Untersuchungsgebiet: Klinische Chemie

Untersuchungsart:
Durchflusszytometrie (Partikeleigenschaftsbestimmungen)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Retikulozyten	EDTA-Blut	Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V011	Sysmex XN1000, Sysmex	x	
Großes Blutbild	EDTA-Blut	Partikelzählung, Partikelgrößenbestimmung, Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V011	Sysmex XN1000, Sysmex	x	
Kleines Blutbild	EDTA-Blut	Partikelzählung, Partikelgrößenbestimmung, Bestimmung zytochemisch-zytometrischer Merkmale	AA-0178-V011	Sysmex XN1000, Sysmex	x	
Thrombozyten	EDTA-Blut	Partikelzählung, optisch-elektronisch	AA-0178-V011	Sysmex XN1000, Sysmex	x	

Untersuchungsart:
Elektrochemische Untersuchungen*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chlorid	Serum, Urin	Ionenselektive Elektrode (ISE)	AA-1480-V007	Cobas pro, Roche	x	
Kalium	Serum, Urin	Ionenselektive Elektrode (ISE)	AA-1480-V007	Cobas pro, Roche	x	
Natrium	Serum, Urin	Ionenselektive Elektrode (ISE)	AA-1480-V007	Cobas pro, Roche	x	

Untersuchungsart:
Elektrophorese*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Monoklonale Gammopathie	Serum	Kapillarelektrophorese	AA-1530-V004	Sebia Capillarys 3 Octa, Sebia	x	
Serumeiweiß	Serum	Kapillarelektrophorese	AA-1486-V006	Sebia Capillarys 3 Octa, Sebia	x	

Untersuchungsart:
Koagulometrie*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Antithrombin III	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
D-Dimer	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Fibrinogen	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
INR	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Partielle Thromboplastinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Plasmathrombinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Faktor 9	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
freies Protein S	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Protein C	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
APC-Resistenz, FV Leiden	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Faktor 8	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Faktor 12	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
Thromboplastinzeit	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	
MCLA HSS/LSS	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	

LA DRVVT S/C	Citratplasma	optische Detektionsverfahren	AA-1528-V008	ACLTop 750, Werfen	x	

Untersuchungsart:
Ligandenassays*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
AFP	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Anti-Müller-Hormon	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Beta-HCG	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
CA 125	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
CA 15-3	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
CA 19-9	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
CEA	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Cortisol	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
C-Peptid	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
DHEA-S	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Folsäure	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
FSH	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
FT3	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
FT4	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Holotranscobalamin (Active B12)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Humanes Wachstumshormon (hGH)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Insulin	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
insulinähnlicher Wachstumsfaktor-1 (IGF-1)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
LH	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
N-terminales pro-B-Typ natriuretisches Peptid (NPROBNP)	Serum	ECLIA	AA-1487-V006	Cobas e411, Roche	x	
Osteocalcin	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Östradiol	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Parathormon (PTH)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Progesteron	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Prolaktin	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
PSA	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
SHBG	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
ß-CrossLaps	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Testosteron	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Troponin T	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
TSH	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Vitamin B12	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Vitamin D	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Humanes Thyreoglobulin (HTG)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
Prokollagen Typ 1 N-terminales Propetid (P1NP)	Serum	ECLIA	AA-1487-V006	Cobas pro, Roche	x	
AFP	Fruchtwasser	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, ThermoFisher	x	
Freies Beta-HCG	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, ThermoFisher	x	
PAPP-A	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, ThermoFisher	x	
Procalcitonin	Serum	Immunometrie (CLIA)	AA-1517-V005	Kryptor compact plus, ThermoFisher	x	
Chromogranin A (CGA)	Serum	CLIA	AA-1517-V005	Kryptor compact plus, ThermoFisher	x	
17-OH-Progesteron	Serum	ELISA	AA-1703-V001	Dynex DSX, R-Biopharm	x	

5alpha-Dihydrotestosteron (DHT)	Serum	ELISA	AA-1703-V001	Dynex DSX, R-Biopharm	x	
DHEA	Serum	ELISA	AA-1703-V001	Dynex DSX, R-Biopharm	x	
Inhibin B	Serum	ELISA	AA-1703-V001	Dynex DSX, R-Biopharm	x	
Okkultes Blut im Stuhl (iFOBT) - Hämoglobin im Stuhl	Stuhl	ELISA	AA-MR-QM-1813	Dynex DSX, R-Biopharm	x	
1,25 Dihydroxivitamin D	Serum	Immunometrie (CLIA)	AA-1624-V004	LIAISON XL, DiaSorin	x	
Aldosteron	Serum	CLIA	AA-1624-V004	LIAISON XL, DiaSorin	x	
Androstendion	Serum	Immunometrie (CLIA)	AA-1624-V004	LIAISON XL, DiaSorin	x	
BAP	Serum	Immunometrie (CLIA)	AA-1624-V004	LIAISON XL, DiaSorin	x	
Calcitonin	Serum	Immunometrie (CLIA)	AA-1624-V004	LIAISON XL, DiaSorin	x	
SFLT1	Serum	Immunometrie (CLIA)	AA-1517-V006	Kryptor compact plus, ThermoFisher	x	
PLGF	Serum	Immunometrie (CLIA)	AA-1517-V006	Kryptor compact plus, ThermoFisher	x	

Untersuchungsart:

Mikroskopie*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Erythrozyten	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Erythrozytenzylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Granulierte Zylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Hyaline Zylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Leukozyten	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Leukozytenzylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Plattenepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Rundepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Übergangsepithelien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Wachszylinder	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
atypische Lymphozten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Basophile Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Blasten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Eosinophile Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Kernschatten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Lymphozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Metamyelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Monozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Morphologie Erythrozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Morphologie Leukozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Morphologie Thrombozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Myelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Normoblasten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	

Plasmazellen	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Promyelozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Segmentkernige Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	
Stabkernige Granulozyten	Vollblut	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-0167-V010	Mikroskop	x	

Untersuchungsart:
Osmometrie*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Osmolalität	Serum, Urin	Kyroskopie	AA-1678-V002	Osmometer 3000D, Gonotec	x	

Untersuchungsart:
Qualitative Untersuchungen (einfache) mit visueller Auswertung*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Bilirubin	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Glucose	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Hämoglobin/Erythrozyten	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Ketone	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Leukozyten	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Nitrit	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
pH	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Proteine	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	
Urobilinogen	Urin	Teststreifen	AA-1467-V004	Teststreifen	x	

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

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Antistreptolysin O (ASLO)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Complement C3c (C3)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Complement C4 (C4)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Albumin	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Albumin	Urin	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Alkalische Phosphatase	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Amylase	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Bilirubin, direkt	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Bilirubin, gesamt	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Calcium	Serum, Urin	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
CHE	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Cholesterin	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
CK	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
CK-MB	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Cystatin C (CYSC)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Eisen	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Eiweiß	Urin	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Eiweiß, gesamt	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
GGT	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Glucose	Serum, NaF-Plasma	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	

Glutamat-Oxalacetat-Transaminase (GOT) bzw. Aspartat-Aminotransferase (AST)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Glutamat-Pyruvat-Transaminase (GPT) bzw. Alanin-Aminotransferase (ALT)	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Harnsäure	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Harnstoff	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
HDL Cholesterin	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Homocystein	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Kreatinin	Serum, Urin	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
LDH	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
LDL	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Lipase	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Magnesium	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Phosphat	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Triglyceride	Serum	UV-/VIS-Photometrie	AA-1480-V007	Cobas pro	x	
Blutsenkung (in mm/h)	EDTA-Blut	Stopped-Flow-Technik	AA-1595-V001	Roller 20LC, Sysmex	x	

Untersuchungsart:
Spektrometrie (Turbidimetrie / Immunturbidimetrie)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
alpha 1 Antitrypsin	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Coeruloplasmin	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
C-reaktives Protein	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Ferritin	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Haptoglobin	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
HbA1c	EDTA-Blut	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Transferrin	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Lp (a)	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	

Untersuchungsgebiet: Immunologie

Untersuchungsart:
Durchflusszytometrie**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Lymphozytentypisierung	CPDA1-Blut, EDTA-Blut	Durchflusszytometrie	AA-0173-V012	FACS Canto II, BD		x

Untersuchungsart:
Ligandenassays*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
IgE	Serum	ECLIA	AA-1487-V007	Cobas pro, Roche	x	
MAK	Serum	ECLIA	AA-1487-V007	Cobas pro, Roche	x	
TAK	Serum	ECLIA	AA-1487-V007	Cobas pro, Roche	x	
TSH-Rezeptor-Antikörper	Serum	ECLIA	AA-1487-V007	Cobas e41, Roche	x	
Myositis-Blot (Mi-2ß-AAk, Ku-AAk, PM-Scl-AAk, Jo-1-AAk, PL-7-AAk, PL-12-AAk, Ro-52-AAk)	Serum	Immunoblot (Westernblot)	AA-MR-QM-1409-V5	Teststreifen, Euroimmun	x	

Sklerodermie-Blot (Scl-70-AAk, CENP-A-AAk, CENP-B-AAk, RP11-AAk, RP155-AAk, Fibrillarin-AAk, NOR 90-AAk, Th/To-AAk, PM-Scl100-AAk, PM-Scl75-AAk, Ku-AAk, PDGFR-AAk, Ro-52-AAk)	Serum	Immunoblot (Westernblot)	AA-MR-QM-1409-V5	Teststreifen, Euroimmun	x	
Leberblot (AMA M2 (nativ) IgG, AMA M2-3E BPO (rekomb) IgG, Sp100 IgG, PML IgG, gp210 IgG, LKM-1 IgG, LC-1 IgG, SLA/LP IgG, F-Aktin IgG, Ro-52 IgG)	Serum	Immunoblot (Westernblot)	AA-MR-QM-1409-V5	Teststreifen, Euroimmun	x	

Untersuchungsart:
Mikroskopie*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Antigen der Mikrosomenfraktion aus Leber und Niere (LKM-1)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V004	IFT, Euroimmun	x	
Antikörper gegen glatte Muskulatur (ASMA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V004	IFT, Euroimmun	x	
Antimitochondriale Antikörper (AMA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V004	IFT, Euroimmun	x	
Antineutrophile cytoplasmatische Antikörper (ANCA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V004	IFT, Euroimmun	x	
Autoantikörper gegen Zellkerne (ANA)	Vollblut, Serum, EDTA-Plasma	Indirekte Immunfluoreszenzmikroskopie	AA-1351-V004	IFT, Euroimmun	x	

Untersuchungsart:
Spektrometrie (Turbidimetrie / Immunturbidimetrie)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Ig A	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Ig G	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
IgM	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	
Rheumafaktor	Serum	Turbidimetrie	AA-1480-V007	Cobas pro, Roche	x	

