

Untersuchungen im Bereich:

Medizinische Laboratoriumsdiagnostik

Untersuchungsgebiete:

Klinische Chemie

Immunologie

Humangenetik (Molekulare Humangenetik)

Humangenetik (Zytogenetik)

Mikrobiologie

Virologie

Transfusionsmedizin

Untersuchungsgebiet: Klinische Chemie

Untersuchungsart:

Chromatographie (Flüssigkeitschromatographie (LC-MS/MS)) ^[Flex C]

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-0065-01.2
Amiodaron	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0105-00
Amisulprid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01
Amitriptylin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0107-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Atomoxetin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Benperidol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01
Benzodiazepine-Bestätigung	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Benzoyllecgonin	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
BDB	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Drogen-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
Bupropion	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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
Cannabinoide-Bestätigung (THC, 11-OH-THC, THC-COOH, CBD)	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0120-00.2
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-0106-01
Ciclosporin	EDTA-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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Desmethylozapin	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
Desmethyflunitrazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Desmethylnortazepam	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Desmethylertralin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
O-Desmethylnaloxon	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
Duloxetine	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-0061-01.1
Ethylsulfat	Urin, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0061-01.1
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
Felbamate	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
Fluoxetine	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Flupentixol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01
Flurazepam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Hydroxy-Bupropion	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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
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-0106-01
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
Melperon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01
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-0002-01 BIO-ING-CHROM-SAA-0115-00.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Methylphenidat	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Mianserin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Midazolam	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3
Milnacipran	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Mirtazapin	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0104-00
Moclobemid	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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
Nicotinamid	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0122-00
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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Opipramol	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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-0106-01
Phosphatidylethanol (PEth)	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0121-00.1
Pipamperon	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01
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
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-0106-01
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, Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3 BIO-ING-CHROM-SAA-0124-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Rufinamid	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0092-02
Serotonin	EDTA-Blut	LC-MS/MS	BIO-ING-CHROM-SAA-0117-01
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	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0087-00.4
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
TMAO (Trimethylammonium-N-Oxid)	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0119-00
Tianeptin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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
Tranylcypromin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Trazodon	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
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
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
Vortioxetin	Serum	LC-MS/MS	BIO-ING-CHROM-SAA-0124-00
Zaleplon	Urin	LC-MS/MS	BIO-ING-CHROM-SAA-0096-00.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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-0106-01
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
Zuclopenthixol	Serum, EDTA-Plasma	LC-MS/MS	BIO-ING-CHROM-SAA-0106-01

Untersuchungsart:

Chromatographie (Gaschromatographie (GC)) ^[Flex C]

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)) ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Vitamin B 1	EDTA-Blut	HPLC-FD	BIO-ING-CHROM-SAA-0069-02.2
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-0099-00.4
Vitamin K1	Serum	HPLC-FD	BIO-ING-CHROM-SAA-0088-01.3
Aminosäuren	Urin, Liquor	HPLC-UV/VIS	BIO-ING-SPCH-SAA-0007-01
Ampicillin	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
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-0090-02
Cefepim	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
Ceftazidim	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
Coenzym Q10	Serum, EDTA-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0026-01.1
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-0098-00.2

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Linezolid	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
Meropenem	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
Piperacillin	Serum	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0118-00
Vitamin A	Serum, EDTA-Plasma	UHPLC	BIO-ING-CHROM-SAA-0009-02.2
Vitamin C	Lithium-Heparin-Plasma	HPLC-UV/VIS	BIO-ING-CHROM-SAA-0077-01.2
Vitamin E	Serum, EDTA-Plasma	UHPLC	BIO-ING-CHROM-SAA-0009-02.2

Untersuchungsart:

Durchflusszytometrie (inkl. Partikeleigenschaftsbestimmungen) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Großes Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößen- bestimmung, elektronisch Bestimmung zytochemisch- zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.3
Kleines Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößen- bestimmung, elektronisch Bestimmung zytochemisch- zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.3
Retikulozyten	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößen- bestimmung, elektronisch Bestimmung zytochemisch- zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.3
Thrombozyten	EDTA-Blut, Spezialröhrchen Thromboexact	Partikelzählung, optisch-elektronisch Partikelgrößen- bestimmung, elektronisch Bestimmung zytochemisch- zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0021-03.3
Zellzahl	Körperflüssigkeiten wie Ascites, Dialysat, Liquor, Pleura-Punktat, Synovia	Partikelzählung, optisch-elektronisch Partikelgrößen- bestimmung, elektronisch Bestimmung zytochemisch- zytometrischer Merkmale	BIO-ING-HÄMA-SAA-0002-00.6

Untersuchungsart:

Elektrochemische Untersuchungen ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Calcium (ionisiert)	Serum	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
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 ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Eiweisselektrophorese	Serum	Kapillarelektrophorese	BIO-ING-IMMU-SAA-0050-04
Alkalische Phosphatase-Isoenzyme	Serum	Agarose-Elektrophorese	BIO-ING-SPCH-SAA-0024-01
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

Untersuchungsart:

Koagulometrie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
APC-Resistenz	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0010-01.1
Fibrinogen	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0023-02.2
Lupus Antikoagulans (Lupus Inhibitoren)	Citratplasma	optisches Detektionsverfahren	BIO-ING-HÄMA-SAA-0018-01.1
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 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 ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
17-Hydroxy(OH)-Progesteron	Serum	RIA	BIO-ING-RIA-SAA-0010-00.8
Adalimumab	Serum	EIA	BIO-ING-SPCH-SAA-0093-00
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-0024-00.7
Androstaniol Glucuronid	Serum	RIA	BIO-ING-RIA-SAA-0003-00.6

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Androstendion	Serum	ECLIA	BIO-ING-AUTO-SAA-0212-00.1
Anti-Müller-Hormon	Serum, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0146-01
Barbiturate	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
Benzodiazepine	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
Benzoyllecgonin	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
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-0070-01.5
Beta-Amyloid 1-42	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.5
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-0024-00.7
CA 50	Serum	RIA	BIO-ING-RIA-SAA-0009-00.7
Calcitonin	Serum, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0144-00.1
Cannabinoide	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
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-0221-00
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-0032-00.1
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-0024-00.7
EDDP	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
Erythropoetin (EPO)	Serum, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0150-00
Ethylglucuronid	Urin	ELISA	BIO-ING-CHROM-SAA-0057-02.4
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-0055-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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
Hämoglobin-Haptogl.-Komplex (immunol.)	Faeces	ELISA	BIO-ING-SPCH-SAA-0063-01
hCG/ β -hCG	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0056-00
Infliximab	Serum	EIA	BIO-ING-SPCH-SAA-0094-00
Inhibin A (Dimer)	Serum	ELISA	BIO-ING-SPCH-SAA-0050-00.3
Inhibin B	Serum	ELISA	BIO-ING-SPCH-SAA-0055-00
Insulin	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0058-00.1
Interleukin-2-Rezeptor, löslicher	Serum	ELISA	BIO-ING-RIA-SAA-0035-00
Leptin	Serum	RIA	BIO-ING-RIA-SAA-0028-00.4
Luteinisierendes Hormon (LH)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0059-00.2
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-0060-00.4
Opiate	Urin	CEDIA	BIO-ING-CHROM-SAA-0024-00.7
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-0017-00.8
Pankreas-Elastase	Serum	ELISA	BIO-ING-SPCH-SAA-0027-00.2
PAPP-A	Serum	ECLIA	BIO-ING-AUTO-SAA-0183-00.2
Parathormon, intakt (PTH)	EDTA-Plasma, Serum	ECLIA	BIO-ING-AUTO-SAA-0064-01
Parathormon-related Protein	EDTA-Plasma	RIA	BIO-ING-RIA-SAA-0004-00.10
Phenobarbital	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0196-00
Phenytoin (Diphenylhydantoin)	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0192-01
Phospho-Tau	Liquor	ELISA	BIO-ING-IMMU-SAA-0070-01.5
PIGF	Serum	ECLIA	BIO-ING-AUTO-SAA-0200-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Plazenta-AP	Serum	ELISA	BIO-ING-SPCH-SAA-0053-01.1
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	EDTA-Plasma	ELISA	BIO-ING-SPCH-SAA-0063-01
Prokollagen-III-Peptid	Serum	RIA	BIO-ING-RIA-SAA-0013-00.7
Prolactin	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0062-00
Protein S-100	Serum	ECLIA	BIO-ING-AUTO-SAA-0201-00.2
Renin	EDTA-Plasma	CLIA	BIO-ING-AUTO-SAA-0136-00
Sexual-Hormon-bindendes Globulin (SHBG)	Serum, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0065-01
sFlt-1	Serum	ECLIA	BIO-ING-AUTO-SAA-0199-00.1
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-0070-01.5
Testosteron	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0067-01
Testosteron, freies	Serum	ELISA	BIO-ING-SPCH-SAA-0063-01
Theophyllin	Serum, EDTA-, Heparin-Plasma	KIMS	BIO-ING-AUTO-SAA-0209-00
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-0086-01
Tissue polypeptide specific antigen (TPS)	Serum	RIA	BIO-ING-RIA-SAA-0024-00.8
Trijodthyronin, freies (FT3)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0085-01
Troponin T	Serum, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0132-00.4
TSH basal	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0081-01
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
Wachstumshormon (HGH)	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0219-00.1

Untersuchungsart:

Mikroskopie ^[Flex B]

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-0031-00.9
Synovialanalyse	Punktat	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-HÄMA-SAA-0031-00.9
Urinsediment	Urin	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-HÄMA-SAA-0029-01.4

Untersuchungsart:

Osmometrie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Osmolalität	Serum, Urin	Kryoskopie	BIO-ING-RIA-SAA-0007-00.10

Untersuchungsart:

Sedimentationsuntersuchungen ^[Flex A]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Blutkörperchensenkungs- geschwindigkeit	EDTA-Blut	kinetische Durchflussanalyse	BIO-ING-HÄMA-SAA-0026-00.3

Untersuchungsart:

Spektrometrie (UV-/VIS-Photometrie) ^[Flex C]

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-0015-01.2
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-0020-02.1
Anti-Faktor Xa Aktivität (Apixaban)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0020-02.1
Anti-Faktor Xa (LMW-Heparin)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0020-02.1
Antithrombin	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0010-01.1
Beta-Hydroxybuttersäure	Serum	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
C1-Esterase-Inhibitor (Aktivität)	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0015-01.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Calcium	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0019-00.1
Calcium	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Cholesterin, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0021-00.3
Cholinesterase (Pseudocholinesterase)	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0022-01
CK	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0023-01
CK-MB	Serum, EDTA-Plasma	Photometrie	BIO-ING-AUTO-SAA-0003-00.2
Carboxyhämoglobin (COHb)	Heparinblut, EDTA-Blut	Photometrie	BIO-ING-SPCH-SAA-0005-01.1
Creatinin	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Creatinin , enzymatisch	Serum, EDTA-, Heparin-Plasma, Urin	Photometrie	BIO-ING-AUTO-SAA-0024-01
Eisen	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0025-00
Eiweiß, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0027-00.2
Ethanol	Serum, Urin, EDTA-Plasma	Photometrie	BIO-ING-CHROM-SAA-0021-00.6
Fettsäuren, frei	Serum	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Fructosamin	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0026-00
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, Hämolysat, Urin	Photometrie	BIO-ING-AUTO-SAA-0030-01 BIO-ING-AUTO-SAA-0007-01
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-0032-00.2
Hämoglobin, frei	Heparin-Plasma, Serum	Photometrie	BIO-ING-SPCH-SAA-0003-01.2
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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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-0213-01
Magnesium	Serum, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0039-00
Magnesium	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Methämoglobin	EDTA-Blut	Photometrie	BIO-ING-SPCH-SAA-0005-01.1
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-0041-00.1
Phosphat, anorganisch	Urin	Photometrie	BIO-ING-SPCH-SAA-0021-01.3
Protein C-Aktivität	Citratplasma	Photometrie	BIO-ING-HÄMA-SAA-0010-01.1
Saure Phosphatase, gesamt	Serum	Photometrie	BIO-ING-AUTO-SAA-0012-00.1
Tobramycin	Serum, EDTA-, Citrat-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0115-00.1
Triglyceride	Serum, EDTA-, Heparin-Plasma	Photometrie	BIO-ING-AUTO-SAA-0042-00.2
Urinstatus	Urin	Photometrie	BIO-ING-HÄMA-SAA-0029-01.4
Zink	Serum, Heparin-Plasma	Photometrie	BIO-ING-SPCH-SAA-0021-01.3

