

Akkreditierung



Die Deutsche Akkreditierungsstelle bestätigt mit dieser **Akkreditierungsurkunde**, dass die

Bioscientia Institut für Medizinische Diagnostik GmbH
Konrad-Adenauer-Straße 17, 55218 Ingelheim

ein Medizinisches Laboratorium betreibt, das die Anforderungen gemäß DIN EN ISO 15189:2024 für die in der nachfolgend aufgeführten Anlage näher spezifizierten Konformitätsbewertungstätigkeiten erfüllt. Dies schließt zusätzlich bestehende gesetzliche und normative Anforderungen an das Medizinische Laboratorium ein, einschließlich solcher in relevanten sektoralen Programmen, sofern diese in der nachfolgend aufgeführten Anlage ausdrücklich bestätigt wird.

D-ML-13452-01-01 Gültig ab: 21.07.2025

Die Anforderungen an das Managementsystem in der DIN EN ISO 15189 sind in einer für medizinische Laboratorien relevanten Sprache verfasst und stehen insgesamt in Übereinstimmung mit den Prinzipien der DIN EN ISO 9001.

Diese Akkreditierung wurde gemäß Art. 5 Abs. 1 Satz 2 VO (EG) 765/2008, nach Durchführung eines Akkreditierungsverfahrens unter Beachtung der Mindestanforderungen der DIN EN ISO/IEC 17011 und auf Grundlage einer Bewertung und Entscheidung der eingesetzten Akkreditierungsausschüsse ausgestellt.

Diese Akkreditierungsurkunde gilt nur in Verbindung mit dem Bescheid vom 21.07.2025. Sie besteht aus diesem Deckblatt, der Rückseite des Deckblatts und der dazugehörigen Anlage.

Registrierungsnummer der Akkreditierungsurkunde: **D-ML-13452-01-00**

Berlin, 21.07.2025

Im Auftrag
Dipl.-Ing. Anna Lewandowski | Fachbereichsleitung

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Deutsche Akkreditierungsstelle GmbH

Standort Berlin
Spittelmarkt 10
10117 Berlin

Die Deutsche Akkreditierungsstelle GmbH (DAkKS) ist die beliehene nationale Akkreditierungsstelle der Bundesrepublik Deutschland gemäß § 8 Absatz 1 AkkStelleG i. V. m. § 1 Absatz 1 AkkStelleGBV. Die DAkKS ist als nationale Akkreditierungsbehörde gemäß Art. 4 Abs. 4 VO (EG) 765/2008 und Tz. 4.7 DIN EN ISO/IEC 17000 durch Deutschland benannt.

Die Akkreditierungsurkunde ist gemäß Art. 11 Abs. 2 VO (EG) 765/2008 im Geltungsbereich dieser Verordnung von den nationalen Behörden als gleichwertig anzuerkennen sowie von den WTO-Mitgliedsstaaten, die sich in bilateralen- oder multilateralen Gegenseitigkeitsabkommen verpflichtet haben, die Urkunden von Akkreditierungsstellen, die Mitglied bei ILAC oder IAF sind, als gleichwertig anzuerkennen.

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Der aktuelle Stand der Mitgliedschaft kann folgenden Webseiten entnommen werden:

EA: www.european-accreditation.org
ILAC: www.ilac.org
IAF: www.iaf.nu

Deutsche Akkreditierungsstelle

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01 nach DIN EN ISO 15189:2024

Gültig ab: 21.07.2025

Ausstellungsdatum: 21.07.2025

Diese Urkundenanlage ist Bestandteil der Akkreditierungsurkunde D-ML-13452-01-00.

Inhaber der Akkreditierungsurkunde:

**Bioscientia Institut für Medizinische Diagnostik GmbH
Konrad-Adenauer-Straße 17, 55218 Ingelheim**

mit dem Standort

**Bioscientia Institut für Medizinische Diagnostik GmbH
Bioscientia MVZ Ingelheim
Konrad-Adenauer-Straße 17, 55218 Ingelheim**

Das Medizinische Laboratorium erfüllt die Anforderungen gemäß DIN EN ISO 15189:2024, um die in dieser Anlage aufgeführten Konformitätsbewertungstätigkeiten durchzuführen. Das Medizinische Laboratorium erfüllt gegebenenfalls zusätzliche gesetzliche und normative Anforderungen, einschließlich solcher in relevanten sektoralen Programmen, sofern diese nachfolgend ausdrücklich bestätigt werden.

Die Anforderungen an das Managementsystem in der DIN EN ISO 15189 sind in einer für medizinische Laboratorien relevanten Sprache verfasst und stehen insgesamt in Übereinstimmung mit den Prinzipien der DIN EN ISO 9001.

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Untersuchungen im Bereich:

Medizinische Laboratoriumsdiagnostik

Untersuchungsgebiete:

Klinische Chemie

Immunologie

Humangenetik (Molekulare Humangenetik)

Humangenetik (Zytogenetik)

Mikrobiologie

Virologie

Transfusionsmedizin

Flexibler Akkreditierungsbereich:

Dem Medizinischen Laboratorium ist innerhalb der gekennzeichneten Untersuchungsbereiche, ohne dass es einer vorherigen Information und Zustimmung der DAkKS bedarf,

[Flex A] die Anwendung der hier aufgeführten genormten oder ihnen gleichzusetzenden Untersuchungsverfahren mit unterschiedlichen Ausgabeständen gestattet. Im Bereich medizinischer Laboratorien sind in dieser Kategorie unter gleichzusetzenden Untersuchungsverfahren auch vom Labor validierte und durch Akkreditierungsentscheidung bestätigte Untersuchungsverfahren zu verstehen. Diese gilt ausschließlich für neue Ausgabestände (Revisionen) bestätigter Untersuchungsverfahren ohne dass Analyt, Matrix oder Untersuchungstechnik verändert werden.

[Flex B] die freie Auswahl von genormten oder ihnen gleichzusetzenden Untersuchungsverfahren gestattet.

[Flex C] die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet.

Die aufgeführten Untersuchungsverfahren sind beispielhaft. Das Medizinische Laboratorium verfügt über eine aktuelle Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich. Die Liste ist öffentlich verfügbar auf der Webpräsenz des Medizinischen Laboratoriums.

Untersuchungsgebiet: Klinische Chemie

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
2C-B	Urin	LC-MS/MS
2C-I	Urin	LC-MS/MS
3-Hydroxybromazepam	Urin	LC-MS/MS
5-Hydroxyindolessigsäure	Urin	LC-MS/MS
6-Acetylcodein	Urin	LC-MS/MS
6-MAM (6-Monoacetylmorphin)	Urin	LC-MS/MS
7-Aminoclonazepam	Urin	LC-MS/MS
7-Aminoflunitrazepam	Urin	LC-MS/MS
7-Aminonitrazepam	Urin	LC-MS/MS
9-OH-Risperidon/Paliperidon	Serum, EDTA-Plasma	LC-MS/MS
10-OH-Carbazepin	Serum, EDTA-Plasma	LC-MS/MS
Alpha-Hydroxyalprazolam	Urin	LC-MS/MS
Alpha-Hydroxymidazolam	Urin	LC-MS/MS
Alpha-Hydroxytriazolam	Urin	LC-MS/MS
Alprazolam	Urin	LC-MS/MS
Aminosäuren	EDTA-Plasma	LC-MS/MS
Amiodaron	Serum, EDTA-Plasma	LC-MS/MS
Amisulprid	Serum, EDTA-Plasma	LC-MS/MS
Amitriptylin	Serum, EDTA-Plasma	LC-MS/MS
Amphetamin	Urin	LC-MS/MS
Amphetamine-Bestätigung	Urin	LC-MS/MS
Aripiprazol	Serum, EDTA-Plasma	LC-MS/MS
Benzodiazepine-Bestätigung	Urin	LC-MS/MS
Benzoyllecgonin	Urin	LC-MS/MS
BDB	Urin	LC-MS/MS
Drogen-Bestätigung	Urin	LC-MS/MS
Brivaracetam	Serum, EDTA-Plasma	LC-MS/MS
Bromazepam	Urin	LC-MS/MS
Buprenorphin	Urin	LC-MS/MS
Brotizolam	Urin	LC-MS/MS
Butylon	Urin	LC-MS/MS
Cannabinoide-Bestätigung	Urin	LC-MS/MS
Cathinon	Urin	LC-MS/MS
Chlordiazepoxid	Urin	LC-MS/MS
Chlorprothixen	Serum, EDTA-Plasma	LC-MS/MS
Ciclosporin	EDTA-Blut	LC-MS/MS
Citalopram	Serum, EDTA-Plasma	LC-MS/MS
Clobazam	Urin	LC-MS/MS

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Clomipramin	Serum, EDTA-Plasma	LC-MS/MS
Clonazepam	Urin	LC-MS/MS
Clozapin	Serum, EDTA-Plasma	LC-MS/MS
Cocaethylen	Urin	LC-MS/MS
Cocain	Urin	LC-MS/MS
Cocain-Bestätigung	Urin	LC-MS/MS
Codein	Urin	LC-MS/MS
Dehydroaripiprazol	Serum	LC-MS/MS
Demoxepam	Urin	LC-MS/MS
Desalkylflurazepam	Urin	LC-MS/MS
Desethylamiodaron	Serum, EDTA-Plasma	LC-MS/MS
Desipramin	Serum, EDTA-Plasma	LC-MS/MS
Desmethylcitalopram	Serum	LC-MS/MS
Norclomipramin	Serum, EDTA-Plasma	LC-MS/MS
Desmethylclozapin	Serum, EDTA-Plasma	LC-MS/MS
Nordoxepin	Serum, EDTA-Plasma	LC-MS/MS
Desmethylflunitrazepam	Urin	LC-MS/MS
Desmethylmirtazapin	Serum, EDTA-Plasma	LC-MS/MS
Desmethylertralin	Serum, EDTA-Plasma	LC-MS/MS
O-Desmethylvenlafaxin	Serum, EDTA-Plasma	LC-MS/MS
Diazepam	Urin	LC-MS/MS
Dihydrocodein	Urin	LC-MS/MS
Doxepin	Serum, EDTA-Plasma	LC-MS/MS
Duloxetine	Serum, EDTA-Plasma	LC-MS/MS
EDDP	Urin	LC-MS/MS
Escitalopram	Serum, EDTA-Plasma	LC-MS/MS
Estazolam	Urin	LC-MS/MS
Ethosuximid	Serum, EDTA-Plasma	LC-MS/MS
Ethylglucuronid	Urin, Serum	LC-MS/MS
Ethylsulfat	Urin, Serum	LC-MS/MS
Everolimus	EDTA-Blut	LC-MS/MS
Fentanyl	Urin	LC-MS/MS
Felbamat	Serum, EDTA-Plasma	LC-MS/MS
Flecainid	Serum, EDTA-Plasma	LC-MS/MS
Fluconazol	Serum, EDTA-Plasma	LC-MS/MS
Flunitrazepam	Urin	LC-MS/MS
Fluoxetin	Serum, EDTA-Plasma	LC-MS/MS
Flupentixol	Serum, EDTA-Plasma	LC-MS/MS
Flurazepam	Urin	LC-MS/MS
Fluvoxamin	Serum, EDTA-Plasma	LC-MS/MS
Gabapentin	Serum, EDTA-Plasma, Urin	LC-MS/MS
Haloperidol	Serum, EDTA-Plasma	LC-MS/MS

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Homocystein	NaF-Plasma	LC-MS/MS
Hydrocodon	Urin	LC-MS/MS
Hydromorphon	Urin	LC-MS/MS
Imipramin	Serum, EDTA-Plasma	LC-MS/MS
Itraconazol	Serum, EDTA-Plasma	LC-MS/MS
Ketamin	Urin	LC-MS/MS
Lacosamid	Serum, EDTA-Plasma	LC-MS/MS
Lamotrigin	Serum, EDTA-Plasma	LC-MS/MS
Levetiracetam	Serum, EDTA-Plasma	LC-MS/MS
Levomepromazin	Serum, EDTA-Plasma	LC-MS/MS
Lorazepam	Urin	LC-MS/MS
Lormetazepam	Urin	LC-MS/MS
LSD	Urin	LC-MS/MS
Maprotilin	Serum, EDTA-Plasma	LC-MS/MS
MBDB	Urin	LC-MS/MS
MDA	Urin	LC-MS/MS
MDEA	Urin	LC-MS/MS
MDMA	Urin	LC-MS/MS
MDPV	Urin	LC-MS/MS
Meconin	Urin	LC-MS/MS
Medazepam	Urin	LC-MS/MS
Meperidin	Urin	LC-MS/MS
Mephedron	Urin	LC-MS/MS
Mescaline	Urin	LC-MS/MS
Methadon	Urin	LC-MS/MS
Metanephrin / Normetanephrin / 3-Methoxythyramin	EDTA-Plasma, Urin	LC-MS/MS
Methamphetamin	Urin	LC-MS/MS
Methaqualon	Urin	LC-MS/MS
Methylmalonsäure	Serum, Urin	LC-MS/MS
Methylon	Urin	LC-MS/MS
Midazolam	Urin	LC-MS/MS
Mirtazapin	Serum, EDTA-Plasma	LC-MS/MS
Morphin	Urin	LC-MS/MS
Mycophenolsäure	Serum, EDTA-Plasma	LC-MS/MS
Naltrexon	Urin	LC-MS/MS
Naloxon	Urin	LC-MS/MS
N-Desmethylnesuximid	Serum, EDTA-Plasma	LC-MS/MS
N-Desmethylnaloxon	Serum	LC-MS/MS
Nitrazepam	Urin	LC-MS/MS
Norbuprenorphin	Urin	LC-MS/MS
Norclobazam	Urin	LC-MS/MS
Norcocain	Urin	LC-MS/MS

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Norcodein	Urin	LC-MS/MS
Nordiazepam	Urin	LC-MS/MS
Norfentanyl	Urin	LC-MS/MS
Desmethylfluoxetin	Serum, EDTA-Plasma	LC-MS/MS
Norketamin	Urin	LC-MS/MS
Normeperidin	Urin	LC-MS/MS
Norquetiapin	Serum	LC-MS/MS
Nortapentadol	Urin	LC-MS/MS
Nortilidin	Urin	LC-MS/MS
Nortriptylin	Serum, EDTA-Plasma	LC-MS/MS
O-Desmethyltramadol	Urin	LC-MS/MS
O-Desmethyl-Venlafaxin	Serum, EDTA-Plasma	LC-MS/MS
Olanzapin	Serum, EDTA-Plasma	LC-MS/MS
Opiate-Bestätigung	Urin	LC-MS/MS
Opioid-Bestätigung	Urin	LC-MS/MS
Oxazepam	Urin	LC-MS/MS
Oxcarbazepin	Serum, EDTA-Plasma	LC-MS/MS
Oxycodon	Urin	LC-MS/MS
Oxymorphon	Urin	LC-MS/MS
Papaverin	Urin	LC-MS/MS
Paroxetin	Serum, EDTA-Plasma	LC-MS/MS
PCP	Urin	LC-MS/MS
Perampanel	Serum, EDTA-Plasma	LC-MS/MS
Perazin	Serum, EDTA-Plasma	LC-MS/MS
Pipamperon	Serum, EDTA-Plasma	LC-MS/MS
PMA	Urin	LC-MS/MS
Posaconazol	Serum, EDTA-Plasma	LC-MS/MS
Prazepam	Urin	LC-MS/MS
Pregabalin	Serum, EDTA-Plasma	LC-MS/MS
Pregabalin	Urin	LC-MS/MS
Primidon	Serum, EDTA-Plasma	LC-MS/MS
Promethazin	Serum, EDTA-Plasma	LC-MS/MS
Promethazin	Urin	LC-MS/MS
Propafenon	Serum, EDTA-Plasma	LC-MS/MS
Propoxyphen	Urin	LC-MS/MS
Psychedelika-Bestätigung	Urin	LC-MS/MS
Quetiapin	Serum, EDTA-Plasma, Urin	LC-MS/MS
Risperidon	Serum, EDTA-Plasma	LC-MS/MS
Ritalinsäure	Urin	LC-MS/MS
Rufinamid	Serum, EDTA-Plasma	LC-MS/MS
Serotonin	EDTA-Blut	LC-MS/MS
Sertralin	Serum, EDTA-Plasma	LC-MS/MS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Sirolimus	EDTA-Blut	LC-MS/MS
Sufentanil	Urin	LC-MS/MS
Sultiam	Serum, EDTA-Plasma	LC-MS/MS
Synthetische Cannabinoide	Urin, Serum	LC-MS/MS
Tabakalkaloide	Urin	LC-MS/MS
Tacrolimus	EDTA-Blut	LC-MS/MS
Tapentadol	Urin	LC-MS/MS
Temazepam	Urin	LC-MS/MS
THC-COOH	Urin	LC-MS/MS
Thebain	Urin	LC-MS/MS
Tilidin	Urin	LC-MS/MS
Topiramat	Serum, EDTA-Plasma	LC-MS/MS
Toxikologische Suchanalyse	Urin, Serum	LC-MS/MS
Tramadol	Urin	LC-MS/MS
Triazolam	Urin	LC-MS/MS
Trimipramin	Serum, EDTA-Plasma	LC-MS/MS
Trizyklische Antidepressiva (TCA Suchanalyse)	Serum	LC-MS/MS
Trizyklische Antidepressiva (TCA Suchanalyse)	Urin	LC-MS/MS
Venlafaxin	Serum, EDTA-Plasma	LC-MS/MS
Vigabatrin	Serum, EDTA-Plasma	LC-MS/MS
25-OH-Vitamin D (gesamt)	Serum, EDTA-Plasma	LC-MS/MS
Voriconazol	Serum, EDTA-Plasma	LC-MS/MS
Zaleplon	Urin	LC-MS/MS
Z-Hypnotika-Bestätigung	Urin	LC-MS/MS
Ziprasidon	Serum, EDTA-Plasma	LC-MS/MS
Zolpidem	Urin	LC-MS/MS
Zonisamid	Serum, EDTA-Plasma	LC-MS/MS
Zopiclon	Urin	LC-MS/MS

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Organische Säuren	Urin	GC-MS
Ultralangkettige Fettsäuren	Serum	GC-MS

Untersuchungsart:

Chromatographie (Hochleistungsflüssigkeitschromatographie (HPLC)) ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Serotonin	EDTA-Blut	HPLC-ECD
Vitamin B 1	EDTA-Blut	HPLC-FD

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Vitamin B 2	EDTA-Blut	HPLC-FD
Vitamin B 6	EDTA-Plasma, Serum	HPLC-FD
Vitamin K1	Serum	HPLC-FD
Aminosäuren	Urin, Liquor	HPLC-UV/VIS
Beta-Carotin	Serum, EDTA-Plasma	HPLC-UV/VIS
CDT (Carbohydrate-Deficient-Transferrin)	Serum	HPLC-UV/VIS
Coenzym Q10	Serum, EDTA-Plasma	HPLC-UV/VIS
HbA	EDTA-Blut	HPLC-UV/VIS
HbA2	EDTA-Blut	HPLC-UV/VIS
HbF	EDTA-Blut	HPLC-UV/VIS
HbX	EDTA-Blut	HPLC-UV/VIS
Hippursäure/Methylhippursäure	Urin	HPLC-UV/VIS
Vitamin A	Serum, EDTA-Plasma	HPLC-UV/VIS
Vitamin C	Lithium-Heparin-Plasma	HPLC-UV/VIS
Vitamin E	Serum, EDTA-Plasma	HPLC-UV/VIS

Untersuchungsart:

Durchflussszytometrie (inkl. Partikeleigenschaftsbestimmungen) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Großes Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale
Kleines Blutbild	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale
Retikulozyten	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale
Thrombozyten	EDTA-Blut	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale
Zellzahl	Körperflüssigkeiten, Ascites, Dialysat, Liquor, Punktat, Synovia	Partikelzählung, optisch-elektronisch Partikelgrößenbestimmung, elektronisch Bestimmung zytochemisch-zytometrischer Merkmale

Untersuchungsart:

Elektrochemische Untersuchungen ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Calcium (ionisiert)	Serum	ionenselektive Elektroden
Chlorid	Serum, Heparin-Plasma	ionenselektive Elektroden
Chlorid	Urin	ionenselektive Elektroden
Kalium	Urin	ionenselektive Elektroden
Kalium	Serum, Heparin-Plasma	ionenselektive Elektroden
Natrium	Urin	ionenselektive Elektroden
Natrium	Serum, Heparin-Plasma	ionenselektive Elektroden

Untersuchungsart:

Elektrophorese ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Eiweisselektrophorese	Serum	Kapillarelektrophorese
Alkalische Phosphatase-Isoenzyme	Serum	Agarose-Elektrophorese
CK-Isoenzyme	Serum	Agarose-Elektrophorese
HB-Differenzierung im alkalischen Milieu	EDTA-Blut	Agarose Elektrophorese
HB-Differenzierung im sauren Milieu	EDTA-Blut	Agarose-Elektrophorese
LDH-Isoenzyme	Serum	Agarose-Elektrophorese
Mucopolysaccharide	Urin	Agarose-Elektrophorese

Untersuchungsart:

Koagulometrie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
APC-Resistenz	Citratplasma	optisches Detektionsverfahren
Fibrinogen	Citratplasma	optisches Detektionsverfahren
Lupus Antikoagulans (Lupus	Citratplasma	optisches Detektionsverfahren
Quick	Citratplasma	optisches Detektionsverfahren
Quick	Citratplasma	mechanisches Detektionsverfahren
INR	Citratplasma	Berechnung
Thromboplastinzeit (aPTT)	Citratplasma	mechanisches Detektionsverfahren
Thrombinzeit	Citratplasma	optisches Detektionsverfahren
Thromboplastinzeit (aPTT)	Citratplasma	optisches Detektionsverfahren

Untersuchungsart:

Ligandenassays ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
17-Hydroxy(OH)-Progesteron	Serum	RIA
Adiponectin	Serum	ELISA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Adrenocorticotropes Hormon (ACTH)	EDTA-Plasma	CLIA
Aldosteron	Serum, EDTA-Plasma	CLIA
Alpha-Fetoprotein (AFP)	Serum	ECLIA
Amphetamine	Urin	CEDIA
Androstaniol Glucuronid	Serum	RIA
Androstendion	Serum	ECLIA
Anti-Müller-Hormon	Serum, Heparin-Plasma	ECLIA
Benzodiazepine	Urin	CEDIA
Benzoylgonin	Urin	CEDIA
Beta-2-Mikroglobulin	Serum, Urin, EDTA-, Citrat-, Heparin-Plasma	CLIA
Beta-Amyloid 1-40	Liquor	ELISA
Beta-Amyloid 1-42	Liquor	ELISA
Beta-CrossLaps (CTX)	Serum, EDTA-, Heparin-Plasma	ECLIA
Beta-hCG, freies	Serum	ECLIA
Biotin	Serum	ELISA
Buprenorphin	Urin	CEDIA
CA 50	Serum	RIA
Calcitonin	Serum, Heparin-Plasma	CLIA
Cannabinoide	Urin	CEDIA
Carbamazepin	Serum, EDTA-, Heparin-Plasma	KIMS
Ciclosporin A	EDTA-Blut	ECLIA
Cortisol	Serum, EDTA-, Heparin-Plasma	ECLIA
Cortisol	Urin	ECLIA
C-Peptid	Serum, EDTA-, Heparin-Plasma	ECLIA
Dehydroepiandrosteron-Sulfat (DHEA-S)	Serum, EDTA-, Heparin-Plasma	ECLIA
Diaminoxidase Aktivität	Serum	REA
Diaminoxidase Konzentration	Serum	ELISA
Digitoxin	Serum, EDTA-, Heparin-Plasma	ECLIA
Digoxin	Serum, EDTA-, Heparin-Plasma	ECLIA
Drogenscreening	Urin	CEDIA
EDDP	Urin	CEDIA
Erythropoetin (EPO)	Serum, Heparin-Plasma	CLIA
Ethylglucuronid	Urin	ELISA
Ferritin	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Follikelstimulierendes Hormon (FSH)	Serum, EDTA-, Heparin-Plasma	ECLIA
Folsäure in Erythrozyten	EDTA-Blut, Heparinblut	ECLIA
Gastrin	Serum	CLIA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Gentamicin	Serum , EDTA-, Heparin-Plasma	KIMS
Hämoglobin (immunologisch)	Faeces	ELISA
Hämoglobin-Haptogl.-Komplex (immunologisch)	Faeces	ELISA
hCG/ β -hCG	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Inhibin A (Dimer)	Serum	ELISA
Inhibin B	Serum	ELISA
Insulin	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Interleukin-2-Rezeptor, löslicher	Serum	CLIA
Leptin	Serum	RIA
Luteinisierendes Hormon (LH)	Serum, EDTA-, Heparin-Plasma	ECLIA
M2-PK	Faeces	ELISA
Methotrexat	Serum	EIA
Neopterin	Serum, EDTA-Plasma	ELISA
NT-pro BNP	Serum, EDTA-, Heparin-Plasma	ECLIA
Opiate	Urin	CEDIA
Ostase	Serum	CLIA
Osteocalcin	Serum, EDTA-, Heparin-Plasma	ECLIA
Östradiol	Serum, EDTA-, Heparin-Plasma	ECLIA
Östriol, freies	Serum	RIA
Östron	Serum	RIA
Pankreas-Elastase	Serum	ELISA
PAPP-A	Serum	ECLIA
Parathormon, intakt (PTH)	EDTA-Plasma, Serum	ECLIA
Parathormon-related Protein	EDTA-Plasma	RIA
Phencyclidin	Urin	CEDIA
Phenobarbital	Serum, EDTA-, Heparin-Plasma	KIMS
Phenytoin (Diphenylhydantoin)	Serum, EDTA-, Heparin-Plasma	KIMS
Phospho-Tau	Liquor	ELISA
PIGF	Serum	ECLIA
Plazenta-AP	Serum	ELISA
Procalcitonin (PCT)	Serum, EDTA-, Heparin-Plasma	ECLIA
Progesteron	Serum, EDTA-, Heparin-Plasma	ECLIA
Proinsulin, intaktes	Serum, EDTA-Plasma	ELISA
Prokollagen-III-Peptid	Serum	RIA
Prolactin	Serum, EDTA-, Heparin-Plasma	ECLIA
Protein S-100	Serum	ECLIA
Renin	EDTA-Plasma	CLIA
Sexual-Hormon-bindendes Globulin (SHBG)	Serum, Heparin-Plasma	ECLIA
sFlt-1	Serum	ECLIA
Squamous cell carcinoma (SCC)-Ag	Serum	ECLIA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Tacrolimus	EDTA-Blut	ECLIA
Tau-Protein	Liquor	ELISA
Testosteron	Serum, EDTA-, Heparin-Plasma	CLIA
Testosteron, freies	Serum	ELISA
Theophyllin	Serum, EDTA-, Heparin-Plasma	KIMS
Thrombin/Antithrombin-III-Komplex	Citratplasma	ELISA
Thymidinkinase (TK)	Serum, EDTA-Plasma	CLIA
Thyreoglobulin-Antikörper	Serum, EDTA-Plasma	ECLIA
Thyroxin, freies (FT4)	Serum, EDTA-, Heparin-Plasma	ECLIA
Tissue Polypeptide Antigen (TPA)	Serum	CLIA
Tissue polypeptide specific antigen (TPS)	Serum	RIA
Trijodthyronin, freies (FT3)	Serum, EDTA-, Heparin-Plasma	ECLIA
Troponin T	Serum, EDTA-, Heparin-Plasma	ECLIA
TSH basal	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
TSH Rezeptor Antikörper (TRAK)	Serum	ECLIA
Valproinsäure	Serum, EDTA-, Heparin-Plasma	EIA
Vancomycin	Serum, EDTA-, Heparin-Plasma	KIMS
1,25 Dihydroxy-Vitamin D	Serum, EDTA-, Heparin-Plasma	CLIA
Wachstumshormon (HGH)	Serum, EDTA-, Heparin-Plasma	ECLIA

Untersuchungsart:

Mikroskopie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Differentialblutbild	EDTA-Blut, Ausstriche	Hellfeldmikroskopie mit Anfärbung
Leukozyten	Punktat, Liquor	Hellfeldmikroskopie ohne Anfärbung (Kammerzählung)
Synovialanalyse	Punktat	Hellfeldmikroskopie ohne Anfärbung
Urinsediment	Urin	Hellfeldmikroskopie ohne Anfärbung

Untersuchungsart:

Osmometrie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Osmolalität	Serum, Urin	Kryoskopie

Untersuchungsart:

Sedimentationuntersuchungen ^[Flex A]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Blutkörperchensenkungs- geschwindigkeit	EDTA-Blut	kinetische Durchflussanalyse

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Alkalische Phosphatase	Serum, Heparin-Plasma	Photometrie
Alpha-Amylase	Serum, Heparin-Plasma	Photometrie
Alpha-Amylase, Pankreas	Serum, Heparin-Plasma	Photometrie
Ammoniak	EDTA-Plasma	Photometrie
Angiotensin-Converting-Enzyme (ACE)	Serum	Photometrie
Anti-Faktor Xa Aktivität (Rivaroxaban)	Citratplasma	Photometrie
Anti-Faktor Xa Aktivität (Apixaban)	Citratplasma	Photometrie
Anti-Faktor Xa (LMW-Heparin)	Citratplasma	Photometrie
Antithrombin	Citratplasma	Photometrie
Bilirubin, direkt	Serum, EDTA-, Heparin-Plasma	Photometrie
Bilirubin, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie
Bromid	Serum	Photometrie
C1-Esterase-Inhibitor (Aktivität)	Citratplasma	Photometrie
Calcium	Serum, Heparin-Plasma	Photometrie
Calcium	Urin	Photometrie
Cholesterin, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie
Cholinesterase (Pseudocholinesterase)	Serum, EDTA-, Heparin-Plasma	Photometrie
CK	Serum, EDTA-, Heparin-Plasma	Photometrie
CK-MB	Serum, EDTA-Plasma	Photometrie
Carboxyhämoglobin (CO-Hb)	Heparinblut, EDTA-Blut, Citrat-Blut, Kapillarblut	Photometrie
Creatinin	Urin	Photometrie
Creatinin , enzymatisch	Serum, EDTA-, Heparin-Plasma, Urin	Photometrie
Eisen	Serum, Heparin-Plasma	Photometrie
Eiweiß, gesamt	Serum, EDTA-, Heparin-Plasma	Photometrie
Ethanol	Serum, Urin, EDTA-Plasma	Photometrie

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fettsäuren, frei	Serum	Photometrie
Fructosamin	Serum, EDTA-, Heparin-Plasma	Photometrie
Gallensäuren, gesamt	Serum	Photometrie
Gamma-Hydroxybuttersäure	Urin	Photometrie
GGT	Serum, EDTA-, Heparin-Plasma	Photometrie
GLDH	Serum, EDTA-, Heparin-Plasma	Photometrie
Glucose	NaF-Citrat-Plasma, Serum, EDTA-, Heparin-Plasma, Hämolyt, Urin	Photometrie
Glucose	Liquor, Urin	Photometrie
GOT	Serum, EDTA-, Heparin-Plasma	Photometrie
GPT	Serum, EDTA-, Heparin-Plasma	Photometrie
Hämoglobin, frei	Heparin-Plasma, Serum	Photometrie
Harnsäure	Serum, Heparin-Plasma	Photometrie
Harnsäure	Urin	Photometrie
Harnstoff	Serum, EDTA-, Heparin-Plasma	Photometrie
Harnstoff	Urin	Photometrie
Harnstoff-N	Urin	Photometrie
HBDH	Serum, EDTA-, Heparin-Plasma	Photometrie
HDL-Cholesterin	Serum, EDTA-, Heparin-Plasma	Photometrie
Kupfer	Serum, Heparinplasma	Photometrie
Lactat	NaF-Plasma, EDTA-Plasma, Liquor	Photometrie
LDH	Serum, Heparin-Plasma	Photometrie
LDL-Cholesterin	Serum, EDTA-, Heparin-Plasma	Photometrie
Lipase	Serum, Heparin-Plasma	Photometrie
Lipoprotein (a)	Serum, EDTA-, Heparin-Plasma	Photometrie
Lithium	Serum, EDTA-, Heparinplasma	Photometrie
Magnesium	Serum, Heparin-Plasma	Photometrie
Magnesium	Urin	Photometrie
Methämoglobin	Heparinblut, EDTA-Blut, Citrat-Blut, Kapillarblut	Photometrie
N-Acetyl-Glucosaminidase	Urin	Photometrie
Oxalat	Urin	Photometrie
Phosphat, anorganisch	Serum, EDTA-, Heparin-Plasma	Photometrie
Phosphat, anorganisch	Urin	Photometrie
Protein C-Aktivität	Citratplasma	Photometrie
Saure Phosphatase, gesamt	Serum	Photometrie
Tobramycin	Serum, EDTA-, Citrat-, Heparin-Plasma	Photometrie
Triglyceride	Serum, EDTA-, Heparin-Plasma	Photometrie
Urinstatus	Urin	Photometrie
Zink	Serum, Heparin-Plasma	Photometrie

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Albumin	Serum, EDTA-, Heparin-Plasma, Liquor, Urin	Nephelometrie
Alpha 1-Mikroglobulin	Urin	Nephelometrie
Alpha-1-Antitrypsin	Serum	Nephelometrie
Apolipoprotein A-1	Serum	Nephelometrie
Apolipoprotein B	Serum	Nephelometrie
Coeruloplasmin	Serum, Heparin-Plasma	Nephelometrie
Cystatin C	Serum, Heparin-Plasma	Nephelometrie
Haptoglobin	Serum, Heparin-, EDTA-Plasma	Nephelometrie
Myoglobin	Serum	Nephelometrie
Proteinurie-Diagnostik	Urin	Nephelometrie
Serum-Amyloid-A (SAA)	Serum, Heparin-Plasma	Nephelometrie

Untersuchungsart:

Spektrometrie (Turbidimetrie/Immunturbidimetrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
CRP	Serum, EDTA-, Heparin-Plasma	Turbidimetrie
CRP high sensitive	Serum, EDTA-, Heparin-Plasma	Turbidimetrie
D-Dimer	Citratplasma	Immunturbidimetrie
Eiweiß	Urin, Liquor	Turbidimetrie
freies Protein S-Antigen	Citratplasma	Immunturbidimetrie
HbA1C	EDTA-Blut	Immunturbidimetrie
Transferrin	Serum, Heparin-Plasma	Turbidimetrie
Transferrin Rezeptor, löslicher (sTfR)	Serum, Heparin-Plasma	Turbidimetrie
vWF:Ag (Faktor VIII-assoziiertes Ag)	Citratplasma	Immunturbidimetrie
vWF Ac (Aktivität)	Citratplasma	Immunturbidimetrie

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Quecksilber	EDTA-Blut, Heparinblut, Urin	AAS
Zink	EDTA-Plasma	AAS

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Antimon	Urin	ICP-MS
Arsen	Urin, EDTA-, Heparin-Blut	ICP-MS
Barium	Urin	ICP-MS
Blei	Urin, EDTA-Blut, Heparinblut	ICP-MS
Cadmium	Urin, EDTA-, Heparin-Blut	ICP-MS
Chrom	EDTA-, Heparin-Plasma, Urin	ICP-MS
Jod	Urin, Serum	ICP-MS
Kobalt	Serum, Urin, EDTA-, Heparin-Blut	ICP-MS
Kupfer	Urin	ICP-MS
Mangan	EDTA-, Heparin-Plasma, Urin, Serum, Heparinblut, EDTA-Blut	ICP-MS
Molybdän	Urin, EDTA-, Heparin-Plasma	ICP-MS
Nickel	Urin, EDTA-Blut, Heparinblut Heparin-, EDTA-Plasma	ICP-MS
Selen	Urin, EDTA-Blut, Heparinblut, EDTA-Plasma, Heparinplasma, Serum	ICP-MS
Thallium	Serum, Urin	ICP-MS
Vanadium	Urin	ICP-MS
Zink	Urin	ICP-MS
Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Zinn	Urin	ICP-MS

Untersuchungsgebiet: Immunologie

Untersuchungsart:

Durchflusszytometrie ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Lymphozytendifferenzierung	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung
Zelluläre Allergiediagnostik	EDTA-Blut	zytochemisch-zytometrische Zellklassifizierung (FLOW CAST)
Lymphozytendifferenzierung und Aktivierungsmarker	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen
Bestimmung des Anteils TCR-γδ-positiver T-Zellen im peripheren	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen
Bestimmung der Leichtketten-Expression auf B-Zellen oder Plasmazellen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen
Immunphänotypisierung bei Non-Hodgkin-Lymphomen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Immunphänotypisierung bei chronisch-myeloischen Erkrankungen	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen
Immunphänotypisierung bei MDS und akuten Leukämien	Vollblut (EDTA, Heparin), Knochenmark	Immunphänotypisierung von hämatopoetischen Zellen
Immunphänotypisierung bei PNH	Vollblut (EDTA, Heparin)	Immunphänotypisierung von hämatopoetischen Zellen

Untersuchungsart:
Elektrophorese ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Paraprotein, monoklonale Gammopathie	Serum, Urin	Immunfixationselektrophorese
Oligoklonale Banden	Serum, Liquor	Isoelektrische Fokussierung
Proteine	Urin	Zonenelektrophorese (SDS-Gelelektrophorese)

Untersuchungsart:
Ligandenassays ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Acetylcholin-Rezeptor-Ak	Serum	Radiorezeptorassay
Cyclische citrulliniertes Peptid-Ak (CCP-Ak)	Serum	ECLIA
Eosinophiles cationisches Protein (ECP)	Serum	CLIA
C1q (C1C-C1q)-Ak	Serum	ELISA
Beta-2-Glycoprotein-Ak	Serum, Citrat-Plasma	ELISA
Cardiolipin-Ak	Serum, Citrat-Plasma	ELISA
Calcium-Kanal-Protein-Ak (P/Q-Typ)	Serum	Radioimmunpräzipitation
Doppelstrang-DNA (dsDNA)-Ak (IgG)	Serum	ELISA
Einzelstrang-DNA (ssDNA)-Ak	Serum	ELISA
Gangliosid-Ak	Serum	ELISA
Glutamat-Decarboxylase (GAD65)-Ak	Serum	ELISA
Histone-Ak (IgG)	Serum	ELISA
IA2-Ak	Serum	ELISA
Immunglobulin E	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Intrinsic-Faktor-Ak	Serum	ELISA
Insulin-Ak gegen Humaninsulin (IgG)	Serum	Radioimmunpräzipitation

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Leber-Nieren-Mikrosomen (LKM 1)-Ak	Serum	ELISA
MCV-Ak	Serum	ELISA
Mikrosomale Schilddrüsen-Ak (MAK, TPO-Ak)	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Mitochondriale-Ak, Subtyp 1 (IgG)	Serum, Citrat-Plasma	ELISA
Mitochondriale-Ak, Subtyp 1 (IgM)	Serum, Citrat-Plasma	ELISA
Mitochondriale-Ak, Subtyp 2	Serum	ELISA
Muskelspezifische Rezeptor-Tyrosinkinase-Ak	Serum	Radioimmunpräzipitation
Myeloperoxidase (MPO)-Ak (IgG)	Serum	ELISA
Parietalzellen-Ak	Serum	ELISA
PF4-Heparin-Ak	Serum	Lateral-Flow-Assay
PMSCL-Ak	Serum	ELISA
Proteinase 3-Ak (IgG)	Serum	ELISA
Saccharomyces-cerevisiae-Ak	Serum	ELISA
SLA/LP-Ak	Serum	ELISA
Basalmembran-Ak (glomerulär)	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
Gliadin DP-IgA und IgG-Ak	Serum, EDTA-, Heparin-Plasma	FEIA
Jo-1-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
Scl-70-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
Sm-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
SSA (Ro)-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
SSB (La)-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
Transglutaminase (Endomysium)-IgA-, IgG-Ak	Serum, EDTA-, Citrat-Plasma	FEIA
U1-snRNP-Ak	Serum, EDTA-, Citrat-, Heparin-Plasma	FEIA
Allergenspezifische IgE Ak	Serum	EAST (Enzym-Allergo-Sorbent-Test)
Mitochondriale-Ak, Subtyp 4	Serum	Immunoblot
Mitochondriale-Ak, Subtyp 9	Serum	Immunoblot
Zentromer-Ak (IgG)	Serum	ELISA
Zinktransporter-8-Ak (ZnT8)	Serum	ELISA

Untersuchungsart:

Mikroskopie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
ANCA (Granulozytenzytoplasma-Ak)	Serum	IFT
Antinukleäre-Ak (inkl. Subtypen)	Serum	IFT
Aquaporin 4-Ak	Serum, Liquor	IFT
Basalmembran-Ak (tubulär)	Serum	IFT
Doppelstrang-DNA (dsDNA)-Ak	Serum	IFT
DPPX-Ak (Anti-Dipeptidyl aminopeptidase-like protein 6)	Serum	IFT
Endomysium-Ak	Serum	IFT
Glatte Muskulatur-Ak (ASMA)	Serum	IFT
Glutamat-Rezeptor (TypNMDA)-Ak	Serum, Liquor	IFT
VGKC-Komplex (Typ CASPR2/Lgi1)-	Serum, Liquor	IFT
Herzmuskel-Ak	Serum	IFT
Inselzell-Ak (ICA)	Serum	IFT
Leber-Nieren-Mikrosomen (LKM)-2-Ak	Serum	IFT
Mitochondriale-Ak (AMA)	Serum	IFT
MOG-Ak (Myelin-Oligodendrozyten-Glykoprotein)	Serum	IFT
Nebennierenrinden-Ak	Serum	IFT
Neuronale Auto-Ak	Serum, Liquor	IFT
Phospholipase-A ₂ -Rezeptor (PLA ₂ R)-Ak	Serum	IFT
Skelettmuskel-Ak	Serum	IFT
Spermatozoen-Ak	Serum	IFT
Thrombospondin type-1 domain-containing protein 7A-Ak (THSD7A)	Serum	IFT

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Komplement-Gesamtaktivität (CH50)	Serum	Photometrie

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
C1-Esterase-Inhibitor (Proteinbestimmung)	Serum, Citrat-Plasma	Nephelometrie
C3-Komplement-Komponente	Serum	Nephelometrie
C4-Komplement-Komponente	Serum	Nephelometrie
Immunglobulin A	Serum, Liquor	Nephelometrie
Immunglobulin D	Serum	Nephelometrie
Immunglobulin G	Serum, Liquor	Nephelometrie
Immunglobulin G-Subklassen	Serum	Nephelometrie
Immunglobulin M	Serum, Liquor	Nephelometrie

Untersuchungsart:

Spektrometrie (Turbidimetrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Immunglobulin A (IgA)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie
Immunglobulin G (IgG)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie
Immunglobulin M (IgM)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie
Rheumafaktor (RF)	Serum, EDTA-, Heparin-Plasma	Turbidimetrie

