Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Lymphozytentypisierung	CPDA1-Blut, EDTA-Blut	Durchflusszytometrie	AA-0173-V011	FACS Canto II, BD	flexibel
Immunglobulin (Ig) E	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Anti-TPO (MAK)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Anti-TG (TAK)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
TSH-Rezeptor-Antikörper	Serum	ECLIA	AA-1487-V006	Cobas e41	flexibel
Antigen der Mikrosomenfraktion aus Leber und Niere (LKM-1)	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Antimitochondriale Antikörper gegen den Pyruvatdehydrogenase Komplex (AMA-M2)	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Cytosolisches Leber-Antigen Typ 1 (LC-1)	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Fusionsprotein der E2-Untereinheiten der alpha-Ketosäure-Dehydrogenasen der inneren Mitochondrienmembran (M2-3E(BPO))	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Lösliches Leber-Antigen/Leber-Pankreas Antigen (SLA/LP)	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Kerngranulaprotein (Sp100, "nuclear dots")	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Promyelocytic Leukaemia Protein (PLM, "nuclear dots")	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Integrales Protein der Zellkernmembran (gp210, "nuclear pore complex")	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel
Ro-52	Serum, EDTA-, Heparin- oder Citrat-Plasma	Immunoblot (Teststreifen)	AA-1409-V003	Teststreifen	flexibel

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Allergie: d1 Dermatophagoides pteronyssinus; e1 Katzenschuppen; e5 Hundeschuppen; d205 Milbenkomponente Tromomyosin (rDer p 10); f1 Hühnereiweiß; f2 Milcheiweiß; f3 Kabeljau; f4 Weizenmehl; f13 Erdnuss; f14 Sojabohne; f17 Haselnuss; f31 Karotte; f85 Sellerie; f353 rGly m 4 Sojabohne: PR-10 Protein; fx5 Nahrungsmittelscreen (f1, f2, f3, f4, f13, f14) Hühnereiweiß, Milcheiweiß, Dorsch (Kabeljau), Weizenmehl, Erdnuss, Sojabohne; g6 Lieschgras; g12 Roggen; m 2 Cladosporium herbarum; mx1 Schimmelpilzmischung 1 (m1, m2, m3, m6) Penicillium chrysogenum, Cladosporium herbarum, Aspergillus fumigatus, Alternaria alternata; sx1 Inhalationsscreen (d1, e1, g6, g12, m 2, t3, w6) Dermatophagoides pternoysinus, Katzenschuppen, Hundeschuppen, Lieschgras, Roggen, Cladosporium herbarum, Bike, Beifuß; t3 Birke; t215 Birkenkomponente PR-10 Protein (rBet v 1); t216 Birkenkomponente, Profilin (rBet v 2); wx209 Kräutermischung Ambrosien (w1, w2, w3) Beifußblättrige Ambrosie, Ausdauernde Ambrosie, Dreilappige Ambrosie; g213 Lischgraskomponenten (rPHl p 1, rPHl p 5b)	Serum	FEIA	AA-1628-V002	Phadia	flexibel
RF Ig A	Serum	FEIA	AA-1628-V002	Phadia	flexibel
RF Ig M	Serum	FEIA	AA-1628-V002	Phadia	flexibel
U1-snRNP AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
CENP-B AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
SS-A/Ro AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
SmD AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Scl-70 AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
RNP70 AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
SS-B/La AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Jo-1 AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
dsDNA AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
ENA-Screen	Serum	FEIA	AA-1628-V002	Phadia	flexibel

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PR3 AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
MPO AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
AMA M2 AAK	Serum	FEIA	AA-1628-V002	Phadia	flexibel
β2-Glycoprotein IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
β2-Glycoprotein IgM	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Cardiolipin IgM	Serum	FEIA	AA-1628-V002	Phadia	flexibel
CCP	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Parietalzell IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Cardiolipin IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Intrinsic Factor IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Gewebstransglutaminase IgA	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Gewebstransglutaminase IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Anti-LKM-1-Antikörper	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Gliadin IgG	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Gliadin IgA	Serum	FEIA	AA-1628-V002	Phadia	flexibel
DFS70	Serum	FEIA	AA-1628-V002	Phadia	flexibel
d1	Serum, EDTA-Plasma	FEIA	AA-1628-V002	Phadia	flexibel
e1	Serum, EDTA-Plasma	FEIA	AA-1628-V002	Phadia	flexibel
t3	Serum, EDTA-Plasma	FEIA	AA-1628-V002	Phadia	flexibel
GBM	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Anti-ssDNA-Antikörper	Serum	FEIA	AA-1628-V002	Phadia	flexibel
Antigen der Mikrosomenfraktion aus Leber und Niere (LKM-1)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V003	IFT, Euroimmun	flexibel
Antikörper gegen glatte Muskulatur (ASMA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V003	IFT, Euroimmun	flexibel
Antimitochondriale Antikörper (AMA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V003	IFT, Euroimmun	flexibel
Antineutrophile cytoplasmatische Antikörper (ANCA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V003	IFT, Euroimmun	flexibel
Autoantikörper gegen Zellkerne (ANA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V003	IFT, Euroimmun	flexibel

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Immunglobulin (Ig) A	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Immunglobulin (Ig) G	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Immunglobulin (Ig) M	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Rheumafaktor	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
sTfR	Serum / Plasma	Turbidimetrie	AA-1480-V006	cobas pro	flexibel
Retikulozyten	EDTA-Blut	Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V010	Sysmex XN1000	flexibel
Großes Blutbild	EDTA-Blut	Partikelzählung, Partikelgrößenbestimmung, Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V010	Sysmex XN1000	flexibel
Kleines Blutbild	EDTA-Blut	Partikelzählung, Partikelgrößenbestimmung, Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V010	Sysmex XN1000	flexibel
Chlorid	Serum, Urin	ISE	AA-1480-V006	Cobas pro	flexibel
Kalium	Serum, Urin	ISE	AA-1480-V006	Cobas pro	flexibel
Natrium	Serum, Urin	ISE	AA-1480-V006	Cobas pro	flexibel
Monoklonale Gammopathie	Serum	Kapillarelektrophorese	AA-1530-V003	Sebia Capillarys 3 Octa	flexibel
Serumeiweiß	Serum	Kapillarelektrophorese	AA-1486-V006	Sebia Capillarys 3 Octa	flexibel
Antithrombin III	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel

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D-Dimer	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Fibrinogen	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
INR	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Partielle Thromboplastinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Plasmathrombinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Faktor 9	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Anti-Xa	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Faktor 13	Citratplasma	Turbidimetrischer Immunoassay	AA-1528-V006	ACLTop 350	flexibel
freies Protein S	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
von Willebrand-Faktor Antigen	Citratplasma	Turbidimetrischer Immunoassay	AA-1528-V006	ACLTop 350	flexibel
von Willebrand-FaktorAktivität	Citratplasma	Turbidimetrischer Immunoassay	AA-1528-V006	ACLTop 350	flexibel
Protein C	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
APC-Resistenz, FV Leiden	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Faktor 8	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Faktor 12	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350	flexibel
Thromboplastinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V006	ACLTop 350 (Quick Test)	flexibel
AFP	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
Anti-Müller-Hormon	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel

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Beta-HCG	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
CA 125	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
CA 15-3	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
CA 19-9	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
Cacinogenic embryonic antigen (CEA)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Cortisol	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
C-Peptid	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
cyclischen Citrullin Peptid-Antikörper (Anti-CCP)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
DHEA-Sulfat	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Folsäure	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Follikelstimulierendes Hormon (FSH)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
FT3	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
FT4	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Holotranscobalamin (Active B12)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Humanes Wachstumshormon (hGH)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Insulin	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
insulinähnlicher Wachstumsfaktor-1 (IGF-1)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Luteinisierendes Hormon (LH)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
N-terminales pro-B-Typ natriuretisches Peptid (NPROBNP)	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
Osteocalcin	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Östradiol	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Parathormon	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Progesteron	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Prolaktin	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Totales prostataspezifisches Antigen (PSA)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Sexualhormonbindendes Globulin (SHBG)	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
ß-CrossLaps	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Testosteron	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Troponin T	Serum	ECLIA	AA-1487-V006	Cobas e411 / Roche	flexibel
Thyroidea stimulierendes Hormon (TSH)	Serum	ECLIA	AA-1487-V006	Cobas e411	flexibel

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Vitamin B12	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Vitamin D	Serum	ECLIA	AA-1487-V006	Cobas pro	flexibel
Inhibin B	Serum	ELISA	AA-1703-V001	Dynex DSX	flexibel
AFP	Fruchtwasser	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, Brahms	flexibel
Freies Beta-HCG	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, Brahms	flexibel
PAPP-A	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, Brahms	flexibel
Procalcitonin	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, Brahms	flexibel
Erythrozyten	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Erythrozytenzylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Granulierte Zylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Hyaline Zylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Leukozyten	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Leukozytenzylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Plattenepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Rundepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Übergangsepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Wachszylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
atypische Lymphozten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Basophile Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Blasten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel

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Eosinophile Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Kernschatten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Lymphozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Metamyelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Monozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Morphologie Erythrozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Morphologie Leukozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Morphologie Thrombozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Myelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Normoblasten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Plasmazellen	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Promyelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel

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Segmentkernige Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Stabkernige Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	flexibel
Bilirubin	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Glucose	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Hämoglobin/Erythrozyten	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Ketone	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Leukozyten	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Nitrit	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
pH	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Proteine	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Urobilinogen	Urin	Teststreifen	AA-1467-V004	Teststreifen	flexibel
Antistreptolysin O (ASLO)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Complement C3c (C3)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Complement C4 (C4)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Albumin	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Albumin	Urin	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Alkalische Phosphatase	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Amylase	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Bilirubin, direkt	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Bilirubin, gesamt	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Calcium	Serum, Plasma, Urin	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Cholinesterase (CHE)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Cholesterin	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Kreatinkinase (CK)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
CK-MB	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Cystatin C (CYSC)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Eisen	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Eiweiß	Urin	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Eiweiß, gesamt	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Gamma-GT (GGT)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Glucose	Serum, NaF-Plasma	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
GOT (AST)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel