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

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-0060-05.1
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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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/Immunturbidimetrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
CRP	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0004-01
CRP high sensitive	Serum, EDTA-, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0203-00.1
D-Dimer	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0039-00.2
Eiweiß	Urin, Liquor	Turbidimetrie	BIO-ING-SPCH-SAA-0021-01.3
freies Protein S-Antigen	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0010-01.1
HbA1C	EDTA-Blut	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0022-00.8
Transferrin	Serum, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0175-00
Transferrin Rezeptor, lösl. (sTfR)	Serum, Heparin-Plasma	Turbidimetrie	BIO-ING-AUTO-SAA-0013-01.1
VWF:Ag (Faktor VIII-assoziiertes Ag)	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0017-02.1
VWF Ac (Aktivität)	Citratplasma	Immunturbidimetrie	BIO-ING-HÄMA-SAA-0017-02.1

Untersuchungsart:

Spektrometrie (Atomabsorptionsspektrometrie (AAS)) ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Quecksilber	EDTA-Blut, Heparinblut, Urin	AAS	BIO-ING-SPCH-SAA-0049-00.5

Untersuchungsart:

Spektrometrie (Induktiv gekoppelte Plasma-Massenspektrometrie (ICP-MS)) ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Antimon	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2
Arsen	Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0085-00.2
Arsen-Spezies	Urin	ICP-MS	BIO-ING-SPCH-SAA-0090-00.1
Barium	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2
Blei	Urin, EDTA-Blut, Heparinblut	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0085-00.2
Bromid	Serum	ICP-MS	BIO-ING-SPCH-SAA-0089-00.2
Cadmium	Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0085-00.2

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Chrom	EDTA-, Heparin-Plasma, Urin	ICP-MS	BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Jod	Urin, Serum	ICP-MS	BIO-ING-SPCH-SAA-0086-00.3
Kobalt	Serum, Urin, EDTA-, Heparin-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Kupfer	Urin, Heparinblut	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2 BIO-ING-SPCH-SAA-0091-00
Mangan	EDTA-, Heparin-Plasma, Urin, Serum, Heparinblut, EDTA-Blut	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Molybdän	Urin, EDTA-, Heparin-Plasma	ICP-MS	BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Nickel	Urin, EDTA-Blut, Heparinblut Heparin-, EDTA-Plasma	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Selen	Urin, EDTA-Blut, Heparinblut, EDTA-Plasma, Heparinplasma, Serum	ICP-MS	BIO-ING-SPCH-SAA-0064-01.2 BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Thallium	Serum, Urin	ICP-MS	BIO-ING-SPCH-SAA-0081-00.2 BIO-ING-SPCH-SAA-0085-00.2
Vanadium	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2
Zink	Urin, Heparinblut	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2 BIO-ING-SPCH-SAA-0091-00
Zinn	Urin	ICP-MS	BIO-ING-SPCH-SAA-0085-00.2

Untersuchungsgebiet: Immunologie

Untersuchungsart:

Durchflusszytometrie ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Lymphozytendifferenzierung	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung	BIO-ING-HÄMA-SAA-0033-00.4
Zelluläre Allergiediagnostik	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung (FLOW CAST)	BIO-ING-HÄMA-SAA-0032-00.1 BIO-ING-HÄMA-VAW-0004-01.2
Lymphozytendifferenzierung und Aktivierungsmarker	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Bestimmung des Anteils TCR-γδ-positiver T-Zellen im peripheren Blut	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Bestimmung der Leichtketten-Expression auf B-Zellen oder Plasmazellen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Immunphänotypisierung bei Non-Hodgkin-Lymphomen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Immunphänotypisierung bei chronisch-myeloischen Erkrankungen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Immunphänotypisierung bei MDS und akuten Leukämien	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2
Immunphänotypisierung bei PNH	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen	BIO-ING-HÄMA-SAA-0024-02.2

Untersuchungsart:

Elektrophorese ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Paraprotein, monoklonale Gammopathie	Serum, Urin	Immunfixationselektrophorese	BIO-ING-IMMU-SAA-0066-04.1
Oligoklonale Banden	Serum, Liquor	Isoelektrische Fokussierung	BIO-ING-IMMU-SAA-0054-02.6

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Acetylcholin-Rezeptor-Ak	Serum	Radiorezeptorassay	BIO-ING-RIA-SAA-0002-00.9
Adalimumab-Ak	Serum	EIA	BIO-ING-SPCH-SAA-0095-00
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 (CIC-C1q)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0045-02.3
Beta-2-Glycoprotein-Ak	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0034-01.2
Cardiolipin-Ak	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0031-02.3
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-0032-01.2
Einzelstrang-DNA (ssDNA)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0043-03.3
Gangliosid-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0052-03.1
Glutamat-Decarboxylase (GAD65)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0033-03.1
Histone-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0039-02.3
IA2-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0051-02.3
Immunglobulin E	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0057-00.1
Infliximab-Ak	Serum	EIA	BIO-ING-SPCH-SAA-0096-00
Intrinsic-Faktor-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0042-02.4
Insulin-Ak gegen Humaninsulin (IgG)	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0019-01.5
Leber-Nieren-Mikrosomen (LKM 1)-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0062-04
MCV-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0053-02.3
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-0031-02.3
Mitochondriale-Ak, Subtyp 1 (IgM)	Serum, Citrat-Plasma	ELISA	BIO-ING-IMMU-SAA-0031-02.3
Mitochondriale-Ak, Subtyp 2	Serum	ELISA	BIO-ING-IMMU-SAA-0040-02.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Muskelspezif. Rezeptor-Tyrosinkinase-Ak	Serum	Radioimmunpräzipitation	BIO-ING-RIA-SAA-0001-00.5
MAG-Ak (Myelin-assoziiertes-Glycoprotein) (IgM)	Serum	ELISA	BIO-ING-IMMU-SAA-0080-00.1
Myeloperoxidase (MPO)-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0037-01.3
Parietalzellen-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0010-04.2
PF4-Heparin-Ak	Serum	Lateral-Flow-Assay	BIO-ING-IMMU-SAA-0047-03.3
PMSCL-Ak	Serum	ELISA	BIO-ING-IMMU-SAA-0035-01.4
Proteinase 3-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0038-02.3
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-0059-06
Mitochondriale-Ak, Subtyp 4	Serum	Immunoblot	BIO-ING-IMMU-SAA-0014-01.5
Mitochondriale-Ak, Subtyp 9	Serum	Immunoblot	BIO-ING-IMMU-SAA-0014-01.5
Zentromer-Ak (IgG)	Serum	ELISA	BIO-ING-IMMU-SAA-0075-00.2
Zinktransporter-8-Ak (ZnT8)	Serum	ELISA	BIO-ING-IMMU-SAA-0073-00.2

Untersuchungsart:

Mikroskopie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
ANCA (Granulozytenzytoplasma-Ak)	Serum	IFT	BIO-ING-IMMU-SAA-0016-04.3
Antinukleäre-Ak (inkl. Subtypen)	Serum	IFT	BIO-ING-IMMU-SAA-0011-03
Aquaporin 4-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0003-02.5
Basalmembran-Ak (tubulär)	Serum	IFT	BIO-ING-IMMU-SAA-0015-02
Doppelstrang-DNA (dsDNA)-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0078-00.2
DPPX-Ak (Anti-Dipeptidyl aminopeptidase-like protein 6)	Serum, Liquor	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-0005-04
Glutamat-Rezeptor (TypNMDA)-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0006-04.1
VGKC-Komplex (Typ CASPR2/Lgi1)-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0067-02.5
Inselzell-Ak (ICA)	Serum	IFT	BIO-ING-IMMU-SAA-0017-03
Mitochondriale-Ak (AMA)	Serum	IFT	BIO-ING-IMMU-SAA-0005-04
MOG-Ak (Myelin-Oligodendrozyten-Glykoprotein)	Serum	IFT	BIO-ING-IMMU-SAA-0071-00.4

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Nebennierenrinden-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0001-04
Neuronale Auto-Ak	Serum, Liquor	IFT	BIO-ING-IMMU-SAA-0013-02
Phospholipase-A ₂ -Rezeptor (PLA ₂ R)-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0007-04.2
Skelettmuskel-Ak	Serum	IFT	BIO-ING-IMMU-SAA-0021-01.5
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-0007-04.2

Untersuchungsart:

Spektrometrie (UV-/VIS-Photometrie) ^[Flex B]

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

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

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-0060-05.1
Immunglobulin D	Serum	Nephelometrie	BIO-ING-IMMU-SAA-0079-02.2
Immunglobulin G	Serum, Liquor	Nephelometrie	BIO-ING-IMMU-SAA-0060-05.1 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-0060-05.1

Untersuchungsart:

Spektrometrie (Turbidimetrie) ^[Flex B]