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Achondroplasie (FGFR3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Alpha-Galaktosidase-A-Mangel (GLA) / Morbus Fabry	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Alpha-Thalassämie (HBA1, HBA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Amyloidpolyneuropathie, familiär (TTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Beta-Thalassämie (HBB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Ehlers-Danlos-Syndrom (TNXB) - Exon 32 bis 44	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Familiäres Mittelmeerfieber (MEFV, Exons 2, 3, 10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Geschlechtsdifferenzierung (SRY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
hereditäre Neuropathie mit Neigung zu Drucklähmungen (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
hereditäre Persistenz des fetalen Hämoglobin (HPFH)-Syndrom (HBG1, HBG2 -> nur Promotorbereiche)	EDTA-Blut, DNA, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Hypercholesterinämie (ApoB)	EDTA-Blut, DNA, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Hypochondroplasie (FGFR3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Lebersche hereditäre Optikusneuropathie (LHON)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
MTHFR-Mangel (A1298C)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Neoplasie, multiple endokrine Typ 2A (RET, Hotspots)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)
Pankreatitis, hereditär (SPINK1, PRSS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
Schwerhörigkeit, nicht-syndromale sensorineurale (GJB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
SHOX(-Defizienz)/Leri-Weill-Syndrom	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)
von Hippel Lindau Syndrom (VHL)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)
alpha-1-Antitrypsin-Mangel (SERPINA1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
Hyperlipoproteinämie Typ 3 (APOE)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
Irinotecan-Therapie (UGT1A1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
Morbus Meulengracht (UGT1A1)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
MTHFR-Mangel (MTHFR, C677T)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
Plasminogen-Aktivator-Inhibitor-1-Mangel (PAI1,4G/5G)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
Steatohepatitis, nicht-alkoholische (PNPLA3)	EDTA-Blut, DNA; DNA	Sequencing by Synthesis (Pyrosequenzierung)
adrenogenitales Syndrom (CYP21A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	DNA Sequenzierung (Sanger)
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
Achromatopsie (ACHM) (ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Acrocallosales Syndrom (GLI3, KIF7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Adenomatöse Polyposis (APC, MUTYH, NTHL1, POLD1, POLE)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Adipositas (ALMS1, APC2, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CARTPT, CCDC28B, CEP164, CEP19, CEP290, CUL4B, DYRK1B, GNAS, IFT172, IFT27, IFT74, KDM6A, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, NFIX, NR0B2, NSD1, NTRK2, PCSK1, PHF6, POMC, PPARG, PTEN, PYY, RAB23, SDCCAG8, SH2B1, SIM1, TMEM67, TRIM32, TTC8, UCP3, VPS13B, WDPCP)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Ahornsirupkrankheit (BCKDHA, BCKDHB, DBT, DLD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
akutes Leberversagen (ALDOB, BCS1L, DGUOK, DLD, FARS2, GALT, GBA, GFM1, HMGCL, IARS1, LARS1, LIPA, MPV17, MRPS16, NBAS, NPC1, NPC2, POLG, SCO1, SMPD1, SUCLG1, TRMU, TSFM, TWNK, TYMP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Albinismus (GPR143, LRMDA, MC1R, MITF, OCA2, SLC24A5, SLC45A2, TYR, TYRP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Arthrogrypose- Nierenfunktionsstörung-Cholestase- Syndrom (VIPAS39, VPS33B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ashkenazi Panel (ASPA, BLM, CFTR, ELP1, FANCC, GBA, HEXA, MCOLN1, SMPD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ataxie (ABCB7, ABHD12, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP1A3, ATP2B3, BEAN1, CA8, CACNA1A, CAPN1, COQ8A, CP, CWF19L1, CYP27A1, DARS2, DNMT1, ELOVL4, FGF14, FLVCR1, FXN, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MARS2, MRE11, NKX6-2, PDYN, PHYH, PMPCA, PNKP, PNPLA6, POLG, PRKCG, RARS2, RNF216, SACS, SCN2A, SETX, SIL1, SLC17A5, SLC1A3, SNX14, SPG7, SPTBN2, STUB1, SYNE1, TGM6, TMEM240, TTBK2, TTC19, TTPA, TWNK, VAMP1, VLDLR, VPS13D, WDR81)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ataxie, autosomal-dominant (AFG3L2, FGF14, ITPR1, KCNC3, PDYN, PRKCG, SPTBN2, TMEM240)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Ataxie, autosomal-rezessiv (ANO10, APTX, ATM, FXN, SETX, SIL1, SNX14, TTPA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Auditorische Neuropathie (AIFM1, DIAPH3, OTOF, PJKV)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Autosomal-dominante Polyzystische Nierenerkrankung (ADPKD) (ALG9, DNAJB11, GANAB, PKD1, PKD2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
autosomal-dominante nächtliche Frontallappenepilepsie (CHRNA2, CHRNA4, CHRN2, DEPDC5, KCNT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Autosomal-rezessive Polyzystische Nierenerkrankung (ARPKD) (DZIP1L, PKD1, PKHD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Bardet-Biedl-Syndrom (ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8orf37, CEP164, CEP19, CEP290, IFT172, IFT27, IFT74, LZTFL1, MKKS, MKS1, SCAPER, SDCCAG8, TMEM218, TRIM32, TTCE, WDRCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Bartter-/Gitelman-Syndrom (AP2S1, BSND, CASR, CFTR, CLCNKA, CLCNKB, CLDN10, CTNS, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3, SLC26A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
Benigne Neugeborenenkrämpfe (KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Benigne rezurrente intrahepatische Cholestase (ABCB11, ATP8B1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Birt-Hogg-Dube-Syndrom (FLCN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Bloom-Syndrom (BLM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
Brustkrebs (BRCA1, BRCA2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Burkitt-Lymphom (ARID1A, CCND3, GNA13, ID3, MYC, RHOA, SMARCA4, TCF3, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Charcot-Marie-Tooth-Krankheit (AARS1, ABHD12, AIFM1, ATL1, ATP1A1, ATP7A, BICD2, BSCL2, CNTNAP1, COA7, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, KIF5A, LITAF, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPZ, MTMR2, MYH14, NDRG1, NEFH, NEFL, PLEKHG5, PMP2, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCO2, SETX, SH3TC2, SIGMAR1, SLC12A6, SORD, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, WARS1, YARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
CHARGE-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Cholestatische Hepatopathie (ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP7B, ATP8B1, CFTR, CYP7B1, DCDC2, HSD3B7, MYO5B, NR1H4, SERPINA1, TJP2, UTP4, VIPAS39, VPS33B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Chondrodysplasia punctata (AGPS, ANOS1, ARSL, EBP, GNPAT, LBR, MGP, NSDHL, PEX5, PEX7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Chronisch progressive externe Ophthalmoplegie (ACADS, DGUOK, DNA2, MGME1, OPA1, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, SPG7, TK2, TOP3A, TWNK, TYMP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Chronische Neutrophilenleukämie (ASXL1, CALR, CBL, CSF3R, ELANE, NRAS, SETBP1, SRSF2, TET2, U2AF1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
chronisch-entzündlichen Darmerkrankungen (CED) (ADA, ADA2, ADAM17, AICDA, AP3B1, AP3D1, ATG16L1, BACH2, CASP10, CASP8, CD27, CD70, CTLA4, CYBA, CYBB, DKC1, EGFR, FADD, FAS, FASLG, G6PD, IL10RA, IL10RB, IL21, IL21R, IL2RA, ITK, KRAS, LRBA, LYST, MAGT1, MEFV, MVK, NCF1, NCF2, NCF4, NFAT5, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PIK3R1, PLCG2, PRF1, PRKCD, PSMB8, PSTPIP1, RBCK1, RNF31, RTEL1, SH2D1A, SLC29A3, STAT1, STAT3, STX11, TNFAIP3, TNFRSF11A, TNFRSF1A, TTC7A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Chylomikronämie (APOA5, APOC2, GPIHBP1, LMF1, LPL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Cobalamin (Vitamin B12)-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
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
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
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
Costello-Syndrom (BRAF, HRAS, KRAS, MAP2K1, NRAS, PTPN11, RAF1, SOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
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
Demenz (APOE, APP, CHCHD10, CHMP2B, CSF1R, FUS, GLA, GRN, MAPT, NOTCH3, PRNP, PSEN1, PSEN2, SQSTM1, TARDBP, TBK1, TREM2, TUBA4A, UBQLN2, VCP)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Diabetes mellitus, neonataler (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
Diabetes und Autoimmunität (AIRE, CTLA4, FOXP3, IL2RA, ITCH, JAK1, LRBA, SIRT1, STAT1, STAT3, STAT5A, STAT5B, TNFAIP3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Diamond-Blackfan-Syndrom (GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, TSR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
DICER1-assoziiertes Tumorsyndrom (DICER1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Diffus großzelliges B-Zell-Lymphom (ARID1A, BCL2, BCL6, CARD11, CREBBP, EZH2, FOXO1, GNA13, KMT2D, MEF2B, MYD88, PCLO, PIM1, TET2, TNFRSF14, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Distale Myopathie (ACTA1, ACTN2, ADSS1, ANO5, CAV3, COL12A1, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, LRP12, MATR3, MYH14, MYH7, MYOT, NEB, PABPN1, RYR1, SLC5A7, SQSTM1, TCAP, TIA1, TMD2, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Duchenne Muskeldystrophie (DMD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Dysostose mit vorwiegend vertebraler und kostaler Beteiligung (BMPER, COG1, DLL3, FGFR3, GDF3, GDF6, HES7, LFNG, MEOX1, MESP2, MNX1, NKX3-2, RIPPLY2, SNRPB, TBX6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Dysostosen (ABCC9, ALX1, ALX3, ALX4, BMPER, CDC45, CDC6, CDT1, CILK1, DHODH, DLL3, EDN1, EDNRA, EFN1B, EFTUD2, EIF4A3, EVC, EVC2, GDF3, GDF6, GMNN, GNAI3, HAAO, HES7, KAT6B, KYNU, LFNG, LMX1B, MEOX1, MESP2, MNX1, MYO18B, NKX3-2, OFD1, ORC1, ORC4, ORC6, PLCB4, POLR1A, POLR1C, POLR1D, RIPPLY2, SF3B4, SH3PXD2B, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Dysplasien der langen Knochen und/oder der Wirbelsäule (ACAN, ACP5, AIFM1, B3GALT6, B3GAT3, BGN, BPNT2, CANT1, CCN6, CFAP410, CHST3, COL10A1, COL11A1, COL11A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, DDR2, DDRGK1, DNAJC21, DYM, EFL1, EIF2AK3, EXTL3, FLNB, FN1, GPX4, HSPA9, HSPG2, IARS2, INPPL1, KIF22, LONP1, LTBP3, MAP3K7, MATN3, MMP13, MMP9, NANS, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, PTH1R, RAB33B, RMRP, RSPRY1, RUNX2, SBDS, SLC26A2, SLC35D1, SLC39A13, SMARCAL1, SRP54, TMEM165, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, XYLT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Dysplasien mit Verkürzung der langen Knochen (akro-meso-rhizomele Dysplasien) (ADAMTS10, ADAMTS17, ADAMTSL2, BMPR1B, COL11A1, COL11A2, DVL1, DVL3, EXT1, FBN1, FGFR2, FGFR3, FLNA, FZD2, GDF5, GPC6, GSC, IFT122, IFT140, IFT43, IHH, LIFR, LRP4, LTBP2, LTBP3, MAB21L2, MIR140, NPR2, NXN, PDE4D, PRKAR1A, ROR2, SHOX, SLC26A2, SLC05A1, SMAD4, SOX9, SULF1, TRPS1, WDR35, WNT5A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Dystonie (ADCY5, ANO3, ATP1A3, COL6A3, DNAJC12, FTL, GCH1, GNAL, GNAO1, HPCA, KMT2B, MECR, PLA2G6, PRKRA, RELN, SGCE, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ehlers-Danlos-Syndrom (ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Endokriner Kleinwuchs (ACAN, FGFR3, GH1, GHR, GHRHR, GHSR, HESX1, IGF1, IGF1R, IGFALS, IHH, LHX3, LHX4, NPR2, OTX2, POU1F1, PROP1, SHOX, SOX3, STAT5B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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, TGMF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Epilepsie, Ataxie, Sensorineurale Taubheit und Tubulopathie Syndrom (KCNJ10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Epilepsie, fokale (CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1, LGI1, NPRL2, NPRL3, RELN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Epilepsie, metabolische (ABAT, ABCD1, ACY1, ADK, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, AUH, BCKDK, BTD, CAD, CLN3, CLN5, CLN6, CLN8, CNNM2, COQ4, COQ8A, CPT1A, CPT2, CTSD, CTSF, D2HGDH, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GALC, GAMT, GATM, GBA, GCDH, GLDC, GLUL, GPHN, GRN, HEXA, HEXB, HLCS, HPD, ITPA, L2HGDH, LIAS, MDH2, MFSD8, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Epileptische Enzephalopathie (AARS1, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, ARHGEF9, ARX, ATP6V1A, ATRX, BRAT1, BTBD, CACNA1A, CACNA1E, CACNA2D2, CACNB4, CDK19, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CNNM2, CNPY3, CNTNAP2, CPA6, CYFIP2, DENND5A, DEPDC5, DNM1, DOCK7, EEF1A2, EHMT1, FOXG1, FRRS1L, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GLDC, GNAO1, GPHN, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HCN2, HNRNPU, IQSEC2, ITPA, KCNA2, KCNB1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, LGI1, MBD5, MDH2, MECP2, MEF2C, MOCS1, MOCS2, NEXMIF, NRXN1, PCDH19, PHF6, PIGA, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PURA, RHOBTB2, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SEMA6B, SETD1A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SPTAN1, STAMBP, STXB1, 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
Episodische Ataxie (CACNA1A, CACNB4, KCNA1, PNKD, PRRT2, SCN2A, SLC1A3, SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Erblicher Brust-und Eierstockkrebs (BRCA1, BRCA2, CHEK2, PALB2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Erblicher Brust-und Eierstockkrebs (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MRE11, NBN, PALB2, RAD51D, STK11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Erythrozytose, familiäre 1 (EPOR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Exsudative Vitreoretinopathie (inkl. Wagner- und Norrie-Syndrom) (CAPN5, CTNNA1, 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
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Familiäres Silver-Russell-Syndrom (HMG2, IGF2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
FAMMM Syndrom (BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Fanconi-Anämie (BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, PALB2, SLX4, UBE2T, XRCC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Fokal segmentale Glomerulosklerose (ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAT1, FN1, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, LRP2, MAGI2, MT-TL1, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCA1, TBC1D8B, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5, YRDC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Follikuläres Lymphom (BCL2, CREBBP, EZH2, KMT2D, MEF2B, TNFRSF14, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Fructoseintoleranz (ALDOB)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Galaktosämie (GALE, GALK1, GALT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Gastrointestinale Stromatumoren (KIT, NF1, PDGFRA, SDHA, SDHB, SDHC, SDHD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Gefleckte-Retina Syndrom (CHM, EFEMP1, PLA2G5, PRPH2, RDH5, RHO, RLBP1, RPE65, RS1, VPS13B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFS+) (GABRG2, SCN1A, SCN1B, SCN2A, STX1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Glanzmann-Thrombasthenia (ITGA2B, ITGB3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Gliedergürteldystrophie (ALG2, ANO5, BVES, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DMD, DNAJB6, DOK7, DPM3, DYSF, FKRP, FKTN, FLNC, GAA, GMPPB, HNRNPDL, LAMA2, LIMS2, LMNA, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RYR1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VCP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Gliom (BRCA2, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Glukokortikoid-Mangel (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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Glukoneogenese-Störung (CA5A, FBP1, G6PC, PC, PCK1, PCK2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Glykogenspeicherkrankheit (AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Glykosylierungs-Defekt Typ 2 (ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, FCSK, FUT8, GANAB, MAN1B1, MAN2B2, MGAT2, MOGS, PRKCSH, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SLC9A7, ST3GAL3, TMEM165, TMEM199, VMA21)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Glykosylierungs-Defekt, kongenitaler (ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, GMPPA, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PGM3, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, ST3GAL3, STT3A, STT3B, TMEM165, TMEM199, TUSC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Gorlin-Goltz-Syndrom (PTCH1, PTCH2, SUFU)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Haarzelleukämie (BRAF, CDKN1B, KLF2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hamartomatöse Polyposis (BMPR1A, PTEN, SMAD4, STK11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hämochromatose (BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TFR2, TMPRSS6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hämophagozytische Lymphohistiozytose (PRF1, SH2D1A, STX11, STXBP2, UNC13D, XIAP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hemiplegische Migräne, familiäre (ATP1A2, CACNA1A, NOTCH3, SCN1A)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hepatozelluläres Karzinom (ABCB11, AGL, APC, CDKN2A, G6PC, HMBS, PPOX, PRKAR1A, SPRTN)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hereditäre hämorrhagische Teleangiektasie (ACVRL1, ENG, GDF2, SMAD4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hereditäre motorisch-sensorische Neuropathien (axonale) (AARS1, GARS1, GDAP1, GJB1, HINT1, HSPB1, KIF1B, MFN2, MME, MPZ, NEFH, NEFL, RAB7A, TRPV4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hereditäre motorisch-sensorische Neuropathien (demyelinisierend) (EGR2, FGD4, FIG4, GDAP1, GJB1, LITAF, MPZ, MTMR2, NEFL, PMP22, PRX, SBF2, SH3TC2, TTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hereditäre Lymphödeme (ADAMTS3, CCBE1, CELSR1, EPHB4, FAT4, FLT4, FOXC2, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, VEGFC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hereditäre motorisch-sensorische Neuropathie (AARS1, ABHD12, AIFM1, ATL1, ATP1A1, ATP7A, BICD2, BSCL2, CNTNAP1, COA7, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, KIF5A, LITAF, LRSAM1, MARS1, MCM3AP, MFN2, MME, MORC2, MPZ, MTMR2, MYH14, NDRG1, NEFH, NEFL, PLEKHG5, PMP2, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCO2, SETX, SH3TC2, SIGMAR1, SLC12A6, SORD, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, WARS1, YARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hereditäre sensorische und autonome Neuropathie (ATL1, ATL3, DNMT1, ELP1, FLVCR1, KIF1A, NGF, NTRK1, PRDM12, RAB7A, RETREG1, SCN10A, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hereditäres nicht-polypöses kolorektales Karzinom (EPCAM, MLH1, MSH2, MSH6, PMS2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Heterotaxie (ACVR2B, ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP52, CFAP53, CFC1, CRELD1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXH1, FOXJ1, GAS2L2, GAS8, GDF1, HYDIN, LMLN2, LRRC56, LRRC6, MMP21, MNS1, NFATC1, NKX2-5, NME8, NODAL, NPHP4, OFD1, PKD1L1, RSPH1, RSPH3, RSPH4A, RSPH9, SMAD2, SPAG1, TTC25, ZIC3, ZMYND10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hodgkin Lymphome, hereditär (DICER1, POT1, PTPN11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Holoprosenzephalie (CDON, CENPF, DHCR7, DISP1, FGF8, FGFR1, GLI2, KMT2D, PTCH1, RAD21, SHH, SIX3, SMC1A, SMC3, STAG2, STIL, TGIF1, ZIC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hornhautdystrophie (AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCN, FOXE3, GSN, KRT12, KRT3, LOXHD1, MIR184, OVOL2, PAX6, PIKFYVE, PRDM5, 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
Hörverlust bei Erwachsenen (COCH, COL11A2, EYA4, KCNQ4, MIR96, SLC17A8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Hyperammonämie (ACAD9, ACADVL, ACAT1, ALDH18A1, ARG1, ASL, ASS1, AUH, CA5A, CPS1, GLUD1, GLUL, HADH, HADHA, HADHB, HLCS, HMGCL, IVD, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMADHC, MMUT, MRPS22, NAGS, OAT, OTC, OXCT1, PC, PCCA, PCCB, SLC25A13, SLC25A15, SLC7A7, TAZ, TMEM70)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Hyperekplexie (HKPX) (GLRA1, GLRB, SLC6A5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hyper-IgE-Syndrom (DOCK8, DSG1, DSP, PGM3, SPINK5, STAT3, TYK2, ZNF341)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
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
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
Hypertriglyzeridämie (APOA5, APOC2, APOC3, APOE, GPD1, GPIHBP1, LIPA, LMF1, LPL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hypertrophe Kardiomyopathie (ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, FHL1, FLNC, FXN, GAA, GLA, HRAS, JPH2, KLF10, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RASA2, RIT1, RRAS, RYR2, SLC25A4, SOS1, SOS2, TAZ, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Hypogonadotroper Hypogonadismus (ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GALT, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SLC29A3, SPRY4, TAC3, TACR3, TCF12, WDR44)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hypomagnesiämie (ATP1A1, BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, EGFR, FAM111A, FXRD2, HNF1B, KCNA1, KCNJ10, MT-TI, PCBD1, SARS2, SLC12A3, TRPM6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Hypopituitarismus (ARNT2, CDON, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RBM28, SOX2, SOX3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ichthyose und verwandte Keratinisierungsstörungen (AAGAB, ABCA12, ABHD5, ALDH3A2, ALMS1, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, ATP2C1, CARD14, CASP14, CDSN, CERS3, CLDN1, CSTA, CTSC, CYP4F22, DSG1, DSP, EBP, ELOVL4, ENPP1, ERCC2, ERCC3, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2H5, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LIPN, LORICRIN, MBTPS2, MPLKIP, NIPAL4, NSDHL, OSMR, PEX7, PHYH, PNPLA1, POMP, RHBDF2, SDR9C7, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, ST14, STS, SUMF1, TAT, TGM1, TGM5, TRPV3, VPS33B, WNT10A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Immundefekt, kombinierter schwerer (ADA, AK2, CD247, CD3D, CD3E, CORO1A, DCLRE1C, DOCK8, IL2RG, IL7R, JAK3, PRKDC, PTPRC, RAG1, RAG2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Insulin-Resistenz (AGPAT2, AKT1, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, FBN1, INSR, LIPE, LMNA, LMNB2, MT-TL1, NSMCE2, PCNT, PIK3R1, PLIN1, POC1A, POLD1, PPARG, PPP1R3A, SH2B1, TBC1D4, TCF7L2, WRN, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Intrahepatische Cholestase bei Schwangerschaft (ABCB11, ABCB4, ABCC2, ATP8B1, NR1H4, TJP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Jervell-Lange-Nielsen-Syndrom (KCNE1, KCNQ1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Jeune-Syndrom (CEP120, CFAP410, CILK1, DPH1, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, INTU, KIAA0586, KIAA0753, LBR, NEK1, TCTEX1D2, TRAF3IP1, TTC21B, WDR19, WDR34, WDR35, WDR60)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Joubert-Syndrom (AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CEP55, CPLANE1, CSPP1, FAM149B1, GLI3, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, POC1B, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Kallmann-Syndrom (ANOS1, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SPRY4, TAC3, TACR3, TCF12, WDR11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kardio-fazio-kutanes Syndrom (BRAF, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, SHOC2, SOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Kardiomyopathie, dilatative (ABCC9, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EMD, EYA4, FHL1, FKTN, GLA, HFE, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEXN, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, SCN5A, SDHA, SGCD, TAZ, TBX20, TCAP, TMPO, TNNC1, TNNI3, TNNT2,	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kardiomyopathie-Panel erweitert (ABCC9, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, BRAF, CALR3, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FKTN, FLNC, FXN, GAA, GLA, HFE, HRAS, ILK, JPH2, JUP, KLF10, KRAS, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NKX2-5, NNT, NRAS, NSD1, OBSCN, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RASA2, RBM20, RIT1, RPS6KA3, RRAS, RYR2, SCN5A, SDHA, SGCD, SLC25A4, SOS1, SOS2, TAZ, TBX20, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Katarakt (ABCA3, ABHD12, AGK, AKR1E2, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, EYA1, FAM126A, FAR1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GJA3, GJA8, GNPAT, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, NHS, P3H2, PAX6, PEX7, PITX3, PXDN, RAB3GAP1, RNLS, SIL1, SIPA1L3, SLC16A12, SLC33A1, TBC1D20, TDRD7, TMEM114, UNC45B, VIM, VSX2, WRN, XYLT2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Ketogenese-Störung (ACADM, ACADVL, HADHA, HADHB, 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
Ketolyse-Störung (ACAD9, ACAT1, ACSF3, AUH, BAAT, BCAT1, BCAT2, BCKDHA, BCKDHB, BCS1L, COQ9, DBT, DLD, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, G6PC, GLYCTK, GSS, GYS2, HADHB, HIBADH, HIBCH, HMGCL, HSD17B10, IVD, MCCC1, MCCC2, MLYCD, MMAA, MMAB, MMADHC, MMUT, OXCT1, PCCA, PCCB, PYGL, SLC16A1, SLC37A4, SUCLA2,	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Knochenmarkdysfunktions-Syndrom (AP3B1, BRCA2, BRIP1, CSF3R, CXCR4, DKC1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, MPL, NHP2, NOP10, PALB2, RAB27A, RAC2, RBM8A, RMRP, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS7, RTEL1, SBDS, SLC37A4, SLX4, SRP72, TAZ, TERC, TERT, TINF2, USB1, VPS13B, WAS, WIPF1,	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kolorektales Karzinom (APC, BMPR1A, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TGFB2, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kongenitale Anomalien der Nieren und ableitenden Harnwege (ACE, AGT, AGTR1, ANOS1, BICC1, BMP4, CDC5L, CHD1L, CHRNA3, CRKL, DSTYK, EYA1, FANCB, FGF20, FOXP1, FRAS1, FREM1, FREM2, GATA3, GDNF, GLIS3, GREB1L, GREM1, GRIP1, HNF1B, ITGA8, KIF14, LHX1, LIFR, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SALL4, SIX1, SIX2, SIX5, SLIT2, SOX17, SRGAP1, TBC1D1, TBX18, TBX6, TRAP1, UMOD, UPK3A, WNT4, WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kongenitale Fibrose der äußeren Augenmuskeln (COL25A1, KIF21A, PHOX2A, TUBB3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Kongenitale Muskeldystrophie (ACTA1, ANO5, B3GALNT2, B4GAT1, BIN1, CAPN3, CAVIN1, CCDC78, CFL2, CHKB, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNM2, DYSF, FHL1, FKRP, FKTN, GBE1, GMPPB, INPP5K, KBTBD13, KLHL40, KLHL41, LAMA2, LARGE1, LMNA, MEGF10, MTM1, NEB, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, RYR1, SELENON, TCAP, TNNT1, TPM2, TPM3, TRIM32, TRIP4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Kongenitales Myasthenes Syndrom (AGRN, CHAT, CHRNA1, CHRN1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, MYO9A, RAPSIN, SLC18A3, SLC5A7, VAMP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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, TGFB1, TGFB2, 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
Kutanes T-Zell-Lymphom (BRD9, CHD3, CREBBP, KMT2D, PLCG1, SMARCA4, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Lebersche kongenitale Amaurose (LCA) (ACBD5, ADAMTS18, AIPL1, ALMS1, BBS2, BBS4, CABP4, CCT2, CEP290, CRB1, CRX, DHX38, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MPDZ, MYO7A, NMNAT1, OTX2, POC1B, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Lebersche hereditäre Optikusneuropathie (MT-ND1, MT-ND4, MT-ND6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, akute myeloische (ASXL1, CCND3, CEBPA, DNMT3A, FLT3, GATA2, IDH1, IDH2, KIT, KMT2A, KRAS, NPM1, NRAS, RUNX1, TET2, TP53, WT1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, atypische chronische myeloische (ASXL1, CALR, CBL, CSF3R, ETNK1, EZH2, JAK2, KRAS, MPL, NRAS, RUNX1, SETBP1, SRSF2, TET2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, B-Zell akute lymphatische (CREBBP, CRLF2, FLT3, IKZF1, NT5C2, PRDM9, SETD2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, chronische myeloische (ABL1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Leukämie, chronische myelomonozytäre (ASXL1, BCOR, CBL, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KRAS, NF1, NPM1, NRAS, PHF6, RUNX1, SETBP1, SF3B1, SRSF2, TET2, U2AF1, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, juvenile myelomonozytische (ARHGAP26, CBL, KRAS, NRAS, PTEN, PTPN11)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, Ph-ähnliche akute lymphatische (ARID2, ASXL1, BLNK, CHD2, CRLF2, DGKH, EPOR, IKZF1, IL2RB, ITPKB, JAK1, JAK2, KRAS, LRP1B, LYN, NRAS, NTRK3, PDGFRA, PTK2B, PTPN11, TP53, TVK2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, T-Zell akute lymphatische (CDKN2A, DNMT3A, FBXW7, FLT3, IDH1, IDH2, IL7R, JAK1, JAK3, KRAS, NOTCH1, NRAS, PHF6, PTEN, RUNX1, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Leukämie, chronische lymphatische (ATM, BCL2, BIRC3, BRAF, BTK, DDX3X, EGR2, FBXW7, KRAS, MED12, MYD88, NFKBIE, NOTCH1, NRAS, PLCG2, POT1, SAMHD1, SETD2, SF3B1, TP53, XPO1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Leukoenzephalopathie (AARS1, ABCD1, ACOX1, ADAR, AIMP1, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, CSF1R, CTC1, DARS1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FAM126A, GALC, GFAP, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, L2HGDH, MLC1, NAXE, NKX6-2, NOTCH3, PLAA, PLP1, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SLC16A2, SNORD118, SOX10, SUMF1, TMEM106B, TREX1, TUBB4A, UFM1, VPS11, ZNHIT3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Linksventrikuläre Noncompaction Kardiomyopathie (ACTC1, ACTN2, DTNA, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, NNT, NSD1, PRDM16, RPS6KA3, SCN5A, TAZ, TNNI3, TNNT2, TPM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Linsenluxation (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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Lissenzephalie (ADGRG1, ARX, CCND2, CRADD, CRPPA, DCX, DYNC1H1, FKR1, FKTN, KATNB1, KIF2A, LAMB1, LAMC3, LARGE1, MACF1, PAFAH1B1, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RELN, RTTN, RXYLT1, TMTC3, TUBA1A, TUBA8, TUBB2B, WDR62)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Long-QT-Syndrom (AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRPM4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Lymphozyten-Leukämie, große granuläre (STAT3, STAT5B)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Lysosomale Speichererkrankungen (AGA, ARSA, ARSB, ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CSTB, CTNS, CTSB, CTSD, CTSF, CTSK, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GRN, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Magenkarzinom (APC, CDH1, CTNNA1, EPCAM, MAP3K6, MLH1, MSH2, MSH6, PMS2, SMAD4, STK11, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Makrozephalie (ABCC9, AKT1, AKT2, AKT3, AMER1, APC2, ASPA, ASXL2, BRAF, BRWD3, CCDC88C, CCND2, CDKN1C, CHD3, CHD4, CHD8, CRADD, CUL4B, DHCR24, DNMT3A, EED, EZH2, FIBP, GCDH, GFAP, GLI3, GPC3, GRIA3, H1-4, HEPACAM, HERC1, HRAS, HUWE1, IGF2, KIF7, KPTN, KRAS, L1CAM, MAP2K2, MED12, MLC1, MPDZ, MTOR, NFIB, NFIX, NRAS, NSD1, OFD1, PAK1, PHF6, PIGA, PIGN, PIGT, PIK3CA, PIK3R2, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RIN2, RNF125, RNF135, SEC24D, SETD2, SNX10, SOS1, STRADA, SUZ12, SYN1, TBC1D7, TCF20, UPF3B, ZBTB20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Männliche Infertilität (AMH, AMHR2, ANOS1, AR, ARMC2, AURKC, BNC2, CATSPER1, CCDC39, CFAP251, CFAP43, CFAP44, CFAP47, CFAP65, CFAP69, CFAP70, CFTR, CYP11B1, CYP17A1, CYP21A2, DMRT1, DNAH1, DNAH17, DNAH2, DNAH5, DNAH6, DNAH8, DNAH9, DNAJB13, DPY19L2, FANCM, FGF8, FGFR1, FSHB, FSHR, FSIP2, GNRH1, GNRHR, HSD17B3, HSD3B2, INSL3, KISS1, KISS1R, KLHL10, LHB, LHCGR, LRRC6, M1AP, MAMLD1, NANOS1, NR0B1, NR5A1, NSMF, PLCZ1, PMFBP1, PPP2R3C, PROK2, PROKR2, QRIC2, RSPO1, SEMA3A, SEPTIN12, SLC26A8, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPEF2, SRD5A2, SRY, SUN5, SYCP2, SYCP3, TACR3, TEX11, TEX14, TEX15, TRIM37, TTC21A, TTC29, USP26, WDR11, XRCC2)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mantellzell-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
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
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, ZDHHC9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Marginalzonen-Lymphom (KLF2, NOTCH1, NOTCH2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mastozytose, systemische (ASXL1, CBL, EZH2, IDH1, IDH2, JAK2, KIT, KRAS, NRAS, RUNX1, SRSF2, TET2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
McCune-Albright-Syndrom (GNAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Meckel-Gruber-Syndrom (AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CEP55, CPLANE1, CSPP1, GLI3, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, POC1B, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Medulloblastom (APC, BRCA2, MSH2, MSH6, PALB2, PMS2, PTCH1, SUFU, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mentale Retardierung - männlich (ARX, ATRX, CUL4B, GPC3, HUWE1, IL1RAPL1, IQSEC2, KDM5C, LAS1L, MAOA, MECP2, MED12, OPHN1, PHF6, PQBP1, PTEN, RPS6KA3, SLC16A2, SLC2A1, SLC6A8, SLC9A6, STXBP1, TCF4, ZC4H2, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mentale Retardierung - weiblich (CDKL5, CLCN4, EHMT1, FOXG1, GDI1, HNRNP2, IL1RAPL1, MECP2, MEF2C, PCDH19, PTEN, SHANK3, SLC2A1, SLC6A8, SLC9A6, STXBP1, TCF4, THOC2, ZEB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Metaphysäre Dyplasie (COL10A1, MMP13, MMP9, POP1, PTH1R, RMRP, RUNX2, SBDS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mikroangiopathie (APP, COL4A1, COL4A2, CTSA, FOXC1, GLA, HTRA1, NOTCH3, SNORD118, TREX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mikrophthalmie/Anophthalmie (ABCB6, ALDH1A3, ATOH7, BCOR, BMP4, C12orf57, CHD7, FOXE3, FREM1, GDF3, GDF6, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, OTX2, PAX6, PORCN, PRSS56, RARB, RAX, RBP4, SALL2, SHH, SIX6, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mikrozephalie (ADARB1, ADGRG1, AGMO, ANKLE2, ANKRD11, AP4M1, ARCN1, ARFGEF2, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATR, ATRIP, ATRX, BUB1B, CARS1, CASK, CDC6, CDK5RAP2, CDK6, CDKL5, CDON, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP164, CEP63, CHMP1A, CIT, CLN5, CLN6, CLN8, COG6, CREBBP, CRIPT, CTSD, CTSF, DCX, DDX11, DHCR7, DIAPH1, DNAJC5, DNM1L, DONSON, DYM, DYNC1H1, DYNC1I2, EFTUD2, EP300, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, EXOSC3, FKR1, FKTN, FLNA, FOXG1, GLI2, GRN, HDAC8, IER3IP1, IGF1, IGF1R, KAT6A, KATNB1, KCTD7, KIF11, KIF14, KIF2A, KIF5C, KNL1, LARGE1, LIG4, MCPH1, MECP2, MED17, MEF2C, MFSD2A, MFSD8, MIR17HG, MSMD1, MTHFS, MYCN, NAA10, NDE1, NIN, NIPBL, NR2E1, ORC1, ORC4, ORC6, PAFAH1B1, PARS2, PCNT, PHC1, PHF6, PHGDH, PIEZO2, PLAA, PLK4, PNKP, POMGNT1, POMT1, POMT2, PPP1R15B, PPT1, PQBP1, PRUNE1	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
PSAT1, PTCH1, PUS7, RAB18, RAD21, RARS2, RBBP8, RELN, RNU4ATAC, RTTN, SASS6, SHH, SIX3, SLC1A4, SLC25A19, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMPD4, SOX11, STAMBP, STIL, TCF4, TGIF1, TMX2, TPP1, TRAIP, TRAPPC9, TRIO, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, UBR1, VARS1, VRK1, WDR4, WDR62, XIRP1, XRCC4, ZEB2, ZIC2, ZNF335)		