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GPT (ALT)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Harnsäure	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Harnstoff	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
HDL Cholesterin	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Homocystein	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Kreatinin	Serum,Urin	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Laktatdehydrogenase (LDH)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Low densitiy lipoprotein (LDL)	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Lipase	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Magnesium	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Phosphat	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Triglyceride	Serum	UV-/VIS-Photometrie	AA-1480-V006	Cobas pro	flexibel
Blutsenkung (in mm/h)	EDTA-Blut	VIS-Photometrie	AA-1595-V001	Roller 20LC	flexibel
Lipoprotein (Lp) a	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
TG	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
alpha 1 Antitrypsin	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Coeruloplasmin	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
C-reaktives Protein (Globaltest und high sensitive)	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Ferritin	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Haptoglobin	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
HbA1c	EDTA-Blut	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
Transferrin	Serum	Turbidimetrie	AA-1480-V006	Cobas pro	flexibel
17-OH-Progesteron	Serum	ELISA	AA-1703-V001	Dynex DSX	flexibel
5alpha-Dihydrotestosteron (DHT)	Serum	ELISA	AA-1703-V001	Dynex DSX	flexibel
DHEA	Serum	ELISA	AA-1703-V001	Dynex DSX	flexibel
Osmolalität	Serum, Urin	Kyroskopie	AA-1678-V001	Osmometer 3000D	flexibel
Bakterien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
Pilze	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	flexibel
HLA-Crossmatch	CPDA1-Blut, Serum	Durchflusszytometrie	AA-0176-V009	FACS Canto II, BD	flexibel
1,25 Dihydroxivitamin D	Serum	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
ACTH	EDTA-Plasma	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Aldosteron	Serum	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Androstendion	Serum	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
BAP	Serum	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Calcitonin	Serum	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Renin	EDTA-Plasma	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Agammaglobulinämie hereditäre (BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A, PIK3R1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hereditäre periodische Fiebersyndrome (HPF) (ELANE, IL1RN, IL36RN,LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Immundefekte im Kindesalter primäre (ADA, AK2, AP3B1, BLNK, BTK, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD79A, CD79B, CD8A, CIITA, CLPB, CORO1A, CSF3R, CXCR4, DCLRE1C, DOCK8, ELANE, FOXN1, G6PC3, GATA1, GATA2, GFI1, HAX1, IGHM, IGLL1, IKZF1, IL2RG, IL7R, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LRRC8A, LYST, MAGT1, NHEJ1, ORAI1, PIK3R1, PNP, PRKDC, PTPRC, RAB27A, RAG1, RAG2, RFX5, RFXANK], RFXAP, RHOH , RMRP, SBDS , SLC37A4, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP , TAZ, TRAC, UNC119 , USB1, VPS13B ,	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Kombinierte T- und B-Zellimmundefekte (ADA, AK2 , CD247, CD3D , CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DOCK8, FOXN1, IKZF1, L2RG, IL7R, ITK, JAK3, LCK, LIG4, MAGT1, NHEJ1, ORAI1 , PNP, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TRAC, UNC119, ZAP70)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Neutropenie, kongenital (AP3B1, CLPB, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, JAGN1, LAMTOR2, LYST, RAB27A, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Omenn-Syndrom (OS) (ADA, AK2, DCLRE1C, IL2RG, IL7R, JAK3, LIG4, RAG1, RAG2, RMRP)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Schwere kombinierte Immundefekte T-B-) (ADA, AK2, DCLRE1C, LIG4, NHEJ1, PRKDC, RAG1, RAG2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Schwere kombinierte Immundefekte T-B+) (CD247, CD3D, CD3E, CORO1A, FOXN1, IL2RG, IL7R, JAK3, PTPRC)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Agammaglobulinämie Bruton (XLA) (BTK-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Autoimmun-Polyendokrinopathie-Candidiasis-Ektodermaldystrophie-Syndrom Typ I (APECED) (AIRE-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
CINCA-Syndrom (NLRP3-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Cryopyrin-assoziierte periodische Syndrome (CAPS) (NLRP3-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Familiäres Kälte-assoziiertes autoinflammatorisches Syndrom Typ I (NLRP3-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hyper-IgD-und-periodisches-Fiebersyndrom (HIDS) (MVK-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hyper-IgM-Syndrom (AICDA-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hyper-IgM-Syndrom (CD40-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hyper-IgM-Syndrom (CD40LG-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hyper-IgM-Syndrom (UNG-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Mevalonazidurie (MVK-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Mittelmeerfieber, familiäre Form (FMF) (MEFV-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Muckle-Wells-Syndrom (NLRP3-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
TNF-Rezeptor-1-assoziiertes periodisches Syndrom (TRAPS) (TNFRSF1A-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Wiskott-Aldrich-Syndrom (WAS) (WAS-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
X-gebundener schwerer kombinierter Immundefekt (X-SCID) (IL2RG-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
X-gebundenes lymphoproliferatives Syndrom (XLP1) (SH2D1A-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Zyklische Neutropenie (CyN) / schwere kongenitale Neutropenie (SCN) (ELANE)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Rheumatoide Arthritis (IL4R (dbSNP rs1805010))	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Rheumatoide Arthritis (TNF-α-Promotor (dbSNP rs361525, rs1800629, rs1800750, rs1799724, rs1800630, rs1799964))	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730	flexibel
Shwachman-Bodian-Diamond-Syndrom (SBDS-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V007, AA-1313-V007	ABI Sequencer 3730	flexibel
X-gekoppelte Agammaglobulinämie Typ Bruton, XLA (BTK-Gen)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
AB0-Blutgruppenbestimmung	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
Antikörpersuchtest	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
direkter Coombstest	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
Kellsystem	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
Rh-D-Bestimmung	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
Rhesusformel	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
Serumgegenprobe zur AB0-Bestimmung	EDTA-Blut / Vollblut	Hämagglutinationstest / Geltechnik	AA-1475-V009	Banjo ID-Reader	flexibel
KIR	Genomische DNA	PCR-SSO	AA-0207-V008	Luminex 200, LabScan 3D	flexibel
KIR	Genomische DNA	PCR-SSP	AA-0207-V008	Helmberg Score, Genovision	flexibel
HLA-Antikörper	Serum, EDTA-Plasma	Festphasenassay	AA-0394-V010	Luminex 200, LabScan 3D	flexibel
HLA-Klasse I	Genomische DNA	Sanger-Sequenzierung	AA-0201-V0111, AA-0206-V007	ABI3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
HLA-Klasse I, II	Genomische DNA	Sequencing-by synthesis (Illumina), MiSeq/NovaSeq Illumina, IMGT HLA-Datenbank	AA-1391-V010	Illumina Series (NovaSeq, Miseq, etc)	flexibel
HLA-Klasse I, II	Genomische DNA	longrange-PCR, Sequencing-by synthesis (Illumina), MiSeq/NovaSeq Illumina, IMGT HLA-Datenbank	AA-1550-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
HLA-Klasse I, II	Genomische DNA	PCR-SSO	AA-0211-V017	Luminex 200, LABScan OneLambda	flexibel
HLA-Klasse I, II	Genomische DNA	PCR-SSP	AA-0192-V016	Helmberg Secore, Genovision	flexibel
HLA-Klasse I, II	Genomische DNA	Sanger-Sequenzierung	AA-0215-V011	ABI3730	flexibel
Nachweis der Exone 5,7 und 10 des RHD Gens (Nachweis der Exone 5,7 und 10 des RHD Gens)	fetale cfDNA aus mütterlichem Plasma (EDTA)	Real-time PCR	AA-1721-V003	CFX96/384 Touch, Bio-Rad	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Augenerkrankungen (ABCA4, ADAM9, ADGRV1, AGBL5, AIPL1, ALMS1, ARL2BP, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C8orf37, CA4, CCDC28B, CDH23, CDH23, CDHR1, CEP290, CERKL, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FSCN2, GUCA1A, GUCA1B, GUCY2D, HARS, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IMPDH1, IMPG1, IMPG2, INVS, IQCB1, KCNV2, KIAA1549, KIZ, KLHL7, LCA5, LRAT, LRP5, LZTFL1, MAK, MERTK, MKKS, MKS1, MYO7A, MYO7A, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OFD1, OPA1, PAX6, PCARE, PCDH15, PCDH15, PDE6A, PDE6B, PDE6G, PDZD7, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RS1, SAG, SDCCAG8, SEMA4A, SLC7A14, SNRNP200, SPATA7, TMEM67, TOPORS, TRAF3IP1, TRIM32, TRPM1, TTC8, TULP1, USH1C, USH1G, USH2A, USH2A, WDPCP, WDR19, WHRN, ZNF408, ZNF513)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Typ-1-Fibrillinopathien (FBN1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Ehlers-Danlos-Syndrom (EDS), dominante Subtypen (COL1A1, COL1A2, COL3A1, COL5A1, COL5A2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Ehlers-Danlos-Syndrom, rezessive Subtypen (ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, SLC39A13, TNXB)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Ehlers-Danlos-Syndrom, classic like Typ 1 (TNXB)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Ehlers-Danlos-Syndrom, classic like Typ 1 (TNXB)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Ehlers-Danlos-Syndrom, seltene Formen, Differenzialdiagnosen (C1R, C1S, COL12A1, FLNA, COL6A1, COL6A2, COL6A3, EMILIN1, PHYKPL, PIEZO2, PLOD3, PRDM5, SLC2A10, ZNF469)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Cutis laxa (ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Kollagen 4-assoziierte intrazerebrale Blutungen (COl4A1, COL4A2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Loeys-Dietz-Syndrom (LDS) (SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Bikuspide Aortenklappe, mit Risiko für Aortenaneurysma und Aortenstenose/-dilatation (GATA5, NOTCH1, ROBO4, SMAD6)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Thorakale Aortenerweiterung mit dem Risiko der Aortendissektion (ACTA2, BGN, COL1A1, COL3A1, COL4A5, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FLNA, FOXE3, GATA5, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Marfan-ähnliche Erkrankungen (ADAMTS10, ADAMTS17, ADAMTSL2, ADAMTSL4, EFEMP1, FBN1,FBN2, LTBP2, LTBP3, MED12, SKI, UPF3B, ZDHHC9)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Osteogenesis imperfecta (OI) ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, MBTPS2, P3H1, , P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Kraniosynostosen (ALPL, ALX4, CDC45, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT140, IFT43, IL11RA, IMPAD1, MYH3, P4HB, POR, RAB23, RECQL4, SCARF2, SEC24D, SMAD6, TCF12, TWIST1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Jeune-/Kurzrippen-Polydaktylie-Syndrom (CEP120, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, KIAA0586, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Stickler-Syndrom (COL11A1, COL11A2, COL2A1, COL9A1, COL9A2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
sonstige Bindegewebserkrankungen/Skelettdysplasien (COL2A1, COL11A1, COL11A2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Metaphysäre Chondrodysplasie Typ Schmid (MCDS) (COL10A1)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Achondroplasie / Hypochondroplasie / Thanatophore Dysplasie (FGFR3)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Léri-Weill Dyschondrosteose (LWD), Langer mesomele Dysplasie (LMD) (SHOX)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Léri-Weill Dyschondrosteose (LWD), Langer mesomele Dysplasie (LMD) (SHOX)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hereditäre Sphärozytose (ANK1, EPB42, SLC4A1, SPTA1, SPTB)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Glucose-6-Phosphat-Dehydrogenase-Defizienz (Favismus) (G6PD-Gen)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Angeborene Herzfehler (ACTC1, ACVR2B, ADAMTS10, ARHGAP31, BMPR2, BRAF, CBL, CFAP53, CHD7, CITED2, CREBBP, CRELD1, DNAH11, DNAH5, DNAI1, DOCK6, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HRAS, JAG1, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, LZTR1, MAP2K1, MAP2K2, MED12, MED13L, MGP, MMP21, MRAS, MYH11, MYH6, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NPHP4, NR2F2, NRAS, NSD1, PITX2, PKD1L1, PPP1CB, PTPN11, RAF1, RBM10, RBPJ, RIT1, RRAS, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SOS1, SOS2, SPRED1, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGFBR1, TGFBR2,	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Arrhythmogene Erkrankungen (ABCC9, ACTC1, ACTN2, AKAP9, ALPK3, ANK2, ANKRD1, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CRYAB, CSRP3, DES, DSC2, DSG2, DSP, FHL1, FHOD3, FLNC, GAA, GLA, GPD1L, HCN4, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PKP2, PLN, PRDM16, PRKAG2, RAF1, RANGRF, RBM20, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLC4A3, SNTA1, TAZ, TCAP, TECRL, TGFB3, TMEM43, TNNC, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hämophilie A (Geninversionen int-22h/int-1h)	EDTA-Blut, DNA aus Blut	Long Range PCR	AA-1413-V003	--	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Gerinnungsstörung Blutungsneigung (F7, FXIII, VWF)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Gerinnungsstörung Thromboseneigung (PROC, PROS1, SERPINC1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Thrombophilie (Faktor V Leiden, F5, dbSNP rs6025)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	LC480II (Roche), BioRad CFX96	flexibel
Thrombophilie (Prothrombin, F2, dbSNP rs1799963)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	LC480II (Roche), BioRad CFX96	flexibel
Methylentetrahydrofolatreduktase- (MTHFR-) Defizienz (MTHFR-Gen: dbSNP rs1801133, rs1801131)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	BioRad CFX96	flexibel
Apolipoprotein B-Defizienz (FLDB) (APOB-Gen: dbSNP rs5742904)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel
APOE-Genotypisierung (APOE-Gen, dbSNP rs429358, rs7412)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hämochromatose (HFE-Gen: dbSNP rs1800562, rs1799945, rs1800730)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	BioRad CFX96	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Alpha-1-Antitrypsin-Mangel (SERPINA1-Gen:dbSNP rs17580, rs28929474)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	BioRad CFX96	flexibel
Arzneimittelunverträglichkeit von CYP2C9-Substraten (CYP2C9, dbSNP rs1799853, rs1057910)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001; DB-0181-V008	BioRad CFX96	flexibel
Arzneimittelunverträglichkeit, Cytochrom P-450-bedingte (CYP2D6, CYP2C19, CYP1A2, CYP2B6, CYP2C8, CYP3A4, CYP3A5)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0590-V011; DB-0183-V007, AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Laktoseintoleranz (LCT-Gen: dbSNP rs182549)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	BioRad CFX96	flexibel
5 Fu-Toxizität (DPYD-Gen c.[1236G>A; 1129-5923G>C, 483 DPYD-Gen c.[1236G>A;1129-5923G>C 483+18G>A] (Haplotyp B) c.1679T>G c.1905+1G>A (Exon 14 Skipping Mutation) c.2846A>T)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-1679-V001	ABI Sequencer 3130XL	flexibel
HCV-Therapie (IL28B dbSNP rs12979860)	EDTA-Blut, DNA aus Blut	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	BioRad CFX96	flexibel
Butyrylcholinesterase (BCHE)-Defizienz und postoperative Apnoe (BCHE-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Kongenitaler Laktasemangel (LCT)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1391-V010, AA-1648-V002	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Arzneimittelunverträglichkeit (NAT2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Azathioprin-Therapie (TPMT)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Multi Drug resistance (ABCB1 (MDR1), dbSNP rs1045642)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel
Verträglichkeit catecholaminerger Neurotransmitter (COMT dbSNP rs4680)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel
Detoxifizierungsstörung (CYP1A1 , dbSNP rs4646903, rs1048943)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel
Detoxifizierungsstörung (GSTM1, GSTP1, GSTT1)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel
HCV-Therapie (HCV-Therapie ITPA, dbSNP rs1127354, rs7270101)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Statin-Unverträglichkeit, Myopathie unter Hochdosis-Therapie (SLCO1B1, dbSNP rs4149056)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
SULT1A1-bedingter verzögerten Phase II-Metabolismus (SULT1A1, dbSNP rs9282861)	EDTA-Blut, DNA aus Blut	Restriktionsanalyse	AA-0143-V004	Thermocycler	flexibel