Untersuchungsgebiet: Humangenetik

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart:
Chromosomenanalyse**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung (siehe Zeile 3), sofern zutreffend
angeborener Chromosomensatz	Blut, Fruchtwasser, Fibroblasten, Chorionzotten, und andere Gewebeproben	Chromosomenbänderungsanalyse	AA-MR-QM-0335-V14			x	

erworbener Chromosomensatz	Blut, Knochenmark	Chromosomenbänderungsanalyse	AA-MR-QM-0251-V7, AA-MR-QM-1319-V5, AA-MR-QM-0221-V8, AA-MR-QM-0256-V10, AA-MR-QM-1596-V2, AA-MR-QM-1621-V3			x	
angeborener Chromosomensatz	Native oder kultivierte Zellen aus Blut, Fibroblasten, Amnionzellen, Chorionzotten oder andere Gewebeproben	Fluoreszenz-in-situ-Hybridisierung (FISH)	AA-MR-QM-0335-V14 AA-MR-QM-0356-V10 AA-MR-QM-1390-V6			x	
erworbener Chromosomensatz	Blut, Knochenmark	Fluoreszenz-in-situ-Hybridisierung (FISH)	AA-MR-QM-0257-V13, AA-MR-QM-0251-V7, AA-MR-QM-0335-V14, AA-MR-QM-0239-V7, AA-MR-QM-0242-V5, AA-MR-QM-0244-V3, AA-MR-QM-0249-V4, AA-MR-QM-1675-V2, AA-MR-QM-0245-V7, AA-MR-QM-1319-V5, AA-MR-QM-0221-V8, AA-MR-QM-0256-V10, AA-MR-QM-1596-V2, AA-MR-QM-1621-V3, AA-MR-QM-1675-V2			x	
angeborener Chromosomensatz	Blut, native Zellen und Zellkultur von Zellen aus Fruchtwasser, Chorionzotten oder andere Gewebeproben; DNA	Molekulare Karyotypisierung mittels Array-CGH	AA-MR-QM-0351-V14	Scanner, Agilent		x	
angeborener Chromosomensatz	Blut, native Zellen und Zellkultur von Zellen aus Fruchtwasser, Chorionzotten; DNA	Molekulare Karyotypisierung mittels optischer Genomkartierung	AA-MR-QM-1651-V4	Saphyr, Bionano		x	
Chromosomensatz (zur Abklärung einer de novo Chromosomenveränderung, PGT-A)	Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD; DNA	NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA, Embryomap Sample Prep), eMap	AA-MR-QM-1773-V1, AA-MR-QM-1780-V1, AA-MR-QM-1787-V2	Illumina Series (MiSeq)		x	
Chromosomensatz (zur Abklärung einer bekannten familiären Chromosomenveränderung, PGT-SR)	Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD, ; DNA	NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA; Embryomap Sample Prep), eMap	AA-MR-QM-1773-V1, AA-MR-QM-1780-V1, AA-MR-QM-1787-V2	Thermocycler, Illumina Series (MiSeq)		x	
Chromosomensatz (zur Abklärung einer bekannten familiären Chromosomenveränderung, PGT-M	Blut, Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD; DNA	PCR, Minisequenzierung, Fragmentanalyse	AA-MR-QM-1378-V9	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730		x	
Fazioskapulohumerale Muskeldystrophie (FSHD1)	EDTA-Blut, Lithium-Heparin-Blut, DNA; DNA	Molekulare Karyotypisierung mittels optischer Genomkartierung	AA-1651-V003	Bionano Saphyr		x	

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart:

Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version_x000D_ Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung (siehe Zeile 3), sofern zutreffend

Hypogonadotroper Hypogonadismus / Kallmann-Syndrom (NGS-Panelanalyse; ANOS1 [OMIM #300836], CHD7 [OMIM #608892], DUSP6 [OMIM #602748], FEZF1 [OMIM #613301], FGF8 [OMIM #612702], FGF17 [OMIM #603725], FGFR1 [OMIM #147950], FLRT3 [OMIM #604808], FSHB [OMIM #136530], GENRH1 [OMIM #152760], GENRHR [OMIM #138850], HS6ST1 [OMIM #604846], IL17RD [OMIM #606807], KISS1 [OMIM #603286], KISS1R [OMIM #604161], LHB [OMIM #152780], NSMF [OMIM #608137], PROK2 [OMIM #607002], PROKR2 [OMIM #607123], SEMA3A [OMIM # 603961], SOX10 [OMIM #602229], SPRY4 [OMIM #607984], TAC3 [OMIM #162330], TACR3 [OMIM #162332], WDR11 [OMIM #606417]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Ovarialdysgenesie (NGS-Panelanalyse; BMP15 [OMIM #300247], FSHR [OMIM #136435], MCM9 [OMIM #610098], NR5A1 [OMIM #184757], PSMC3IP [OMIM #608665]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Vorzeitige Ovarialinsuffizienz (NGS-Panelanalyse; BMP15 [OMIM #300247], DIAPH2 [OMIM #300108],ESR1 [OMIM #133430], FIGLA [OMIM #608697], FOXL2 [OMIM #605597], FSHR [OMIM #136435], GDF9 [OMIM #601918], INHA [OMIM #147380], LHCGR [OMIM #152790], NOBOX [OMIM #610934], NR5A1 [OMIM #184757], SOHLH1 [OMIM #610224], SOHLH2 [OMIM #616066], STAG3 [OMIM #608489], MCM9, PSMC3IP, SYCE1, ERCC6)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Spermiendefekte (NGS-Panel; AR, DMRT1, M1AP, NR5A1, TEX11, TEX14); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
46,XY-DSD Varianten der Geschlechtsdifferenzierung (NGS-Panel; AR, DHH, DMRT1, HSD17B3, HSD3B2, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX9, SRD5A2, SRY, TSPYL1, WNT4, WT1, WWOX); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Angelman-Syndrom (UBE3A-Gen [OMIM #601623])	EDTA-Blut, DNA; DNA	MS MLPA	AA-MR-QM-0370-V5, AA-MR-QM-0269-V7, AA-MR-QM-0272-V5	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730		x	

Androgeninsensitivität (AIS) (AR-Gen [OMIM #313700])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen-Pipeline, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
FSH-Rezeptor-Defizienz / (FSHR-Gen [OMIM #136435])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen-Pipeline, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Azoospermie (AZF-Mikrodeletionen)	EDTA-Blut, DNA; DNA	PCR, Gelelektrophorese	AA-MR-QM-0272-V5, AA-MR-QM-0284-V10	Thermocycler, Agarosegelelektrophoresekammer		x	
V. a. Fertilitätsstörung, wiederholte Fehlgeburten (ANXA5-M2 Genotyp: dbSNP rs112782763, rs28717001, rs28651243, rs113588187)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V1, AA-MR-QM-0269-V7, AA-MR-QM-0272-V5	ABI Sequencer 3730XL, 3130XL, 3730,		x	
Non-invasive prenatal testing (VERACITY-NIPT)	BCT-Blut (Streck); cfDNA	Sequence Capture, Sequencing-by-synthesis (Illumina), Software Polaris	AA-MR-QM-1640-V3, AA-MR-QM-1641-V4, AA-MR-QM-1642-V2, AA-MR-QM-1643-V5, AA-MR-QM-1644-V1	Illumina Series (NovaSeq)	x		
Prader-Willi-Syndrom/Angelman-Syndrom (Deletionen, Duplikationen, Methylierung in Chromosomenregion 15q11.2-q13, SNRPN)	DNA aus Blut, Chorionzotten, Amniozyten, Mundschleimhaut, DNA; DNA	(MS) MLPA	AA-MR-QM-1756-V1	ABI Sequencer 3730XL; ABI Sequencer 3730	x		
Segregationsanalyse, CNV-Bestätigung/-Ausschluss, STRC-CNV-Analyse	EDTA-Blut, DNA, Gewebeproben[a], kultivierte Zellen aus Fruchtwasser und Chorionzotten; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1420-V7	CFX96/384Touch, BioRad, Easy PGX (Diatech)		x	
Uniparentale Disomie 14 / 15 (UPD 14/15)	Blut, kultivierte Zellen aus Fruchtwasser und Chorionzotten, Mundschleimhaut, DNA; DNA	Fragmentlängenanalyse	AA-MR-QM-1526-V2	ABI Sequencer 3730XL, 3130XL, 3730		x	
ABL1 (OMIM # 189980); SNV	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA, cDNA; RNA, cDNA	Amplikonbasiertes NGS; Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1617-V3 AA-MR-QM-1709-V2	Illumina Series (NovaSeq)		x	
Glioblastom (MGMT-Promotormethylierung); SNV	Gewebeproben, DNA; DNA	Bisulfitkonvertierung; amplikonbasiertes NGS; Sequencing-by synthesis; Dragen, JSI medical systems SeqNext	AA-MR-QM-1376-V5 AA-MR-QM-1617-V3 AA-MR-QM-1709-V2	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Lynch-Syndrom (HNPCC) (MLH1-Promotormethylierung (OMIM #120436)); SNV	Gewebeproben, EDTA-Blut, DNA; DNA	Bisulfitkonvertierung, amplikon-basiertes NGS Sequencing-by synthesis; Dragen, JSI medical systems SeqNext	AA-MR-QM-1376-V5 AA-MR-QM-1617-V3 AA-MR-QM-1709-V2	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen

Somatische Varianten bei Endometriumkarzinomen, (POLE (OMIM #174762)); SNV	Gewebeproben, DNA; DNA	Amplikon-basiertes NGS, Sequencing-by synthesis; Dragen, JSI medical systems SeqNext	AA-MR-QM-1617-V3 AA-MR-QM-1709-V2	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Solide Tumore, hämatologische Neoplasien (TP53 (OMIM #191170)); SNV	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, DNA, Knochenmark, Gewebeproben; DNA	Amplikonbasiertes NGS; Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-MR-QM-1733-V4 AA-MR-QM-1617-V3	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Hämatologische Neoplasien (ABL1, ACTG1, ANGPT2, ANKRD26, ARID1A, ARID1B, ARID2, ARID5B, ASXL1, ASXL2, ATM, BCL10, BCL11B, BCL2, BCL6, BCOR, BCORL1, BIRC3, BRAF, BTK, CALR, CARD11, CBL, CCND1, CCND3, CCR4, CCR7, CD177, CD40LG, CD79A, CD79B, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDH2, CDH8, CEBPA, CREBBP, CSF3R, CUX1, CXCR4, CYLD, DDX3X, DDX41, DHX15, DIS3, DNMT1, DNMT3A, DNMT3B, EGR1, EGR2, ELANE, EP300, EPHA7, ETNK1, ETV6, EZH2, FAS, FBXW7, FGFR3, FLT3, FOXO1, FYN, GATA1, GATA2, GATA3, GNA13, GNAS, HIST1H1E, HNRNPA2B1, HRAS, IDH1, IDH2, IKZF1, ILR7, JAK1, JAK2, JAK3, KDM1A, KDM6A, KDR, KLF2, KIT, KMT2A, KMT2C, KMT2D, KRAS, LTB, MAG, MALT1, MAP2K1, MAP3K14, MAX, MECOM, MED12, MEF2B, MPL, MYBBP1A, MYC, MYD88, NCOR1, NF1, NF2, NFKB1, NFKB2, NFKBIB, NFKBIE, NFKBIZ, NOTCH1, NOTCH2, NPM1, NRAS, NSD2, NSD3, PDGFRA, PDGFRB, PHF6, PLCG1, PLCG2, POT1, PRDM1, PRKCB, PRPF8, PTEN, PTPN11, PTPN13, PTPN14, PTPN23, PTPRD, PTPRT, RAD21, RB1, RHOA, RIMS1, RPS15, RUNX1, S1PR1, SCRIB, SETBP1, SETDB1, SETD2, SF3B1,	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1617-V3 AA-MR-QM-1709-V2 AA-MR-QM-1733-V4 Panel TPv3b	Illumina Series (NovaSeq)		x	