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Mitochondriale DNA (MT-ATP6, MT-ATP8, MT-CO1, MT- CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT- ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT- TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT- TS1, MT-TS2, MT-TT, MT-TV, MT- TW, MT-TY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mito-Nuklear (AARS2, ABAT, ABCB7, ACAD9, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, AK2, ALDH18A1, APTX, ATAD3A, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12orf65, C19orf12, CARS2, CHCHD10, CHKB, CLPB, CLPP, COA3, COA5, COA8, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6B1, COX7A1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DBT, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, ELAC2, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, FXN, GARS1, GDAP1, GFER, GFM1, GFM2, GLRX5, GLUD1, GTPBP3, HAAO, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HMGCL, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH3A, ISCA2, ISCU, KARS1 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mito-Nuklear Fortsetzung KIF5A, KYNU, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM4, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MICU2, MIPEP, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MTFMT, MTO1, MTPAP, NADK2, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NMT1, NSUN3, NUBPL, OPA1, OPA3, PANK2, PARS2, PC, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PLA2G6, PMPCA, PNPLA2, PNPLA8, PNPT1, POLG, POLG2, PPA2, PPARG, PTRH2, PUS1, QRSL1, RARS2, RMND1, RMRP, RNASEH1 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mito-Nuklear Fortsetzung RRM2B, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC33A1, SLC39A8, SLC6A8, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TK2, TMEM126A, TMEM70, TPK1, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCRB, UQCRC2, 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
Mitochondriopathien (mito- chondriale und nukleäre Gene) (AARS2, ABAT, ABCB7, ACAD9, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, AK2, ALDH18A1, APTX, ATAD3A, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12orf65, C19orf12, CARS2, CHCHD10, CHKB, CLPB, CLPP, COA3, COA5, COA8, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6B1, COX7A1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DBT, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNAJC3, DNM1L, EARS2, ECHS1, ELAC2, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, FXN, GARS1, GDAP1, GFER, GFM1 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mitochondriopathien (mito- chondriale und nukleäre Gene) (Fortsetzung GFM2, GLRX5, GLUD1, GTPBP3, HAAO, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HMGCL, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH3A, ISCA2, ISCU, KARS1, KIF5A, KYNU, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM4, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MICU2, MIPPEP, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MT- ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT- ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT- RNR2, MT-TA, MT-TC, MT-TD, MT- TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT- TS1, MT-TS2, MT-TT, MT-TV, MT- TW, MT-TY, MTFMT, MTO1, MTPAP, NADK2, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2 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