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Cumarin-und Cumarinderivat-Sensitivität (VKORC1, dbSNP rs9934438, rs28527768)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hypophosphatämie (CLCN5, DMP1, ENPP1, FAM20C, FGF23, PHEX, SLC34A1, SLC34A3, SLC9A3R1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hypophosphatasie (ALPL)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Kongenitale Defekte der Glykosylierung (CDG-Syndrome) (ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, CAD, CCDC115, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TM5M165, TM5M199, TUSC3)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

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Mukopolysaccharidosen (MPS) (ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Maligne Hyperthermie (RYR1, CACNA1S)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hyperoxalurie (AGXT, GRHPR, HOGA1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Fettstoffwechselstörungen (ABCA1, ANGPTL3, APOA1, APOA5, APOB, APOC2, APOE, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LMF1, LPL, MTTP, PCSK9, SAR1B)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
MODY-Diabetes (Maturity-Onset Diabetes of the Young) (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Porphyrien (ALAD, CPOX, HMBS, PPOX, ALAS2, FECH, UROD, UROS)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Adipositas, monogene (KSR2, LEP, LEPR, MC3R, MC4R, MRAP2, NTRK2, PCSK1, POMC, SH2B1, SIM1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Sphingolipidosen (GLA, GAA, GM2A, HEXA, HEXB, GALC, PSAP, NPC1, NPC2, SMPD1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/ Version**	**Gerät**	**Akkreditierungs-status**
Morbus Gaucher (GBA)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Morbus Gaucher (GBA)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Harnstoffzyklusdefekte (ARG1, ASL, ASS1, CPS1, NAGS, OTC, SLC25A13, SLC25A15)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Störungen der Fettsäure-Oxidation (ACADM, HADHA, HADHB, ACADVL, ETFA, ETFB, ETFDH)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Fruktose-Intoleranz, hereditäre (ALDOB, FBP1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hämochromatose, hereditäre (BMP6, HAMP, HFE, HJV, SLC40A1, TFR2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Adrenogenitales Syndrom (AGS) (CYP11B1, CYP11B2, CYP17A1, HSD3B2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Adrenogenitales Syndrom (AGS) (CYP21A2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1599-V003; AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Adrenogenitales Syndrom (AGS) (CYP21A2)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-1599-V003, AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Ahornsirupkrankheit (MSUD) (BCKDHA, BCKDHB, DBT)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Alkoholintoleranz (ADH1B, ALDH2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Alpha-1-Antitrypsin-Mangel (SERPINA1)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Biotinidasemangel (BTD)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Carnitinzyklusdefekte (CPT1A, CPT2, SLC25A20)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Crigler-Najjar-Syndrom (UGT1A1)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Meulengracht- (Gilbert-) Syndrom (UGT1A1, dbSNP rs3064744)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Galaktosämie (GALT)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Glutarazidurie Typ 1 (GCDH)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Isovalerianazidämie (IVD)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Methylmalonazidurie, Vitamin B12-resistent (MMUT)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Morbus Wilson (ATP7B)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Pädiatrische Neurotransmitterstörungen (DBH, DDC, GCH1, MAOA, PCBD1, PTS, QDPR, SLC18A2, SLC6A3, SPR, TH, TPH2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Phenylketonurie (PAH)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Propionazidämie (PCCA, PCCB)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Smith-Lemli-Opitz-Syndrom (DHCR7)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Tyrosinämie Typ I (FAH)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Basalzellnävus-Syndrom (BCNS) (PTCH1, PTCH2, SUFU)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Legius-Syndrom (SPRED1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Neurofibromatose Typ 1 (NF1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Schwannomatose (LZTR1, NF2, SMARCB1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Tuberöse Sklerose Complex (TSC) (TSC1, TSC2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
TSC2/PKD1-Contiguous-Gene-Syndrom (TSC2-, PKD1-Gen)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Ziliopathien (ACVR2B, AHI1,ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, C2CD3, C8orf37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP298, CFAP53, CHD1L, CPLANE1, CRELD1, CSPP1, DCDC2, DDX59, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, EVC, EVC2, FRAS1, GANAB, GAS8, GDF1, GLIS2, HNF1B, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LEFTY2, LRRC6, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MMP21, MUC1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PDE6D, PIH1D3, PKD1, PKD1L1, PKD2, PKHD1, POC1B, ROBO2, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCLT1, SDCCAG8, SIX2, SLC41A1, SPAG1, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TTC21B, TTC25, TTC8, UMOD, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZIC3, ZMYND10, ZNF423)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Morbus Osler (ACVRL1, ENG, GDF2, SMAD4)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Thalassämie (HBB, HBA1, HBA2, HBD-, HBG1, HBG2-Promoter)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	NovaSeq6000	flexibel
Sideroblastische Anämie, X-gebunden (XLSA) (ALAS)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Cystische Fibrose (CF) (CFTR)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Congenitale bilaterale Aplasie des Vas deferens (CBAVD) (CFTR)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Alopezie (HR)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Morbus Crohn (NOD2/CARD15, dbSNP rs2066844, rs2066845, rs2066847)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Interstitielle Lungenerkrankungen im Kindesalter (chILD) (ABCA3, CSF2RA, CSF2RB, FLNA, FOXF1, NKX2-1, SFTPB, SFTPC)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Pulmonale alveoläre Mikrolithiasis (PAM) (SLC34A2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Pulmonale arterielle Hypertonie (PAH) (ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNA5, KCNK3, SMAD1, SMAD4, SMAD9, TBX4)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Nierenerkrankungen (ACE, ACTA2, ACTG2, ACTN4, AGT,AGTR1, AGXT, AHI1, ANKS6, ANLN, ANOS1, APOL1, ARHGAP24, ARHGDIA, BICC1, BMP4, BMP7, CC2D2A, CD2AP, CDC5L, CEP164, CEP290, CEP83, CFH, CHD1L, CHRM3, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DACH1, DCDC2, DGKE, DSTYK, DZIP1L, EMP2, ETV4, ETV5, EYA1, FGF20, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GANAB, GATA3, GDNF, GLA, GLIS2, REM1, GRHPR, GRIP1, HNF1B, HOGA1, HPSE2, IFT172, INF2, INVS, IQCB1, ITGA3, ITGA8, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, LRIG2, MAPKBP1, MUC1, MYH9, MYO1E, NEIL1, NEK8, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, PAX2, PAX8, PDSS2, PKD1, PKD2, PKHD1, PLCE1, PTPRO, REN, RET, ROBO2, RPGRIP1L, SALL1, SCARB2, SDCCAG8, SIX1, SIX2, SIX5, SLC41A1, SMARCAL1, SOX17, TMEM216, TMEM237, TMEM67, TRAP1, TRPC6, TTC21B, UMOD, UPK2, UPK3A, WDR19, WNT4, WT1, XPNPEP3, ZIC3, ZNF423)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Pankreatitis, chronisch (hereditäre) (CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/ Version**	**Gerät**	**Akkreditierungs-status**
Pankreatitis, chronisch (PRSS1, SPINK1)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Pankreatitis, chronisch (PRSS1)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
RASopathien (BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/ Version**	**Gerät**	**Akkreditierungs-status**
Schwerhörigkeit/Taubheit (ABCC1, ABHD12, ACTG1, ADCY1, ADGRV1, AIFM1, ATP6V1B1, BDP1, BSND, CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP250, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COL11A1, COL11A2, COL4A6, COX1, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ERAL1, ESPN, ESRP1, ESRRB, EYA1, EYA4, FAM189A2, FOXI1, GAB1, GATA3, GIPC3, GJB2,GJB3, GJB6, GPRASP2, GPSM2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MIR182, MIR183, MIR96, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7, PJVK, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, RNR1, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLITRK6, SMPX, SNAI2, SOX10, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRNE, TRNL1, TRNS1, TRNS2, TRRAP, TSPEAR, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hörverlust, autosomal-rezessiv, nicht-syndromal (GJB2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hörverlust, autosomal-rezessiv, nicht-syndromal (GJB2, GJB6)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hörverlust, autosomal-rezessiv, nicht-syndromal (GJB6, Deletion D13S1830))	EDTA-Blut, DNA aus Blut	Multiplex-PCR, Agarosegelelektrophorese	AA-0272-V005	Thermocycler	flexibel
Taubheit, autosomal-rezessiv 16, DFNB16 (STRC)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Taubheit, autosomal-rezessiv (STRC, OTOA)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Pseudoxanthoma Elasticum (ABCC6)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Pseudoxanthoma Elasticum (ABCC6)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Ataxien (ABCB7, ABHD12, ADGRG1, AFG3L2, AHI1, AMACR, ANO10, APTX, ARL13B, ARSA, ATCAY, ATG5, ATM, ATP13A2, ATP1A3, ATP8A2, ATXN10, B4GALNT1, BTD, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CC2D2A, CCDC88C, CEP290, CEP41, CHP1, CLCN2, CLN5, CLN6, COA7, COQ8A, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EEF2, EIF2B1,EIF2B2,EIF2B3, EIF2B4,EIF2B5, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, GALC, GBA, GBA2, GCLC, GDAP2, GJB1, GJC2, GOSR2, GRID2, GRM1, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0586, KIF1C, KIF26B, KIF7, MARS2, MICU1, MME, MRE11, MTPAP, NEU1, NKX6-2, NPC1, NPC2, NPHP1, OFD1, OPA1, OPA3, PANK2, PDE10A, PDE6D, PDHX, PDYN, PEX10, PEX2, PIK3R5, PLA2G6, PLD3, PLP1, PMPCA, PNKP, PNPLA6, POC1B, POLG, POLR3A, PRKCG, PRNP, PUM1, RNF216, RPGRIP1L, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC17A5, SLC1A3, SLC9A1, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, THG1L, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRPC3, TTBK2, TTC21B, TTPA, TUBB4A, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR81, WFS1, WWOX, XRCC1, ZNF423)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Ataxien, spinocerebelläre autosomal-dominante (SCA) (ATXN 1 und/oder 2,3,7, CACNA1A, TBP)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse, TP-PCR	AA-1300-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Ataxie, Friedreich'sche (FRDA1) (FXN)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse, TP-PCR; Long-Range PCR	AA-0313-V004, AA-1300-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Epilepsien (AARS, ACTL6B, ADAM22, ADGRV1, ADRA2B, ALDH7A1, ALG13, AP3B2, ARHGEF15, ARHGEF9, ARV1, ARX, ATP1A2, BRAT1, CACNA1A, CACNA1E, CACNA1H, CACNB4, CAD, CASR, CDK19, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CNPY3, CNTN2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DCX, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EPM2A, FGF12, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GLUL, GNAO1, GOSR2, GOT2, GPHN, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HDAC4, HNRNPU, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH5, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MBD5, MDH2, MECP2, MEF2C, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PRDM8, PRICKLE1, PRICKLE2, PRRT2, RANBP2, RANGAP1, RELN, RHOBTB2, RNF13, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SMC1A, SPTAN1, SRPX2, ST3GAL3, STX1B, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Rett Syndrom (RTT) (MECP2)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