BRCA-Diagnostik, Therapie PARP-Inhibitor (BRCA1 [OMIM #113705], BRCA2 [OMIM #600185]), SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1637-V3 Panel TPv3a	Illumina Series (NovaSeq)		x	
Familiäres Mamma-/Ovarialkarzinom (HBOC) (ATM [OMIM #607585], BARD1 [OMIM #601593], BRCA1, BRCA2, BRIP1 [OMIM #605882], CDH1 [OMIM #192090], CHEK2, PALB2, PTEN, RAD51C [OMIM #179617], RAD51D [OMIM #602954], STK11, TP53 [OMIM #191170]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 Panel TPv3a	Illumina Series (NovaSeq)		x	
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	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 PanelTPv3b	Illumina Series (NovaSeq)		x	

Hereditäre Tumorsyndrome (AIP [OMIM #605555], AKT1 [OMIM #164730], ANKRD26 [OMIM #610855], AP2S1, APC [OMIM #611731], ATM [OMIM #607585], AXIN2, BAP1 [OMIM #603089], BARD1 [OMIM #601593], BLM, BMPR1A [OMIM #601299], BRCA1 [OMIM #113705], BRCA2 [OMIM #600185], BRIP1 [OMIM #605882], BUB1, CASR, CDC73 [OMIM #607393], CDH1 [OMIM #192090], CDH23, CDK12 [OMIM #615514], CDK4 [OMIM #123829], CDKN1A, CDKN1B [OMIM #600778], CDKN2A [OMIM #600160], CDKN2B, CDKN2C, CEBPA [OMIM #116897], CHEK1 [OMIM #603078], CHEK2 [OMIM #604373], CTNNA1 [OMIM #116805], DDB2, DDX41 [OMIM #608170], DICER1 [OMIM #606241], DLST, EGLN1 [OMIM #606425], EPAS1, EPCAM [OMIM #185535], EPOR, ERCC2, ERCC3, ERCC4 [OMIM #133520], ERCC5, ETV6 [OMIM #600618], FANCA [OMIM #607139], FANCB [OMIM #300515], FANCC [OMIM #613899], FANCD2 [OMIM #613984], FANCE [OMIM #613976], FANCF [OMIM #613897], FANCG [OMIM #602956], FANCI [OMIM #611360], FANCL [OMIM #608111],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
Qualitativer Nachweis Fusionsgene NSCLC (ALK [OMIM #105590], RET [OMIM #164761], ROS1 [OMIM #165020])); SNV	DNA, RNA, cDNA, Gewebeproben; RNA, cDNA	Amplikonbasiertes NGS, Sequencing-by synthesis; Dragen	AA-MR-QM-1733-V4 AA-MR-QM-1617-V3	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen

Qualitativer Nachweis Fusionsgene Sarkome (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,	DNA, RNA, cDNA, Gewebeproben; RNA, cDNA	Amplikonbasiertes NGS, Sequencing-by synthesis; Dragen	AA-MR-QM-1733-V4 AA-MR-QM-1617-V3	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Solide Tumore (TSO500-Panel)	DNA, RNA, Gewebeproben; cDNA	Sequence capture (TSO500), Sequencing-by synthesis; Dragen, VarSeq (Golden Helix), MH Guide (Molecular Health)	AA-MR-QM-1732-V1 Panel TSO500	Illumina Series (NovaSeq)	x		ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Solide Tumoren (AKT1, ALK, APC, AR, ARID1A, ARID1B, ATM, ATR, ATRX, BAP1, BARD1, BMPR1A, BRAF, BRCA1, BRCA2, BRINP1, CBL, CCAR2, CCND1, CDK12, CDK4, CDKN2A, CDKN2B, CHEK1, CHEK2, CREBBP, CTNNB1, CPYD, E2F3, EIF1AX, EP300, EPCAM, ERBB2, ERBB3, ERCC2, ESR1, FANCA, FANCC, FANCL, FAT1, FGFR1, FGFR2, FGFR3, FOXA1, GNA11, GNAQ, GNAS, H3F3A, HIST1H3B, HIST1H3C, HRAS, IDH1, IDH2, JAK1, KDM6A, KIT, KRAS, LTK, MAP2K1, MDM2, MEN1, MGMT, MITF, MLH1, MLH3, MSH2, MSH6, MTHFR, MYC, NBN, NCOR1, NCOR2, NF1, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PDGFRA, PIK3CA, PIK3CB, PMS2, PRKN, PTCH1, PTEN, RAD51, RAD51B, RAD51C, RAD51D, RB1, RET, ROS1, SDHA, SDHB, SDHC, SDHD, SMAD4, SPOP, STAG2, STK11, SUFU, TERT, TP53, TSC1, TYMS, VEGFA,	DNA, Gewebeproben, BCT-Blut; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin); Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by synthesis, JSI medical systems SeqNext	AA-MR-QM-1617-V3 AA-MR-QM-1709-V2 AA-MR-QM-1733-V4 Panel TPv3b	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen

Solide Tumoren (ALK, ATM, BRAF, BRCA1, BRCA2, CDK12, CHEK2, EGFR, ERBB2, FANCA, FGFR2, FGFR3, KIT, KRAS, MET, NF1, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PDGFRA, PIK3CA, PTEN, RAD51L3, RET, ROS1, SDHA, SMAD4); SNV	Gewebeproben, DNA; DNA	Amplikonbasiertes NGS, Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1617-V3 AA-MR-QM-1733-V4	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
Ovarialkarzinom, Mammakarzinom, Pankreaskarzinom, Prostatakarzinom (BRCA1, BRCA2); SNV	Gewebeproben, DNA; DNA	Amplikonbasiertes NGS, Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1617-V3 AA-MR-QM-1733-V4	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
qualitativer Nachweis Fusionsgene AML/ALL-Panel (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,	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA, Knochenmark, cDNA; RNA, cDNA	Amplikonbasiertes NGS; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1733-V4 AA-MR-QM-1617-V3	Illumina Series (NovaSeq)		x	

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,	Gewebeproben, DNA, RNA, cDNA; DNA, RNA, cDNA	Amplikonbasiertes NGS; Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-MR-QM-1733-V4 AA-MR-QM-1617-V3	Illumina Series (NovaSeq)		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen
quantitativer Nachweis Fusionsgene AML, ALL, CML, MPN (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, cDNA; RNA, cDNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1433-V9 AA-MR-QM-0238-V6 AA-MR-QM-1470-V5 AA-MR-QM-1555-V5	QX200 Droplet Digital PCR System (BioRad) QuantStudio 7 Pro		x	
Leukämien, solide Tumoren (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, DNA, BCT-Blut; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1433-V9	QX200 Droplet Digital PCR System (BioRad)		x	
Chimärismusanalyse	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1541-V3	ABI Sequencer 3730XL, 3130XL, 3730, QX200 Droplet Digital PCR System (BioRad)		x	
Chimärismusanalyse	EDTA-Blut, EDTA-Knochenmark,Heparin-Blut, Heparin-Knochenmark, DNA; DNA	Amplikon-basiertes NGS, Sequencing-by-synthesis, Advyser-Software	AA-MR-QM-1770-V1	Illumina Series (NovaSeq)		x	
Hereditäre Alpha-Tryptasämie (TPSAB1-Kopienzahlveränderungen)	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1728-V1	QX200 Droplet Digital PCR System (BioRad)		x	
Lynch-Syndrom (HNPCC); (EPCAM [OMIM #185535], MLH1 [OMIM #120436], MSH2 [OMIM #609309], MSH6 [OMIM #600678], PMS2 [OMIM #600259]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	

Endokrinologische Tumorerkrankungen (endokrinologische Neoplasien) (AIP [OMIM #605555], AP2S1, CASR, CDC73, CDKN1B [OMIM #600778], GCM2, GNA11, GNAS, MEN1 [OMIM #613733], PRKAR1A, PTH, RET [OMIM #164761]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
Familiäres Phäochromozytom-/Paragangliomsyndrom (PGL/PCC) (MAX [OMIM #154950], RET [OMIM #164761], SDHA [OMIM #600857], SDHAF2 [OMIM #613019], SDHB [OMIM #185470], SDHC [OMIM #602413], SDHD [OMIM #602690], TMEM127, VHL [OMIM #608537]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
Gastrointestinale Tumorerkrankungen (Polyposis-Syndrome; hereditäres Magenkarzinomsyndrom) (APC [OMIM #611731], BMPR1A, CDH1, CHEK2, CTNNA1, MLH3, MSH3, MUTYH [OMIM #604933], NTHL1 [OMIM #602656], POLD1 [OMIM #174761], POLE [OMIM #174762], PTEN, RNF43, SMAD4, STK11, TP53, GREM1 (regulatorische Region)); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
Hereditäres Nierenzellkarzinom (BAP1 [OMIM #603089], FH [OMIM #136850], FLCN [OMIM #607273], MET [OMIM #164860], PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, VHL [OMIM #608537]); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
Pankreas-/Prostatakarzinomsyndrom (ATM, BRCA1, BRCA2, CDK4, CDKN2A, CHEK2, HOXB13, PALB2, POT1, STK11, TP53); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis; Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1617-V3 AA-MR-QM-1662-V1 Panel TPv3a	Illumina Series (NovaSeq)		x	
IgHV-Mutationsstatus	EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA, cDNA; RNA, cDNA	Sanger-Sequenzierung	AA-MR-QM-0228	ABI Sequencer 3730XL, 3130XL, 3730		x	
Solide Tumoren (Mikrosatelliteninstabilität)	Gewebeprobe, DNA; DNA	Fragmentlängenanalyse	AA-MR-QM-0269-V7	ABI Sequencer 3730XL, 3130XL, 3730		x	ja, für erhaltene Gewebeproben ist eine Gewebeauswahl und eine Bestimmung des Tumoranteils notwendig; erfolgt durch eigenen Pathologen