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Mitochondriopathien (mitochondriale und nukleäre Gene) (Fortsetzung NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAB1, NDUFAB2, NDUFAB3, NDUFAB4, NDUFAB5, NDUFAB6, NDUFAB10, NDUFAB11, NDUFAB3, NDUFAB8, NDUFAB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFX1, 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	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
SLC6A8, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TK2, TMEM126A, TMEM70, TPK1, TRAK1, TRIT1, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCRB, UQCRC2, UQCRCF1, UQCRCQ, VARS2, WARS2, WFS1, YARS2, YME1L1)		
MNGIE-Syndrom (MGME1, POLG, RRM2B, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
MODY-Diabetes (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
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
Monogener Diabetes (ABCC8, AGPAT2, AIRE, ALMS1, APPL1, BLK, BSCL2, CAV1, CAVIN1, CEL, CIDEA, CISD2, CTLA4, CYC1, DCAF17, DNAJC3, DUT, DYRK1B, EIF2AK3, EIF2S3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, ITCH, KCNJ11, KLF11, LMNA, LMNB2, LRBA, MNX1, MT-CO1, MT- CO3, MT-ND1, MT-ND4, MT-ND6, MT-RNR1, MT-RNR2, MT-TE, MT-TI, MT-TK, MT-TL1, MT-TR, MT-TS1, NEUROD1, NEUROG3, NKX2-2, PAX4, PAX6, PCBD1, PDX1, PLAGL1, PPARG, PPP1R15B, PPP1R3A, PTF1A, RFX6, SIRT1, SLC19A2, SLC29A3, SLC2A2, STAT1, STAT3, STAT5A, STAT5B, TRMT10A, TWNK, UCP2, WFS1, WRN, ZFP57, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Morbus Waldenström (ARID1A, CXCR4, MYD88)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Morbus Wilson (ATP7B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mukopolysaccharidose (ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Mukoviszidose (CFTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
Multiple epiphysäre Dysplasie (COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, PTH1R, SLC26A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Multiples Myelom (BRAF, DIS3, KRAS, NRAS, TENT5C, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Myelodysplastisches Syndrom (ASXL1, BCOR, CBL, DNMT3A, ETV6, EZH2, FLT3, GATA2, IDH1, IDH2, KRAS, NRAS, RUNX1, SF3B1, SRSF2, STAG2, TET2, TP53, U2AF1, ZRSR2)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Myopathie, metabolische (ABHD5, ACAD9, ACADS, ACADVL, AGK, AGL, ALDOA, AMACR, AMPD1, ATP2A1, CPT2, DGUOK, ENO3, ETFA, ETFB, ETFDH, FBXL4, FHL1, FLAD1, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMA2, LDHA, LPIN1, MGME1, MPV17, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, PNPLA8, POLG, POLG2, PUS1, PYGM, RBCK1, RRM2B, SLC16A1, SLC22A5, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, TYMP, YARS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Myopie (ARR3, COL11A1, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPSF1, LRPAP1, NYX, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Neoplasie, myeloproliferative (ASXL1, CALR, CBL, DNMT3A, EZH2, IDH1, IDH2, JAK2, MPL, RUNX1, SF3B1, SH2B3, SRSF2, TET2, TP53, U2AF1)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Nephroblastom (ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CTR9, DICER1, DIS3L2, GPC3, GPC4, NLRP2, PALB2, PIK3CA, REST, TP53, TRIM37, WT1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Nephronophthise (NPHP) (AGXT, ANKS6, CEP164, CEP290, CEP83, DCDC2, FAN1, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TRAF3IP1, TTC21B, WDR19, XPNPEP3, ZNF423)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Nephrotisches-Syndrom (ACTN4, ANLN, APOL1, ARHGAP24, ARHGDI, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAT1, FN1, GON7, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, LRP2, MAGI2, MT-TL1, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCA1, TBC1D8B, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5, YRDC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Netzhautablösung (CAPN5, CHM, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, EFEMP1, FBN1, FZD4, KCNJ13, LRP5, NDP, P3H2, PLA2G5, RDH5, RLB1, RS1, TSPAN12, TUBGCP4, VCAN, VPS13B, ZNF408)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Neuroblastom (ALK, BARD1, BLM, CHEK2, FANCD2, KIF1B, PALB2, PHOX2B, PINK1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Neurofibromatose und Schwannomatose (LZTR1, NF1, NF2, SMARCB1, SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Neuronale Ceroid-Lipofuszinose (ATP13A2, 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
Neutropenie, schwere kongenitale (CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Neutropenie, somatische Mutationen (CSF3R, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Nicht-syndromische Schwerhörigkeit (ACTG1, ADCY1, AIFM1, ATP2B2, BSND, CABP2, CCDC50, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, CLPP, COCH, COL11A2, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MIR96, MPZL2, MSRB3, MT-RNR1, MT-TS2, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDZD7, PJVK, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, S1PR2, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, WFS1, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Nierenkrebs (BAP1, EPCAM, FH, FLCN, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCB1, TP53, TRIP13, TSC1, TSC2, VHL, WT1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Nierensteinerkrankung (ABCC6, ADCY10, AGXT, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CYP24A1, CYP27B1, FAH, FAM20A, FOXI1, G6PC, GCM2, GNA11, GRHPR, HNF4A, HOGA1, HPRT1, HSD11B2, INSR, KCNJ1, MOCOS, OCRL, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC7A9, SLC9A3R1, VDR, VIPAS39, VPS33B, WDR72, WNK1, WNK4, XDH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
NK-/T-Zell-Lymphom (ARID1A, ASXL3, BCOR, DDX3X, EP300, FAS, JAK3, KMT2D, MGA, STAT3, STAT5B, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Non-Langerhans-Zell-Histiozytosen (CBL, KRAS, NF1, NF2, NRAS, PTPN11, TP53)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Noonan-Syndrom (A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Noonan-Syndrom mit multiplen Lentiginen (BRAF, MAP2K1, NRAS, PTPN11, RAF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Norrie-Syndrom (NDP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Oligosaccharidose (AGA, CTSA, FUCA1, MAN2B1, MANBA, NAGA, NEU1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Optikusatrophie (inkl. LHON- Hotspots) (ACO2, AFG3L2, ANTXR1, ATP1A3, C12orf65, CISD2, DNAJC30, DNM1L, FA2H, MCAT, MFN2, MT-ND1, MT- ND4, MT-ND6, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Organische Azidämie (ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTB, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC13A5, SLC25A1, SLC25A19, SUCLA2, SUCLG1, TAZ, TMEM70)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Oro-fazio-digitales-Syndrom (C2CD3, CPLANE1, DDX59, FAM149B1, IFT172, IFT57, INTU, KIAA0753, NEK1, OFD1, SCLT1, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Osteolysegruppe (LMNA, MAFB, MMP14, MMP2, NOTCH2, TNFRSF11A, TREM2, TYROBP, ZMPSTE24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Osteosarkom (APC, BLM, RB1, RECQL4, TP53, WRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Otopalatodigitales Syndrom (FLNA, MAP3K7, SH3PXD2B, TAB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
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, TPOV2, 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
Pankreaskarzinom (ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, PALB2, PRSS1, STK11)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Pankreatitis (CASR, CFTR, CLDN2, CPA1, CTSC, SBDS, SPINK1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Paragangliom-Phäochromozytom-Syndrom (FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Parkinson Krankheit (ATP13A2, CHCHD2, DNAJC13, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, LRRK2, PARK7, PDE8B, PINK1, PODXL, PRKN, SLC18A2, SLC6A3, SNCA, SYNJ1, TAF1, VPS13C, VPS35)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Paroxysmale Dyskinesie (ADCY5, KCNMA1, PNKD, PRRT2, SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Patellare Dysostosen (CDC6, CDT1, KAT6B, LMX1B, ORC1, ORC4, ORC6, PITX1, TBX4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Pena-Shokeir-Syndrom Typ I (ADCY6, ADGRG6, CHRNA1, CHRND, CHRNA1, CNTN1, CNTNAP1, DNMT2, DOK7, ERBB3, GBE1, GLE1, KIAA1109, KLHL40, MUSK, MYBPC1, NUP88, PIP5K1C, RASGEF1B, RYR1, ZBTB42)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Periodische Fiebersyndrome (ADA, ADA2, ADAM17, AP1S3, BACH2, CARD14, COPA, ELANE, FOXD3, HAX1, HSPA1L, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL36RN, LPIN2, LRBA, MEFV, MVK, NFAT5, NLRC4, NLRP1, NLRP12, NLRP3, NLRP7, NOD2, OTULIN, PLCG2, PSMB8, PSTPIP1, RAB27A, RBCK1, RNF31, SH3BP2, SLC29A3, STING1, TNFAIP3, TNFRSF11A, TNFRSF1A, TTC7A, WDR1, XIAP, ZBTB24)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Peroxisomen-Biogenese-Störungen (PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Perrault-Syndrom (CLPP, HARS2, HSD17B4, LARS2, TWNK)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Phenylketonurie und Hyperphenylalaninämie (DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Phosphatdiabetes (ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Polyzystische Lebererkrankung (PCLD) (ALG8, ALG9, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Pontocerebelläre 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
Potentiell letale Skeletterkrankungen (AGPS, ALPL, ARSL, BMPER, CANT1, CEP120, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DHCR7, DLL3, DYNC2H1, EBP, EVC, EVC2, FAM111A, FAM20C, FGFR2, FGFR3, FLNA, FLNB, GLI3, GNPAT, IFT140, IFT172, IFT80, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX7, PPIB, PTH1R, RNU4ATAC, SLC26A2, SLC35D1, SOX9, TRIP11, TTC21B, WDR19, WDR34, WDR60, WNT7A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Primäre ciliäre Dyskinesie (ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, LRRC6, MCIDAS, NME8, OFD1, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, SPEF2, TTC25,	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Primärer Aldosteronismus (ARMC5, CACNA1D, CACNA1H, CLCN2, CTNNB1, CYP11B1, CYP11B2, HSD11B2, KCNJ5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Progressive familiäre intrahepatische Cholestase (ABCB11, ABCB4, ABCC2, ABCD3, ACOX2, AKR1D1, ATP8B1, DCDC2, HSD3B7, KIF12, LSR, MPV17, MYO5B, NR1H4, PPM1F, TJP2, USP53, VIPAS39, VPS33B, WDR83OS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Prolaktinom (AIP, DICER1, MEN1, SDHB)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Prostatakarzinom (ATM, BRCA1, BRCA2, CHEK2, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Proximale Tubulopathie (ABCC6, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BCS1L, CLCN5, COQ9, COX10, CTNS, CUBN, CYP27B1, CYP2R1, DMP1, EHHADH, ENPP1, FAH, FGF23, G6PC, GATM, GLA, GRHPR, HNF4A, HOGA1, HPRT1, LRP2, MOCOS, MRPS22, NDUFAF6, OCRL, PHEX, RRM2B, SLC20A1, SLC20A2, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A4, SLC7A9, SLC9A3R1, VDR, XDH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
Pulmonale Hypertonie (ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, KCNK3, SMAD4, SMAD9, TBX4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Pyoderma gangrenosum - Akne - Hidradenitis suppurativa (FGFR2, GJB2, NCSTN, PSEN1, PSENEN, PSTPIP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Renale Glukosurie (SLC5A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Renalen tubulären Azidose (ATP6V0A4, ATP6V1B1, 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
Renales Fanconi-Syndrom (ATP6V0A4, ATP6V1B1, ATP7B, CLCN5, CTNS, EHHADH, FAH, G6PC, GATM, HNF4A, NDUFAF6, OCRL, SLC34A1, SLC4A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Retinitis pigmentosa, autosomal- dominant (ARL3, BEST1, CA4, CRX, CTNNA1, FSCN2, GUCA1B, HK1, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RPE65, SEMA4A, SNRNP200, TEAD1, TOR1AIP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Retinitis pigmentosa, autosomal-rezessiv (ABCA4, ABHD12, ADGRA3, ADIPOR1, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, ASRGL1, BBS2, BBS4, BEST1, C8orf37, CC2D2A, CDH16, CEP290, CERKL, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, DNAJC17, DYNC2H1, EMC1, EYS, FAM161A, FLVCR1, HGSNAT, IDH3A, IDH3B, IFT140, IFT172, IMPG1, IMPG2, KIAA1549, KIZ, LAMA1, LRAT, MAK, MERTK, MFSD8, NR2E3, NRL, PCARE, PDE6A, PDE6B, PDE6G, PLA2G5, PNPLA6, POMGNT1, PRCD, PROM1, PRPF31, PRPH2, RBP3, RBP4, RCBTB1, RDH11, RDH12, RDH5, REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RPE65, RPGRIP1, SAG, SCAPER, SLC7A14, SPATA7, TRNT1, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, WDR19, ZNF408, ZNF513)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Retinitis pigmentosa, syndromal (ABHD12, CC2D2A, CEP164, CEP19, COL9A1, ERCC6, EVC, EVC2, GNPTG, IFT140, INVS, IQCB1, KIF11, MFSD8, NPHP3, OAT, PANK2, PHYH, PLK4, POC1A, POC5, RB1, RDH11, TUBGCP6, WDR19, XPNPEP3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Retinitis pigmentosa, X-gekoppelt (CHM, OFD1, PGK1, RP2, RPGR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Retinoblastom (RB1)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
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
SANDD-Syndrom (CACNA1D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Schilddrüsenerkrankung (CDCA8, DUOX1, DUOX2, DUOX2A2, 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
Schilddrüsenkarzinom (APC, CDKN1B, DICER1, FOXE1, HABP2, MEN1, MINPP1, NKX2-1, PRKAR1A, PTEN, RET, SDHB, SDHD, SRGAP1, TP53)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Schwerhörigkeit (ACTG1, ACTN4, ADCY1, ADGRV1, AIFM1, AMACR, ANLN, APOL1, ARHGAP24, ARHGDI, ATP2B2, AVIL, BSND, CABP2, CACNA1D, CCDC50, CD2AP, CDC14A, CDH23, CEACAM16, CEP250, CHD7, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COQ2, COQ6, COQ8B, CRB2, CRYM, CUBN, DCDC2, DGKE, DIABLO, DIAPH1, DIAPH3, DLC1, DSPP, EDN3, EDNRB, ELMOD3, EMP2, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAT1, FN1, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HSD17B4, ILDR1, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LAGE3, LAMB2, LARS2, LHFPL5, LMX1A, LMX1B, LOXHD1, LOXL3, LRP2, LRTOMT, MAGI2, MARVELD2, MIR96, MITF, MPZL2, MSRB3, MT-RNR1, MT-TL1, MT-TS2, MYH14 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Schwerhörigkeit (Fortsetzung MYH9, MYO15A, MYO1E, MYO3A, MYO6, MYO7A, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSBPL2, OSGEP, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX2, PAX3, PCDH15, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PJVK, PLCE1, PLS1, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRO, PTPRQ, RDX, S1PR2, SCARB2, SERPINB6, SGPL1, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMARCA1, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TBC1D8B, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TNS2, TP53RK, TPRKB, TPRN, TRIOBP, TRPC6, TRRAP, TSPEAR, TTC21B, TWNK, USH1C, USH1G, USH2A, WDR73, WFS1, WHRN, WT1, XPO5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Schwerhörigkeit, autosomal- dominante nicht-syndromale (ACTG1, CCDC50, CEACAM16, COCH, COL11A2, CRYM, DIABLO, DIAPH1, DIAPH3, ESPN, EYA4, GJB2, GJB3, GJB6, GRHL2, GSDME, KCNQ4, KITLG, LMX1A, MIR96, MYH14, MYH9, MYO6, MYO7A, OSBPL2, P2RX2, PLS1, POU4F3, PTPRQ, SIX1, SLC17A8, TBC1D24, TECTA, TJP2, TMC1, TNC, TRRAP, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Schwerhörigkeit, autosomal-rezessive nicht syndromale (ADCY1, ATP2B2, BSND, CABP2, CDC14A, CDH23, CIB2, CLDN14, CLIC5, CLPP, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PNPT1, PTPRQ, RDX, S1PR2, SERPINB6, SLC26A4, SLC26A5, SLITRK6, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, TSPEAR, USH1C, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Schwerhörigkeit, mitochondriale (MT-RNR1, MT-TS2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Senior-Loken-Syndrom (CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1, WDR19)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Sensenbrenner-Syndrom (DPH1, DYNC2H1, IFT122, IFT43, WDR19, WDR35)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Septo-optische Dysplasie - Spektrum (ALG13, ARNT2, FGF8, FGFR1, FLNA, HESX1, OTX2, PAX6, PROKR2, SOX2, SOX3, STAG2, TCF7L1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Single Nucleotide Polymorphisms for Gender Determination (AMELX, AMELY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Skelettaler Kleinwuchs (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
Skelettdysplasie mit abnormer Mineralisierung (ALPL, ANKH, AP2S1, CASR, CDC73, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAM20C, FGF23, GCM2, GNA11, HRAS, NRAS, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, TRPV6, VDR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Skelettdysplasie mit erhöhter Knochendichte (AMER1, ANKH, CA2, CLCN7, COL1A1, CSF1R, CTSK, DHCR24, DLX3, FAM20C, FERMT3, FGF23, GALNT3, GJA1, HPGD, IKBKG, KL, LEMD3, LRP4, LRP5, LRRK1, MAP2K1, MTAP, OSTM1, PLEKHM1, POLR3B, PTDSS1, PTH1R, SFRP4, SLC29A3, SLC02A1, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Skelettdysplasien mit reduzierter Knochendichte (ALPL, ANO5, ATP6V0A2, B3GALT6, B4GALT7, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, DDX58, FKBP10, GORAB, IFIH1, IFITM5, LRP5, MBTPS2, NBAS, NOTCH2, P3H1, P4HB, PLOD2, PLS3, POLR3A, PPIB, PYCR1, SEC24D, SERPINF1, SERPINH1, SGMS2, SP7, SPARC, TENT5A, TMEM38B, TNFRSF11B, WNT1, 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
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
Spastische Paraplegie, hereditäre (ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GFAP, GJC2, HSPD1, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NT5C2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TH, VAMP1, VPS37A, WASHC5, ZFYVE26, ZFYVE27)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Spinale Muskelatrophie (SMA), distale Form (AARS1, ASAH1, ATP7A, BICD2, BSCL2, DCTN1, DNAJB2, DYNC1H1, EXOSC3, GARS1, HSPB1, HSPB8, IGHMBP2, PLEKHG5, SIGMAR1, SMN1, TRIP4, TRPV4, UBA1, VRK1, WARS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Stargardt-Krankheit (ABCA4, CTNNA1, ELOVL4, IMPG1, IMPG2, PROM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Stickler-Syndrom (COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Störung der Fettsäureoxidation (ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADHA, HADHB, HSD17B10, MLYCD, SLC22A5, SLC25A20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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, NR0B1, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Störungen der Geschlechtsentwicklung XY (AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, BMP15, CBX2, CHD7, CLPP, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DIAPH2, DMRT1, DMRT2, DUSP6, EIF4ENIF1, ESR1, FEZF1, FGF17, FGF8, FGFR1, FIGLA, FLRT3, FOXL2, FSHB, FSHR, GALT, GATA4, GDF9, GNRH1, GNRHR, HARS2, HFM1, HHAT, HNF1B, HOXA13, HOXA7, HS6ST1, HSD17B3, HSD17B4, HSD3B2, IL17RD, KISS1, KISS1R, KLHL4, LARS2, LHB, LHCGR, LHX1, MAMLD1, MAP3K1, MCM9, NOBOX, NROB1, NR5A1, NSMF, PAX2, POF1B, POR, PROK2, PROKR2, PSMC3IP, RPL10, RSPO1, SAMD9, SEMA3A, SF1, SGPL1, SOX3, SOX9, SPRY4, SRD5A2, SRY, STAG3, STAR, SYCE1, TAC3, TACR3, TBX6, TOE1, WDR11, WNT4, WNT9B, WT1, ZFPM2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Surfactant-Protein-Mangel (ABCA3, CSF2RA, CSF2RB, NKX2-1, SFTPB, SFTPC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Syndrom, autoimmun- lymphoproliferatives (AIRE, CASP10, CASP8, FADD, FAS, FASLG, FOXP3, IL2RA, ITCH, PRKCD, STAT3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Syndromaler Insulinmangel (CISD2, DCAF17, DNAJC3, DUT, EIF2AK3, EIF2S3, FOXP3, GATA4, GATA6, GLIS3, IER3IP1, MT-TL1, PAX6, PPP1R15B, RFX6, SLC19A2, SLC29A3, STAT3, TRMT10A, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Syndromaler Kleinwuchs (ACAN, ANKRD11, ASXL1, ATR, ATRX, BLM, BRAF, CCDC8, CDKN1C, CDT1, CEP152, CHD7, CREBBP, CUL7, DHCR7, DVL1, DVL3, EP300, ERCC6, ERCC8, EVC, EVC2, FAM111A, FANCA, FANCC, FANCG, FGD1, FGFR3, GLI2, GLI3, HDAC8, HRAS, IARS1, IGF1R, KDM6A, KMT2D, KRAS, LARP7, LIG4, LMNA, MAP2K1, MAP2K2, NBN, NIPBL, OBSL1, ORC1, ORC4, ORC6, PCNT, PIK3R1, POC1A, PTPN11, RAD21, RAF1, RNU4ATAC, ROR2, RPS6KA3, SEMA3E, SMARCA1, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Syndromische Schwerhörigkeit (ACTN4, ADGRV1, AMACR, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CACNA1D, CD2AP, CDH23, CEP250, CHD7, CIB2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EDN3, EDNRB, EMP2, EYA1, FAT1, FN1, FOXI1, HARS1, HARS2, HSD17B4, INF2, ITGA3, ITGB4, ITSN1, KANK1, KANK2, KANK4, KCNE1, KCNJ10, KCNQ1, KIT, LAGE3, LAMB2, LARS2, LMX1B, LOXL3, LRP2, MAGI2, MITF, MT-RNR1, MT-TL1, MT-TS2, MYH9, MYO1E, MYO7A, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OSGEP, PAX2, PAX3, PCDH15, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PLCE1, POLR1C, POLR1D, PTPRO, SCARB2, SGPL1, SIX1, SIX5, SLC26A4, SMARCA1, SNAI2, SOX10, TBC1D8B, TCOF1, TNS2, TP53RK, TPRKB, TRPC6, TTC21B, TWNK, USH1C, USH1G, USH2A, WDR73, WHRN, WT1, XPO5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Tachykardie, katecholaminerge polymorphe ventrikuläre (ANK2, CALM1, CALM2, CALM3, CASQ2, GNAI2, KCNJ2, RYR2, SCN5A, TECRL, TRDN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Thrombozytopenie (ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CYCS, DIAPH1, ETV6, FCGR2C, FLI1, FLNA, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, ORAI1, PRKACG, RBM8A, RUNX1, SLFN14, SRC, STIM1, THBD, THPO, TPM4, TUBB1, VWF, WAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Treacher-Collins-Syndrom (POLR1C, POLR1D, TCOF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Tuberöse Sklerose Komplex (TSC) (TSC1, TSC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Tubulointerstitielle Nierenerkrankung, autosomal- dominant (DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Tumorerkrankung mit unbekanntem Primärtumor (ARID1A, BRAF, KMT2D, KRAS, MYC, PTEN, SMARCA4, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Tyrosinämie (FAH, HPD, TAT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
T-Zell-Lymphom, peripheres (DNMT3A, FAS, IDH2, JAK1, PLCG1, PRDM1, PRF1, RHOA, STAT3, STIM2, TET2, TP53)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Next Generation Sequencing (NGS), Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Usher Syndrom Typ 1 (CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Usher-Syndrom (ADGRV1, CDH23, CEP250, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Usher-Syndrom Typ 2 (ADGRV1, USH2A, WHRN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Vaskulopathie (ACVRL1, COL4A1, EFEMP2, ENG, FLNA, GDF2, NOTCH1, PKD1, SLC2A10, SMAD4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Vorderkammerdysgenesie (ASPH, B3GLCT, BMP4, COL4A1, CPAMD8, CYP1B1, ELP4, EYA1, FOXC1, FOXC2, FOXE3, JAG1, LAMB2, MIR184, PAX6, PITX2, PITX3, PXDN, SLC38A8, VSX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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
Walker-Warburg-Syndrom (B3GALNT2, B4GAT1, CRPPA, FKRP, FKN, GMPFB, 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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Warburg-Mikro-Syndrom (RAB18, RAB3GAP1, RAB3GAP2, TBC1D20)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Weibliche Infertilität (AR, BMP15, BTG4, BUB1B, C11orf80, CBX2, CFTR, CLPP, CPEB1, CYP11A1, CYP21A2, DHH, EIF2B1, EIF2B2, EIF2B4, EIF2B5, EIF4ENIF1, ESR1, ESR2, F2, F5, FGF8, FGFR1, FIGLA, FOXL2, FSHB, FSHR, GNRH1, GNRHR, HARS2, HFM1, HSD17B3, HSD17B4, INHA, KHDC3L, KISS1, KISS1R, LARS2, LHB, LHCGR, LHX8, MAP3K1, MCM8, MCM9, MEI1, MRPS22, MTHFR, NANOS3, NLRP2, NLRP5, NLRP7, NOBOX, NR0B1, NR5A1, NUP107, PADI6, PANX1, PATL2, PROC, PROKR2, PROS1, PSMC3IP, REC114, SEMA3A, SERPINC1, SERPINE1, SMC1B, SOHLH1, SPIDR, SRD5A2, SRY, STAG3, SYCE3, SYCP3, TAC3, TACR3, THBD, TRIP13, TUBB8, WDR11, WEE2, WNT4, ZP1, ZP2, ZP3)	EDTA-Blut, DNA; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Wilms-Tumor-Aniridia-Syndrom (ELP4, PAX6, TRIM44, WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
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
X-gekoppelte, rezessive nicht syndromaler Schwerhörigkeit (XLNSHL) (AIFM1, POU3F4, PRPS1, SMPX, TIMM8A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Zapfen-Stäbchen-Dystrophie (ABCA4, ACBD5, ADAM9, AIPL1, BEST1, C1QTNF5, C8orf37, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP78, CERKL, CFAP410, CLN5, CNGA3, CNGB3, CNNM4, CRX, DRAM2, ELOVL4, FBLN5, GUCA1A, GUCA1B, GUCY2D, HMCN1, IFT81, ITM2B, KCNV2, MERTK, MFSD8, MT-ATP6, MT-TL1, PCYT1A, PDE6C, PITPNM3, PLK4, POC1B, PRDM13, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RP1L1, RPGR, RPGRIP1, SEMA4A, TEAD1, TIMP3, TTLL5, TUBGCP4, TUBGCP6, UNC119, WASF3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
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