MECP2-Duplikationsyndrom (MECP2)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Rett-Syndrom (MECP2)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hereditäre Hyperekplexie (Startle disease) (ARHGEF9, ATAD1, GLRA1, GLRB, SLC6A5)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Choreatiforme Bewegungsstörungen (ADCY5, ARSA, ATM, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, FRRS1L, FTL, GM2A, GNAO1, KCNA1, NKX2-1, PANK2, PDE10A, PRNP, RNF216, SETX, TBP, VPS13A, XK)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Neuropathien, hereditäre (AARS, ABCD1, ABHD12, AFG3L2, AIFM1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARHGEF10, ARL6IP1, ATL1, ATL3, ATP13A2, ATP1A1, ATP7A, B4GALNT1, BAG3, BICD2, BSCL2, C12orf65, C19orf12, CAPN1, CAPN3, CCT5, COX6A1, CPT1C, CTDP1, CYP2U1, CYP7B1, DCTN1, DDHD1, DDHD2, DES, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DPM3, DSTYK, DYNC1H1, EGR2, ELP1, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GBA2, GDAP1, GJB1, GJC2, GNB4, HARS, HINT1, HK1, HOXD10, HPDL, HSPB1, HSPB3, HSPB8, HSPD1, IBA57, IGHMBP2, INF2, JPH1, KARS, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, L1CAM, LAMA2, LITAF, LMNA, LRSAM1, MAG, MARS, MATR3, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NIPA1, NKX6-2, NT5C2, NTRK1, OPA1, PCYT2, PDK3, PLEKHG5, PLP1, PMP22, PNKP, PNPLA6, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, REEP2, RETREG1, RTN2, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SELENOI, SEPT9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC16A2, SLC25A46, SLC33A1, SLC5A7, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTLC1, SPTLC2, SURF1, SYT2, TDP1, TECPR2, TFG, TRIM2, TRPV4, TTR, TUBB4A, UBAP1, UCHL1, VCP, VPS37A, WASHC5, WNK1, YARS, ZFYVE26, ZFYVE27)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Charcot-Marie-Tooth-Neuropathien, CMT (ATL1, DNM2, GARS, GDAP1, GJB1, HINT1, IGHMBP2, MFN2, MPZ, NEFL, NGF, PMP22, SH3TC2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Charcot-Marie-Tooth Neuropathie Typ 1 (PMP22)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hereditäre Neuropathie mit Neigung zu Drucklähmung (HNPP) (PMP22)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Hereditäre Neuropathie mit Neigung zu Drucklähmung (HNPP) (PMP22)	EDTA-Blut, DNA aus Blut	MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
CADASIL (HTRA1, NOTCH3)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Alzheimer Erkrankung, Frühform (AD1) (APP, PSEN1, PSEN2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Chorea Huntington (HTT)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse, TP-PCR; Long-Range PCR	AA-0316-V003, AA-1300-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Creutzfeldt-Jakob Erkrankung, familiäre Form (CJD) (PRNP)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Dentatorubrale Pallidoluysische Atrophie (DRPLA) (ATN1)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-1300-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Gehirnfehlbildungen (AMPD2, ARX, CDK5, CEP85L, CHMP1A, CLP1, COASY, DCX, EXOSC3, EXOSC8, EXOSC9, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, PCLO, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TBCD, TMTC3, TOE1, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VPS51, VPS53, VRK1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Leukoenzephalopathie mit Verlust der weißen Substanz (EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Spastische Paraplegien (AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, C19orf12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENOI, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TUBB4A, UBAP1, UCHL1, VPS37A, WASHC5, [illegible])	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Entwicklungsstörungen und Komorbiditäten, Wachstumsstörungen (AARS, ABCC9, ABCD1, ACSL4, ACTB, ACTG1, ADAT3, ADNP, AFF2, AHDC1, AIFM1, AKT3, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMER1, AMPD2, ANK3, ANKLE2, ANKRD11, AP1S2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASPA, ASPM, ATP6AP2, ATP7A, ATRX, AUTS2, B4GALT1, BCAP31, BCOR, BDNF, BRAF, BRD4, BRWD3, C12orf4, C12orf57, CA8, CACNA1C, CACNG2, CAD, CAMK2A, CAMK2B, CAMK2G, CASK, CBL, CC2D1A, CCDC115, CCDC22, CCND2, CDH15, CDK13, CDK5, CDK5RAP2, CDK6, CDKL5, CDKL5, CDKN1C, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP85L, CHAMP1, CHD4, CHD7, CHD8, CHMP1A, CIC, CIT, CLCN4, CLIC2, CLP1, CLTC, CNKSR2, CNOT3, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COL4A3BP, COLGALT1, COPB2, CRADD, CRBN, CREBBP, CSNK2A1, CTCF, CTNNB1, CUL4B, DBH, DCX, DDC, DDOST, DDX3X, DEAF1, DHCR24, DHCR7, DIS3L2, DKC1, DLG3, DLG4, DNAJC12, DNM1, DNMT3A, DOCK7, DOCK8, DOLK, DONSON, DPAGT1, DPF2, DPM1, DPM2, DPM3, DPP6, DVL1, DVL1, DVL3, DVL3, DYNC1H1, DYRK1A, EBP, EDC3, EED, EEF1A2, EHMT1, EIF2B5, EIF2S3, EIF3F, ELP2, EP300, EPB41L1, EXOSC3, EXOSC8, EXOSC9, EZH2, FANCB, FBXO31, FGD1, FLNA, FMN2, FMR1, FOXG1, FOXG1, FOXP1, FOXP2, FRMPD4, FTSJ1, GABRA1, GALT, GATAD2B, GCDH, GCH1, GDI1, GFAP, GIMAP1, GK, GLI3, GNAI1, GNAO1, GNB1, GPAA1, GPC3, GPKOW, GPSM2, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, HCCS, HCFC1, HCN1, HDAC4, HDAC6, HDAC8, HEPACAM, HERC1,	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Myopathien, kongenitale (ACTA1, BIN1, CCDC78, CFL2, CNTN1, DNM2, GNE, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MICU1, MTM1, MTMR14, MYF6, MYH2, MYH7, MYL1, MYO18B, MYPN, NEB, ORAI1, RYR1, SELENON, SPEG, SPTBN4, STAC3, STIM1, TNNT1, TPM2, TPM3, TTN, VCP)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Myopathien, nemaline (ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Core-Myopathien (ACTA1, BIN1, DNM2, MTM1, RYR1, SELENON, TPM2, TPM3, TTN)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Myopathien, myofibrilläre (BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PLEC, PYROXD1, TTN)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hypokaliämische Periodische Paralyse (HypoPP) (CACNA1S, KCNJ2, SCN4A)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Muskelatrophie, spinobulbär (SBMA, Kennedy Krankheit) (AR)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-1300-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Muskelatrophie, spinale Typ I – III (IV) (SMA1,2,3,4) (SMN1, SMN2)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V008; AA-1298-V004	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Muskelatrophie, spinale Typ I – III (IV) (SMA1,2,3,4) (SMN1, SMN2)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-0143-V004	Thermocycler	flexibel
Muskelatrophien, spinale (SMA) (ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HSPB8, IGHMBP2, PLEKHG5, REEP1, SLC5A7, TFG, TRIP4, TRPV4, UBA1, VAPB, VRK1)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Muskeldystrophien, kongenitale (CHKB, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DNM2, DPM3, FHL1, FKRP, FKTN, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMT1, POMT2, SELENON, TCAP)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Muskeldystrophien, progrediente (ANO5, BVES, CAPN3, CAV3, CRPPA, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, HNRNPDL, LAMA2, LIMS2, LMNA, MATR3, MYOT, PLEC, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TRAPPC11, TRIM32, TTN)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Muskeldystrophie Duchenne / Becker (DMD)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Muskeldystrophie Duchenne / Becker (DMD)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Myotone Dystrophie Typ 1 (Curschmann-Steinert-Syndrom) (DMPK)	EDTA-Blut, DNA aus Blut	Southern-Blot-Hybridisierung	AA-1299-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Myotone Dystrophie Typ 1 (Curschmann-Steinert-Syndrom) (DMPK)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-1300-V003, AA-1299-V003	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Nicht-dystrophische Myotonien und periodische Paralysen (CACNA1S, CLCN1, HSPG2, KCNJ2, SCN4A)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Stoffwechselmyopathien (ACADVL, AGK, ALDOA, CPT2, DGUOK, ETFA, ETFB, ETFD, FBXL4, GAA, HADHA, HADHB, INIP, ISCU, LAMA2, LDHA, LPIN1, MGNE1, MPV17, PFKM, PGAM2, PHKA1, PHKB, POLG, PYGM, RRM2B, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Fragiles X-Syndrom (FMR1-Gen)	EDTA-Blut, DNA aus Blut	Southern-Blot-Hybridisierung	AA-0277-V013	Thermocycler, Hybridisierungs ofen	Einzelgenanalyse
Fragiles-X-Syndrom (FMR1)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-0277-V013	ABI Sequencer 3730XL	Einzelgenanalyse
Amyloidose, familiäre Form (TTR)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
HIV-1-Wirtsresistenz (CCR5-Gen: dbSNP rs333, CCR2-Gen: dbSNP rs1799864, SDF1-Gen: dbSNP rs1801157)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-0143-V004	Thermocycler	flexibel
Short Tandem repeats-/Mikrosatelliten-Analyse (Short Tandem repeats-/Mikrosatelliten-Analyse)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-1730-V001	ABI Sequencer 3130XL	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Zieldiagnostik / Bestätigungsanalyse SNV	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Zieldiagnostik / Bestätigungsanalyse CNV	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730	flexibel
Whole Exome Sequencing	EDTA-Blut, DNA aus Blut, Chorionzotten, Amniozyten	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1654-0002, AA-1637-V003, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
mTORopathien (AKT1, AKT1S1, AKT3, CCND2, DEPDC5, DEPTOR, DOCK7, G3BP1, G3BP2, MLST8, MTOR, NPRL2, NPRL3, PAK2, PIK3CA, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PTEN, RICTOR, RPTOR, STK11, STRADA, TBC1D7, TSC1, TSC2)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1391-V010, AA-1648-V002	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Prader-Willi-Syndrom/Angelman-Syndrom (Deletionen, Duplikationen, Methylierung in Chromosomenregion 15q11.2-q13)	DNA aus Blut, extrahierte DNA	(MS) MLPA	AA-1756-V001	Thermocycler; ABI Sequencer 3730XL; ABI Sequencer 3730	flexibel
Pankreas Elastase	Stuhl	ELISA	AA-1590-V001	Dynex DSX	flexibel
Calprotectin	Stuhl	ELISA	AA-1598-V001	Dynex DSX	flexibel
Staphylokokken (Koagulase, clumping factor, Protein A, Polysaccharide)	Bakterienkultur	Partikelagglutination	AA-1478-V001		flexibel
Streptokokken (Lancefield-Antigen)	Bakterienkultur	Partikelagglutination	AA-1452-V002		flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Bakterien (Resistenztestung)	Keimkolonien in Reinkultur	Agardiffusionstest, partielles Buillondilutionsverfahren als minimale Hemmkonzentration (MHK) mit Extrapolation	AA-1473-V004, AA-1518-V003	manuell & Vitek2, Biomerieux	flexibel
Hefen	Pilzisolat	biochemisch, aufwendig	AA-1518-V003	Vitek2, Biomerieux	flexibel
Anaerobier, Corynebakterien	Bakterienisolat	biochemisch, aufwendig	AA-1518-V003	Vitek2, Biomerieux	flexibel
gram-negative aerobe Bakterien	Bakterienisolat	biochemisch, aufwendig	AA-1518-V003	Vitek2, Biomerieux	flexibel
gram-positive aerobe Bakterien	Bakterienisolat	biochemisch, aufwendig	AA-1518-V003	Vitek2, Biomerieux	flexibel
Neisseria sp., Haemophilus sp.	Bakterienisolat	biochemisch, aufwendig	AA-1518-V003	Vitek2, Biomerieux	flexibel
Bakterien (Orientierungs-/Differenzierungsteste)	Keimkolonien in Reinkultur	biochemisch, orientierend (Katalase, Oxidase, Nitrocefin (Beta-Lactamase))	AA-1449-V003, AA-1455-V003		flexibel
Pneumokokken	Keimkolonien in Reinkultur	biochemisch, orientierend (Optochin)	AA-1455-V003		flexibel
Staphylococcus saprophyticus	Keimkolonien in Reinkultur	biochemisch, orientierend (Novobiocin)	AA-1472-V007		flexibel
Bakterien, Pilze (Anlage und ablesen)	Abstrich (urogenital, HNO), Blut, Haut, Wunde, Punktat, Stuhl,	in CO2-Atmosphäre, mikroaerobe/anaerobe Atmosphäre, spezifisch, unspezifisch	AA-1482-V004, AA-1490-V004, AA-1531-V002. AA-1472-V007		flexibel
Bakterien, Pilze	Urin	spezifisch, unspezifisch, Keimzahlbestimmung	AA-1472-V007		flexibel