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, [illegible])	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V9	ABI Sequencer 3730XL, 3130XL, 3730	x		
Agammaglobulinämie hereditäre (in-house) (BLNK [OMIM #604515], BTK [OMIM #300300], CD79A [OMIM #112205], CD79B [OMIM #147245], IGHM [OMIM #147020], IGLL1 [OMIM #146770], LRRC8A [OMIM #608360], PIK3R1 [OMIM #171833]); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	
Autoinflammatorische Syndrome (in-house) (ELANE [OMIM #130130], IL1RN [OMIM #147679], IL36RN [OMIM #605507],LPIN2 [OMIM #605519], MEFV [OMIM #608107], MVK [OMIM #251170], NLRC4 [OMIM #606831], NLRP12 [OMIM #609648], NLRP3 [OMIM #606416], NOD2 [OMIM #605956], PSMB8 [OMIM #177046], PSTPIP1 [OMIM #606347], TMEM173 [OMIM #612374], TNFRSF1A [OMIM #191190], ADA2, CARD14, LACC1, NLRP1, OTULIN, PLCG2, RBCK1, RIPK1, TNFAIP3, UBA1, WDR1); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen; Genoox (Franklin) Für UBA1: Amplikon basiert; Sequencing-by synthesis (Illumina); SeqPilot (JSI medical systems)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel TPv3b / Pipeline	Illumina Series (NovaSeq)		x	

Immundefekte im Kindesalter primäre (ADA [OMIM #608958], AK2 [OMIM #103020], AP3B1 [OMIM #603401], BLNK [OMIM #604515], BTK [OMIM #300300], CD247 [OMIM #186780], CD3D [OMIM #186790], CD3E [OMIM #186830], CD3G [OMIM #186740], CD40 [OMIM #109535], CD40LG [OMIM #300386], CD79A [OMIM #112205], CD79B [OMIM #147245], CD8A [OMIM #186910], CIITA [OMIM #600005], CLPB [OMIM #616254], CORO1A [OMIM #605000], CSF3R [OMIM #138971], CXCR4 [OMIM #162643], DCLRE1C [OMIM #605988], DOCK8 [OMIM #611432], ELANE [OMIM #130130], FOXN1 [OMIM #600838], G6PC3 [OMIM #611045], GATA1 [OMIM #305371], GATA2 [OMIM #137295], GFI1 [OMIM #600871], HAX1 [OMIM #605998], IGHM [OMIM #147020], IGLL1 [OMIM #146770], IKZF1 [OMIM #603023], IL2RG [OMIM #308380], IL7R [OMIM #146661], ITK [OMIM #186973], JAGN1 [OMIM #616012], JAK3 [OMIM #600173], LAMTOR2 [OMIM #610389], LCK [OMIM #153390], LIG4 [OMIM #601837], LRRC8A [OMIM #608360], LYST [OMIM #606897], MAGT1 [OMIM	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	

Kombinierte T- und B-Zellimmundefekte (ADA [OMIM #608958], AK2 [OMIM #103020], CD247 [OMIM #186780], CD3D [OMIM #186790], CD3E [OMIM #186830], CD3G [OMIM #186740], CD40 [OMIM #109535], CD40LG [OMIM #300386], CD8A [OMIM #186910], CIITA [OMIM #600005], CORO1A [OMIM #605000], DCLRE1C [OMIM #605988], DOCK8 [OMIM #611432], FOXN1 [OMIM #600838], IKZF1 [OMIM #603023], IL2RG [OMIM #308380], IL7R [OMIM #146661],ITK [OMIM #186973], JAK3 [OMIM #600173], LCK [OMIM #153390], LIG4 [OMIM #601837], MAGT1 [OMIM #300715], NHEJ1 [OMIM #611290], ORAI1 [OMIM #610277], PNP [OMIM #164050], PRKDC [OMIM #600899], PTPRC [OMIM #151460], RAG1 [OMIM #179615], RAG2 [OMIM #179616], RFX5 [OMIM #601863], RFXANK [OMIM #603200], RFXAP [OMIM #601861], RHOH [OMIM #602037], RMRP [OMIM #157660], STAT5B [OMIM #604260], STIM1 [OMIM #605921], STK4 [OMIM #604965], TAP1 [OMIM #170260], TAP2 [OMIM #170261], TAPBP [OMIM #601962],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	
Neutropenie, kongenital (AP3B1 [OMIM #603401], CLPB [OMIM #616254], CSF3R [OMIM #138971], CXCR4 [OMIM #162643], ELANE [OMIM #130130],G6PC3 [OMIM #611045], GATA1 [OMIM #305371], GATA2 [OMIM #137295], GFI1 [OMIM #600871], HAX1 [OMIM #605998], JAGN1 [OMIM #616012], LAMTOR2 [OMIM #610389], LYST [OMIM #606897], RAB27A [OMIM #603868], SBDS [OMIM #607444], SLC37A4 [OMIM #602671], TAZ [OMIM #300394], USB1 [OMIM #613276], VPS13B [OMIM #607817], VPS45 [OMIM #610035], WAS [OMIM	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel TPv3a / Pipeline	Illumina Series (NovaSeq)		x	

Omenn-Syndrom (OS) (ADA [OMIM #608958], AK2 [OMIM #103020], DCLRE1C [OMIM #605988], IL2RG [OMIM #308380], IL7R [OMIM #146661], JAK3 [OMIM #600173], LIG4 [OMIM #601837], RAG1 [OMIM #179615], RAG2 [OMIM #179616], RMRP [OMIM #157660]); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	
Schwere kombinierte Immundefekte (T-B-) (ADA [OMIM #608958], AK2 [OMIM #103020], DCLRE1C [OMIM #605988], LIG4 [OMIM #601837], NHEJ1 [OMIM #611290], PRKDC [OMIM #600899], RAG1 [OMIM #179615], RAG2 [OMIM #179616]); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	
Schwere kombinierte Immundefekte T-B+) (CD247 [OMIM #186780], CD3D [OMIM #186790], CD3E [OMIM #186830], CORO1A [OMIM #605000], FOXN1 [OMIM #600838], IL2RG [OMIM #308380], IL7R [OMIM #146661], JAK3 [OMIM #600173], PTPRC [OMIM #151460])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3 AA-MR-QM-1313 AA-MR-QM-1617-V3 Panel Exom / Pipeline	Illumina Series (NovaSeq)		x	
Agammaglobulinämie Bruton (XLA) (BTK-Gen [OMIM #300300]); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1313-V9 AA-MR-QM-1617-V3 AA-MR-QM-1637-V3 Panel TPv3a / Pipeline	Illumina Series (NovaSeq)		x	
Autoimmun-Polyendokrinopathie-Candidiasis-Ektodermaldystrophie-Syndrom Typ I (APECED) (AIRE-Gen [OMIM #607358]); CNV, SNV	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-MR-QM-1313-V9 AA-MR-QM-1617-V3 AA-MR-QM-1637-V3 Panel TPv3a / Pipeline	Illumina Series (NovaSeq)		x	
CINCA-Syndrom (NLRP3-Gen [OMIM #606416]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Cryopyrin-assoziierte periodische Syndrome (CAPS) (NLRP3-Gen [OMIM #606416]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Familiäres Kälte-assoziiertes autoinflammatorisches Syndrom Typ I (NLRP3-Gen [OMIM #606416]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Hyper-IgD-und-periodisches-Fiebersyndrom (HIDS) (MVK-Gen [OMIM #251170]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Mevalonazidurie (MVK-Gen [OMIM #251170]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Mittelmeerfieber, familiäre Form (FMF) (MEFV-Gen [OMIM #608107]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Muckle-Wells-Syndrom (NLRP3-Gen [OMIM #606416]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	

TNF-Rezeptor-1-assoziiertes periodisches Syndrom (TRAPS) (TNFRSF1A-Gen [OMIM #191190]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Wiskott-Aldrich-Syndrom (WAS) (WAS-Gen [OMIM #300392]); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Zyklische Neutropenie (CyN) / schwere kongenitale Neutropenie (SCN) (ELANE (OMIM #130130)); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Shwachman-Bodian-Diamond-Syndrom (SBDS-Gen); SNV	EDTA-Blut, DNA; DNA	PCR; Sequencing-by synthesis (PacBio); Seqpilot (JSI medical systems)	AA-MR-QM-1313-V9 AA-MR-QM-1769	PacBio, Sequel IIe		x	
Hereditäre Neuropathien (AARS [OMIM #601065], ABCD1, ABHD12 [OMIM #613599], AFG3L2, AIFM1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARHGEF10 [OMIM #608136], ARL6IP1, ATL1, ATL3, ATP13A2, ATP1A1, ATP7A [OMIM #300011], B4GALNT1, BAG3 [OMIM #603883], BICD2, BSCL2 [OMIM #606158], C12orf65, C19orf12, CAPN1, CAPN3, CCT5 [OMIM #610150], COX6A1, CPT1C, CTDP1 [OMIM #604927], CYP2U1, CYP7B1, DCTN1 [OMIM #601143], DDHD1, DDHD2, DES, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DPM3, DSTYK, DYNC1H1 [OMIM #600112], EGR2, ELP1, ENTPD1, ERLIN1, ERLIN2, FA2H, FAM134B [OMIM #613114], FARS2, FBLN5, FBXO38, FGD4 [OMIM #611104], FIG4 [OMIM #609390], GAN [OMIM #605379], GARS, GBA2, GDAP1, GJB1 [OMIM #304040], GJC2, GNB4, HARS, HINT1, HK1, HOXD10 [OMIM #142984], HPDL, HSPB1, HSPB3 [OMIM #604624], HSPB8 [OMIM #608014], HSPD1, IBA57, IGHMBP2, INF2, JPH1, KARS [OMIM #601421], KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, L1CAM, LAMA2, LITAF, LMNA [OMIM #150330], LRSAM1 [OMIM #610933],	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Entwicklungsstörungen (Makrozephalie; GPI-Ankerdefekte/Hyperphosphatasie-mentale Retardierungssyndrom; Cornelia-de-Lange-Syndrom; Großwuchssyndrome; Primäre Mikrozephalien; Coffin-Siris-Syndrom; Rett-Syndrom und Rett-ähnliche Erkrankungen; Kabuki-Syndrom; Sprachentwicklungsstörungen; X-gebundene mentale Retardierung; Pädiatrische Neurotransmitterstörungen; CDG-Syndrome; Robinow-Syndrom; Autismus; Gehirnfehlbildungen; Rubinstein-Taybi-Syndrom (RTS); Mowat-Wilson-Syndrom) (AARS [OMIM #601065], ABCC9, ABCD1, ACSL4 [OMIM #300157], ACTB [OMIM #102630], ACTG1 [OMIM #102560], ADAT3 [OMIM #615302], ADNP [OMIM #611386], AFF2, AGTR2 [OMIM #300034], AHDC1 [OMIM #615790], AIFM1 [OMIM #300169], AKT3 [OMIM #611223], ALDH5A1 [OMIM #610045], ALG1 [OMIM #605907], ALG11 [OMIM #613666], ALG12 [OMIM #607144], ALG13 [OMIM #300776], ALG2 [OMIM #607905], ALG3 [OMIM #608750], ALG6 [OMIM #604566], ALG8 [OMIM #608103], ALG9 [OMIM #606941],	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Stoffwechselmyopathien (Glycogenosen mit muskulärer Symptomatik und Carnitinstoffwechselstörungen), (Defekte der mitochondrialen ß-Oxidation und Mitochondriale Deletionssyndrome (MTDPS) und Mypathie) (ACADVL [OMIM #609575], AGK [OMIM #610345], ALDOA [OMIM #103850], C10orf2 [OMIM #606075], CPT2 [OMIM #600650], DGUOK [OMIM #601465], ETFA [OMIM #608053], ETFB [OMIM #130410], ETFDH [OMIM #231675], FBXL4 [OMIM #605654], GAA [OMIM #606800], HADHA [OMIM #600890], HADHB [OMIM #143450], INIP, ISCU, LAMA2 [OMIM #156225], LDHA [OMIM #150000], LPIN1 [OMIM #605518], MGME1 [OMIM #615076], MPV17 [OMIM #137960], NOTCH2 [OMIM #600275], PFKM [OMIM #610681], PGAM2 [OMIM #612931], PHKA1 [OMIM #311870], PHKB [OMIM #172490], POLG [OMIM #174763], PYGM [OMIM #608455], RRM2B [OMIM #604712], SALL4 [OMIM #607343], SLC25A20 [OMIM #613698], SLC25A4 [OMIM #103220], SUCLA2 [OMIM #603921], SUCLG1 [OMIM #611224], TBX3 [OMIM #601621], TBX5 [OMIM #601620], TK2 [OMIM	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-MR-QM-1637-V3, AA-MR-QM-1617-V3, AA-MR-QM-1648-V3, AA-MR-QM-1662-V1, AA-MR-QM-1652-V1, AA-MR-QM-1504-V7, AA-MR-QM-1635-V5 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Carnitinzyklusdefekte (Carnitin-Palmitoyltransferase 1A (CPT1A)-Defizienz: Carnitin-Palmitoyltransferase 2 (CPT2)-Defizienz; Carnitin-Acylcarnitin-Translokase (CACT)-Defizienz) (CPT1A, CPT2, SLC25A20)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Plattform (NovaSeq)		x	