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 ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Achromatopsie (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Acne inversa, familiäre (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Acrocallosal-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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adenomatöse Polyposis (APC, MUTYH, NTHL1, POLD1, POLE, RNF43)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Adipositas (ADCY3, ALMS1, APC2, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CARTPT, CCDC28B, CEP164, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, GNAS, IFT172, IFT27, IFT74, KDM6A, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, NFIX, NR0B2, NSD1, NTRK2, PCSK1, PHF6, POMC, PPARG, PTEN, PYY, RAB23, SDCCAG8, SH2B1, SIM1, TMEM67, TRIM32, TTC8, UCP3, VPS13B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Adrenogenitales Syndrom (CYP21A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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 BIO-ING-HUM-SAA-0146-04.1
Ahornsirupkrankheit (BCKDHA, BCKDHB, BCKDK, DBT, PPM1K)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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-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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Albinismus, okulokutaner (DCT, GPR143, LRMDA, MC1R, MITF, MYO5A, OCA2, PMEL, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Aldosteronismus, primärer (ARMCS, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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
Alpha-Thalassämie (HBA1, HBA2 (alpha-Globingen-Cluster))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Amyloidose, hereditäre (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Amyotrophe Lateralsklerose (ALS2, ANG, ANXA11, C9orf72, CCNF, CHCHD10, CHGB, CHMP2B, DAO, DCTN1, FIG4, FUS, HNRNPA1, MATR3, NEFH, NEK1, OPTN, PFN1, SETX, SIGMAR1, SOD1, SQSTM1, TARDBP, TIA1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alström-Syndrom (ALMS1)	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
Altersabhängige Makuladegeneration-ähnlich/Drusen (ABCA4, ABCC6, BEST1, C1QTNF5, CDHR1, CFH, CHM, EFEMP1, ELOVL4, IMPG1, IMPG2, MFRP, PRDM13, PRPH2, RDH5, TIMP3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Alzheimer-Krankheit (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Anämie, kongenitale dyserythroetische (CDAN1, CDIN1, GATA1, KIF23, KLF1, RACGAP1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Angelman-ähnliche Syndrome (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Anämie, Aplastische (ASXL1, BCOR, BCORL1, DNMT3A, PIGA)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Anämie, hämolytische (ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, ATP11C, CD59, CDAN1, COL4A1, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GPX1, GSR, GSS, HK1, KCNN4, KIF23, KLF1, LPIN2, NT5C3A, PFKL, PFKM, PGK1, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1, SLC4A1, SPTA1, SPTB, TPI1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Aortopathie / Aortenaneurysma (ACTA2, ADAMTS10, BGN, COL11A1, COL12A1, 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, TGFBRI1, TGFBRI2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Arthrogrypose (ACTA1, ADGRG6, ALG3, BIN1, CHRNA1, CHRND, CHRNA1, CHST14, CNTN1, COL6A1, COL6A2, COL6A3, CRLF1, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKTN, GBA1, GBE1, GLDN, GLE1, IBA57, LGI4, MEGF10, MPZ, MTM1, MYBPC1, MYH2, MYH3, MYH8, NALCN, NEB, PIEZO2, PIP5K1C, PLOD2, PMM2, RAPSIN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Angelman-Syndrom (SNRPN-Genregion (15q11-q13); UBE3A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MS-MLPA	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-0146-04.1
Ataxie, episodische (ATP1A2, CACNA1A, CACNB4, KCNA1, OTC, PDHA1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ataxie, hereditäre (ABCB7, ABHD12, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CAMTA1, CAPN1, CCDC88C, CLCN2, CLCN6, COQ8A, CWF19L1, DNMT1, ELOVL4, ELOVL5, FAT2, FGF14, FXN, GDAP2, GRID2, GRM1, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KIF1C, MARS2, MME, MRE11, MTPAP, NKX6-2, NPTX1, PDYN, PI4KA, PMPKA, PNKP, PNPLA6, PRKCG, PRNP, RNF216, SACS, SCN2A, SCYL1, SETX, SLC1A3, SNX14, SPG7, SPTBN2, STUB1, SYNE1, TGM6, TMEM240, TPP1, TTBK2, TTPA, TUBA1A, TUBB4A, TWNK, VAMP1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Autoimmunerkrankungen, polyendokrine (AIRE, CTLA4, FOXP3, IL2RA, ITCH, JAK1, LRBA, STAT1, STAT3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Azidämie, organische (ACAD8, ACADSB, ACAT1, ACSF3, ALDH6A1, ASPA, AUH, BCKDHA, BCKDHB, BTD, 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, TFAFAZZIN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Atypisches hämolytisch-urämisches Syndrom (ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, PIGT, THBD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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, CEP164, CEP19, CEP290, CFAP418, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bartter- und Gitelman-Syndrom (AP2S1, BSND, CASR, CFTR, CLCNKA, CLCNKB, CLDN10, CTNS, GNA11, KCNJ1, MAGED2, MT-TF, MT-TI, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Benigne infantile 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Beckwith-Wiedemann-Syndrom (11p15.5; CDKN1C)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Beta-Thalassämie (HBB (beta- Globingen-Cluster))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Biotinidase-Mangel (BTD)	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
Biotin-responsive Basalganglienstörung (SLC19A3)	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
Birt-Hogg-Dube-Syndrom (FLCN)	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
Blepharophimose-Ptois-Epikanthus inversus-Syndrom (FOXL2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Bloom-Syndrom (BLM/RECQL3; BLM)	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
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Brust-/Eierstockkrebs, familiärer (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Burkitt-Lymphom (ARID1A, BACH2, BCL6, CCND3, DDX3X, DNMT1, FBXO11, ID3, IGLL5, MYC, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cholestase, progressive familiäre intrahepatische (ABCB11, ABCB4, ABCC2, ABCD3, ACOX2, AKR1D1, ATP8B1, DCDC2, HNF1B, HSD3B7, KIF12, LSR, MPV17, MYO5B, NR1H4, PPM1F, TJP2, USP53, VIPAS39, VPS33B, WDR83OS, ZFYVE19)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cerebrale autosomal dominante Arteriopathie mit subkortikalen Infarkten und Leukenzephalopathie (NOTCH3)	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
Cholestatische Hepatopathie (ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP7B, ATP8B1, CFTR, CYP7B1, DCDC2, HNF1B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
chronische Darmentzündung (ADA, ADAM17, AICDA, AIRE, ANKZF1, ARPC1B, BACH2, BTK, CARD8, CARMIL2, CASP8, CD19, CD3G, CD40, CD40LG, CD55, CD81, CHD7, CIITA, COL7A1, CR2, CTLA4, CYBA, CYBB, CYBC1, DCLRE1C, DKC1, DOCK8, DUOX2, FCHO1, FERMT1, FOXP3, FUT2, G6PC3, GUCY2C, HPS1, HPS4, HPS6, ICOS, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL21, IL23R, IL2RA, IL2RB, IL2RG, IL7R, ITCH, ITGB2, JAK1, LCK, LIG4, LRBA, LYST, MALT1, MEFV, MVK, MYO5A, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NLRC4, NLRP12, NOD2, NOP10, NPC1, PIK3CD, PIK3R1, PLCG2, POLA1, PRF1, PTEN, RAB27A, RAC1, RAC2, RAG1, RAG2, RET, RFX5, RFXANK, RFXAP, RIPK1, RIPK2, RTEL1, SH2D1A, Si, SLC37A4, SLC9A3, SLCO2A1, STAT1, STAT3, STAT5B, STIM1, STX3, STXBP2, STXBP3, TAP1, TAP2, TERC, TERT, TGFB1, TGFB1, TGFB2, TINF2, TNFAIP3, TNFRSF13B, TRAF3, TRIM22, TTC7A, UNC13D, UNG, WAS, XIAP, ZAP70, ZBTB24, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Chylomikronämie-Syndrom, familiäres (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cobalamin-Mangel (ABCD4, ACSF3, AMN, CBLIF, CBS, CD320, CUBN, HCFC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
chronische Eosinophilenleukämie (ASXL1, CBL, DNMT3A, EZH2, IDH1, IDH2, NF1, SETBP1, SF3B1, SH2B3, SRSF2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
CINCA-Syndrom (CIAS1/NLRP3) (NLRP3)	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
CM (FGF3, FOXI3, GREB1L)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Cohen-Syndrom (VPS13B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-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-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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
DICER1-assoziiertes Tumorsyndrom (DICER1)	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
DiGeorge-Syndrom (22q11.2)	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-0146-04.1 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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
Dravet-Syndrom (SCN1A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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, KYN1, LFNG, LMX1B, MEOX1, MESP2, MNX1, MYO18B, NKX3-2, OFD1, ORC1, ORC4, ORC6, PLCB4, POLR1A, POLR1C, POLR1D, RIPPLY2, SF3B4, SH3PXD2B, SNRPB, 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-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, SMARCA1, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dysplasien mit Verkürzung der langen Knochen (akro-meso-rhizome Dysplasien) (ADAMTS10, ADAMTS17, ADAMTS2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Dystonie, hereditäre (ADCY5, ANO3, AOEPE, ATP1A3, COL6A3, DNAJC12, ECHS1, EIF2AK2, FTL, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECP, NKX2-1, PLA2G6, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TSPOAP1, TUBB4A, VPS16)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ehlers-Danlos-Syndrom (ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Dystonie, Dopa-responsive (GCH1; TH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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, IKBKG, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Epilepsie und epileptische Enzephalopathie (AARS1, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, ARHGEF9, ARX, ATP6V1A, ATRX, BRAT1, BTBD, CACNA1A, CACNA1E, CACNA2D2, CACNB4, CDK19, CDKL5, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN4, CNNM2, CNPY3, CNTNAP2, CPA6, CYFIP2, DENND5A, DEPDC5, DNM1, DOCK7, EEF1A2, EHMT1, FOXG1, FRRS1L, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GABRG3, GATM, GLDC, GNAO1, GPHN, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HCN2, HNRNP, IQSEC2, ITPA, KCNA2, KCNB1, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, LGI1, MBD5, MDH2, MECP2, MEF2C, MOCS1, MOCS2, NEXMIF, NPRL2, NPRL3, NRXN1, PCDH19, PHF6, PIGA, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PURA, RHOBTB2, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SEMA6B, SETD1A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SPTAN1, STAMBP, STX1B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Erythrozytose, familiäre (BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
episodische Ataxie Typ 2 (CACNA1A)	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
Familiäre idiopathische 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Familiäre hemiplegische Migräne (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fanconi-Syndrom, renales (ATP6V0A4, ATP6V1B1, ATP7B, CLCN5, CTNS, EHHADH, FAH, G6PC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Familiäres atypisches multiples Muttermal- und Melanomsyndrom (ACD, BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERF2IP, TERT, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
familiäres Mittelmeefieber (MEFV)	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
Fiebersyndrome, periodische (ADA, ADA2, ADAM17, AP1S3, BACH2, CARD14, COPA, ELANE, FOXD3, HAX1, HSPA1L, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL36RN, LPIN2, LRBA, MEFV, MVK, NFAT5, NLR4, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Fokale Epilepsie (CHRNA2, CHRNA4, CHRN2, DEPD3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Galaktosämie (GALE, GALK1, GALM, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Generalisierte Epilepsie mit Fieberkrämpfen plus (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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
frühkindliche Netzhautdegeneration (RPE65)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
frühkindliche Schwerhörigkeit DFNA3 (GJB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Gliedergürtelmuskeldystrophie (ALG2, ANO5, 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, POPDC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glukokortikoid-Mangel, familiär (AIRE, CYP11A1, MC1R, MC2R, MCM4, MRAP, NNT, NROB1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glukoneogenese-Störung (FBP1, G6PC1, GK, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glucose-6-Phosphat-Dehydrogenase- Defizienz (G6PD)	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
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Glukosurie, renale (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
GLUT1-Defekt (SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Glutarazidurie und verwandte Erkrankungen (D2HGDH, ETFA, ETFB, ETFDH, FLAD1, GCDH, HADH, HMGCL, IDH1, IDH2, L2HGDH, OGDH, SLC25A1, SLC25A32, SUGCT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Glykosylierungs-Defekt Typ 2 (ATP6AP1, ATP6AP2, B4GALT1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, FCSK, FUT8, GANAB, MAN1B1, MAN2B2, MGAT2, MOGS, PRKCSH, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SLC9A7, ST3GAL3, TMEM165, VMA12, VMA21, VMA22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hamartomatösen Polyposis Syndromen (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hämochromatose und Eisenstoffwechselstörungen (BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC11A2, SLC40A1, STEAP3, TF, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Haarzell-Leukämie (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Harnstoffzyklus-Störung und 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, TFAZZIN, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Heterotaxie-Syndrome (ACVR2B, CCDC39, CCDC40, CCNO, CFAP298, CFAP52, CFAP53, CFC1, CIROP, CRELD1, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DRC2, DRC4, FOXH1, FOXJ1, GAS2L2, GDF1, HYDIN, LRRC56, MMP21, MNS1, NFATC1, NKX2-5, NME8, NODAL, NPHP4, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, PKD1L1, RSPH1, RSPH3, RSPH4A, RSPH9, SMAD2, SPAG1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hepatozelluläres Karzinom (ABCB11, AGL, APC, CDKN2A, G6PC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hereditäre Fruktoseintoleranz (ALDOB)	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
Hereditäre motorische und sensorische Neuropathie Typ 1A (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Hereditäre Neuropathie mit Neigung zu Drucklähmungen (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Hereditäre Pankreatitis (PRSS1; SPINK1; CFTR)	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
HLH (PRF1, STX11, STXP2, UNC13D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Holoprosenzephalie (CDON, CENPF, DHC7, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hornhautdystrophie (AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCM, EPYC, FOXE3, GRHL2, GSN, KERA, KRT12, KRT3, LOXHD1, LUM, MCOLN1, MIR184, OVOL2, PAX6, PIKFYVE, PRDM5, PRDX3, SLC4A11, TACSTD2, TCF4, TGFB1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyperekplexie (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-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-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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hyper-IgD-Syndrom (MVK)	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
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypertriglyceridämie (APOA5, APOC2, APOC3, APOE, CREB3L3, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Hypertonie, arterielle (ATP1A1, ATP2B3, CACNA1D, CACNA1H, CLCN2, CLCNKA, CLCNKB, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD11B2, HSD3B2, KCNJ1, KCNJ5, KLHL3, MAGED2, NR3C2, PDE3A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC12A1, SLC12A3, VHL, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (ANOS1, AXL, CHD7, DLG2, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypomagnesiämie (ATP1A1, BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, EGFR, FXD2, HNF1B, KCNA1, KCNJ10, RRAGD, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypothyreose (CDCA8, DUOX1, DUOX2, DUOXA2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Hypophysenhormon-Defizienz (ARNT2, CDON, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RBM28, RNPC3, ROBO1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Imerslund-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Insulinresistenz (AGPAT2, AKT1, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, FBN1, INSR, LIPE, LMNA, LMNB2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Jeune asphyxierende Thoraxdystrophie (CEP120, CFAP410, CILK1, DPH1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2L1, DYNLT2B, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, INTU, KIAA0586, KIAA0753, LBR, NEK1, TRAF3IP1, TTC21B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Joubert-Syndrom und Meckel-Gruber-Syndrom (AHL1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CEP55, CIMIP2B, CPLANE1, CSPP1, FAM149B1, GLI3, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RNU4ATAC, RPGRIP1L, SLC30A7, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM17, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie, arrhythmogene rechtsventrikuläre (CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, RYR2, TGFβ3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie, dilatative (ABCC9, ACADVL, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, C10orf71, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FBXO32, FHL1, FKTN, FLNC, GCOM1, GLA, HFE, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, MYZAP, NEBL, NEXN, NPPA, PKP2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, RRAGC, SCN5A, SDHA, SGCA, SGCB, SGCD, SOD2, TAFAZZIN, TBX20, TCAP, TMPO, TNNC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kardiomyopathie, hypertrophe (ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CACNA1C, CALR3, CAV3, CSRP3, FHL1, FHOD3, FLNC, FXN, GAA, GLA, HRAS, JPH2, KLF10, KLHL24, 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, RYYR2, SLC25A4, SOS1, SOS2, TAFAZZIN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kardiomyopathie, non-compaction (ACTC1, ACTN2, DSP, DTNA, FLNC, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, NNT, NSD1, PRDM16, RPS6KA3, RYYR2, SCN5A, TAFAZZIN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Katarakt (ABCA3, ABHD12, AGK, AKR1E2, AQP5, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, DNMBP, EPG5, EPHA2, EYA1, FAR1, FOXE3, FTL, FYCO1, GALK1, GALM, GALT, GCNT2, GJA3, GJA8, GNPAT, HSF4, HYCC1, JAM3, LEMD2, LIM2, LSS, MAF, MIP, MIR184, NHS, P3H2, PANK4, PAX6, PEX7, PITX3, PXDN, RAB3GAP1, RNLS, SIL1, SIPA1L3, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, TRPM3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Katecholaminerge polymorphe ventrikuläre Tachykardie (ANK2, CALM1, CALM2, CALM3, CASQ2, GNAI2, KCNJ2, RYYR2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Ketogenese-Störung (HMGCL, HMGCS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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, BCAT2, BCKDHA, BCKDHB, BCS1L, COQ9, DBT, DLD, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, G6PC1, GLYCTK, GSS, GYS2, HADHB, HIBADH, HIBCH, HMGCL, HSD17B10, IVD, MCCC1, MCCC2, MLYCD, MMAA, MMAB, MMADHC, MMUT, OXCT1, PCCA, PCCB, PYGL, SLC16A1, SLC37A4, SUCLA2, SUCLG1, TFAZZIN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kleinwuchs, endokriner (ACAN, FGFR3, GH1, GHR, GHRHR, GHRSR, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kleinwuchs, skelettaler (ACAN, BMP2, BMPR1B, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, FBN1, FGFR3, GDF5, GNAS, IHH, MATN3, NPR2, PAPSS2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kleinwuchs (ACAN, ANKRD11, ASXL1, ATR, ATRX, BLM, BMP2, BMPR1B, BRAF, CCDC8, CDKN1C, CDT1, CEP152, CHD7, COL10A1, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREBBP, CUL7, DHCR7, DVL1, DVL3, EP300, ERCC6, ERCC8, EVC, EVC2, FAM111A, FANCA, FANCC, FANCG, FBN1, FGD1, FGFR3, GDF5, GH1, GHR, GHRHR, GHSR, GLI2, GLI3, GNAS, HDAC8, HESX1, HMGA2, HRAS, IARS1, IGF1, IGF1R, IGF2, IGFALS, IHH, KDM6A, KMT2D, KRAS, LARP7, LHX3, LHX4, LIG4, LMNA, MAP2K1, MAP2K2, MATN3, NBN, NIPBL, NPR2, OBSL1, ORC1, ORC4, ORC6, OTX2, PAPSS2, PCNT, PIK3R1, PLAG1, POC1A, POU1F1, PRKAR1A, PROP1, PTH1R, PTHLH, PTPN11, RAD21, RAF1, RNU4ATAC, ROR2, RPS6KA3, SEMA3E, SHOX, SLC26A2, SMARCAL1, SMC1A, SMC3, SOS1, SOX3, SRCAP, STAT5B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kleinwuchs, syndromaler (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Knochenmarkdysfunktionssyndrom (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, RPS19, RPS24, RPS26, RPS7, RTEL1, SBDS, SLC37A4, SLX4, SRP72, TAFAZZIN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kongenitale Anomalien der Nieren und ableitenden Harnwege (ACE, AGT, AGTR1, ANOS1, ARHGEF6, BICC1, BMP4, BNC2, CDC5L, CHD1L, CHRM3, CHRNA3, COL4A1, CRKL, DSTYK, ETV4, EYA1, FANCB, FAT4, FGF20, FOXC1, FOXC2, FOXP1, FRAS1, FREM1, FREM2, GATA3, GDNF, GFRA1, GLIS3, GREB1L, GREM1, GRIP1, HNF1B, HOXA11, HPSE2, ITGA8, KIF14, LHX1, LIFR, LRIG2, MUC1, MYOCD, NPNT, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SALL4, SIX1, SIX2, SIX5, SLIT2, SOX17, SRGAP1, TBC1D1, TBX18, TBX6, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, VWA2, WNT4, WT1, ZMYM2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Klonale Hämatopoese von unbestimmtem Potential (ASXL1, BCOR, BCORL1, BRCC3, CBL, CTCF, DNMT3A, GNAS, GNB1, IDH1, IDH2, JAK2, KRAS, NRAS, PPM1D, PTPN11, SF3B1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Klonale Zytopenie unbestimmter Signifikanz (ASXL1, BCOR, BCORL1, BRCC3, CBL, CTCF, DNMT3A, GNAS, GNB1, IDH1, IDH2, JAK2, KRAS, NRAS, PPM1D, PTPN11, SF3B1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kranioektodermale Dysplasie (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Kolobom (ABCB6, ACTB, ACTG1, ALDH1A3, ALG3, ALX3, BCOR, BMP7, C12orf57, CC2D2A, CEP290, CHD7, CREBBP, CRIM1, CRYAA, DPYD, EYA1, FADD, FLNA, FOXE3, FZD5, GATA3, GDF3, GDF6, GZF1, HMGB3, HMX1, IGBP1, INPP5E, KCTD1, KIFBP, KMT2D, LCP1, LRP2, MAB21L2, MAF, MIR204, MITF, MKS1, NAA10, NRAS, OTX2, PAX2, PAX6, PBX1, PDE6D, PIGL, PITX2, POMT1, PORCN, PQBP1, PTCH1, PTCH2, PTPN11, PUF60, RAB3GAP1, RARA, RAX, RBP4, RPGRIP1L, SALL1, SALL2, SALL4, SEMA3E, SHH, SIX3, SIX6, SMCHD1, SMO, SOX2, SRD5A3, STRA6, TBX1, TBX22, TENM3, TFAP2A, TMEM216, TMEM237, TMEM67, TWF1, UBE3B, VSX2, WDR11, YAP1, ZEB2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolonkarzinom, hereditäres nicht polypöses (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kolorektales Karzinom (APC, BMPR1A, EPCAM, MBD4, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Kraniosynostosen (ALPL, ALX4, BMP4, CCBE1, CDC45, COLEC11, CYP26B1, EFN1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lebersche hereditäre Optikusneuropathie (DNAJC30, MCAT, MECP, MT-ND1, MT-ND4, MT-ND6, NDUFA12, NDUFA7, NDUFS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Lebersche kongenitale Amaurose (ACBD5, ADAMTS18, AIPL1, ALMS1, BBS2, BBS4, CABP4, CCT2, CEP290, CRB1, CRX, DHX38, DTHD1, GDF6, GPATCH11, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MPDZ, NMNAT1, OTX2, POC1B, RD3, RDH12, RPE65, RPGRI1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Krebserkrankung mit unbekanntem Primärtumor (ARID1A, ATM, BRAF, EGFR, JAK2, KRAS, PTEN, SMAD4, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukoencephalopathie (AARS1, ABCD1, ACOX1, ADAR, AIMP1, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, CSF1R, CTC1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, GALC, GFAP, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, HYCC1, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lesch-Nyhan-Syndrom (HPRT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Leukämie (BCR-ABL1), qualitativ und quantitativ (BCR-ABL1, t(9;22)(q34;q11))	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR; größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	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 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-SAA-0063-02.2