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Chlamydia trachomatis IgG/IgA	Serum	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Toxoplasma gondii, IgG, IgM	Serum	CLIA	AA-1401-V009	Cobas e411	flexibel
Treponema pallidum Antikörper	Serum, EDTA-Plasma	Partikelagglutination	AA-1674-V002	Sensititer Manual Viewer	flexibel
Treponema pallidum Infektion assoziierte, nichtspezifische Lipoidantikörper (IgG, IgM)	Serum, EDTA-Plasma	Partikelagglutination	AA-1673-V001	Kartenschüttler	flexibel
Treponema pallidum, Ig	Serum	CLIA	AA-1401-V009	Cobas e411, cobas pro	flexibel
Borrelia burgdorferi sensu lato IgG, IgM	Serum, EDTA-Plasma	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Diphtherie-Toxoid, IgG	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Yersinien Antikörper IgA/IgG/IgM	Serum	Immunoblot	AA-1538-V002	manuell (Mikrogen)	flexibel
Borrelia burgdorferi sensu lato IgG, IgM	Serum, EDTA-Plasma	Immunoblot	AA-1538-V002	manuell (Mikrogen)	flexibel
Bakterien, Pilze	Abstrich	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-1444-V005		flexibel
Bordetalla pertussis, Bordetella parapertussis, Bordetella holmesii, DNA	Abstrich, Sputum	Real-Time PCR	AA-1511-V004	Rotor-Gene, Qiagen, CFX Opus Biorad	flexibel
Chlamydia trachomatis, Neisseria gonorrhoeae, DNA	Abstrich, Urin	Real-Time PCR	AA-1602-V003	cobas 6800, Roche	flexibel
Chlamydia trachomatis, Neisseria gonorrhoeae, DNA	Abstrich, Urin	Real-Time PCR (Duplex-PCR)	AA-1458-V005, AA-1659-V003	Alinity m, Rotor-Gene, Qiagen	flexibel
Chlamydia pneumoniae, Mycoplasma pneumoniae, Legionella spp, DNA	Abstrich, Sputum	Real-time PCR (Oligoplex-PCR)	AA-1523-V003	Rotor-Gene, Qiagen, CFX Opus Biorad	flexibel
Chlamydia trachomatis, Mycolpasma hominis, genitalium, Neisseria gonorrhoeae, Trichomonas vaginalis, Ureaplasma urealyticum, parvum (STIs, sexually transmitted infections)	Abstrich, Urin, Ejakulat	Real-time PCR (Duplex-PCR, Oligoplex-PCR, Multiplex-PCR)	AA-1659-V003, AA-1458-V005	Alinity m, Abbott, CFX Opus Biorad	flexibel