Kongenitale Myopathien (ACTA1 [OMIM #102610], BIN1 [OMIM #601248], CCDC78 [OMIM #614666], CEP120 [OMIM #613446], CFL2 [OMIM #601443], CNTN1 [OMIM #600016], DNM2 [OMIM #602378], KBTBD13 [OMIM #613727], KLHL40 [OMIM #615340], KLHL41 [OMIM #607701], LMOD3 [OMIM #616112], MEGF10 [OMIM #612453], MICU1, MSX2 [OMIM #123101], MTM1 [OMIM #300415], MTMR14 [OMIM #611089], MYF6 [OMIM #159991], MYH2 [OMIM #160740], MYH3 [OMIM #160720], MYH7 [OMIM #160760], MYL1, MYO18B, MYPN, NEB [OMIM #161650], ORAI1, RYR1 [OMIM #180901], SELENON, SPEG [OMIM #615950], SPTBN4, STAC3, TNNT1 [OMIM #191041], TPM2 [OMIM #190990], TPM3 [OMIM #191030], TTN [OMIM #188840], VCP [OMIM #601023])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
mTOR Gesamt (AKT1 [OMIM #164730], AKT1S1 [OMIM #610221], AKT3 [OMIM #611223], DEPDC5 [OMIM #614191], DEPTOR [OMIM #612974], DOCK7 [OMIM #615730], FKRP [OMIM #606596], FKTN [OMIM #607440], GMPPB [OMIM #615320], ISPD [OMIM #614631], LARGE1 [OMIM #603590], MLST8 [OMIM #612190], MTOR [OMIM #601231], NPRL2 [OMIM #607072], NPRL3 [OMIM #600928], PAK2 [OMIM #605022], PIK3CA [OMIM #171834], PIK3CD [OMIM #602839], PIK3R2 [OMIM #603157], POMGNT1 [OMIM #606822], POMGNT2 [OMIM #614828], POMK [OMIM #615247], POMT1 [OMIM #607423], POMT2 [OMIM #607439], PTEN [OMIM #601728], RPTOR [OMIM #607130], STRADA [OMIM #608626], TBC1D7 [OMIM #612655], TMEM5 [OMIM #605862], TSC1 [OMIM #605284], TSC2 [OMIM #191092])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Kraniosynostosen (Antley-Bixler-Syndrom mit Genitalanomalien und Steroidsynthesestörungen; Apert-Syndrom; Baller-Gerold-Syndrom; BGS; Carpenter-Syndrom Typ 1 (CRPT1); Chondrodysplasie mit Gelenkdislokation, Typ Grapp; Cole-Carpenter-Syndrom Typ 1 und 2; Cranioektodermale Dysplasie Typ 1 und 3; Craniofrontonasales Syndrom (CFNS); Crouzon-Syndrom; Distale Arthrogrypose Typ 8 (DA8); Greig Cephalosyndaktylie-Syndrom (GCPS); Jackson-Weiss Syndrom; Kraniosynostose Typ 3, 4, 5, 7; Kraniosynostose und Zahnanomalien (CRSDA); Kurzrippen-Thoraxdysplasie Typ 9 mit oder ohne Polydaktylie; Meier-Gorlin-Syndrom; Muenke Syndrom (MNKES); Pfeiffer-Syndrom; Roberts-Syndrom; Robinow-Sorauf-Syndrom; Seathre-Chotzen-Syndrom; Trigonozephalie Typ 1 (TRIGNO1); Van Den Ende-Gupta-Syndrom (VDEGS)) (ALPL, ALX4 [OMIM #605420], CDC45, CEP120 [OMIM #613446], EFNB1 [OMIM #300035], ERF [OMIM #611888], ESCO2 [OMIM #609353], FGFR1 [OMIM #136350], FGFR2 [OMIM #176943], FGFR3 [OMIM #134934], GLI3 [OMIM #165240],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Mukopolysaccharidosen (Mukopolysaccharidose I (Hurler-Syndrom); Mukopolysaccharidose II (Hunter); Mukopolysaccharidose IVA; Mukopolysaccharidose Typ IIIA (Sanfilippo A); Mukopolysaccharidose Typ IIIB (Sanfilippo B); Mukopolysaccharidose Typ IIIC (Sanfilippo C); Mukopolysaccharidose Typ IVB (Morquio); Mukopolysaccharidose Typ IX; Mukopolysaccharidose Typ VI (Maroteaux-Lamy); Mukopolysaccharidose VII) (ARSB [OMIM #611542], GALNS [OMIM #612222], GLB1 [OMIM #611458], GNS, GUSB [OMIM #611499], HGSNAT [OMIM #610453], HYAL1 [OMIM #607071], IDS [OMIM #300823], IDUA [OMIM #252800], NAGLU [OMIM #609701], SGSH [OMIM #605270])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Osteogenesis imperfecta (OI) (KLF3, BMP1 [OMIM #112264], COL1A1 [OMIM #120150], COL1A2 [OMIM #120160], CREB3L1, CRTAP [OMIM #605497], FKBP10 [OMIM #607063], IFITM5 [OMIM #614757], MBTPS2, P3H1 [OMIM #610339], P4HB, PLOD2 [OMIM #601865], PLS3, PPIB [OMIM #123841], SEC24D, SERPINF1 [OMIM #172860], SERPINH1 [OMIM #600943], SP7 [OMIM #606633], SPARC, TENT5A, TMEM38B [OMIM #611236], WNT1 [OMIM #164820])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Stickler-Syndrom (COL11A1 [OMIM #120280], COL11A2 [OMIM #120290], COL2A1 [OMIM #120140], COL9A1 [OMIM #120210], COL9A2)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Ataxien (spastische Ataxien, episodische Ataxien; Ataxien syndromale Formen; Ataxie mit okulomotorischer Apraxie) (ABCB7, ABHD12 [OMIM #613599], ADGRG1, AFG3L2 [OMIM #604581], AHI1 [OMIM #608894], AMACR, ANO10, APTX, ARL13B [OMIM #608922], ARSA [OMIM #607574], ATCAY [OMIM #608179], ATG5, ATM [OMIM #607585], ATP13A2, ATP1A3 [OMIM #182350], ATP8A2 [OMIM #605870], ATXN10 [OMIM #611150], B4GALNT1, BTD [OMIM #609019], CA8 [OMIM #114815], CACNA1A, CACNA1G, CACNAB4, CAPN1 [OMIM #114220], CC2D2A [OMIM #612013], CCDC88C, CEP290 [OMIM #610142], CEP41 [OMIM #610523], CHP1, CLCN2 [OMIM #600570], CLN5 [OMIM #608102], CLN6 [OMIM #606725], COA7, COQ8A [OMIM #606980], CP [OMIM #117700], CPLANE1, CSPP1 [OMIM #611654], CWF19L1, CYP27A1 [OMIM #606530], DARS2 [OMIM #610956], DLAT [OMIM #608770], DNAJC19 [OMIM #608977], DNMT1 [OMIM #126375], EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1 [OMIM #609144], GALC [OMIM #606890], GBA [OMIM #606463],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.V1	Illumina Series (NovaSeq)	-	x	

