeae (NG), Mycoplasma genitalium (MG), Mycoplasma hominis (MH), Trichomonas vaginalis (TV), Ureaplasma urealyticum (UU), Ureaplasma parvum (UP), DNA	Urin, Endozervikal- und Urethral-Abstriche	Real-time PCR	BIO-ING-MOLE-SAA-0049-00.2
16-S-rRNA-Gensequenzidentifizierung von Bakterien	Bakterielle Reinkulturen	Real-time PCR und Sequenzierung	BIO-ING-MOLE-SAA-0002-00.5
Trichomonas vaginalis, DNA	Urin, Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0052-00.4

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Aeromonas spp, Campylobacter spp., Clostridium difficile toxin B, Clostridium difficile hypervirulent, E. coli enteroaggregativ (EAEC), E. coli enteropathogen (EPEC), E. coli enterotoxigenic (ETEC), E. coli Shiga toxin-prod. (STEC), E. coli 0157, Salmonella spp., Shigella spp. / Enteroinvasive E. coli (EIEC), Vibrio spp., Yersinia enterocolitica, Blastocystis hominis, Cryptosporidium spp., Cyclospora cayetanensis, Dientamoeba fragilis, Entamoeba histolytica, Giardia lamblia, DNA	Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0092-00.1

### Untersuchungsgebiet: Virologie

#### Untersuchungsart: Ligandenassays\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Cytomegalovirus, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0205-00.1
Cytomegalovirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Cytomegalovirus, IgG, Avidität	Serum	Immunoblot	BIO-ING-SERO-SAA-0004-02
Cytomegalovirus, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0004-02
Dengue-Virus, IgG oder IgM	Serum	ELISA	BIO-ING-SERO-SAA-0057-02
Epstein-Barr-Virus, EBNA1, IgG	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0123-00
Epstein-Barr-Virus, IgG oder IgM	Serum, Citrat-Plasma	CLIA	BIO-ING-AUTO-SAA-0123-00
Epstein-Barr-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Epstein-Barr-Virus, IgG, Avidität	Serum	Immunoblot	BIO-ING-SERO-SAA-0006-03
Frühsommer-Meningoenzephalitis-Virus, IgG oder IgM	Serum	ELISA	BIO-ING-SERO-SAA-0061-02.1
Frühsommer-Meningoenzephalitis-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Hantaviren, IgG oder IgM	Serum	ELISA	BIO-ING-SERO-SAA-0011-00.6
Hantaviren, IgG oder IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0013-01
Hepatitis-A-Virus, IgG oder IgM	Serum	ECLIA	BIO-ING-AUTO-SAA-0099-02, BIO-ING-AUTO-SAA-0100-01
Hepatitis-B-Virus, HBc-Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0102-01.1
Hepatitis-B-Virus, HBc-IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0101-01
Hepatitis-B-Virus, HBe-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0097-01.1
Hepatitis-B-Virus, HBe-Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0096-01.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Hepatitis-B-Virus, HBs-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0103-01.1
Hepatitis-B-Virus, HBs-Antikörper	Serum, EDTA-Plasma	ECLIA	BIO-ING-AUTO-SAA-0104-01.1
Hepatitis-C-Virus, Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0095-01.1
Hepatitis-C-Virus, IgG	Serum	Immunoblot	BIO-ING-SERO-SAA-0062-01
Hepatitis-D-Virus, Antikörper	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0180-00
Hepatitis-E-Virus, IgG und IgM	Serum	EIA	BIO-ING-SERO-SAA-0063-02.1
Hepatitis-E-Virus, IgG und IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0017-01
Herpes-Simplex-Virus Typ 1, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Herpes-Simplex-Virus, IgG oder IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0124-00
Humane Immundefizienz-Viren Typ 1/2, Antikörper	Serum	Immunoblot	BIO-ING-SERO-SAA-0065-02
Humane Immundefizienzviren Typ 1/2, Antikörper und p24-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0098-00.1
Humane T-Zell-Leukämie-Viren Typ 1/2, Antikörper	Serum	ELISA	BIO-ING-SERO-SAA-0066-03
Humane T-Zell-Leukämie-Viren Typ 1/2, Antikörper	Serum	Immunoblot	BIO-ING-SERO-SAA-0019-02.1
Masernvirus, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0127-00
Masernvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
EBV -Mononukleose Schnelltest	Serum	Lateralfluss- Immunoassay	BIO-ING-SERO-SAA-0023-002
Mumpsvirus, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0125-00
Mumpsvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Parvovirus B19, IgG oder IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0128-00.1
Parvovirus B19, IgG oder IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0044-00.3
Rötelnvirus, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0206-00
Rötelnvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2
Varicella-Zoster-Virus, IgA	Serum	ELISA	BIO-ING-SERO-SAA-0077-01
Varicella-Zoster-Virus, IgG oder IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0122-00.1
Varicella-Zoster-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.2