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Leukämie, akute myeloische (ANKRD26, ASXL1, BCOR, BCORL1, BRAF, CBL, CCND3, CEBPA, CSF3R, DDX41, DNMT3A, DNMT3B, ETV6, EZH2, FLT3, GATA2, IDH1, IDH2, JAK2, KIT, KMT2A, KRAS, NF1, NPM1, NRAS, PHF6, PPM1D, PTPN11, RAD21, RUNX1, SETBP1, SF3B1, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, akute myeloische (FLT3- ITD)	EDTA-Blut, EDTA-Knochenmark, 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
Leukämie, akute promyelozytische (PML-RARA), qualitativ und quantitativ (PML-RARA, t(15;17)(q22;q21))	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR; größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)	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 BIO-ING-HUM-SAA-0147-02 BIO-ING-HUM-SAA-0063-02.2
Leukämie, atypische chronische myeloische (ASXL1, BRAF, CALR, CBL, CSF3R, DNMT3A, ETNK1, EZH2, JAK2, KRAS, MPL, NRAS, RUNX1, SETBP1, SF3B1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, B-Zell akute lymphatische (CREBBP, CRLF2, FLT3, IKZF1, KRAS, NF1, NRAS, NSD2, NTSC2, PAX5, PRDM9, PTPN11, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, chronische myeloische (ABL1)	EDTA-Blut, EDTA-Knochenmark, 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
Leukämie, Chronische Myelomonozytäre (ASXL1, BCOR, CBL, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KRAS, NF1, NPM1, NRAS, PHF6, PTPN11, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, Juvenile Myelomonozytäre (ASXL1, CBL, EZH2, JAK3, KRAS, NF1, NRAS, PTPN11, SETBP1)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Leukämie, myeloische (ASXL1, CALR, CBL, CEBPA, CSF3R, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KRAS, MPL, NPM1, NRAS, RUNX1, SETBP1, SF3B1, SH2B3, SRSF2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, Ph-ähnliche akute lymphatische (CRLF2, FLT3, IL7R, JAK1, JAK2, JAK3, KRAS, NRAS, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, T-Zell akute lymphatische (DNM2, FBXW7, NOTCH1, PHF6, PTEN, TP53, USP7)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, T-Zell-Prolymphozyten (ATM, BCOR, EZH2, JAK1, JAK3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämie, chronische lymphatische (ATM, BIRC3, BRAF, DDX3X, EGR2, FBXW7, KRAS, MED12, MYD88, NFKBIE, NOTCH1, NRAS, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Leukämien und Lymphome (TP53)	EDTA-Blut, EDTA-Knochenmark, 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
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lissenzephalie (ADGRG1, ARX, CCND2, CRADD, CRPPA, DCX, DYNC1H1, FKRP, 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-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, CALM3, CAV3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TECRL, TRDN, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
LL (ARID1A, ATM, BCL2, BIRC3, BRAF, BTK, CARD11, CCND1, CD79B, CREBBP, CXCR4, DNMT3A, EP300, ETV6, EZH2, FBXW7, FOXO1, ID3, IDH2, KMT2D, KRAS, MAP2K1, MEF2B, MYC, MYD88, NOTCH1, NOTCH2, NRAS, PAX5, PLCG2, POT1, PTEN, RHOA, RPS15, SF3B1, SGK1, SOCS1, STAT3, STAT5B, STAT6, TET2, TNFAIP3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphödeme, hereditär (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lysosomale Speichererkrankungen (AGA, ARSA, ARSB, ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CSTB, CTNS, CTSA, CTSD, CTSF, CTSK, FUCA1, GAA, GALC, GALNS, GBA1, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Lymphom, diffus-großzelliges B-Zell- (ARID1A, B2M, BCL2, BCL6, CARD11, CD58, CD79B, CREBBP, EP300, EZH2, FOXO1, GNA13, KLHL6, KMT2D, MEF2B, MYD88, NOTCH1, PCLO, PIM1, SGK1, SOCS1, STAT3, STAT6, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphom, follikuläres (ARID1A, BCL2, CARD11, CREBBP, EP300, EZH2, FOXO1, MEF2B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphom, kutanes T-Zell (ARID1A, CARD11, CD28, CREBBP, DNMT3A, KMT2D, PLCG1, TET2, TP53, TRRAP)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphom, peripheres T-Zell- (CCL22, CD28, FYN, IDH2, PLCG1, RHOA, STAT3, STAT5B, TET2, VAV1)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Lymphozyten-Leukämie, große granuläre (CCL22, STAT3, STAT5B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Magenkarzinom, familiäres (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Makrozephalie (ABCC9, AKT1, AKT2, APC2, ASXL2, BRAF, CCDC88C, CDKN1C, CHD3, CHD4, CHD8, CRADD, CUL4B, DHCR24, DNMT3A, EED, FIBP, GLI3, H1-4, HERC1, HRAS, HUWE1, IGF2, KRAS, L1CAM, MAP2K2, MPDZ, MTOR, NFIB, NRAS, OFD1, PAK1, PHF6, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, RIN2, RNF135, SETD2, SOS1, STRADA, SUZ12, TCF20, 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-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-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-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, ZHHHC9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Männliche Infertilität (ACTL7A, ADAD2, ADGRG2, AK7, AMH, AMHR2, ANOS1, AR, ARMC2, AURKC, BNC2, C14orf39, CATSPER1, CATSPER2, CATSPER3, CCDC146, CCDC39, CCDC40, CCNO, CEP135, CFAP251, CFAP298, CFAP300, CFAP43, CFAP44, CFAP47, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFTR, CHD7, CYP11B1, CYP17A1, CYP21A2, DDX3Y, DMC1, DMRT1, DMXL2, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH10, DNAH11, DNAH12, DNAH17, DNAH2, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DNHD1, DPY19L2, DRC1, DRC2, DRC4, DUSP6, FANCM, FGF17, FGF8, FGFR1, FKBP6, FLRT3, FSHB, FSHR, FSIP2, GAS2L2, GCNA, GNRH1, GNRHR, HESX1, HFM1, HSD17B3, HSD3B2, HYDIN, IL17RD, INSL3, KASH5, KCTD19, KISS1, KISS1R, KLB, KLHL10, LHB, LHCGR, LRRC56, M1AP, MAMLD1, MCIDAS, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MNS1, MSH4, MSH5, NANOS1, NME8, NR0B1, NR5A1, NSMF, ODAD1, ODAD2, ODAD3, PICK1, PLCZ1, PMFBP1, PNLD1, PNPLA6, POLR3A, POLR3B, PPP2R3C, PROK2, PROKR2, QRIH2, RAD21L1, RSPH1, RSPH3, RSPH4A, RSPH9, RSPO1, SEMA3A, SEMA7A, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPAG1, SPAG6, SPATA16, SPEF2, SPO11, SPRY4, SRD5A2, SRY, SSX1, STAG3, STK36, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAC3, TACR3, TDRD9, TERB1, TERB2, TEX11, TEX14, TEX15, TRIM37, TSGA10, TTC21A, TTC29, USP26, WDR11, XRCC2, YTHDC2, ZMYND10, ZMYND15, ZPBP, ZSWIM7)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Metabolische Epilepsie (ABAT, ABCD1, ACY1, ADK, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, AUH, BCKDK, BTB, CAD, CLN3, CLN5, CLN6, CLN8, CNM2, COQ4, COQ8A, CPT1A, CPT2, CTSD, CTSF, D2HGDH, DPYD, ETFB, ETDH, ETHE1, FH, FOLR1, GALC, GAMT, GATM, GBA1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mikrophthalmie und Anophthalmie (ABCB6, ALDH1A3, ATOH7, BCOR, BMP4, C12orf57, CHD7, FOXE3, FOXL2, FREM1, FZD5, GDF3, GDF6, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, OTX2, PAX6, PORCN, PRSS56, RARB, RAX, RBP4, SALL2, SHH, SIX6, SMOC1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Marginalzonen-Lymphom (BIRC3, CARD11, KLF2, KMT2D, MYD88, NOTCH1, NOTCH2, TNFAIP3, TP53, TRAF3)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mastozytose (ASXL1, CBL, DNMT3A, ETNK1, EZH2, IDH1, IDH2, JAK2, KIT, KRAS, NRAS, RUNX1, SF3B1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Maturity-onset Diabetes of the Young (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
McCune-Albright-Syndrom (GNAS)	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
Mikrozephalie (ADARB1, AGMO, ARCN1, BUB1B, CARS1, CIT, COG6, CRIPT, DDX11, DIAPH1, DONSON, DYNC1I2, KAT6A, KATNB1, KIF11, KIF14, LMNB1, LMNB2, MEF2C, MTHFS, PARS2, PLAA, PPP1R15B, PRUNE1, PUS7, RAB18, SLC1A4, SLC9A6, SMPD4, TCF4, TMX2, TRIO, TRMT10A, TUBGCP2, VARS1, WDR4, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mitochondriopathie, mitochondriale und nukleäre DNA (AARS2, ABAT, ABCB7, ACAD9, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, AK2, ALDH18A1, APTX, ATAD3A, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C19orf12, CARS2, CHCHD10, CHKB, CLPB, CLPP, COA3, COA5, COA8, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX11, 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, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mitochondriopathie, mitochondriale und nukleäre DNA Fortsetzung HSPD1, HTRA2, IARS2, IBA57, IDH3A, ISCA2, ISCU, KARS1, KIF5A, KYNU, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM4, MARS2, MDH2, MECP, 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, MTRFR, 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, NDUF51, NDUF52, NDUF53, NDUF54, NDUF56, NDUF57, NDUF58, 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, TFAZZIN, TARS2, TFAM, TIMM50, TIMM8A, TK2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Mitochondriopathie, mitochondriale und nukleäre DNA Fortsetzung TMEM126A, TMEM70, TPK1, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCRB, UQCRC2, UQCRCF1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Monogener Diabetes mellitus (ABCC8, AGPAT2, AIRE, ALMS1, APPL1, BLK, BSCL2, CAV1, CAVIN1, CEL, CIDEC, 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, NROB2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mukopolysaccharidosen (ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, SUMF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Morbus Fabry (GLA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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
Morbus Waldenström (ARID1A, CD79A, CXCR4, MYD88, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Morbus Wilson (ATP7B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
MTHFR-Mangel (MTHFR)	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
Muskeldystrophie, kongenital (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Mukoviszidose (CFTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Muskelfibrose, kongenitale extraokuläre (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Multi-Organ-Syndrome mit fakultativ CAKUT (ACTB, ACTG1, ACTG2, ANOS1, ARID1B, B3GLCT, BCOR, BSC12, CCBE1, CD151, CD96, CENPF, CEP55, CHD7, CHRNA1, CISD2, COL18A1, CREBBP, CTU2, EMG1, EP300, ERCC8, ESCO2, ETFA, ETFB, ETFDH, FANCA, FANCB, FANCD2, FANCE, FANCI, FANCL, FBN1, FGFR1, FGFR3, FIBP, FLNA, FOXF1, GDF6, GLI2, HAAO, HYL1, JAG1, JAM3, KAT6B, KCNQ1OT1, KCTD1, KDM5C, KDM6A, KMT2D, KRAS, KYN1, LMNA, LRP2, MID1, MNX1, MTOR, NADSYN1, NCAPG2, NFIX, NIPBL, NSDHL, PEX1, PEX5, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIGY, PMM2, POC1A, PORCN, PROK2, PROKR2, PTPN11, RAB23, RAF1, RAI1, RBM8A, RECQL4, ROR2, SEMA3E, SETBP1, SF3B4, SMC1A, SNRPB, SOS1, SRCAP, STRA6, TBX1, TBX3, TFAP2A, TMC01, TP63, TSC1, TSC2, TWIST2, UBR1, UPF3B, VANGL1, WFS1, WNT3, ZIC3, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Muskeldystrophie Duchenne/Becker (DMD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Myasthenie Syndrome, kongenitale (AGRN, CHAT, CHRNA1, CHRNA1, CHRND, CHRNE, CHRNA1, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, MYO9A, RASPN, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myopathie, distale (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myelom, multiples (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myopathie, metabolische (AGK, ALDOA, ATP2A1, DGUOK, FBXL4, FHL1, FLAD1, LAMA2, MGME1, MPV17, PNPLA8, POLG, SLC25A4, SUCLA2, SUCLG1, TK2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nävöides Basalzellkarzinom-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neonataler Diabetes mellitus (ABCC8, AGPAT2, BSCL2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Myopie (ARR3, COL11A1, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPSF1, LOXL3, LRPAP1, NYX, P3H2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephrokalzinose und Nephrolithiasis (ABCC6, ADCY10, AGXT, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CYP24A1, CYP27B1, FAH, FAM20A, FOXI1, G6PC1, GCM2, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, HSD11B2, INSR, KCNJ1, MOCOS, NHERF1, OCRL, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC7A9, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephronophtise (AGXT, ANKS6, CEP164, CEP290, CEP83, DCDC2, FAN1, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RRGIP1L, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neoplasie, Myelodysplastische (ASXL1, BCOR, BCORL1, CBL, CSF3R, DNMT3A, ETV6, EZH2, FLT3, GATA2, GNB1, IDH1, IDH2, JAK2, KRAS, NF1, NRAS, PHF6, PPM1D, PRPF8, PTPN11, RAD21, RUNX1, SETBP1, SF3B1, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neoplasie, myeloproliferative (ASXL1, BCOR, BCORL1, CALR, CBL, DNMT3A, EZH2, IDH1, IDH2, JAK2, MPL, NRAS, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neoplasie, myeloproliferative (CALR, JAK2, MPL)	EDTA-Blut, EDTA-Knochenmark, 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
Nephroblastom (ASXL1, BLM, BRCA2, BUB1B, CDC73, CTR9, DICER1, DIS3L2, GPC3, GPC4, PALB2, PAX6, REST, TP53, TRIM28, TRIM37, TRIP13, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nephrotisches Syndrom und Fokal Segmentale Glomerulosklerose (ACTN4, ANLN, APOL1, ARHGAP24, ARHGDI1, ATF3, AVIL, BLVRB, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DAAM2, DGKE, DLC1, EMP2, FAT1, FN1, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, LAGE3, LAMA5, LAMB2, LMX1B, LRP2, MAGI2, MT-TL1, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCA1, SYNPO, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neurofibromatose (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neuronale Ceroid-Lipofuszinose (ATP13A2, CLCN6, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Neuropathie, hereditäre motorisch-sensorische (AARS1, ABHD12, AIFM1, ATL1, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CNTNAP1, COA7, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GAN, GARS1, GBF1, 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, SLC25A46, SORD, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, VRK1, VWA1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neuropathie, hereditäre sensorisch autonome (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neutropenie, kongenitale schwere (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Non-Langerhans-Zell-Histiozytose (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Noonan-Syndrom und klinisch und genetisch überlappende RASopathien (BRAF, CBL, ERF, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Neutrophilenleukämie, Chronische (ABL1, ASXL1, CALR, CBL, CSF3R, CUX1, DNMT3A, ETNK1, EZH2, GATA2, JAK2, KDM6A, NRAS, NTRK2, PTPN11, RUNX1, SETBP1, SF3B1, SH2B3, SRSF2, STAG2, TET2, U2AF1, U2AF2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Nierenkrebs (BAP1, CDKN2B, CHEK2, DICER1, DIS3L2, FBXW7, FH, FLCN, MET, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TP53, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Nierenzellkarzinom (BAP1, FH, FLCN, MET, MITF, PTEN, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
NK-/T-Zell-Lymphom (ARID1A, BCOR, DDX3X, JAK3, KMT2D, STAT3, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Oligosaccharidosen (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Optikusatrophie (ACO2, AFG3L2, ANTXR1, ATP1A3, CACNA1F, CISD2, CRYAB, DNAJC30, DNMI1L, FA2H, MCAT, MECP, MFN2, MT-ND1, MT-ND4, MT-ND6, MTRFR, NBAS, NDUFA12, NDUFA7, NDUFS2, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SNF8, SPG7, SSBP1, TIMM8A, TMEM126A, UCHL1, WDR45, 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-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, RAB34, SCLT1, SCN1M1, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, ZRSR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Ophthalmoplegie, chronisch progressive externe (ACADS, DGUOK, DNA2, MGME1, OPA1, PABPN1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Osteolyse (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-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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pankreatitis (CASR, CELA3B, CFTR, CLDN2, CPA1, CTSC, PNLIP, SPINK1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Paroxysmale Dyskinesie (ADCY5, KCNMA1, PNKD, PRRT2, SLC2A1, TMEM151A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pankreasinsuffizienz, exokrine (CFTR, COX4I2, DNAJC21, EFL1, PDX1, PTF1A, SBDS, SRP54, UBR1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Pankreaskarzinom, familiäres (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pendred-Syndrom (FOXI1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Paragangliome-Phäochromozytome, hereditäre (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Parkinson-Krankheit (ATP13A2, DNAJC13, DNAJC6, FBXO7, GBA1, LRRK2, PARK7, PDE8B, PINK1, PLA2G6, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Peroxisomenbiogenesedefekte-Zellweger-Syndrom-Spektrum (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Paroxysmale nächtliche Hämoglobinurie (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pelizaeus-Merzbacher-Krankheit (PLP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Perrault-Syndrom (CLPP, HARS2, HSD17B4, LARS2, MRPL49, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Phenylketonurie und Hyperphenylalaninämie (DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SPR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Phosphatdiabetes (ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, NHERF1, OCRL, PHEX, SLC34A1, SLC34A3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Peutz-Jeghers-Syndrom (STK11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
PGLHL (DLST, NF1, NF2, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyzystische Lebererkrankung (ALG8, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyzystische Nierenerkrankung, autosomal-rezessiv erbliche (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Plasminogen-Aktivator-Inhibitor-1- Mangel (PAI1)	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
Platelet-Syndrom mit Prädisposition für akute myeloische Leukämie (FDP/AML) (RUNX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1