Angeborene Herzfehler (Alagille-Syndrom; Isolierte Herzfehler; RASopathien mit Herzfehlern; syndromale Herzfehler; Heterotaxie-assoziierte Herzfehler) (ACTC1 [OMIM #102540], ACVR2B [OMIM #602730], ADAMTS10 [OMIM #608990], ARHGAP31 [OMIM #610911], BMPR2 [OMIM #600799], BRAF [OMIM #164757], CBL [OMIM #165360], CFAP53 [OMIM #614759], CFC1 [OMIM #605194], CHD7 [OMIM #608892], CITED2 [OMIM #602937], CREBBP [OMIM #600140], CRELD1 [OMIM #607170], DNAH11 [OMIM #603339], DNAH5 [OMIM #603335], DNAI1 [OMIM #604366], DOCK6 [OMIM #614194], DTNA [OMIM #601239], EHMT1 [OMIM #607001], ELN [OMIM #130160], EOGT [OMIM #614789], EP300 [OMIM #602700], EVC [OMIM #604831], EVC2 [OMIM #607261], FBN1 [OMIM #134797], FBN2 [OMIM #612570], FLNA [OMIM #300017], FOXC1 [OMIM #601090], FOXH1 [OMIM #603621], FOXP1 [OMIM #605515], GATA4 [OMIM #600576], GATA5 [OMIM #611496], GATA6 [OMIM #601656], GDF1 [OMIM #602880], GJx1 [OMIM #121014], GPC3 [OMIM #300037], HRAS [OMIM	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Fettstoffwechselerkrankungen (Familiäre Hypercholesterinämie; Primäre Hypertriglyzeridämie; Gemischte Hyperlipoproteinämie; Hypoalphalipoproteinämie; Hypoalphalipoproteinämie; Hypolipoproteinämie) (ABCA1 [OMIM #600046], ANGPTL3 [OMIM #604774], APOA1 [OMIM #107680], APOA5 [OMIM #606368], APOB [OMIM #107730], APOC2 [OMIM #608083], APOE [OMIM #107741], GPIHBP1 [OMIM #612757], LCAT [OMIM #606967], LDLR [OMIM #606945], LDLRAP1 [OMIM #605747], LIPC [OMIM #151670], LMF1, LPL [OMIM #609708], MTTP [OMIM #157147], PCSK9 [OMIM #607786], SAR1B)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Interstitielle Lungenerkrankungen im Kindesalter (chILD) / diffuse parenchymatöse Lungenerkrankungen (DPLD) / Surfactant-Dysfunktionen (ABCA3 [OMIM #601615], CSF2RA [OMIM #306250], CSF2RB [OMIM #138981], FLNA [OMIM #300017], FOXF1 [OMIM #601089], NKX2-1 [OMIM #600635], SFTPB [OMIM #178640], SFTPC [OMIM #178620])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Augenerkrankungen (Bardet-Biedl Syndrom; Retinitis pigmentosa; Senior-Løken-Syndrom; Usher-Syndrom; Wagner-Syndrom) (ABCA4 [OMIM #601691], ADAM9, ADGRV1 [OMIM #602851], AGBL5, AIPL1, ALMS1 [OMIM #606844], ARL2BP, ARL6 [OMIM #608845], BBIP1 [OMIM #613605], BBS1 [OMIM #209901], BBS10 [OMIM #610148], BBS12 [OMIM #610683], BBS2 [OMIM #606151], BBS4 [OMIM #600374], BBS5 [OMIM #603650], BBS7 [OMIM #607590], BBS9 [OMIM #607968], BEST1 [OMIM #607854], C8orf37 [OMIM #614477], CA4 [OMIM #114760], CCDC28B [OMIM #610162], CDH23 [OMIM #605516], CDHR1 [OMIM #609502], CEP290 [OMIM #610142], CERKL [OMIM #608381], CIB2 [OMIM #605564], CLN3, CLRN1 [OMIM #606397], CNGA1 [OMIM #123825], CNGA3, CNGB1 [OMIM #600724], CNGB3, CRB1 [OMIM #604210], CRX, CYP4V2, DHDDS [OMIM #608172], EYS [OMIM #612424], FAM161A [OMIM #613596], FSCN2 [OMIM #607643], GUCA1A, GUCA1B [OMIM #602275], GUCY2D, HARS [OMIM #142810], IDH3A, IDH3B [OMIM #604526], IFT140, IFT172	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TwistWE.v1	Illumina Series (NovaSeq)	-	x	
MODY-Diabetes (ABCC8 [OMIM #600509], APPL1 [OMIM #604299], BLK [OMIM #191305], CEL [OMIM #114840], GCK [OMIM #138079], HNF1A [OMIM #142410], HNF1B [OMIM #189907], HNF4A [OMIM #600281], INS [OMIM #176730], KCNJ11 [OMIM #600937], KLF11 [OMIM #603301], NEUROD1 [OMIM #601724], PAX4 [OMIM #167413], PDX1 [OMIM #600733])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Arrhythmogene Erkrankungen (Arrhythmogene rechtsventrikuläre Kardiomyopathie; Brugada-Syndrom; Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT); Dilatative Kardiomyopathie (DCM); Hypertrophe Kardiomyopathie (HCM); Long-QT-Syndrom; Non-compaction Kardiomyopathie (NCCM); Restriktive Kardiomyopathie (RCM); Short QT-Syndrom) (ABCC9 [OMIM #601439], ACTC1 [OMIM #102540], ACTN2 [OMIM #102573], AKAP9 [OMIM #604001], ALPK3, ANK2 [OMIM #106410], ANKRD1 [OMIM #609599], BAG3 [OMIM #603883], CACNA1C [OMIM #114205], CACNA2D1 [OMIM #114204], CACNB2 [OMIM #600003], CALM1 [OMIM #114180], CALM2 [OMIM #114182], CALM3, CALR3 [OMIM #611414], CASQ2 [OMIM #114251], CAV3 [OMIM #601253], CRYAB [OMIM #123590], CSRP3 [OMIM #600824], DES [OMIM #125660], DSC2 [OMIM #125645], DSG2 [OMIM #125671], DSP [OMIM #125647], FHL1, FHOD3, FLNC, GAA, GLA, GPD1L [OMIM #611778], HCN4 [OMIM #605206], JPH2 [OMIM #605267], JUP [OMIM #173325], KCND3 [OMIM #605411], KCNE1	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Schwerhörigkeit/Taubheit (Hörverlust, autosomal-rezessiv, nicht-syndromal; Pendred Syndrom; Perrault-Syndrom; Taubheit/Schwerhörigkeit, autosomal-dominant; Taubheit/Schwerhörigkeit, autosomal rezessiv; Taubheit/Schwerhörigkeit, syndromal; Usher Syndrom; Waardenburg-Syndrom) (ABCC1, ABHD12 [OMIM #613599], ACTG1 [OMIM #102560], ADCY1 [OMIM #103072], ADGRV1 [OMIM #602851], AIFM1, ATP6V1B1 [OMIM #192132], BDP1 [OMIM #607012], BSND [OMIM #606412], CABP2 [OMIM #607314], CACNA1D, CCDC50 [OMIM #611051], CD164, CDC14A [OMIM #603504], CDH23 [OMIM #605516], CEACAM16 [OMIM #614591], CEP250, CIB2 [OMIM #605564], CISD2, CLDN14 [OMIM #605608], CLDN9, CLIC5 [OMIM #607293], CLPP [OMIM #601119], CLRN1 [OMIM #606397], CLRN2, COCH [OMIM #603196], COL11A1, COL11A2 [OMIM #120290], COL4A6 [OMIM #303631], COX1, CRYM [OMIM #123740], DCDC2 [OMIM #605755], DIAPH1 [OMIM #602121], DIAPH3 [OMIM #614567], DMXL2, DSPP [OMIM #125485], EDN3, EDNRB, ELMOD3 [OMIM #615427], EPS8	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Core-Myopathien (ACTA1 [OMIM #102610], BIN1 [OMIM #601248], DNM2 [OMIM #602378], MTM1, RYR1, SELENON, TPM2 [OMIM #190990], TPM3 [OMIM #191030], TTN [OMIM #188840])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Nemaline Myopathien (ACTA1 [OMIM #102610], CFL2 [OMIM #601443], KBTBD13 [OMIM #613727], KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Thorakale Aortenerweiterung mit dem Risiko der Aortendissektion (Thorakale Aortenerkrankungen familiär, nicht-syndromal; Thorakale Aortenerkrankungen, seltene, syndromal; Thorakale Aortenerkrankungen, selten, syndromal, rezessiv; Cutis laxa; Loeys-Dietz-Syndrom; Bikuspide Aortenklappe, mit Risiko für Aortenstenose und Aortendilatation/ -dissektion; Kollagen 4-assoziierte intrazerebrale Blutungen) (ACTA2 [OMIM #102620], BGN [OMIM #301870], COL1A1 [OMIM #120150], COL3A1 [OMIM #120180], COL4A5 [OMIM #303630], COL5A1 [OMIM #120215], COL5A2 [OMIM #120190], EFEMP2 [OMIM #604633], ELN [OMIM #130160], EMILIN1 [OMIM #130660], FBLN5 [OMIM #604580], FBN1 [OMIM #134797], FBN2 [OMIM #612570], FLNA [OMIM #300017], FOXE3 [OMIM #601094], GATA5 [OMIM #611496], LTBP3 [OMIM #602090], LOX [OMIM #153455], MAT2A [OMIM #601468], MFAP5 [OMIM #601103], MYH11 [OMIM #160745], MYLK [OMIM #600922], NOTCH1 [OMIM #190198], PLOD1 [OMIM #153454], PRKG1 [OMIM #176894], ROBO4, SKI [OMIM	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Ehlers-Danlos-Syndrom autosomal-rezessive Subtypen; EDS, classical-like (clEDS); EDS mit Herzklappenbeteiligung (cvEDS); EDS, Dermatosparaxis Typ (dEDS); EDS, kyphoskoliotischer Typ (kEDS); EDS, muskulokontraktureller Typ (mcEDS); EDS, Spondylodysplastische Form (ADAMTS2 [OMIM #604539], AEBP1 [OMIM #602981], B3GALT6 [OMIM #615291], B4GALT7 [OMIM #604327],CHST14, COL1A2 [OMIM #120160], , DSE [OMIM #605942], FKBP14 [OMIM #614505], PLOD1 [OMIM #153454], SLC39A13 [OMIM #608735], TNXB)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Pulmonale arterielle Hypertonie (PAH) (ACVRL1 [OMIM #601284], BMPR1B [OMIM #603248], BMPR2 [OMIM #600799], CAV1 [OMIM #601047], EIF2AK4 [OMIM #609280], ENG [OMIM #131195], GDF2 [OMIM #605120], KCNA5 [OMIM #176267], KCNK3 [OMIM #603220], SMAD1 [OMIM #601595], SMAD4 [OMIM #600993], SMAD9 [OMIM #603295], TBX4 [OMIM #601719])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Zilliopathie (Autosomal-dominante polyzystische Nierenerkrankung (ADPKD); Autosomal-rezessive polyzystische Nieren- und Lebererkrankung (ARPKD); Bardet-Biedl-Syndrom; Heterotaxie; Jeune-/ Kurzrippen-Polydaktylie-Syndrom; Joubert-Syndrom; Meckel-Gruber-Syndrom; Nephronophthise; Oro-fazio-digitales Syndrom ; Primäre ziliäre Dyskinesie; Senior-Løken-Syndrom) (ACVR2B [OMIM #602730], AHI1 [OMIM #608894], ALMS1 [OMIM #606844], ANKS6 [OMIM #615370], ARL13B [OMIM #608922], ARL6 [OMIM #608845], ARMC4 [OMIM #615408], ATXN10 [OMIM #611150], B9D1 [OMIM #614144], B9D2 [OMIM #611951], BBIP1 [OMIM #613605], BBS1 [OMIM #209901], BBS10 [OMIM #610148], BBS12 [OMIM #610683], BBS2 [OMIM #606151], BBS4 [OMIM #600374], BBS5 [OMIM #603650], BBS7 [OMIM #607590], BBS9 [OMIM #607968], BICC1 [OMIM #614295], BMP4 [OMIM #112262], C2CD3 [OMIM #615944], C8orf37, CC2D2A [OMIM #612013], CCDC103 [OMIM #614677], CCDC114 [OMIM #615038], CCDC151 [OMIM #615956], CCDC28B [OMIM #610162], CCDC39 [OMIM	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Marfan-ähnliche Erkrankungen (Akromikrische Dysplasie (ACMICD); Ectopia lentis; Geleophysische Dysplasie Typ 1-3; Kongenitale kontrakturale Arachnodaktylie (CCA); Lujan-Fryns-Syndrom; Marfanoider Habitus - Intelligenzminderung, autosomal-rezessiv; Megalokornea, Mikrosphärophakie, progressive Ectopia lentis, Myopie und sekundäres Glaukom; Shprintzen-Goldberg-Syndrom (SGS); Weill-Marchesani-Syndrom Typ 1-4; XLMR mit marfanoidem Habitus) (ADAMTS10 [OMIM #608990], ADAMTS17, ADAMTSL2 [OMIM #612277], ADAMTSL4 [OMIM #610113], EFEMP1, FBN1, FBN2 [OMIM #612570], LTBP2 [OMIM #602091], LTBP3, MED12 [OMIM #300188], SKI [OMIM #164780], UPF3B [OMIM #300298],ZDHHC9 [OMIM #300646])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
(ADCY5 [OMIM #600293], ARSA [OMIM #607574], ATM [OMIM #607585], ATN1 [OMIM #607462], ATXN1 [OMIM #601556], ATXN2 [OMIM #601517], ATXN3 [OMIM #607047], ATXN7 [OMIM #607640], FRRS1L [OMIM #604574], FTL [OMIM #134790], GM2A [OMIM #613109], GNAO1 [OMIM #139311], KCNA1 [OMIM #176260], PANK2, PDE10A [OMIM #610652], PRNP [OMIM #176640], RNF216 [OMIM #609948], SETX [OMIM #608465], TBP [OMIM #600075], VPS13A [OMIM #605978], XK [OMIM #314850])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a; TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Ehlers-Danlos-Syndrom, dominante Subtypen, klassischer Typ (cEDS), EDS vaskulärer Typ (vEDS); EDS Arthrochalasis Typ (aEDS) (COL1A1 [OMIM #120150], COL1A2, COL3A1, COL5A1 [OMIM #120215], COL5A2 [OMIM #120190])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Hyperoxalurie (AGXT [OMIM #604285], GRHPR [OMIM #604296], HOGA1 [OMIM #613597])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Porphyrien (Akute intermittierende Porphyrie (AIP); Porphyria variegata (PV); Hereditäre Koproporphyrie (HCP); ALA-Dehydratase Defizienz-Porphyrie; Porphyria cutanea tarda (PCT); Erythropoetische Protoporphyrie; Protoporphyrie X-chromosomal; Kongenitale Erythropoetische Porphyrie; Hepatoerythro-poetische Porphyrie (HEP)) (ALAD [OMIM #125270], ALAS2 [OMIM #301300], CPOX [OMIM #612732], FECH [OMIM #612386], HMBS [OMIM #609806], PPOX [OMIM #600923], UROD [OMIM #613521], UROS [OMIM #606938])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Cutis laxa (Cutis laxa, autosomal-dominant, Typ 1 (ADCL1); Cutis laxa, autosomal-dominant, Typ 2 (ADCL2); Cutis laxa, autosomal-dominant, Typ 3 (ADCL3); Cutis laxa, autosomal-rezessiv, Typ 1B (ARCL1B); Cutis laxa, autosomal-rezessiv, Typ 2B (ARCL2B); Cutis laxa, autosomal-rezessiv, Typ 2C (ARCL2C); Cutis laxa, autosomal-rezessiv, Typ 2D (ARCL2D); Wrinkly skin-Syndrom (WSS)) (ALDH18A1 [OMIM #138250], ATP6V0A2 [OMIM #611716], ATP6V1A [OMIM #607027], ATP6V1E1 [OMIM #108746], EFEMP2 [OMIM #604633], ELN [OMIM #130160], FBLN5 [OMIM #604580], LTBP4 [OMIM #604710], PYCR1 [OMIM #179035])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