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Cryptosporidien Antigennachweis	Stuhl	ELISA	AA-1563-V007	r-biopharm DSX	flexibel
Entamoeba histolytica/dispar Antigennachweis	Stuhl	ELISA	AA-1563-V007	r-biopharm DSX	flexibel
Giardia lamblia Antigennachweis	Stuhl	ELISA	AA-1563-V007	r-biopharm DSX	flexibel
Treponema pallidum IgM	Serum, EDTA-Plasma	Immunoblot	AA-1583-V003	manuell (Mikrogen)	flexibel
Chlamydia trachomatis, psittaci, pneumoniae IgA, IgG, IgM	Serum, EDTA-Plasma	Immunoblot	AA-1583-V003	manuell (Mikrogen)	flexibel
Helicobacter pylori Antigennachweis	Stuhl	ELISA	AA-1563-V007	r-biopharm DSX	flexibel
Tetanus-Toxoid IgG	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Interferon-Gamma Release-Assay	EDTA-Plasma	CLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Clostridium difficile Toxin A/B-Antig.	DHT	ELISA	AA-1558-V001	Dynex DSX	flexibel
Cytomegalievirus, IgG, IgM	Serum, EDTA-Plasma	ELISA/CMIA	AA-1401-V009, AA-1437-V010	Cobas e411 / Roche, Dynex DSX	flexibel
Epstein-Barr-Virus, VCA, EA, EBNA, IgG, IgM	Serum, EDTA-Plasma	ECLIA	AA-1624-V002	LIAISON XL / DiaSorin	flexibel
Epstein-Barr-Virus	Serum, EDTA-Plasma	Immunoblot	AA-1538	RemcomScan	flexibel
Hepatitis-A-Virus, Ig	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche	flexibel
Hepatitis-A-Virus, IgM	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche	flexibel
Hepatitis-B-Virus, Anti-HBc Ig	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-B-Virus, Anti-HBc IgM	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-B-Virus, Anti-HBe Ig	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hepatitis-B-Virus, Anti-HBs Ig	Serum, EDTA-Plasma	CMIA/ELISA	AA-1401-V009, AA-1398-V007	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-B-Virus, HBs-Antigen, qualitativ	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-B-Virus, HBs-Antigen, quantitativ	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-C-Virus, Ig	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Hepatitis-C-Virus, Ig	Serum, EDTA-Plasma	Immunoblot	AA-1399-V004	manuell (Mikrogen)	flexibel
Hepatitis-D-Virus, Ig	Serum, EDTA-Plasma	ELISA	AA-1398-V007	manuell (Kit DiaSorin)	flexibel
Hepatitis-E-Virus, IgG, IGM	Serum, EDTA-Plasma	ELISA	AA-1398-V007	r-biopharm DSX	flexibel
Hepatititis-B-Virus, HBe-Antigen	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche; cobas pro	flexibel
Herpes-Simplex-Virus, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Humanes Immundefizienzvirus, Antigen + Antikörper	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche	flexibel
Humanes Immundefizienzvirus, Antikörper	Serum, EDTA-Plasma	Immunoblot	AA-1399-V004	manuell (Mikrogen)	flexibel
Masern IgG/IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Mumps IgG/IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Parvovirus B19, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Rötelnvirus, IgG, IgM	Serum, EDTA-Plasma	CMIA	AA-1401-V009	Cobas e411 / Roche	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Varizella Zoster-Virus, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V010	manuell (EuroImmun)	flexibel
Influenzaviren, Respiratory-Syncytial-Virus, SARS-CoV-2 (nur Alinity)	Abstrich	Real-time PCR (Oligoplex-PCR)	AA-1659-V004, AA-1523-V003,	Alinity m, Abbott; CFX Opus, Biorad	flexibel
Hepatitis-B-Virus, DNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1400-V006, AA-1602-V003, AA-1659-V001	Alinity m, Abbott; cobas 6800, Roche	flexibel
Hepatitis-C-Virus, RNA, Genotyp	Serum, EDTA-Plasma	Real-time PCR	AA-1410-V002	Alinity m, Abbott	flexibel
Hepatitis-C-Virus, RNA, Genotyp Core	Serum, EDTA-Plasma	Real-time PCR	AA-1428-V006	Biometra-Thermocyler	flexibel
Hepatitis-C-Virus, RNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1400-V006, AA-1602-V003, AA-1659-V001	Alinity m, Abbott; cobas pro, Roche	flexibel
Humane Papillomaviren, qualitativ	Abstrich	Real-time PCR	AA-1629-V001, AA-1659-V001	Alinity m, Abbott	flexibel
Humans Immundefizienzvirus, RNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1400-V006, AA-1602-V003, AA-1659-V001	Alinity m, Abbott; cobas 6800, Roche	flexibel
Parvovirus B19 DNA	Biopsat, EDTA-Plasma	Real-time PCR	AA-1570-V003	Rotor-Gene, Qiagen	flexibel
SARS-CoV-2 IgG quantitativ	Serum, EDTA-Plasma	CLIA	AA-1624-V005	Liaison XL	flexibel
HDV-RNA (quantitativ) (HDV-RNA (quantitativ))	Serum, EDTA-Plasma	Real-time PCR	AA-1693-V002	CFX Opus Biorad	flexibel
HEV-RNA (HEV-RNA)	Serum, EDTA-Plasma	Real-time PCR	AA-1692-V002	Rotor-Gene, Qiagen	flexibel
SARS-CoV-2	Abstrich (cobas 6800: nur Nasopharyngeal-Abstrich), Rachenspülung	Real-time PCR	AA-1659-V001, AA-1602-V004	Alinity m, Abbott; cobas 6800, Roche, BioRad CFX384 CFX96	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Herpes-Simplex-Virus-1 und 2 DNA	Abstrich (urogenital)	Real-time PCR	AA-1458-V005	CFX Opus Bio-Rad	flexibel
Adenoviren, DNA, Enteroviren, Rhinoviren, RNA	Abstrich	Real-time PCR	AA-1523-V004	CFX Opus Bio-Rad	flexibel
ABL1	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut,Heparin-Knochenmark, RNA aus Blut und Knochenmark (cDNA wird analysiert)	Amplikon-basiertes NGS, Sequencing-by synthesis, Dragen, JSI medical systems SeqNext	AA-1617-V003, AA-1729-V001, AA-1709-V002	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Glioblastom (MGMT), MGMT-Promotormethylierung	DNA aus FFPE-Gewebe, DNA aus Tumorgewebe	Sanger-Sequenzierung; Amplikon-basiertes NGS, Sequencing-by synthesis, Dragen, JSI medical systems SeqNext	AA-1461-V008, AA-1617-V003, AA-1729-V001, AA-1709-V002	Illumina Series (NovaSeq, Miseq, etc)	flexibel
POLE	DNA aus FFPE-Gewebe, DNA aus Tumorgewebe	Sanger-Sequenzierung; Amplikon-basiertes NGS, Sequencing-by synthesis, Dragen, JSI medical systems SeqNext	-AA-1461-V008, AA-1617-V003, AA-1729-V001, AA-1709-V002	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Fusionen bei soliden Tumoren (NTRK3-ETV6, EWSR1-NR4A3, EWSR1-PBX1, EWSR1-ZNF384, EWSR1-ATF1, EWSR1-PATZ1, EWSR1-DDIT3, EWSR1-SP3, EWSR1-FEV, EWSR1-CREB1, EWSR1-FLI1, EWSR1-ETV4, EWSR1-ETV1, EWSR1-ERG, YY1-EWSR1, EWSR1-ZNF444, EWSR1-SMARCA5, NFATC2-EWSR1, SS18-SSX1, SS18-SSX4, FUS-CREB3L2, FUS-CREB3L1, FUS-DDIT3, FUS-ERG, FUS-ATF1, FUS-FEV)	RNA aus FFPE-Gewebe, RNA aus Tumorgewebe, cDNA	Amplikon-basiertes NGS, Sequencing-by synthesis, Dragen, JSI medical Systems Seqnext	AA-1463-V004, AA-1733-V002, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Fusion bei Leukämien/Lymphomen (ACTN4-MLL, BCR-ABL1, CBFB-MYH11, CDK6-MLL, DEK-CAN, DEK-NUP214, ETV6-ABL1, ETV6-MECOM, ETV6-PDGFRB, ETV6-RUNX1, FUS-ERG, LASP1-MLL, LPP-MLL, MAPRE1-MLL, MLL-ABI1, MLL-ABI2, MLL-ACACA, MLL-ACTN4, MLL-AFF1, MLL-AFF3, MLL-AFF4, MLL-ARHGAP26, MLL-ARHGEF12, MLL-CASC5, MLL-CASP8AP2, MLL-CBL, MLL-CENPK, MLL-CEP170B, MLL-CREBBP, MLL-CT45A2, MLL-DAB2IP, MLL-DCPS, MLL-EEFSEC, MLL-ELL, MLL-EP300, MLL-EPS15, MLL-FLNA, MLL-FOXO3, MLL-FRYL, MLL-GAS7, MLL-GMPS, MLL-GPHN, MLL-KIAA1524, MLL-LASP1, MLL-LPP, MLL-MAML2, MLL-MLLT1, MLL-MLLT10, MLL-MLLT11, MLL-MLLT3, MLL-MLLT4, MLL-MLLT6, MLL-MYO1F, MLL-NCKIPSD, MLL-NRIP3, MLL-PDS5A, MLL-PICALM, MLL-SEPT11, MLL-SEPT2, MLL-SEPT5, MLL-SEPT6, MLL-SEPT9, MLL-SH3GL1, MLL-SORBS2, MLL-TET1, MLL-TOP3A, MLL-ZFYVE19, MN1-ETV6, NPM1-ALK, NPM1-MLF1, NPM1-RARA, NUP98-MLL, PAX5-PML, PML-RARA, RPN1-MECOM, RUNX1-MECOM, RUNX1-RUNX1T1, SET-NUP214, STIL-TAL1, TCF3-HLF, TCF3-PBX1) (Fusion AML-ALL- (BCR), CBFB, DEK, ETV6, FUS, MLL, NPM1, PAX5, PML, MECOM, RUNX1, SET, SIL, TCF3) Fusion bei Leukämien/Lymphomen (ACTN4-MLL, BCR-ABL1, CBFB-MYH11, CDK6-MLL, DEK-CAN, DEK-NUP214, ETV6-ABL1, ETV6-MECOM, ETV6-PDGFRB, ETV6-RUNX1, FUS-ERG, LASP1-MLL, LPP-MLL, MAPRE1-MLL, MLL-ABI1, MLL-ABI2, MLL-ACACA, MLL-ACTN4, MLL-AFF1, MLL-AFF3, MLL-AFF4, MLL-ARHGAP26, MLL-ARHGEF12, MLL-CASC5, MLL-CASP8AP2, MLL-CBL, MLL-CENPK, MLL-CEP170B, MLL-CREBBP, MLL-CT45A2, MLL-DAB2IP, MLL-DCPS, MLL-	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA	Amplikon-basiertes NGS, Sequencing-by synthesis, Dragen, JSI medical Systems Seqnext	AA-1463-V004, AA-1733-V002, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Solide Tumoren (HotSpots: AKT1, ALK, AR, BRAF, CDK4, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR2, FGFR3, GNA11, GNAQ, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, PDGFRA, PIK3CA, RAF1, RET, ROS1, SMO, CNV: ALK, AR, BRAF, CCND1, CDK4, CDK6, EGFR,ERBB2, FGFR1, FGFR2, FGFR3, FGFR4, KIT, KRAS, MET, MYC, MYCN, PDGFRA, PIK3CA, Fusionen: ABL1, AKT3, ALK, AXL, BRAF, EGFR, ERBB2, ERG, ETV1, ETV4, ETV5, FGFR1, FGFR2, FGFR3, MET, NTRK1, NTRK2, NTRK3, PDGFRA, PPARG, RAF1, RET, ROS1)	DNA aus FFPE-Gewebe, DNA und RNA aus Tumorgewebe, DNA und cDNA, RNA aus FFPE-Gewebe (cDNA wird analysiert), RNA aus Tumorgewebe (cDNA wird analysiert)	Amplikon-basiertes NGS, Sequencing-by synthesis , Dragen, VarSeq (Golden Helix)	AA-1463-V005. AA-1733-V002, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
quantitativer Nachweis Fusionsgene (RUNX1::RUNX1T1, CBFB::MYH11, PML::RARA, BCR::ABL1, KMT2A::AFF1, ETV6::RUNX1, TCF3::PBX1, STIL::TAL1)	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1433-V006, AA-0238-V006, AA-1470-V004, AA-1555-V004	ABI 7900HT, QX200 Droplet Digital PCR System (BioRad), QuantStudio 7 Pro	flexibel
quantitativer Nachweis spezifischer Varianten (NPM1 c.860_863dup, c.863_864insCATG, IDH1 c. 395G>A, JAK2 c.1849G>T, MPL c.1544G>T, KIT c.2447A>T, BRAF c.1799T>A, MYD88 c.794T>C, CXCR4 c.1025C>G/A, EGFR c.2573T>G, c.2369C>T, c. 2155G>A, c.2235_2249del15, KRAS c.35G>T, c.35G>C, c.34G>T, c.35G>A, c.34G>C, c.34G>A, c.38G>A, c.183A>C, NRAS c.182A>G	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1433-V006, AA-0238-V006, AA-1470-V004, AA-1555-V004	ABI 7900HT, QX200 Droplet Digital PCR System (BioRad), QuantStudio 7 Pro	flexibel
Chimärismus (Chimärismusanalyse)	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA	Fluoreszenz-markierte Hybridisierungssonden (ddPCR), Fragmentlängenanalyse	AA-1541-V003	ABI Sequencer 3730XL, 3130XL, 3730, QX200 Droplet Digital PCR System (BioRad), QuantStudio 7 Pro	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Phäochromozytom/Paragangliom (MAX)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Mikrosatelliteninstabilität (MSI-Analyse)	DNA aus FFPE-Gewebe, DNA aus Tumorgewebe	Fragmentlängenanalyse	AA-0218, AA-0269	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730, Agarosegelelektrophoreskammer	flexibel
Lynch-Syndrom (HNPCC), z.A. (MLH1-Promotormethylierung)	DNA aus FFPE-Gewebe	Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-0229-V008, AA-1617-V003, AA-1615-V001, AA-1376-V004	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Familiäres Mamma-/Ovarialkarzinomsyndrom (HBOC) (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext)	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Endokrinologische Tumorerkrankungen (endokrinologische Neoplasien) (AIP, AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, GNAS, MEN1, PRKAR1A, PTH, RET)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext)	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Familiäres Paragangliom-/Phäochromozytomsyndrom (PCC/PGL) (MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hereditäre Nierenzellkarzinome (BAP1, FH, FLCN, MET, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, VHL)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Pankreas- /Prostatakarzinomsyndrom (ATM, BRCA1, BRCA2, CDK4, CDKN2A, CHEK2, HOXB13, PALB2, POT1, STK11, TP53)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hereditäre Tumorsyndrome (AIP, AKT1, ANKRD26, AP2S1, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1, CASR, CDC73, CDH1, CDH23, CDK12, CDK4, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CEBPA, CHEK1, CHEK2, CTNNA1, DDB2, DDX41, DICER1, DLST, EGLN1, EPAS1, EPCAM, EPOR, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALTN12, GATA2, GCM2, GNA11, GNAS, GPR101, GREM1, HOXB13, KIF1B, KIT, KITLG, MAD2L2, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PPP2R2A, PTEN, PTH, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54L, RB1, RECQL, RECQL4, RET, RFWD3, RNF43, RPS20, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SLX4, SMAD4, SMARCA4, SAMARCB1, SMARCE1, SRP72, STK11, TERT, TMEM127, TP53, UBE2T, VHL, XPA, XPC,XRCC2, XRCC3)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext)	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hämatologische Neoplasien mit Keimbahnprädisposition (ABCB7, ACD, ADH5, ALAS2, ALDH2, ANKRD26, ATG2B, ATM, BLM, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CSF3R, CTC1, DDX41, DKC1, DNAJC21, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, GLRX5, GSKIP, HAX1, HSPA9, JAGN1, KRAS, LIG4, MAD2L2, MBD4, MDM4, MECOM, MLH1, MSH2, MSH6, MYSM1, NBN, NF1, NHP2, NOP10, PALB2, PARN, PMS2, PTPN11, RAD50, RAD51, RAD51C, RFWD3, RPL11, RPL15, RPL18, RPL23, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLC19A2, SLC25A38, SLX4, SRP54, SRP72, TERC, TERT, THPO, TINF2, TOP3A, TP53, TRNT1, TSR2, UBE2T, VPS45, WAS, WRAP53, WRN, XPC, XRCC2)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext)	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hereditäre Tumorsyndrome (AIP, APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDKN1B, CDKN2A, CDKN2B, CHEK1, CHEK2, DICER1, EPCAM, FH, FLCN, GATA2, GNAS, GREM1, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RB1, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, VHL)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
BRCA-Diagnostik, Therapie PARP-Inhibitor (BRCA1, BRCA2)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext)	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Lynch-Syndrom (HNPCC) (MLH1, MSH2, MSH6, PMS2, EPCAM)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Gastrointestinal Tumorerkrankungen (Polyposis-Syndrome; hereditäres Magenkarzinomsyndrom) (APC, BMPR1A, CDH1, CHEK2, CTNNA1, MLH3, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53, GREM1 (regulatorisceh Region))	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST) Sequencing-by synthesis,Dragen, VarSeq (Golden Helix); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems	AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635	Illumina Series (NovaSeq, Miseq, etc)	flexibel
erworbener Chromosomensatz (Tumorzytogenetik)	Blut, Knochenmark, CD34+ Zellen, CD138+ Zellen	Chromosomenbänderungsanalyse, Fluoreszenz in situ Hybridisierung (FISH)	AA-0257-V013, AA-0251-V006, AA-0335-V013, AA-0239-V007, AA-0242-V005, AA-0244-V003, AA-0249-V004, AA-1675-V001, AA-0245-V007, AA-1319-V005, AA-0221-V008, AA-0256-V010, AA-1596-V001, AA-1621-V002	Metafer (MetaSystems), Zeiss Axio Scope A1, Zeiss Axioskope 2 Plus	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hereditäre Alpha-Tryptasämie (TPSAB1)	EDTA-Blut	Fluoreszenz-markierte Hybridisierungssonden; Chimärismusanalyse; Fragmentlängenanalyse	AA-1433-V006, AA-1541-V003	QX200 Droplet Generator	flexibel
Hypogonadotroper Hypogonadismus / Kallmann-Syndrom (ANOS1, CHD7, DUSP6, FEZF1, FGF8, FGF17, FGFR1, FLRT3, FSHB, GENRH1, GENRHR, HS6ST1 , IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2 , PROKR2, SEMA3A, SOX10, SPRY4, TAC3, TACR3, WDR11)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Ovarialdysgenesie (BMP15, FSHR, MCM9, NR5A1, PSMC3IP)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, VarSeq (Golden Helix)	AA-1637-V002, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Vorzeitige Ovarialinsuffizienz (BMP15, DIAPH2, ESR1, FIGLA, FOXL2, FSHR, GDF9, INHA, LHCGR, NOBOX, NR5A1, SOHLH1, SOHLH2, STAG3)	EDTA-Blut, DNA aus Blut	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, VarSeq (Golden Helix)	AA-1637-V002, AA-1617-V003	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Hypogonadotroper Hypogonadismus 1 mit oder ohne Anosmie (ANOS1-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Androgeninsensitivität (AIS) (AR-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Ovarialdysgenesie Typ 2 /POI (BMP15-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel