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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Polyzystische Nierenerkrankung, autosomal-dominant erbliche (ALG5, ALG9, DNAJB11, GANAB, IFT140, NEK8, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Primäre ziliäre Dyskinesie (BRWD1, CCDC39, CCDC40, CCNO, CFAP298, CFAP300, CFAP47, CFAP74, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH7, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DRC2, DRC4, FOXJ1, GAS2L2, HYDIN, LRRC56, MCIDAS, NME8, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, SPEF2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Prader-Willi-Syndrom (SNRPN- Genregion (15q11-q13))	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-0146-04.1 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
prämatüre Ovarialinsuffizienz (AARS2, ACAD9, AIRE, ALOX12B, AMH, AMHR2, ANKRD31, ANTXR1, ATG7, ATG9A, ATM, BLM, BMP15, BMPR1A, BMPR1B, BNC1, BRCA2, BUB1B, C14orf39, CLPP, COX10, CYP17A1, CYP19A1, DCAF17, DIAPH2, DMC1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4ENIF1, ERCC6, ESR2, EXO1, FANCA, FANCC, FANCG, FANCL, FANCM, FIGLA, FMR1, FOXE1, FOXL2, FSHR, GALT, GDF9, GGPS1, GNAS, HARS2, HAX1, HFM1, HROB, HSD17B4, HSF2BP, INHA, KASH5, KHDRBS1, LARS2, LHCGR, LIG4, LMNA, LRPPRC, MCM8, MCM9, MCMDC2, MEIOB, MRE11, MRPS22, MRPS7, MSH4, MSH5, NANOS3, NBN, NOBOX, NOG, NOTCH2, NR5A1, NSMCE2, NUP107, PANX1, PGRMC1, PMM2, POF1B, POLG, POLR2C, POLR3H, POR, PRDM9, PSMC3IP, RAD51, RCBTB1, RECQL4, SALL4, SGO2, SLC29A3, SOHLH1, SOHLH2, SPATA22, SPIDR, STAG3, STAR, SYCE1, SYCP2L, THOC6, TP63, TWNK, WDR62, WRN, WT1, XRCC2, ZSWIM7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Primäre Hyperoxalurie (AGXT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Prolaktinom (AIP, CDH23, DICER1, GPR101, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Prostatakarzinom, familiäres (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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pulmonale arterielle 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pseudohypoparathyreoidismus, Pseudopseudohypoparathyreoidismus (GNAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Renal-tubuläre Azidose (ATP6V0A4, ATP6V1B1, ATP6V1C2, CA2, FOXI1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Pyridoxin-abhängige Epilepsie (ALDH7A1)	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
Refsum-Syndrom (AMACR, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Renale Glukosurie (SLC5A2)	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
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schwerhörigkeit, nicht-syndromale (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, GIPC3, GJB2, 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, PJKV, PLS1, PNPT1, POLR1C, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, S1PR2, SERPINB6, SIX1, SLC17A8, SLC26A4, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Schwerhörigkeit, syndromale (ACOX1, ACTN4, ADGRV1, AMACR, ANLN, APOL1, ARHGAP24, ARHGDIA, ARSG, ATP6V1B1, 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, ESPN, EYA1, FAT1, FN1, FOXI1, HARS1, HARS2, HSD17B4, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, KCNE1, KCNJ10, KCNQ1, KIT, LAGE3, LAMB2, LARS2, LMX1B, LOXL3, LRP2, MAGI2, MITF, MRPL49, MT-RNR1, MT- TL1, MT-TS2, MYH9, MYO1E, MYO7A, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OPA1, OSGEP, PAX2, PAX3, PBX1, PCDH15, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PLCE1, POLR1C, POLR1D, PTPRO, SALL1, SCARB2, SGPL1, SIX1, SIX5, SLC26A4, SMARCA11, SNAI2, SOX10, TBC1D8B, TCOF1, TFAP2A, TIMM8A, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Retinitis Pigmentosa 3 (RPGR)	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
Retinoblastom (RB1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Retinopathie, mitochondriale (MT- ATP6, MT-ND1, MT-ND4, MT-ND6, MT-TH, MT-TL1, MT-TP, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Senior-Løken-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Schilddrüsenkarzinom, familiäres (APC, CDKN1B, DICER1, DUOX2, FOXE1, HABP2, MEN1, MINPP1, NKX2-1, PRKAR1A, PTEN, RET, SDHB, SDHD, SRGAP1, TINF2, TP53, WDR77)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Silver-Russell-Syndrom (HMGA2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Skeletal dysplasia with abnormal mineralization (ALPL, ANKH, AP2S1, CASR, CDC73, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAM20C, FGF23, GCM2, GNA11, HRAS, NHERF1, NRAS, OCLP, PHEX, SLC34A1, SLC34A3, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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, PTDS1, PTH1R, SFRP4, SLC29A3, SLC20A1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
SHOX-Defizienz, Langer mesomele Dysplasie (SHOX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Sichelzellanämie (HBB (c.20A>T))	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
Silver-Russell Syndrom (7p12.1, 7q32.2)	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-0146-04.1 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Silver-Russell-Syndrom (11p15.5)	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-0146-04.1 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Skelettdysplasien mit reduzierter Knochendichte (ALPL, ANO5, ATP6V0A2, B3GALT6, B4GALT7, BMP1, CCDC134, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, GORAB, IFIH1, IFITM5, KDELR2, LRP5, MBTPS2, MESD, NBAS, NOTCH2, P3H1, P4HB, PHLDB1, PLOD2, PLS3, POLR3A, PPIB, PYCR1, RIGI, SEC24D, SERPINF1, SERPINH1, SGMS2, SP7, SPARC, TAPT1, TENT5A, TMEM38B, TNFRSF11B, UNC45A, WNT1, XYL2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 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-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, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GFAP, GJC2, HSPD1, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MTRFR, NIPA1, NT5C2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SACS, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Small fiber-Neuropathie (SCN9A)	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
spinale Muskelatrophie, distale (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stargardt-ähnlich (ABCA4, ELOVL4, IMPG1, IMPG2, PROM1, PRPH2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stickler-Syndrom (COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, P3H2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Spinale Muskelatrophie (SMN1)	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-0146-04.1 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
spinozerebelläre Ataxie 1, 2, 3, 6, 7, 17 (ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP)	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
Störungen der Geschlechtsentwicklung XY (AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARMC3, 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, HSD17B3, HSD17B4, HSD3B2, IL17RD, KISS1, KISS1R, KLHL4, LARS2, LHB, LHCGR, LHX1, MAMLD1, MAP3K1, MCM9, NOBOX, NR0B1, NR5A1, NSMF, PAX2, POF1B, POR, PROK2, PROKR2, PSMC3IP, RPL10, RSP01, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Sterilität (CBAVD) (CFTR)	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
Störungen des Fettsäuren-, Carnitin- und Ketonkörperstoffwechsels (ACADM, ACADS, ACADVL, ACAT1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, MLYCD, OXCT1, SLC16A1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stoffwechselstörungen schwefelhaltiger Aminosäuren (ADK, AHCY, BHMT, CBS, CTH, ETHE1, GNMT, GPHN, MAT1A, MAT2A, MMADHC, MOCS1, MOCS2, MOCS3, MTHFR, MTR, MTRR, SQOR, SUOX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Stromatumor, gastrointestinaler (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Syndrom der unkämmbaren 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Teleangiektasie, hereditäre hämorrhagische (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thrombasthenie Glanzman (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thrombozytopenie, hereditäre (ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CYCS, DIAPH1, ETV6, FLI1, FLNA, FYB1, GALE, GATA1, GF11B, GNE, GP1BA, GP1BB, GP9, HOXA11, IKZF5, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, ORAI1, PRKACG, RAP1B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Störungen des Peptid- und Aminstoffwechsels (BHMT, BHMT2, CNDP1, DMGDH, FMO3, GCLC, GGCT, GGT1, GNMT, GPX4, GSR, GSS, ODC1, OPLAH, PEPD, SARDH, SELENBP1, SMS, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tuberöse Sklerose Komplex (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Tubulointerstitielle Nierenerkrankung, autosomal- dominante (DNAJB11, HNF1B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Tyrosinämie und verwandte Erkrankungen (FAH, GSTZ1, HGD, HPD, TAT, TH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thiopurin 5-Methyltransferase-Mangel (TPMT)	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
Thiopurin Therapie (NUDT15, TPMT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline	BIO-ING-HUM-VAW-0090-01.1 BIO-ING-HUM-FB-0220-01.1 BIO-ING-HUM-SAA-0094-01 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thiopurin-Mangel (NUDT15)	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
Usher-Syndrom (ADGRV1, ARSG, CDH23, CLRN1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Thrombotisch-thrombozytopenische Purpura (ADAMTS13)	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
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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Tieftonschwerhörigkeit (CCDC50, CENPP, DIAPH1, EYA4, MYO6, MYO7A, SLC26A4, TNC, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Transthyretin-Amyloidose (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
TRAPS (TNFRSF1A) (TNFRSF1A)	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
Vitreoretinopathie, exsudative (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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Vorderkammerdysgenese (ASPH, B3GLCT, BMP4, COL4A1, CPAMD8, CYP1B1, ELP4, EYA1, FOXC1, FOXC2, FOXE3, FOXL2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Undine-Syndrom (PHOX2B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); größen-spezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)	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-0147-02
Uniparentale Disomie 11 (Chromosom 11)	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