CDG-Syndrom congenitale Defekte der Glykosylierung (ALG1 [OMIM #605907], ALG11 [OMIM #613666], ALG12 [OMIM #607144], ALG13 [OMIM #300776], ALG2 [OMIM #607905], ALG3 [OMIM #608750], ALG6 [OMIM #604566], ALG8 [OMIM #608103], ALG9 [OMIM #606941], B4GALT1 [OMIM #137060], CAD [OMIM #114010], CCDC115 [OMIM #613734], COG1 [OMIM #606973], COG4 [OMIM #606976], COG5 [OMIM #606821], COG6 [OMIM #606977], COG7 [OMIM #606978], COG8 [OMIM #606979], DDOST [OMIM #602202], DOLK [OMIM #610746], DPAGT1 [OMIM #191350], DPM1 [OMIM #603503], DPM2 [OMIM #603564], DPM3 [OMIM #605951], MGAT2 [OMIM #602616], MOGS [OMIM #601336], MPDU1 [OMIM #604041], MPI [OMIM #154550], NGLY1 [OMIM #610661], PGM1 [OMIM #171900], PMM2 [OMIM #601785], PMS2 [OMIM #600259], RFT1 [OMIM #611908], RYR1 [OMIM #180901], SLC35A1 [OMIM #605634], SLC35A2 [OMIM #314375], SLC35C1 [OMIM #605881], SLC39A8 [OMIM #608732], SRD5A3 [OMIM #611715], SSR4 [OMIM #300090], STT3A [OMIM #601134],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Gerinnungsstörung Blutungsneigung (Faktor VII- Defizienz, Faktor XIII-Defizienz, von Willebrand-Jürgens-Syndrom) (F13A1 [OMIM #134570], F13B [OMIM #134580], F7 [OMIM #613878], VWF [OMIM #613160])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Charcot-Marie-Tooth-Neuropathien Typ 1 und Typ 2 (CMT1; CMT2) (ATL1 [OMIM #606439], DNM2 [OMIM #602378], GARS [OMIM #600287], GDAP1 [OMIM #606598], GJB1, HINT1, IGHMBP2 [OMIM #600502], MFN2 [OMIM #608507], MPZ [OMIM #159440], NEFL [OMIM #162280], NGF [OMIM #162030], PMM2 [OMIM #601785], PMP22 [OMIM #601097],	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Sonstige Bindegewebserkrankungen / Skelettdysplasien (Achondrogenesie Typ 2 (ACG2, Langer-Saldino); Hypochondrogenesie; Kniest-Syndrom; Marshall-Syndrom; Otospondylomegaepiphysäre Dysplasie (OSMED); Spondyloepiphysäre Dysplasie (SED)) (COL11A1 [OMIM #120280], COL11A2 [OMIM #120290],COL2A1)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Hereditäre Sphärozytose (HS) (Elliptozytose; Pyropoikolozytose) (ANK1 [OMIM #612641], EPB42 [OMIM #177070], SLC4A1 [OMIM #109270], SPTA1 [OMIM #182860], SPTB [OMIM #182870])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Progrediente Progressive Muskeldystrophien (Gliedergürtelmuskeldystrophien; Emery-Dreifuß Muskeldystrophie) (ANO5 [OMIM #608662], BVES, CAPN3 [OMIM #114240], CAV3 [OMIM #601253], CRPPA, DAG1 [OMIM #128239], DES [OMIM #125660], DMD [OMIM #300377], DNAJB6 [OMIM #611332], DYSF [OMIM #603009], EMD [OMIM #300384], FHL1 [OMIM #300163], FKRP [OMIM #606596], FKTN [OMIM #607440], GAA [OMIM #606800], GMPPB [OMIM #615320], HNRNPDL [OMIM #607137], LAMA2, LIMS2 [OMIM #607908], LMNA [OMIM #150330], MATR3, MYOT [OMIM #604103], PLEC [OMIM #601282], POMGNT1 [OMIM #606822], POMK [OMIM #615247], POMT1 [OMIM #607423], POMT2 [OMIM #607439], SGCA [OMIM #600119], SGCB [OMIM #600900], SGCD [OMIM #601411], SGCG [OMIM #608896], SYNE1 [OMIM #608441], SYNE2 [OMIM #608442], TCAP [OMIM #604488], TMEM43 [OMIM #612048], TNPO3 [OMIM #610032], TRAPPC11 [OMIM #614138], TRIM32 [OMIM #602290], TTN [OMIM #188840])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Hereditäre Hyperekplexie (Startle disease) (ARHGEF9 [OMIM #300429], ATAD1 [OMIM #614452], GLRA1 [OMIM #138491], GLRB, SLC6A5)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a; TwistWE.v1	Illumina Series (NovaSeq)	-	x	