Analyt / Indikation (Messgröße; Gen/e, Variante/n)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	Akkreditierungs-status
Hypogonadotroper Hypogonadismus 6 mit oder ohne Anosmie (FGF8-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hypogonadotroper Hypogonadismus 2 mit oder ohne Anosmie (FGFR1-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
FISH-Rezeptor-Defizienz / Ovarialdysgenesie Typ 1 /POI (FSHR-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-0197-V007, AA-1313-V007, AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hypogonadotroper Hypogonadismus 4 mit oder ohne Anosmie (PROK2-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Hypogonadotroper Hypogonadismus 3 mit oder ohne Anosmie (PROKR2-Gen)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Azoospermie (AZF-Mikrodeletionen)	EDTA-Blut, DNA aus Blut	Fragmentlängenanalyse	AA-0272-V005, AA-0284-V008	Thermocycler	flexibel
Ovarielles Hyperstimulationssyndrom (OHSS) (FSHR-Gen: dbSNP rs6166)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	flexibel
V. a. Fertilitätsstörung, wiederholte Fehlgeburten (ANXA5-M2 Genotyp: dbSNP rs112782763, rs28717001, rs28651243, rs113588187)	EDTA-Blut, DNA aus Blut	Sanger-Sequenzierung	AA-1668-V001, AA-0272-V005, AA-0269-V007	ABI Sequencer 3730XL, 3130XL, 3730	flexibel