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Uniparentale Disomie 14 (Chromosom 14)	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
Uniparentale Disomie 15 (Chromosom 15)	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
Uniparentale Disomie 7 (Chromosom 7)	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
Walker-Warburg-Syndrom (B3GALNT2, B4GAT1, CRPPA, FKR, FKTN, GMPPB, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Warburg-Mikro-Syndrom (FOX L2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wilms-Tumor-Aniridia-Syndrom (ELP4, FOX L2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
von Hippel-Lindau-Syndrom (VHL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Von-Willebrand-Syndrom (VWF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wachstumshormonmangel, Kowarski-Syndrom (GH1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger); MLPA	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-0146-04.1
Zapfen-Stäbchen-Dystrophie (ABCA4, ABCG6, ACBD5, ADAM9, AIPL1, AP5B1, AP5M1, AP5Z1, BEST1, C19orf44, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP78, CERKL, CFAP410, CFAP418, CLN5, CNGA3, CNGB3, CNNM4, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FBLN5, FBN2, GUCA1A, GUCA1B, GUCY2D, IFT81, IMPG1, IMPG2, IRX5, IRX6, ITM2B, KCNV2, KIF11, MERTK, MFRP, MFSD8, MMP2, MT-ATP6, MT-TL1, PCYT1A, PDE6C, PITPNM3, PLK4, POC1B, PRDM13, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, 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-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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
Weibliche Infertilität (AARS2, ACAD9, AIRE, ALOX12B, AMH, ANKRD31, ANTXR1, AR, ATG7, ATG9A, ATM, BLM, BMP15, BMPR1A, BMPR1B, BNC1, BRCA2, BTG4, BUB1B, C14orf39, CBX2, CCNB3, CDC20, CFTR, CLPP, COX10, CPEB1, CYP11A1, CYP17A1, CYP19A1, CYP21A2, DCAF17, DHH, DIAPH2, DMC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4ENIF1, ERCC6, ESR1, ESR2, EXO1, F2, F5, FANCA, FANCC, FANCG, FANCL, FANCM, FGF8, FGFRL1, FIGLA, FMR1, FOXE1, FOXL2, FOXO3, FSHB, FSHR, GALT, GDF9, GGPS1, GNAS, GNRH1, GNRHR, HARS2, HAX1, HFM1, HROB, HSD17B3, HSD17B4, HSF2BP, INHA, INSR, KASH5, KHD3L, KHDRBS1, KISS1, KISS1R, LARS2, LHB, LHCGR, LHX8, LIG4, LMNA, LRPPRC, MAP3K1, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MRPS22, MRPS7, MSH4, MSH5, MTHFR, NANOS3, NBN, NLRP2, NLRP5, NLRP7, NOBOX, NOG, NOTCH2, NR0B1, NR5A1, NSMCE2, NUP107, PADI6, PANX1, PATL2, PGRMC1, PMM2, POF1B, POLG, POLR2C, POLR3H, POR, PRDM9, PROC, PROKR2, PROS1, PSMC3IP, RAD51, RCBTB1, REC114, REC8, RECQL4, SALL4, SEMA3A, SERPINC1, SERPINE1, SGO2, SLC29A3, SMC1B, SOHLH1, SOHLH2, SPATA22, SPIDR, SRD5A2, SRY, STAG3, STAR, SYCE1, SYCE3, SYCP2L, SYCP3, TAC3, TACR3, THBD, THOC6, TLE6, TOP6BL, TP63, TRIP13, TUBA1C, TUBA4A, TUBB8, TWNK, WDR11, WDR62, WEE2, WNT4, WRN, WT1, XRCC2, ZP1, ZP2, ZP3, ZSWIM7)	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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
zystische Nierenerkrankung (ALG5, ALG9, ANKS6, BICC1, COL4A1, COL4A3, COL4A4, DICER1, DNABJ11, DZIP1L, GANAB, HNF1B, IFT140, INVS, KAT6B, LRP5, NEK8, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5
Wiskott-Aldrich-Syndrom (WAS)	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
Adrenogenitales-Syndrom (ARMC5, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, MC2R, PDE11A, PDE8B, POR, PRKAR1A, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.5 BIO-ING-HUM-SAA-0071-00.1
X-chromosomale Adrenoleukodystrophie (ABCD1)	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
ALPS (ADA2, CARD11, CASP10, CASP8, CTLA4, DEF6, FADD, FAS, FASLG, IL2RA, IL2RB, ITK, KRAS, LRBA, MAGT1, NRAS, PIK3CD, PIK3R1, PRKCD, RASGRP1, RELA, SH2D1A, STAT3, STK4, TET2, TNFAIP3, TNFRSF9, TPP2, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.6
Retinitis pigmentosa (ABCA4, ABHD12, ADGRA3, ADIPOR1, AGBL5, AHR, ARHGEF18, ARL2BP, ARL3, ARL6, ASRGL1, BBS2, BBS4, BEST1, CA4, C2D2A, CDH16, CEP290, CERKL, CFAP418, CHM, CLCC1, CLRN1, CNGA1, CNGB1, COQ2, CRB1, CREB3, CRX, CTNNA1, CYP4V2, DHDDS, DHX38, DNAJC17, DYNC2H1, EMC1, EYS, FAM161A, FLVCR1, GUCA1B, HGSNAT, HK1, IDH3A, IDH3B, IDH3G, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, IQCB1, KIAA1549, KIZ, KLHL7, LAMA1, LRAT, MAK, MERTK, MFSD8, MRPL49, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, PGK1, PLA2G5, PNPLA6, POC5, POMGNT1, PRC1, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RCBT1, RDH11, RDH12, RDH5, REEP6, RHO, RLB1, RNU4-2, RNU4ATAC, RNU6-1, RNU6-2, RNU6-8, RNU6-9, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RS1, SAG, SCAPER, SEMA4A, SLC7A14, SNRNP200, SPATA7, TEAD1, TOPORS, TRNT1, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, VPS13B, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.6
Störung des Surfactant-Stoffwechsels, pulmonal (ABCA3, CSF2RA, CSF2RB, NKX2-1, SFTPA1, 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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.6
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-FB-0151-00.1 BIO-ING-HUM-SAA-0149-00.6
ApoE	EDTA-Blut; DNA	Real-time PCR (Schmelzpunktanalyse)	BIO-ING-MOLE-SAA-0104-00
CYP2C19 Genotypisierung	EDTA-Blut; DNA	Real-time PCR (Schmelzpunktanalyse)	BIO-ING-MOLE-SAA-0103-00
Faktor V-Genmutation	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0093-00.2
Hämochromatose Genanalyse	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0022-02.2
HLA-B27	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0100-00.1
Prothrombin (Faktor II)-Genmutation	EDTA-Blut; DNA	Real-time PCR	BIO-ING-MOLE-SAA-0093-00.2