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Zystische Nierenerkrankung (ALG9, ANKS6, BICC1, COL4A1, COL4A4, DICER1, DNAJB11, DZIP1L, GANAB, HNF1B, INVS, KAT6B, LRP5, NOTCH2, NPHP3, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, TMEM67, UMOD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Adrenogenitales-Syndrom (ARMC5, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, MC2R, PDE8B, POR, STAR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Whole Exome Sequencing (WES)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Whole Exome Sequencing (WES), Trio-Analyse	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
Zapfendystrophie, X-gekoppelt ORF15 -Region des RPGR-Gens (NGS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Next Generation Sequencing (NGS), Whole-Exome-Sequencing (WES) Sequence capture (Hybridization), Sequencing-by-synthesis, Bioscientia Variant Identification Pipeline
1p36 Deletionssyndrom	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
ABCD-Syndrom (EDNRB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Achondrogenese Typ 2 (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
acrocallosales Syndrom (GLI3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Adipositas (LEPR, MC4R, POMC, LEP, MC2R, SIM1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
ADPKD (PKD1, PKD2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alagille Syndrom (JAG1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Albinismus, okulokutaner (OCA2, TYR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Albinismus, okulärer (GPR143, PITX2, FOXC1, TWIST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alpha-Galaktosidase-A-Mangel (GLA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alpha-Thalassämie (HBA1, HBA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alport-Syndrom (COL4A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alport-Syndrom (COL4A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Alport-Syndrom (COL4A5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Androgeninsensitivitäts-Syndrom (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Angelman Syndrom (SNRPN, UBE3A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Aniridie / WAGR (PAX6, WT1, SOX2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
ARPKD (PKHD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ataxia Teleangiectasia (ATM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Bartter-Syndrom (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Bartter-Syndrom, klassisches (CLCNKB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Beckwith-Wiedemann-Syndrom (H19, KCNQ1OT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Beta-Thalassämie (HBB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
BOR-Syndrom (EYA1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (BARD1)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (BRIP1/CHEK1)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (CHEK2)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (BRCA1)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (BRCA2)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (PALB2, RAD50, RAD51D)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (ATM)	EDTA-Blut, DNA; DNA	MLPA
Brust-/Eierstockkrebs, familiärer (STK11)	EDTA-Blut, DNA; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
CADASIL (NOTCH3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Caffey-Krankheit (COL1A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Charcot-Marie-Tooth-Krankheit Typ 1A (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
CHARGE-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Chorioideremie (CHM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Cohen-Syndrom (VPS13B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Cowden-Syndrom Typ 1 (PTEN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Cystinose (CTNS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Cystinurie (SLC3A1, SLC7A9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Diabetes mellitus, permanenter neonataler (INS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Diabetes mellitus, transienter neonataler (6q24, UPD6, ZFP57, PLAGL1, KCNJ11, INS, ZC2HC1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
DiGeorge-Syndrom (TBX1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Dopa-responsive Dystonie (TH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Dysostose, kleidokraniale (RUNX2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Dysplasie, kampomele (SOX9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Dystonie, Dopa-responsive (GCH1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
EGFR-Mutationsanalyse (EGFR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ehlers-Danlos-Syndrom Typ 6 (PLOD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ehlers-Danlos-Syndrom, hypermobiler Typ (TNXB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ehlers-Danlos-Syndrom, vaskulärer Typ (COL3A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
ektodermale Dysplasie, hypohidrotische (EDARADD, EDAR, EDA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ellis-van-Crevelde-Syndrom (EVC, EVC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
familiäres Mittelmeerfieber (MEFV)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Fanconi-Anämie (FANCA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Platelet-Syndrom mit Prädisposition für akute myeloische Leukämie (FDP/AML) (RUNX1, GATA2, TERC, TERT, CEBPA)	EDTA-Blut, DNA; DNA	MLPA
Feingold-Syndrom Typ 1 und 2 (MYCN, MIR17HG)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Fruktose-1,6-Diphosphatase-Mangel und Fruktose-Intoleranz (ALDOB, FBP1)	EDTA-Blut, DNA; DNA	MLPA
Galaktosämie (GALT)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Geschlechtsdifferenzierung (SRY)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Gitelman-Syndrom (SLC12A3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Gliedergürteldystrophie (LGMD) (SGCA, SGCB, SGCD, SGCG, FKRP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Gliedergürteldystrophie (DYSF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
GLUT1-Defekt (SLC2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Glykogenspeicherkrankheit Typ 2 (GAA)	EDTA-Blut, DNA; DNA	MLPA
Gonadendysgenese (NR5A1, NR0B1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Greig-Syndrom (GLI3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
HDR-Syndrom (GATA3, GATA4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
hereditäre Neuropathie mit Neigung zu Drucklähmungen (PMP22)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Holt-Oram-Syndrom (TBX5)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hyper-IgE-Syndrom (DOCK8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hypercholesterinämie (LDLR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hyperinsulinismus, kongenitaler (ABCC8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hyperkalzämie, familiäre hypokalziurische (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hyperoxaluria, primäre (AGXT, GRHPR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Hypochondrogenese (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hypokalzämie (CASR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hypophosphatasie (ALPL/TNSALP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hypophosphatämie / hypophosphatämische Rachitis (FGF23, PHEX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hypophysenhormon-Defizienz, kombinierte (LHX3, LHX4, GH1, HESX1, POU1F1, PROP1, GHRHR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hämochromatose (HFE, TFR2, HFE2, HAMP, SLC40A1)	EDTA-Blut, DNA; DNA	MLPA
Hämolytisch-urämisches Syndrom, atypisches (MCP/CD46, CFI)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Hämolytisch-urämisches Syndrom, atypisches (CFH und CFH-verwandte Gene(CFHR1-CFHR5))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ichthyose, X-chromosomale (STS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
IGF1-Mangel (IGF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kabuki-Syndrom (KDM6A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kabuki-Syndrom (MLL2/KMT2D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kallmann-Syndrom (CHD7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kallmann-Syndrom (FGFR1,PROK2, PROKR2, GNRHR, KISS1R, GNRH1, NELF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kallmann-Syndrom (KAL1/ANOS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Kardiomyopathie (MYBPC3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kardiomyopathie, arrhythmogene ventrikuläre (PKP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kniest-Dysplasie (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (MLH1, MSH2)	EDTA-Blut, DNA; DNA	MLPA
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (MSH6, EPCAM)	EDTA-Blut, DNA; DNA	MLPA
Kolonkarzinom, hereditäres nicht polypöses, HNPCC (PMS2)	EDTA-Blut, DNA; DNA	MLPA
Kraniosynostosen (FGFR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Lafora-Krankheit (EPM2A, EPM2B/NHLRC1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Laron Syndrom (GHR, IGF1, JAK2, STAT5B), Laron-ähnliches Syndrom	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Leber congenitale Amaurose (GUCY2D, RDH12, RPGRIP1, CEP290 (nur Intron 26))	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Leber congenitale Amaurose (CRX, AIPL1, CRB1, LCA5, RPE65)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Legius-Syndrom (SPRED1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Li-Fraumeni-Syndrom (TP53)	EDTA-Blut, DNA	MLPA
Loeys-Dietz Syndrom (TGFB1, TGFB2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Long-QT-Syndrom & Brugada- Syndrom (SCN5A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Long-QT-Syndrom (KCNQ1, KCNH2, KCNE1, KCNE2, KCNJ2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Lymphohistiozytose, hämophagozytische (PRF1, STX11, UNC13D)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Magenkarzinom, hereditäres diffuses (CDH1)	EDTA-Blut, DNA; DNA	MLPA
Marfan-Syndrom (FBN1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Mayer-Rokitansky-Küster-Hauser- Syndrom (WNT4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Medium-chain-Acyl-CoA- Dehydrogenase-Mangel (ACADM)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Mental retardation-hypotonic facies syndrome, X-linked (ATRX)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Mikrodeletionssyndrom (TSC2, PKD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Mikrozephalie (STIL, ASPM, MCPH1, CDK5RAP2, CENPJ, WDR62)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
MODY (HNF1A, GCK, HNF4A, HNF1B)	EDTA-Blut, DNA; DNA	MLPA
MODY (PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS)	EDTA-Blut, DNA; DNA	MLPA
Morbus Gaucher (GBA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Morbus Wilson (ATP7B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Mukoviszidose (CFTR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Multilocus Methylierungsdefekte (PLAGL1, GRB10, MEST, H19, KCNQ1OT1, DLK1, MEG3, RTL1, SNRPN, PEG3, GNAS, MEG8, GNASXL, NESP55)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Muskeldystrophie Duchenne/Becker (DMD)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Muskeldystrophie, kongenitale (LAMA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
MYH9-zugehörige Erkrankungen (MYH9)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Myotonia congenita Typ Becker/Thomsen (CLCN1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Nagel-Patella-Syndrom (LMX1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Neoplasie, multiple endokrine, Typ 1 (MEN1)	EDTA-Blut, DNA; DNA	MLPA
Nephronophthise (NPHP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Neurodegeneration mit Eisenspeicherung im Gehirn (PANK2 + PLA2G6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Neurofibromatose Typ 1 (NF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Neurofibromatose Typ 2 (NF2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Nijmegen-Chromosomenbruch-Syndrom (NBN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Norrie-Syndrom (NDP)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Optikusatrophie (OPA1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Ornithintranscarbamylase-Mangel (OTC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Osteogenesis imperfecta, autosomal-dominant (COL1A1, COL1A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Parkinson-Krankheit (PARK7, ATP13A2, PINK1, UCHL1, SNCA, PARK2, CAV1/2, LRRK2, GCH1)	EDTA-Blut, DNA; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Pelizaeus-Merzbacher-Krankheit (PLP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Pendred-Syndrom (SLC26A4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Peutz-Jeghers-Syndrom (STK11)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Phelan-McDermid-Syndrom (SHANK3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Phenylketonurie (PAH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Pheochromocytoma-Paraganglioma-Syndrom (SDHB, SDHC, SDHD, SDHAF1, SDHAF2)	EDTA-Blut, DNA; DNA	MLPA
Polyposis coli, familiäre adenomatöse (APC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Polyposis, MUTYH-assoziierte (MUTYH)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Prader-Willi-Syndrom (SNRPN)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Progressive familiäre intrahepatische Cholestase (ABCB4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Propionazidämie (PCCA, PCCB)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Pseudohypoparathyreoidismus 1a und 1b (GNAS, STX16)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
RAS Gene (BRAF, HRAS, KRAS, NRAS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
RCAD, renales Zysten- und Diabetes-Syndrom (HNF1B)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Retinitis pigmentosa (PRPF31, RP1, IMPDH1, RHO)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Retinitis pigmentosa (RP2, RPGR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Retinitis pigmentosa (ABCA4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Retinitis pigmentosa (RP25/EYS)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Retinoblastom (RB1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Rett-Syndrom (MECP2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Rett-Syndrom, atypisches (FOXP1, CDKL5, NTNG1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Robinow-Syndrom, autosomal- rezessiv (ROR2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Rubinstein-Taybi Syndrom (CREBBP, EP300)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Schilddrüsenerkrankung (TPO, PAX8, FOXE1, NKX2-1, TSHR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Schwannomatose (SMARCB1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Schwere myoklonische Epilepsie des Kindesalters (Dravet) (SCN1A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Schwerhörigkeit, nicht-syndromale sensorineurale (GJB2, GJB3, GJB6, WFS1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Schwerhörigkeit, nicht-syndromale sensorineurale (STRC, CATSPER2, OTOA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
SHOX-Defizienz (SHOX)	EDTA-Blut, DNA; DNA	MLPA
Silver-Russell-Syndrom (H19, KCNQ1, KCNQ1OT1, IGF2, CDKN1C)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Silver-Russell-Syndrom (UPD7mat) (GRB10, MEST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Simpson-Golabi-Behmel-Syndrom (GPC3, GPC4)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Smith-Lemli-Opitz-Syndrom (DHCR7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Sotos-Syndrom (NSD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Spastische Paraplegie (ATL1, SPAST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Spinale Muskelatrophie (SMN1, SMN2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Spondyloepiphysäre Dysplasie, kongenitaler Typ (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Stickler-Syndrom Typ 1 (COL2A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Stickler-Syndrom Typ 2 (COL11A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Surfactant-Protein-Defizienz (ABCA3, SFTPC)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Tay-Sachs-Syndrom (HEXA)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Teleangiektasie, hämorrhagische hereditäre (ENG, ACVRL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Temple-Syndrom (UPD14mat) (DLK1, MEG3, RTL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Townes-Brocks-Syndrom (SALL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Treacher-Collins-Syndrom (TCOF1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Tuberöse Sklerose (TSC1, TSC2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Tumor-Prädispositionssyndrom (BAP1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Uniparentale Disomie 7 (GRB10, MEST)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Uniparentale Disomie 14 (DLK1, MEG3, RTL1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MS-MLPA
Usher Syndrom (PCDH15)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Usher Syndrom (USH2A)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Von-Hippel-Lindau-Syndrom (VHL)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Von-Willebrand-Syndrom (VWF)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Waardenburg Syndrom (PAX3, MITF, SOX10)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Waardenburg-Shah Syndrom (EDNRB, NRTN)	EDTA-Blut, DNA; DNA	MLPA
Wachstumshormonmangel (GH1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Weaver-Syndrom (NSD1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Wilms-Tumor (WT1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Zapfen-Stäbchen-Dystrophie (PRPH2, BEST1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Adrenogenitales Syndrom (CYP17A1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Adrenogenitales Syndrom (CYP21A2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	MLPA
Angiotensin 1 konvertierendes Enzym (ACE) Insertions-/Deletions- Polymorphismus	EDTA-Blut, DNA; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Atrophie, dentato-rubro-pallido- luysische (ATN1)	EDTA-Blut, DNA; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Dihydropyrimidin-Dehydrogenase- Mangel (DPYD)	EDTA-Blut, DNA; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Kennedy Syndrom (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)
Leukämie (Fusionsgen-Panel) (AML1- ETO, BCR-ABL, CALM-AF10, CBFB- MYH11, DEK-CAN, MLL-AF6, MLL- AF9, MLL-ELL, MLL-PTD (part. tandem dupl.), NPM1-MLF1, PML- RARA)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Leukämie, akute promyelozytische (PML-RARA, qualitativ)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Mukoviszidose (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Ondine-Syndrom (PHOX2B, Repeat, Stufe 1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Pankreatitis, hereditär (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Partielle Chromosom Y-Deletion, AZF-Faktor (AZF, Devyser-Kit)	EDTA-Blut, DNA; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (ATXN2/SCA2)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (ATXN3/SCA3)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA- Fragmentanalyse in Gelmatrix (Fragmentanalyse)

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Spinocerebelläre Ataxie (ATXN7/SCA7)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (CACNA1A/SCA6)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (SCA1, Fragment)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (SCA1, Restriktion)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (SCA8)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (SCA12)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (TBP/SCA17)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Spinocerebelläre Ataxie (SCA 1,2,3,6,7,17)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Sterilität, CBAVD (CFTR, häufigste Mutationen)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Testung auf verschobene X-Inaktivierung (AR)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)
Trisomie-Schnelltest	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	QF-PCR, Mikro /Mini Satellitenanalyse (Fragmentanalyse)
Leukämie (BCR-ABL1, qualitativ)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (Fragmentanalyse)
Uniparentale Disomie der Chr. 2, 4, 5, 6, 7, 8, 9, 10, 11, 11(BWS), 13, 14, 15, 16, 17, 18, 20, 22, X	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)
Fragiles X-Syndrom (FMR1)	EDTA-Blut, DNA, Chorionzotten, Fruchtwasser, Abortmaterial, Mundschleimhautabstriche; DNA	Mikro /Mini Satellitenanalyse (Fragmentanalyse)
Leukämie (BCR-ABL1, Expression)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
Leukämie, akute promyelozytische (PML-RARA, Expression)	EDTA-Blut, EDTA-Knochenmark, RNA; RNA	Real-time PCR
Neoplasie, myeloproliferative (JAK2, Exon 14, V617F)	EDTA-Blut, EDTA-Knochenmark, DNA; DNA	Real-time PCR
Faktor V-Genmutation	EDTA-Blut; DNA	Real-time PCR
Hämochromatose Genanalyse	EDTA-Blut; DNA	Real-time PCR
HLA-B27	EDTA-Blut; DNA	Real-time PCR
Prothrombin (Faktor II)-Genmutation	EDTA-Blut; DNA	Real-time PCR
HLA-Typisierung	EDTA-Blut; DNA	SSO-PCR
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	PCR, Schmelzpunktanalyse
Lactose-Intoleranz (LCTGEN)	EDTA-Blut; DNA	Sequenzierung

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart:

Chromosomenanalyse ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik
angeborener Chromosomensatz	Blut, Nabelschnurblut, Fibroblasten, Amnionzellen, Chorionzotten, Abortgewebe	Chromosomenbänderungsanalyse
erworbener Chromosomensatz	Blut und/oder Knochenmark	Chromosomenbänderungsanalyse
angeborener Chromosomensatz	Native und/oder kultivierte Zellen aus Blut, Fibroblasten, Amnionzellen, Chorionzotten	Fluoreszenz-in-situ-Hybridisierung (FISH)
erworbener Chromosomensatz	Native und/oder kultivierte Zellen aus Blut und/oder Knochenmark	Fluoreszenz-in-situ-Hybridisierung (FISH)
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
Nicht invasive Pränataldiagnostik (fetale Aneuploidien für Chromosomen 13,18,21,X und Y)	peripheres Vollblut; DNA	Next Generation Sequencing (NGS), VeriSeq NIPT

Untersuchungsgebiet: Mikrobiologie

Untersuchungsart:

Agglutinationsteste ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Haemophilus influenzae-, Escherichia coli-, Neisseria meningitidis-, Streptococcus pneumoniae-, B-Streptokokken-Antigen	Liquor, Blutkulturen, Serum, Urin, Pleurapunktat	Partikelagglutinationstest (Latex)
Staphylococcus aureus, Anti-Staphylolysin, Antikörper	Serum	Partikelagglutinationstest (Latex)

Untersuchungsart:

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Carbapenemase-Bildner	Kulturmateriail	Immunchromatographie
Cryptococcus neoformans, Antigen	Serum, Liquor	Immunchromatographie

Untersuchungsart:

Spektrometrie (Nephelometrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Streptokokken, DNase-B-Antikörper (ADNA)	Serum	Nephelometrie

Untersuchungsart:

Spektrometrie (Turbidimetrie) ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Streptokokken, Streptolysin-O-Antikörper (ASL)	Serum	Turbidimetrie