**Untersuchungsart:**

**Molekularbiologische Untersuchungen (Amplifikationsverfahren)\*\***

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
BK-Polyomavirus, DNA, Viruslast	EDTA-Blut, Urin, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3
Cytomegalovirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, Abstriche, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3
Enterovirus, RNA	Liquor, BAL, Tracheal- oder Bronchialsekret, Abstriche, Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0059-00.1
Epstein-Barr-Virus, DNA, Viruslast	EDTA-Blut, Liquor, Abstriche, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Hepatitis-B-Virus, DNA, Viruslast	EDTA-Blut, Serum, EDTA Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0051-00.4
Hepatitis-B-Virus, genotypische Resistenzbestimmung	EDTA-Blut	DNA-Sequenzierung, Kapillarelektrophorese	BIO-ING-MOLE-SAA-0050-00.3
Hepatitis-C-Virus, Genotypisierung	EDTA-Plasma, Serum	DNA-Sequenzierung, Kapillarelektrophorese	BIO-ING-MOLE-SAA-0048-00.6
Hepatitis-C-Virus, Genotypisierung	EDTA-Blut, Serum	Real-time PCR	BIO-ING-MOLE-SAA-0018-01.3
Hepatitis-C-Virus, RNA, qualitativ	Serum, EDTA-Blut, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0051-00.4
Hepatitis-C-Virus, RNA, Viruslast	Serum, EDTA-Blut, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0051-00.4
Herpes-Simplex-Virus Typen 1/2, DNA	EDTA-Blut, Liquor, Abstriche, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3
Humanes Immundefizienzvirus Typ 1, Env-Gen, Korezeptor-Tropismus	EDTA-Blut	DNA-Sequenzierung, Kapillarelektrophorese	BIO-ING-MOLE-SAA-0027-00.4
Humanes Immundefizienzvirus Typ 1, genotypische Resistenzbestimmung, Integrase	EDTA-Blut, EDTA-Plasma	RT-PCR, NGS Sequenzierung	BIO-ING-MOLE-SAA-0094.00.1
Humanes Immundefizienzvirus Typ 1, genotypische Resistenzbestimmung, Reverse Transkriptase und Protease	EDTA-Blut, EDTA-Plasma	RT-PCR, NGS Sequenzierung	BIO-ING-MOLE-SAA-0094.00.1
Humanes Immundefizienzvirus Typ 1, RNA, Viruslast	EDTA-Blut, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0051-00.4
Humanes Papilloma Virus, DNA	Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0052-00.4
Influenzaviren Typen A/B, RNA	Abstriche	Real-time PCR	BIO-ING-MOLE-SAA-0034-01.2 BIO-ING-MOLE-SAA-0057-00.2
JC-Polyomavirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, BAL	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3
Parainfluenzaviren RNA, Metapneumovirus RNA, Enteroviren RNA, Adenoviren DNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR	BIO-ING-MOLE-SAA-0003-00.6
Coronaviren, Bocavirus, Rhinoviren, RNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR	BIO-ING-MOLE-SAA-0003-00.6
Influenzaviren, Respiratorische Synzytialviren, RNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR	BIO-ING-MOLE-SAA-0003-00.6
SARS-CoV-2 (RNA)	Abstrich, Bal, Bronchialsekret, Sputum	Real-time PCR	BIO-ING-MOLE-SAA-0075-00.3 BIO-ING-MOLE-SAA-0076-00.1
Varicella-Zoster-Virus, DNA	Liquor, Abstriche, EDTA-Blut, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.3
SARS-CoV-2-RNA	Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0090-00
Adenovirus DNA, Astrovirus RNA, Norovirus RNA, Rotavirus RNA, Sapovirus RNA	Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0092-00.1

## Untersuchungsgebiet: Transfusionsmedizin

### Untersuchungsart: Agglutinationsteste\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
AB0-Blutgruppenbestimmung	EDTA-Blut, Vollblut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0022-06
Antikörper- Titer	Vollblut, EDTA-Blut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0026-02
Antikörper-Differenzierung	Vollblut, EDTA-Blut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0026-02
Antikörper-Suchtest	Vollblut, EDTA-Blut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0025-02.1
Coombs-Test, direkt	EDTA-Blut, Vollblut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0024-02
Coombs-Test, indirekt (EB)	EDTA-Blut, Vollblut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0025-02.1
Rhesusformel, inkl. Kell-Antigen	EDTA-Blut, Vollblut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0022-06