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PGT-A (Aneuploidiediagnostik), Chromosomensatz (zur Abklärung einer de novo Chromosomenveränderung)	Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD, Genomische DNA aus Einzelzellen	NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA, Sureplex DNA Amplification System), Bluefuse Multi Software	AA-1585-V003, AA-1700-V001	Illumina Series (NovaSeq, Miseq, etc)	flexibel
PGT-SR (Translokationsdiagnostik), partieller Chromosomensatz (zur Abklärung einer bekannten familiären Chromosomenveränderung)	Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD, Genomische DNA aus Einzelzellen	NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA, Sureplex DNA Amplification System), Bluefuse Multi Software	AA-1585-V003, AA-1700-V001		flexibel
Hohes Risiko einer schwerwiegenden Erbkrankheit für die Nachkommenschaft (PGT-M)	Genomische DNA aus Einzelzellen, Polkörper, Trophektodermzellen	Fragmentlängenanalyse	AA-1378-V007	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Aneuploidiediagnostik (nicht invasiver Pränataltest): Trisomie 21, Trisomie, 18, Trisomie 13, gonosomale Aberrationen, Mikrodeletionen	BCT-Blut (Streck), zellfreie (fetale&maternale) DNA aus Blut	Gesamtgenomsequenzierung, Sequencing-by synthesis, Dragen	AA-1640-V001, AA-1641-V001, AA-1642-V001, AA-1643-V004, AA-1644-V001,	Illumina Series (NovaSeq, Miseq, etc)	flexibel
Kallmann-Syndrom (FGFR1-, GNRHR-, KISS1R-, GNRH1-, NELF-, PROK2-, PROKR2-Gen)	EDTA-Blut, DNA aus Blut	(MS) MLPA	AA-0103-V008	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730	flexibel
Segregationsanalyse, CNV-Bestätigung/-Ausschluss, STRC-CNV-Analyse (Genomische Imbalancen)	EDTA-Blut, Genomische DNA aus Blut, natives Abortgewebe / kultivierte Zellen aus Fruchtwasser und Chorionzotten	Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR)	AA- 1420-V006	LC480II, LC1.2 (Roche), CFX96/384Touch (BioRad)	flexibel

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Uniparentale Disomie 14 (UPD 14)	genomische DNA	Mikrosatellitenanalyse (Fragmentanalyse STR)	AA-1526-V002	Thermocycler, ABI Sequencer 3730	flexibel
Uniparentale Disomie 15 (UPD 15)	genomische DNA	Mikrosatellitenanalyse (Fragmentanalyse STR)	AA-1526-V002	Thermocycler, ABI Sequencer 3730	flexibel
angeborener Chromosomensatz (Array-CGH)	Genomische DNA aus Blut sowie aus nativen Zellen und Zellkultur von Zellen aus Fruchtwasser, Chorionzotten oder Abortmaterial	Array basierte CGH	AA-0351-V014	SureScan Microarray Scanner G2505C (Agilent)	flexibel
angeborener Chromosomensatz (Chromosomenanalyse & FISH)	Peripheres Blut, Fruchtwasser, Chorionzotten, Abortgewebe, Nabelschnurblut, Haut, Knochenmark	Chromosomenbänderungsanalyse, Fluoreszenz in situ Hybridisierung (FISH): Pränataler Schnelltest, Mikrodeletionsdiagnostik, Chromosomenpainting, Subtelomeranalysen, Interphase-Untersuchungen, Vielfarbenkaryotypisierung	AA-0335-V013, AA-0356-V010, AA-1390-V005		flexibel
angeborener Chromosomensatz	Genomische DNA aus Blut sowie aus Zellkultur von Zellen aus Fruchtwasser oder Chorionzotten	Molekulare Karyotypisierung	AA-1651-V003	Bionano Saphyr	flexibel
Fazioskapulohumerale Muskeldystrophie (FSHD1)	EDTA-Blut; DNA	Molekulare Karyotypisierung	AA-1651-V003	Bionano Saphyr	flexibel

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Hämophilie (F8, F9)	EDTA-Blut, DNA aus Blut	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-1413-V003, AA-1637-V003, AA-1391-V010, AA-1648-V002	Illumina Plattform	flexibel