Untersuchungsart:

Empfindlichkeitstestungen von Bakterien und Pilzen ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Schnellwachsende aerobe und anaerobe Bakterien sowie anspruchsvolle Bakterien	Reinkulturen	Agardiffusionstest, Bouillon-dilutionsverfahren als minimale Hemmkonzentration (MHK) Break-Point, teilmechanisiert
Mycobacterium-tuberculosis-Komplex	Kulturmateriail	modifizierte Proportionsmethode (Flüssigmedien)
Candida albicans, non-albicans	Kulturmateriail, Hefen	Bouillon-dilutionsverfahren als minimale Hemmkonzentration (MHK) Break-Point, teilmechanisiert

Untersuchungsart:

Keimdifferentenzierung/-identifizierung/-typisierung*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Schnellwachsende aerobe und anaerobe Bakterien sowie anspruchsvolle Bakterien sowie Pilze	Reinkulturen	biochemisch-aufwändig (Bunte Reihe, Mehrkammerverfahren), einfach, orientierend, massenspektrometrische Erregerdifferentenzierung
Mykobakterien	Reinkulturen	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit
Mykobakterien	Kulturmateriel, Sputum, Bronchialsekret, Bronchial-Lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	biochemisch-einfach, orientierend, physikalisch, Säureempfindlichkeit
Hefen und Hefeähnliche Pilze	Kulturmateriel	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferentenzierung
Dermatophyten und andere Erreger von Dermatomykosen	Kulturmateriel	biochemisch einfach, orientierend, massenspektrometrische Erregerdifferentenzierung
Schimmelpilze	Kulturmateriel	biochemisch-aufwändig (Bunte Reihe, Mehrkammerverfahren), einfach, orientierend, massenspektrometrische Erregerdifferentenzierung
Schimmelpilze wie Zygomyceten: Absidia, Mucor, Rhizomucor; Hyalohyphomyceten: Aspergillus, Penicillium; Phaeohyphomyceten: Exophiala, Cladophialophora, Alternaria	Kulturmateriel	biochemisch einfach, orientierend
Schimmelpilze (Zygomyceten)	Kulturmateriel	biochemisch einfach, orientierend

Untersuchungsart:

Kulturelle Untersuchungen ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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)
Helicobacter pylori	Magenbiopsien	Kultur, selektiv
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)
Hefen und hefeähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	in aerober Atmosphäre, spezifisch (selektiv), unspezifisch (nicht selektiv)
Dermatophyten und andere Erreger von Dermatomykosen	Hautgeschabsel, Haare, Nagelmateriale, Abstriche Hautläsionen, Pus, Biopsien	in aerober Atmosphäre, spezifisch (selektiv),
Schimmelpilze (Zygomyceten, Hyalohyphomyceten, Phaeohyphomyceten)	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	in aerober Atmosphäre, spezifisch (selektiv),

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Borrelia, IgG, IgM	Serum	CLIA
Campylobacter, Antigen	Stuhl	CLIA
Clostridioides difficile, GLDH-Antigen	Stuhl	CLIA
Clostridioides difficile, Toxin A und B, Antigen	Stuhl	CLIA
Treponema pallidum, Gesamt-Antikörper	Serum, EDTA-, Heparin-Plasma	ECLIA
Mycoplasma pneumoniae, IgG, IgM	Serum	CLIA
Toxoplasma gondii, IgG, IgM	Serum, EDTA-, Heparin-Plasma	ECLIA
Aspergillus, Antigen	Serum	ELISA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Bordetella pertussis, Toxin, IgG, IgA	Serum	EIA
Borrelia, IgG, IgM, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Echinococcus multilocularis, Antikörper	Serum	ELISA
Helicobacter pylori, Antigen	Stuhl	CLIA
Helicobacter pylori, IgA, IgG	Serum	ELISA
Plasmodium spp.-Antikörper (Malaria)	Serum	ELISA
Legionella pneumophila Serotyp 1, Antigen	Urin	CLIA
Mycobacterium-tuberculosis-Komplex, MPT64-Antigen	Kulturmateriale aus Flüssigkulturen	ELISA
Mycoplasma pneumoniae, IgA	Serum	ELISA
Streptococcus pneumoniae, Antikörper	Serum	ELISA
Salmonella-IgA Antikörper	Serum	ELISA
Salmonella-Antikörper (polyvalent)	Serum	ELISA
Treponema pallidum, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Borrelia, IgG, IgM	Serum	Immunoblot
Helicobacter, IgA, IgG	Serum	Immunoblot
Treponema pallidum, IgG, IgM	Serum	Immunoblot
Yersinia, IgA, IgG	Serum	Immunoblot

Untersuchungsart:

Mikroskopie ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Bakterien und Pilze	Blut, Serum, Plasma, Urin, Stuhl, extravasale Körperflüssigkeiten (Sputum, Liquor, Aszites, Punktionsflüssigkeiten, Ergüsse, Lavagen), Säfte (Magensaft, Galle)	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen ohne Anfärbung Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen
Bartonella henselae, IgG, IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Bartonella quintana, IgG, IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Cestoden, z.B. Taenia, Diphyllbothrium, Hymenolepis, Dipylidium	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Cryptococcus neoformans	Liquor	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Cryptosporidien	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Dermatophyten	Hautgeschabsel, Haare, Nagelmateriale, Abstriche Hautläsionen, Pus, Biopsien	Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen
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
Enterobius vermicularis	Klebestreifenpräparate	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen, ohne Anfärbung
Hefen und Hefe- ähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Fluoreszenzmikroskopie direkte Fluoreszenzmikroskopie mittels Fluorochromen
Hefen und Hefe-ähnliche Pilze	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Hefen und Hefe-ähnliche Pilze (Candida albicans, Candida non- albicans, Kluyveromyces, Pichia, Saccharomyces spp., Geotrichum, Trichosporon)	Bronchiallavage, Sputum, Liquor, sterile Körperflüssigkeiten, Genitalabstrich, Hautabstrich, Stuhl	Hellfeldmikroskopie ohne Anfärbung
Legionella, non-pneumophila- Spezies, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Legionella pneumophila, Serogruppen 1-6, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Legionella pneumophila, Serogruppen 7-14, Antikörper	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Malaria-Erreger	EDTA-Blut	Hellfeldmikroskopie mit Anfärbung

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Mykobakterien	Sputum, Bronchialsekret, Bronchial-lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Mykobakterien	Sputum, Bronchialsekret, Bronchial-lavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Nematoden, z.B. Enterobius vermicularis, Trichuris, Ascaris, Hakenwürmer, Strongyloides	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Protozoen	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Protozoen (z.B. Giardia lamblia, Entamoeba histolytica/dispar, Entamoeba spp., sonstige Amöben, Blastocystis hominis)	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Rickettsia typhi, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Rickettsia rickettsii, IgG oder IgM	Serum	Fluoreszenzmikroskopie indirekte Immunfluoreszenzmikroskopie
Schimmelpilze	Bronchioalveoläre Lavage, Sputum, Liquor, sterile Körperflüssigkeiten, Ohrabstrich	Fluoreszenzmikroskopie
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
Sporozoen/Coccidien (z.B. Cryptosporidien, Cyclospora, Isospora belli, Sarcocystis)	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen
Trematoden, z.B. Schistosoma	Urin	Hellfeldmikroskopie ohne Anfärbung
Trematoden, z.B. Schistosoma, Clonorchis, Ophisthorchis, Fasciola, Paragonimus	Stuhl	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Würmer, Wurmteile, Ektoparasiten	Stuhl, ausgeschiedene Parasiten	Hellfeldmikroskopie ohne Anfärbung

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Bordetellen, DNA	Abstrich, bronchoalveoläre Lavage	Real-time PCR
Chlamydia pneumoniae, DNA	Abstrich, Sputum, Sekret	Real-time PCR
Chlamydia trachomatis, DNA	Abstrich, Urin, Ejakulat, Punktat	Real-time PCR
Chlamydia trachomatis L-Serovar, DNA	Abstrich, Urin	Real-time PCR
Dermatophyten, DNA	Hautschuppen, Nagelspäne, Haare, Kulturmateri	Real-time PCR; Micro-Array
Mycobacterium-tuberculosis-Komplex, DNA	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl	Polymerasekettenreaktion
Mycobacterium-tuberculosis-Komplex, DNA	Sputum, Bronchialsekret, Bronchiallavage, Magennüchternsekret, Magenspülwasser, Urin, Blut, Liquor, Punktate, Biopsien, Eiter, Sperma, Prostatasekret, Menstrualblut, Stuhl, Kulturmateri	PCR (molekulare Resistenztestung)
Mycobacterium-tuberculosis-Komplex, DNA	Kulturmateri aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	PCR mit nachfolgender fest-flüssig Hybridisierung (Festphasen-gebundene Fangsonden und Enzymimmunoassay basierende Detektion der Hybridisierungsereignisse)
Mykobakterien, nicht-tuberkulös (NTM), DNA	Kulturmateri aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	PCR mit nachfolgender fest-flüssig Hybridisierung (Festphasen-gebundene Fangsonden und Enzymimmunoassay basierende Detektion der Hybridisierungsereignisse)
Mykobakterien, nicht tuberkulös (NTM), DNA	Kulturmateri aus Flüssig- und Festkulturen (Primärkulturen und Isolaten)	DNA-Sequenzierung, Kapillarelektrophorese
Mycoplasma genitalium, DNA	Abstrich, Urin	Real-time PCR

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Mycoplasma genitalium: Genotypische Makrolid-Resistenz	Urin, Abstrich	Sequenzierung
Mycoplasma genitalium: Genotypische Fluorchinolon- Resistenz	Urin, Abstrich	Sequenzierung
Mycoplasma pneumoniae, DNA	Abstrich, bronchoalveoläre Lavage, Sputum, Sekret	Real-time PCR
Neisseria gonorrhoeae, DNA	Urin, Abstrich	Real-time PCR
Pneumocystis jirovecii, DNA	BAL, Sputum, Trachealsekret, Abstriche	Real-time PCR
Streptococcus pneumoniae, Legionella pneumophila, Haemophilus influenzae, Bordetellen, Mycoplasma pneumoniae, Chlamydia pneumoniae, DNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Real-time PCR
Staphylococcus aureus, Methicillin- resistent (MRSA), DNA	Abstrich	Real-time PCR
PVL-Staphylococcus aureus, Methicillin-resistent (MRSA), DNA	Bakterielle Reinkulturen	Real-time PCR
Chlamydia trachomatis, Neisseria gonorrhoeae, Mycoplasma genitalium, Mycoplasma hominis, Trichomonas vaginalis, Ureaplasma urealyticum, Ureaplasma parvum, DNA	Urin, Endozervikal- und Urethral- Abstriche	Real-time PCR
16-S-rRNA- Gensequenzidentifizierung von Bakterien	Bakterielle Reinkulturen	Real-time PCR und Sequenzierung
Trichomonas vaginalis, DNA	Urin, Abstrich	Real-time PCR

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

Untersuchungsart:

Zellfunktionsteste ^[Flex B]

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

Untersuchungsgebiet: Virologie

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Cytomegalovirus, IgG oder IgM	Serum, EDTA-, Heparin-Plasma	ECLIA
Cytomegalovirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Cytomegalovirus, IgG, Avidität	Serum	Immunoblot
Cytomegalovirus, IgM	Serum	Immunoblot
Dengue-Virus, IgG oder IgM	Serum	ELISA
Epstein-Barr-Virus, EBNA1, IgG	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA
Epstein-Barr-Virus, IgG oder IgM	Serum, Citrat-Plasma	CLIA
Epstein-Barr-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Epstein-Barr-Virus, IgG, Avidität	Serum	Immunoblot

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Frühsommer-Meningoenzephalitis-Virus, IgG oder IgM	Serum	ELISA
Frühsommer-Meningoenzephalitis-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Hantaviren, IgG, IgM	Serum	ELISA
Hantaviren, IgG, IgM	Serum	Immunoblot
Hepatitis-A-Virus, IgG, IgM	Serum	ECLIA
Hepatitis-B-Virus, HBc-Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-B-Virus, HBc-IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-B-Virus, HBe-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-B-Virus, HBe-Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-B-Virus, HBs-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-B-Virus, HBs-Antikörper	Serum, EDTA-Plasma	ECLIA
Hepatitis-C-Virus, Antikörper	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Hepatitis-C-Virus, IgG	Serum	Immunoblot
Hepatitis-D-Virus, Antikörper	Serum, EDTA-, Heparin-Plasma	CLIA
Hepatitis-E-Virus, IgG, IgM	Serum	EIA
Hepatitis-E-Virus, IgG, IgM	Serum	Immunoblot
Herpes-Simplex-Virus Typ 1, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Herpes-Simplex-Virus, IgG, IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA
Humane Immundefizienz-Viren Typ 1/2, Antikörper	Serum	Immunoblot
Humane Immundefizienzviren Typ 1/2, Antikörper und p24-Antigen	Serum, EDTA-, Citrat-, Heparin-Plasma	ECLIA
Humane T-Zell-Leukämie-Viren Typ 1/2, Antikörper	Serum	ELISA
Humane T-Zell-Leukämie-Viren Typ 1/2, Antikörper	Serum	Immunoblot
Masernvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	CLIA
Masernvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
EBV	Serum	Lateralfuss-Immunoassay (Mononukleose Schnelltest)
Mumpsvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	CLIA
Mumpsvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA

Anlage zur Akkreditierungsurkunde D-ML-13452-01-01

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Parvovirus B19, IgG, IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA
Parvovirus B19, IgG, IgM	Serum	Immunoblot
Rötelnvirus, IgG, IgM	Serum, EDTA-, Heparin-Plasma	ECLIA
Rötelnvirus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA
Varicella-Zoster-Virus, IgA	Serum	ELISA
Varicella-Zoster-Virus, IgG, IgM	Serum, EDTA-, Citrat-, Heparin-Plasma	CLIA
Varicella-Zoster-Virus, IgG, Antikörper-Spezifitätsindex	Serum, Liquor	ELISA

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex C]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
BK-Polyomavirus, DNA, Viruslast	EDTA-Blut, Urin, BAL, EDTA-Plasma	Real-time PCR
Cytomegalovirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, Abstriche, BAL, EDTA-Plasma	Real-time PCR
Enterovirus, RNA	Liquor, BAL, Tracheal- oder Bronchialsekret, Abstriche, Stuhl	Real-time PCR
Epstein-Barr-Virus, DNA, Viruslast	EDTA-Blut, Liquor, Abstriche, BAL, EDTA-Plasma	Real-time PCR
Hepatitis-B-Virus, DNA, Viruslast	EDTA-Blut, Serum, EDTA Plasma	Real-time PCR
Hepatitis-B-Virus, genotypische Resistenzbestimmung	EDTA-Blut	DNA-Sequenzierung, Kapillarelektrophorese
Hepatitis-C-Virus, Genotypisierung	EDTA-Plasma, Serum	DNA-Sequenzierung, Kapillarelektrophorese
Hepatitis-C-Virus, Genotypisierung	EDTA-Blut, Serum	Real-time PCR
Hepatitis-C-Virus, RNA, qualitativ	Serum, EDTA-Blut, EDTA-Plasma	Real-time PCR
Hepatitis-C-Virus, RNA, Viruslast	Serum, EDTA-Blut, EDTA-Plasma	Real-time PCR
Herpes-Simplex-Virus Typen 1/2, DNA	EDTA-Blut, Liquor, Abstriche, BAL, EDTA-Plasma	Real-time PCR
Humanes Immundefizienzvirus Typ 1, Env-Gen, Korezeptor-Tropismus	EDTA-Blut	DNA-Sequenzierung, Kapillarelektrophorese
Humanes Immundefizienzvirus Typ 1, genotypische Resistenzbestimmung, Integrase	EDTA-Blut, EDTA-Plasma	RT-PCR, NGS Sequenzierung
Humanes Immundefizienzvirus Typ 1, genotypische Resistenzbestimmung, Reverse Transkriptase und Protease	EDTA-Blut, EDTA-Plasma	RT-PCR, NGS Sequenzierung
Humanes Immundefizienzvirus Typ 1, RNA, Viruslast	EDTA-Blut, EDTA-Plasma	Real-time PCR
Humanes Papilloma Virus, DNA	Abstrich	Real-time PCR
Influenzaviren Typen A/B, RNA	Abstriche	Real-time PCR
JC-Polyomavirus, DNA, Viruslast	EDTA-Blut, Liquor, Urin, BAL	Real-time PCR

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Parainfluenzaviren RNA, Metapneumovirus RNA, Enteroviren RNA, Adenoviren DNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR
Coronaviren, Bocavirus, Rhinoviren, RNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR
Influenzaviren, Respiratorische Synzytialviren, RNA	Sputum, bronchoalveoläre Lavage, Nasenrachen-Abstrich	Multiplex-PCR
SARS-CoV-2 (RNA)	Abstrich, Bal, Bronchialsekret, Sputum	Real-time PCR
Varicella-Zoster-Virus, DNA	Liquor, Abstriche, EDTA-Blut, BAL, EDTA-Plasma	Real-time PCR
SARS-CoV-2-RNA	Abstrich	Real-time PCR
Adenovirus DNA, Astrovirus RNA, Norovirus RNA, Rotavirus RNA, Sapovirus RNA	Stuhl	Real-time PCR

Untersuchungsgebiet: Transfusionsmedizin

Untersuchungsart:

Agglutinationsteste ^[Flex B]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
ABO-Blutgruppenbestimmung	EDTA-Blut, Vollblut	Hämagglutinationstest
Antikörper- Titer	Vollblut, EDTA-Blut	Hämagglutinationstest
Antikörper-Differenzierung	Vollblut, EDTA-Blut	Hämagglutinationstest
Antikörper-Suchtest	Vollblut, EDTA-Blut	Hämagglutinationstest
Coombs-Test, direkt	EDTA-Blut, Vollblut	Hämagglutinationstest
Coombs-Test, indirekt (EB)	EDTA-Blut, Vollblut	Hämagglutinationstest
Rhesusformel, inkl. Kell-Antigen	EDTA-Blut, Vollblut	Hämagglutinationstest

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex A]

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fetaler Rhesusfaktor D-Varianten	EDTA-Blut, EDTA-Plasma	Multiplex Real-time PCR