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version
HLA-Typisierung	EDTA-Blut; DNA	SSO-PCR	BIO-ING-MOLE-SAA-0001-00.7
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	PCR, Schmelzpunktanalyse	BIO-ING-MOLE-SAA-0078-00.1
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	Sequenzierung	BIO-ING-MOLE-SAA-0087-00.3

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart:

Chromosomenanalyse ^[Flex C]

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 BIO-ING-HUM-SAA-0191-00 BIO-ING-HUM-VAW-0023-01.6
erworbener Chromosomensatz	Blut und/oder Knochenmark	Chromosomenbänderungs-analyse	BIO-ING-HUM-SAA-0083-00.6 BIO-ING-HUM-SAA-0001-00.3 BIO-ING-HUM-FB-0001.00.2 BIO-ING-HUM-VAW-0038-01.5 BIO-ING-HUM-SAA-0191-00 BIO-ING-HUM-VAW-0023-01.6
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-0151-00.3 BIO-ING-HUM-SAA-0152-00.5 BIO-ING-HUM-VAW-0071-00.4
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-0152-00.5 BIO-ING-HUM-SAA-0153-00.7 BIO-ING-HUM-VAW-0071-00.4
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-0194-00 BIO-ING-HUM-VAW-0102-00 BIO-ING-HUM-FB-0151-00.1 BIO-ING-HUM-SAA-0101-00.2 BIO-ING-HUM-SAA-0183-01
Nicht invasive Pränataldiagnostik (fetale Aneuploidien für	peripheres Vollblut; DNA	Next Generation Sequencing (NGS), VeriSeq NIPT	BIO-ING-HUM-SAA-0187-00

Untersuchungsgebiet: Mikrobiologie

Untersuchungsart:

Chromatographie (Immunchromatographie (IC)) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Carbapenemasen	Kulturmateriel	Immunchromatographie	BIO-ING-MIKR-SAA-0093-00.3
Cryptococcus neoformans, Antigen	Serum, Liquor	Immunchromatographie	BIO-ING-MIKR-SAA-0065-00.3
MRSA	Kulturmateriel	Immunchromatographie	BIO-ING-MIKR-VAW-0072-00

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

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

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Streptokokken, Streptolysin-O-Antikörper (ASL)	Serum	Turbidimetrie	BIO-ING-AUTO-SAA-0014-00.1

Untersuchungsart:

Empfindlichkeitstestungen von Bakterien und Pilzen ^[Flex B]

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-0018-05, BIO-ING-MIKR-VAW-0022-01.3, BIO-ING-MIKR-SAA-0061-01.1, 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-0048-01.1, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.3, BIO-ING-MIKR-LIS-0001-00.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.2, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.3, BIO-ING-MIKR-SAA-0069-00.3, 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-0078-01.2, BIO-ING-MIKR-SAA-0066-00.1, BIO-ING-MIKR-SAA-0045-02, BIO-ING-MIKR-SAA-0046-00.1, BIO-ING-MIKR-SAA-0033-00.2, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-03, BIO-ING-MIKR-SAA-0049-00.1, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00, BIO-ING-MIKR-SAA-0036-00.3, BIO-ING-MIKR-SAA-0043-00.5, BIO-ING-MIKR-SAA-0038-01.1
Mycobacterium-tuberculosis-Komplex	Kulturmateriail	modifizierte Proportionsmethode (Flüssigmedien)	BIO-ING-MIKR-SAA-0096-00
Candida albicans, non-albicans	Kulturmateriail, 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 ^[Flex B]

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	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-0022-01.3, BIO-ING-MIKR-SAA-0051-00, BIO-ING-MIKR-SAA-0060-00.3, BIO-ING-MIKR-SAA-0048-01.1, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.3, BIO-ING-MIKR-SAA-0038-01.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.2, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.3, BIO-ING-MIKR-SAA-0069-00.3, 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-0078-01.2, BIO-ING-MIKR-SAA-0066-00.1, BIO-ING-MIKR-SAA-0045-02, BIO-ING-MIKR-SAA-0046-00.1, BIO-ING-MIKR-SAA-0033-00.2, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-03, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00, BIO-ING-MIKR-SAA-0036-00.3, BIO-ING-MIKR-SAA-0043-00.5
Mykobakterien	Reinkulturen	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit	BIO-ING-MIKR-SAA-0006-01 BIO-ING-MIKR-SAA-0007-01
Mykobakterien	Kulturmateriel, Sputum, Bronchialsekret, Bronchial-Lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punkate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit	BIO-ING-MIKR-SAA-0010-00.2 BIO-ING-MIKR-FB-0082-00
Hefen und Hefeähnliche Pilze	Kulturmateriel	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferenzierung	BIO-ING-MIKR-SAA-0085-00 BIO-ING-MIKR-VAW-0056-00 BIO-ING-MIKR-FB-0062-00
Dermatophyten und andere Erreger von Dermatomykosen	Kulturmateriel	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferenzierung	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	Kulturmateriel	biochemisch-aufwändig (Bunte Reihe, Mehrkammerverfahren), einfach, orientierend, massenspektrometrische Erregerdifferenzierung	BIO-ING-MIKR-VAW-0010-00
Schimmelpilze wie Zygomyceten: Absidia, Mucor, Rhizomucor; Hyalohyphomyceten: Aspergillus, Penicillium; Phaeohyphomyceten: Exophiala, Cladophialophora, Alternaria	Kulturmateriel	biochemisch einfach, orientierend	BIO-ING-MIKR-VAW-0010-00
Schimmelpilze (Zygomyceten)	Kulturmateriel	biochemisch einfach, orientierend	BIO-ING-MIKR-SAA-0017-00

Untersuchungsart:

Kulturelle Untersuchungen ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bakterien und Pilze	Blut, Serum, Plasma, Urin, Stuhl, extravasale Körperflüssigkeiten (Sputum, Liquor, Aszites, Punktionsflüssigkeiten, Ergüsse, Lavagen), Magensaft, Galle, 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-0053-00.2, BIO-ING-MIKR-VAW-0054-00.1, BIO-ING-MIKR-VAW-0051-00, BIO-ING-MIKR-SAA-0031-02, BIO-ING-MIKR-SAA-0082-00.1, BIO-ING-MIKR-SAA-0063-00.3, BIO-ING-MIKR-SAA-0038-01.1, BIO-ING-MIKR-SAA-0064-00, BIO-ING-MIKR-SAA-0075-01, BIO-ING-MIKR-SAA-0074-00.2, BIO-ING-MIKR-SAA-0039-00.2, BIO-ING-MIKR-SAA-0062-00.3, BIO-ING-MIKR-SAA-0069-00.3, 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-0045-02, BIO-ING-MIKR-SAA-0046-00, BIO-ING-MIKR-SAA-0033-00.2, BIO-ING-MIKR-SAA-0035-00.2, BIO-ING-MIKR-SAA-0048-01.1, BIO-ING-MIKR-SAA-0079-01, BIO-ING-MIKR-SAA-0055-00.1, BIO-ING-MIKR-SAA-0047-03, BIO-ING-MIKR-SAA-0080-01, BIO-ING-MIKR-SAA-0081-00, BIO-ING-MIKR-SAA-0036-00.3, BIO-ING-MIKR-SAA-0043-00.5, BIO-ING-MIKR-SAA-0034-00.4, BIO-ING-MIKR-SAA-0050-00.3
Helicobacter pylori	Magenbiopsien	Kultur, selektiv	BIO-ING-MIKR-SAA-0078-01.2

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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 ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Borrelia, IgG, 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, IgM	Serum	CLIA	BIO-ING-AUTO-SAA-0126-00
Toxoplasma gondii, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0207-01
Aspergillus, Antigen	Serum	ELISA	BIO-ING-SERO-SAA-0039-02
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-0041-01.3
Echinococcus-Antikörper (IgG)	Serum	ELISA	BIO-ING-SERO-SAA-0008-02
Helicobacter pylori, Antigen	Stuhl	CLIA	BIO-ING-MIKR-SAA-0028-01
Helicobacter pylori, IgA, IgG	Serum	ELISA	BIO-ING-SERO-SAA-0016-02
Plasmodium ssp.-Antikörper (Malaria)	Serum	ELISA	BIO-ING-IMMU-SAA-0069-01.1
Legionella pneumophila Serotyp 1, Antigen	Urin	CLIA	BIO-ING-MIKR-SAA-0029-01
Mycobacterium-tuberculosis-Komplex, MPT64-Antigen	Kulturmateriale aus Flüssigkulturen	ELISA	BIO-ING-MIKR-SAA-0010-00.2
Mycoplasma pneumoniae, IgA	Serum	ELISA	BIO-ING-SERO-SAA-0068-03
Streptococcus pneumoniae, Antikörper	Serum	ELISA	BIO-ING-SERO-SAA-0069-01.1

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Salmonella-IgA Ak	Serum	ELISA	BIO-ING-SERO-SAA-0071-03
Salmonella- Ak (polyvalent)	Serum	ELISA	BIO-ING-SERO-SAA-0071-03
Treponema pallidum, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
Borrelia, IgG, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0049-01.2
Echinococcus-Antikörper (IgG)	Serum	Immunoblot	BIO-ING-SERO-SAA-0113-00
Helicobacter, IgA, IgG	Serum	Immunoblot	BIO-ING-SERO-SAA-0015-02
Treponema pallidum, IgG, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0029-02.1
Yersinien, IgA, IgG	Serum	Immunoblot	BIO-ING-SERO-SAA-0034-02

Untersuchungsart:

Mikroskopie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Bakterien und Pilze	Blut, Serum, Plasma, Urin, Stuhl, extravasale Körperflüssigkeiten (Sputum, Liquor, Aszites, Punktions-flüssigkeiten, 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, IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0045-01
Bartonella quintana, IgG, IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-SERO-SAA-0045-01
Cestoden, z.B. Taenia, Diphyllobothrium, 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
Dermatophyten	Hautgeschabsel, Haare, Nagelmateriale, Abstriche Hautläsionen, Pus, Biopsien	Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen	BIO-ING-MIKR-SAA-0018-01
Dermatophyten, Trichophyton, Microsporum, Epidermophyton und andere Erreger von Dermatomykosen, Scopulariopsis	Kulturmateriale aus Kulturen aus Hautgeschabsel, Haare, Nagelmateriale, 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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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	BIO-ING-SERO-SAA-0067-02.1
Legionella pneumophila, Serogruppen 1-6, Antikörper	Serum	Fluoreszenzmikroskopie indirekte	BIO-ING-SERO-SAA-0067-02.1
Legionella pneumophila, Serogruppen 7-14, Antikörper	Serum	Fluoreszenzmikroskopie indirekte	BIO-ING-SERO-SAA-0067-02.1
Malaria-Erregernachweis	EDTA-Blut	Hellfeldmikroskopie mit Anfärbung	BIO-ING-HÄMA-SAA-0027-00.6
Mykobakterien	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie	BIO-ING-MIKR-SAA-0007-01
Mykobakterien	Sputum, Bronchialsekret, Bronchiallavage, 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 Amöben, 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
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	Kulturmateriale 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, Ophisthorchis, Fasciola, Paragonimus	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	BIO-ING-MIKR-SAA-0019-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Würmer, Wurmteile, Ektoparasiten	Stuhl, ausgeschiedene Parasiten	Hellfeldmikroskopie ohne Anfärbung	BIO-ING-MIKR-FB-0336-00

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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-0007-01.4
Dermatophyten, DNA	Hautschuppen, Nagelspäne, Haare, Kulturmaterial	Real-time PCR; Micro-Array	BIO-ING-MIKR-SAA-0104-01
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, Kulturmaterial	PCR (molekulare Resistenztestung)	BIO-ING-MIKR-SAA-0095-00
Mycobacterium-tuberculosis-Komplex, DNA	Kulturmaterial aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	PCR mit nachfolgender fest-flüssig Hybridisierung (Festphasen-gebundene Fangsonden und Enzymimmunoassay basierende Detektion der Hybridisierungsergebnisse)	BIO-ING-MIKR-SAA-0009-01
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 Hybridisierungsergebnisse)	BIO-ING-MIKR-SAA-0009-01
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 Fluorchinolon-Resistenz	Urin, Abstrich	Sequenzierung	BIO-ING-MOLE-SAA-0065-00.5
Mycoplasma pneumoniae, DNA	Liquor	Real-time PCR	BIO-ING-MOLE-SAA-0102-00
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-0060-00.5

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Streptococcus pneumoniae, Legionella pneumophila, Haemophilus influenzae, Bordetellen, Mycoplasma pneumoniae, Chlamydia pneumoniae, DNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0101-00
Staphylococcus aureus, Methicillin- resistent (MRSA), DNA	Abstrich, Kulturmateriel	Real-time PCR	BIO-ING-MOLE-SAA-0037-00.6
PVL-Staphylococcus aureus, Methicillin-resistent (MRSA), DNA	Bakterielle Reinkulturen	Real-time PCR	BIO-ING-MOLE-SAA-0089-00.2
Chlamydia trachomatis, Neisseria gonorrhoeae, Mycoplasma genitalium, Mycoplasma hominis, Trichomonas vaginalis, Ureaplasma urealyticum, Ureaplasma parvum, DNA	Urin, Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0049-00.5
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
Campylobacter spp., Clostridioides difficile toxin B, E. coli enteroaggregativ (EAEC), E. coli enteropathogen (EPEC), E. coli enterotoxigenic (ETEC), E. coli Shiga toxin-prod. (STEC), Salmonella spp., Shigella spp. / Enteroinvasive E. coli (EIEC), Yersinia enterocolitica, Blastocystis hominis, Cryptosporidium spp., Cyclospora cayetanensis, Dientamoeba fragilis, Entamoeba histolytica, Giardia lamblia, Strongyloides stercoralis, DNA	Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0092-00.2

Untersuchungsart:

Zellfunktionsteste ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Freisetzung von IFN- γ nach Stimulation mit Antigenen von <i>M.</i> <i>tuberculosis</i> (Quantiferon-TB Gold Plus)	Heparin-Blut in spezial- beschichteten Röhrchen	Zytokinfreisetzung, Messung Zytokin mittels CLIA	BIO-ING-AUTO-SAA-0188-00.1

Untersuchungsgebiet: Virologie

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Cytomegalovirus, IgG oder IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0205-00.2
Cytomegalovirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
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
Dengue-Virus NS1-Antigen	Serum	ELISA	BIO-ING-SERO-SAA-0112-00
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-0041-01.3
Epstein-Barr-Virus, IgG, Avidität	Serum	Immunoblot	BIO-ING-SERO-SAA-0006-04
Frühsommer-Meningoenzephalitis-Virus, IgG oder IgM	Serum	ELISA	BIO-ING-SERO-SAA-0061-03
Frühsommer-Meningoenzephalitis-Virus, IgG, Antikörper-	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
Hantaviren, IgG, IgM	Serum	ELISA	BIO-ING-SERO-SAA-0011-00.6
Hantaviren, IgG, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0013-01.1
Hepatitis-A-Virus, IgG, 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-0101-01.1
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
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-0062-01.1
Hepatitis-D-Virus, Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0180-01
Hepatitis-E-Virus, IgG, IgM	Serum	EIA	BIO-ING-SERO-SAA-0063-02.2
Hepatitis-E-Virus, IgG, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0017-02
Herpes-Simplex-Virus Typ 1, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Herpes-Simplex-Virus, IgG, IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0124-00.1
Humane Immundefizienz-Viren Typ 1/2, Antikörper	Serum	Immunoblot	BIO-ING-SERO-SAA-0065-02.1
Humane Immundefizienzviren Typ 1/2, Antikörper und p24-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0098-00.1
Masernvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0127-00
Masernvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
Mumpsvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0125-01
Mumpsvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
Parvovirus B19, IgG, IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA	BIO-ING-AUTO-SAA-0128-01
Parvovirus B19, IgG, IgM	Serum	Immunoblot	BIO-ING-SERO-SAA-0044-01.1
Rötelnvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	ECLIA	BIO-ING-AUTO-SAA-0206-00
Rötelnvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3
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-0122-01
Varicella-Zoster-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA	BIO-ING-SERO-SAA-0041-01.3

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
BK-Polyomavirus, DNA, Viruslast	EDTA-Blut, Urin, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.4
Cytomegalovirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, Abstriche, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.4
Enterovirus, RNA	Liquor, BAL, Tracheal- oder Bronchialsekret, Abstriche, Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0059-00.3
Epstein-Barr-Virus, DNA, Viruslast	EDTA-Blut, Liquor, Abstriche, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.4
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-0032-01.4
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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
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-0098-00
JC-Polyomavirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, BAL	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.4
Adenoviren DNA, Influenzaviren Typen A/B RNA, Metapneumovirus RNA, Parainfluenzaviren RNA, Coronaviren RNA, Enteroviren RNA, Rhinoviren RNA, Respiratorische Synzytialviren RNA, SARS-CoV-2 RNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Real-time PCR	BIO-ING-MOLE-SAA-0099-00
SARS-CoV-2 (RNA)	Abstrich, Bal, Bronchialsekret, Sputum	Real-time PCR	BIO-ING-MOLE-SAA-0098-00
SARS-CoV-2 (RNA)	Abstrich, Bal, Bronchialsekret, Sputum	Real-time PCR	BIO-ING-MOLE-SAA-0076-00.3
Varicella-Zoster-Virus, DNA	Liquor, Abstriche, EDTA-Blut, BAL, EDTA-Plasma	Real-time PCR	BIO-ING-MOLE-SAA-0032-01.4
Adenovirus DNA, Astrovirus RNA, Norovirus RNA, Rotavirus RNA, Sapovirus RNA, Enteroviren RNA, Parcheoviren RNA	Stuhl	Real-time PCR	BIO-ING-MOLE-SAA-0092-00.2

Untersuchungsgebiet: Transfusionsmedizin

Untersuchungsart:

Agglutinationsteste ^[Flex B]

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-0026-02.1
Antikörper-Differenzierung	Vollblut, EDTA-Blut	Hämagglutinationstest	BIO-ING-IMMU-SAA-0026-02.1
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

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex A]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
Fetaler Rhesusfaktor D-Varianten	EDTA-Blut, EDTA-Plasma	Multiplex Real-time PCR	BIO-ING-MOLE-SAA-0091-00.2