Alzheimer Erkrankung, Frühform (AD1) (APP [OMIM #104760], PSEN1 [OMIM #104311], PSEN2 [OMIM #600759])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a; TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Gehirnfehlbildungen (Lissenzephalien; Tubulinopathien; Pontozerebelläre Hypoplasie) (AMPD2, ARX [OMIM #300382], CDK5 [OMIM #123831], CEP85L, CHMP1A, CLP1, COASY, DCX [OMIM #300121], 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; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Harnstoffzyklus-Defekt (Arginase-1-Mangel (Hyperargininämie); Argininosuccinat-Lyase(ASL)-Mangel (Argininbernsteinsäure-Krankheit); Argininosuccinat-Synthetase(ASS)-Mangel (Citrullinämie Typ 1); Carbamoylphosphat-Synthetase(CPS)-1-Mangel; Citrullinämie Typ 2, neonatal; HHH-Syndrom; N-Acetylglutamat-Synthetase(NAGS)-Mangel; Ornithin-Transcarbamylase(OTC)-Mangel) (ARG1 [OMIM #608313], ASL [OMIM #608310], ASS1 [OMIM #603470], CPS1, NAGS, OTC, SLC25A13 [OMIM #603859], SLC25A15 [OMIM #603861])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Epilepsie (Benigne familiäre Epilepsie (neonatal, infantil); Dravet-Syndrom (frühkindliche Grand-mal-Epilepsie); Epilepsien mit erhöhter Therapierelevanz; Fokale Epilepsien; Frühkindliche epileptische Enzephalopathien; Frühkindliche X-gebundene Epilepsie mit geistiger Behinderung; Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFS+); Generalisierte juvenile myoklonische Epilepsien; Glukosetransporter Typ 1-Defizienz-Syndrom (GLUT1-DS); Migräne, familiäre hemiplegische (FHM); Pyridoxin-abhängige Epilepsie (EPD)) (AARS, ACTL6B, ADAM22, ADGRV1, ADRA2B, ALDH7A1, ALG13, AP3B2, ARHGEF15 [OMIM #608504], ARHGEF9, ARV1, ARX, ATP1A2, BRAT1 [OMIM #614506], CACNA1A, CACNA1E, CACNA1H, CACNB4, CAD, CASR, CDK19, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4 [OMIM #302910], 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,	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Spinale Muskelatrophien (SMA) (Spinale Muskelatrophien (neonatal/frühmanifestierend) und Pontocerebelläre Hypolasie; Spinale Muskelatrophien (spätmanifestierend)) (ASAH1 [OMIM #613468], ATP7A [OMIM #300011], BICD2 [OMIM #609797], BSCL2, CHCHD10 [OMIM #615903], DNAJB2 [OMIM #604139], DYNC1H1, EXOSC3, EXOSC8, FBXO38 [OMIM #608533], GARS [OMIM #600287], HSPB8 [OMIM #608014], IGHMBP2, PLEKHG5, REEP1 [OMIM #609139], SLC5A7 [OMIM #608761], TFG [OMIM #602498], TRIP4, TRPV4 [OMIM #605427], UBA1, VAPB [OMIM #605704], VRK1)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Ehlers-Danlos-Syndrom seltene Formen Differenzialdiagnosen; Arterial Tortuosity Syndrom (ATS); Arthrogrypose mit okulomotorischen und elektroretinalen Anomalien; Bindegewebserkrankung mit peripherer Neuropathie, Arthropathie und Hautelastizität; Brittle-Cornea-Syndrom; EDS mit periventrikulärer Heterotopie (PVNH4); EDS, parodontaler Typ; Ehlers-Danlos-Syndrom, myopathischer Typ; Lysylhydroxylase-3-Defizienz; Muskeldystrophie, kongenitale, Typ Ullrich (UCMD1) (C1R [OMIM #613785], C1S [OMIM #120580], COL12A1 [OMIM #120320], COL6A1 [OMIM #120220], COL6A2 [OMIM #120240], COL6A3, EMILIN1 [OMIM #130660], FLNA, PHYKPL, PIEZO2, PLOD3, PRDM5, SLC2A10, VHL [OMIM #608537], ZNF469)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Jeune-/ Kurzrippen-Polydaktylie-Syndrom (CEP120 [OMIM #613446], CSPP1 [OMIM #611654], DYNC2H1 [OMIM #603297], EVC, EVC2, IFT80 [OMIM #611177], IFT122, IFT140, IFT172, IFT43, IFT52, KIAA0586 [OMIM #610178],NEK1, TCTN3 [OMIM #613847], TTC21B [OMIM #612014], WDR19, WDR34, WDR35, WDR60)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TwistWE.v1	Illumina Series (NovaSeq)	-	x	
Myofibrilläre Myopathien (BAG3 [OMIM #603883], CRYAB [OMIM #123590], DES [OMIM #125660], DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PLEC, PYROXD1, TTN)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Nierenerkrankungen (Polyzystische Nierenerkrankung APKD; Alport-Syndrom; Angeborene Fehlbildung der Nieren und ableitenden Harnwege (CAKUT); Fehlbildungen der ableitenden Harnwege; Diabetes insipidus renalis (NDI); Dünne Basalmembran Nephropathie (TBMN); Gitelman-Syndrom; Hyperoxalurie; LUTO (Lower Urinary Tract Obstruction) / Megacystis / Analatresie; Nephronophthise (NPHP); Nephrotisches Syndrom (NS) / Fokal segmentale Glomerulosklerose (FSGS); Nierenagenesie / -hypoplasie; Renale tubuläre Dysgenesie) (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,	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a, TwistWE.v1	Illumina Series (NovaSeq)			
Hämochromatose, hereditär Stufe II (BMP6 [OMIM #112266], HAMP [OMIM #606464], HFE [OMIM #613609], HJV, SLC40A1 [OMIM #604653], TFR2 [OMIM #604720])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
RASopathien (Noonan-Syndrom; Cardio-Fazio-Cutanes-Syndrom (CFC); LEOPARD-Syndrom; Costello-Syndrom) (BRAF [OMIM #164757], CBL [OMIM #165360], HRAS [OMIM #190020], KRAS [OMIM #190070], LZTR1 [OMIM #600574], MAP2K1 [OMIM #176872], MAP2K2 [OMIM #601263], MRAS, NF1 [OMIM #613113], NRAS [OMIM #164790], PPP1CB [OMIM #600590], PTPN11 [OMIM #176876], RAF1 [OMIM #164760], RASA2 [OMIM #601589], RIT1 [OMIM #609591], RRAS [OMIM #165090], RRAS2, SHOC2 [OMIM #602775], SOS1 [OMIM #182530], SOS2 [OMIM #601247], SPRED1 [OMIM #609291])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Maligne Hyperthermie (MH) (CACNA1S [OMIM #114208], RYR1 [OMIM #180901])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Nicht-dystrophische Myotonien und periodische Paralysen (CACNA1S [OMIM #114208], CLCN1 [OMIM #118425], HSPG2 [OMIM #142461], KCNJ2, SCN4A)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Hypokaliämische Periodische Paralyse (HypoPP) (CACNA1S [OMIM #114208], KCNJ2 [OMIM #600681], SCN4A [OMIM #603967])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Kollagen 4-assoziierte intrazerebrale Blutungen (Zerebrale Hämorrhagie; Porenzephalie 2; Zerebrale Mikroangiopathie mit Hämorrhagie, vermehrte Schlängelung der Netzhautarterien; Porenzephalie 1) (COL4A1 [OMIM #120130], COL4A2 [OMIM #120090])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
(Muskeldystrophien (Dystroglycanopathien Typ A+B); Muskeldystrophien (Kollagen-assoziierte und sonstige)) (CHKB [OMIM #612395], COL12A1, COL6A1 [OMIM #120220], COL6A2 [OMIM #120240], COL6A3 [OMIM #120250], CRPPA, DNM2 [OMIM #602378], DPM3, FHL1 [OMIM #300163], FKRP [OMIM #606596], FKTN [OMIM #607440], ITGA7 [OMIM #600536], LAMA2 [OMIM #156225], LARGE1 [OMIM #603590], LMNA, POMGNT1 [OMIM #606822], POMGNT2 [OMIM #614828], POMT1 [OMIM #607423], POMT2 [OMIM #607439], SELENON, TCAP [OMIM #604488])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Marfan-Syndrom, Typ 1 Fibrillinopathien, Akromikrische Dysplasie (ACMICD); Ektopia lentis; familiär; isoliert (ECTOL1); Geleophysische Dysplasie Typ 1 (GPHYSD1); Geleophysische Dysplasie Typ 2 (GPHYSD2); MASS-Syndrom (MASS); Stiff-Skin-Syndrom (SSKS); Weill-Marchesani-Syndrom (WMS2) (FBN1 [OMIM #134797])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Bikuspide Aortenklappe, mit Risiko für Aortenaneurysma und Aortenstenose/ -dilatation (Biskupide Aortenklappe 1-3 (AVOD 1-3); Multiple Herzfehlbildungen mit Bikuspider Aortenklappe) (GATA5 [OMIM #611496], NOTCH1 [OMIM #190198], ROBO4, SMAD6 [OMIM #602931])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005	Illumina Series (NovaSeq)	-	x	
Gerinnungsstörung Thromboseneigung (Protein C- Mangel, Protein S-Mangel, Antithrombin-Mangel (ATIII-Defizienz), hereditär) (PROC [OMIM #612283], PROS1 [OMIM #176880], SERPINC1)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
CADASIL (HTRA1 [OMIM #602194], NOTCH3 [OMIM #600276])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a; TwistWE v1	Illumina Series (NovaSeq)	-	x	
Legius-Syndrom, Neurofibromatose Typ 1-ähnliches Syndrom (NFLS) (SPRED1 [OMIM #609291])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a	Illumina Series (NovaSeq)	-	x	
Neurofibromatose Typ 1 (NF1)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a, TPv3b	Illumina Plattform (NovaSeq)		x	
Schwannomatose (Neurofibromatose Typ 2; Schwannomatose Typ 1 (SWNTS1); Schwannomatose Typ 2 (SWNTS2)) (LZTR1, NF2, SMARCB1)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a	Illumina Plattform (NovaSeq)			
Loeys-Dietz-Syndrom (LDS) (LDS Typ 1-6, Aneurysmen Osteoarthritis Syndrom (AOS)) (SMAD2 [OMIM #601366], SMAD3 [OMIM #603109], TGFB2, TGFB3 [OMIM #190230], TGFBR1 [OMIM #190181], TGFBR2 [OMIM #190182])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Tuberöse Sklerose Complex (TSC1 [OMIM #605284], TSC2 [OMIM #191092])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a, TPv3b	Illumina Series (NovaSeq)	-	x	
Sphingolipidosen (Morbus Fabry, Morbus Pompe, Morbus Tay-Sachs, Morbus Krabbe, Morbus Niemann-Pick) (GLA, GAA, GM2A, HEXA, HEXB, GALC, PSAP, NPC1, NPC2, SMPD1)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	

Störungen der Fettsäure-Oxidation (Mittelketten-Acyl-CoA-Dehydrogenase-Defizienz (MCADD); Langketten-3-Hydroxyacyl-CoA-Dehydrogenase-Defizienz (LCHADD); Sehr-Langketten-Acyl-CoA-Dehydrogenase-Defizienz (VLCADD); Multiple-Acyl-CoA-Dehydrogenase-Defizienz (MADD)) (ACADM, HADHA, HADHB, ACADVL, ETFA, ETFB, ETFDH)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Morbus Osler (Hereditäre Hämorrhagische Teleangiektasie (HHT)) (ACVRL1, ENG, GDF2, SMAD4)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a; TwistWE.v1	Illumina Series (NovaSeq)		x	
Sideroblastische Anämie, X-gebunden (XLSA) (ALAS)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
Pädiatrische Neurotransmitterstörungen (Parkinsonismus-Dystonie, infantil, 2; Brunner-Syndrom; Dopamintransporter-Defizienz-Syndrom / Parkinsonismus infantiler 1; Hyperphenylalaninämie, BH4-Defizient, A ; Hyperphenylalaninämie, BH4-Defizient, B; Hyperphenylalaninämie, BH4-Defizient, D; Kongenitale Dopamin β-Hydroxylase Defizienz; Tetrahydrobiopterin-Defizienz; Tryptophan-Hydroxylase-2-Defizienz; Tyrosinhydroxylase-Defizienz, Segawa-Syndrom) (DBH, DDC, GCH1, MAOA, PCBD1, PTS, QDPR, SLC18A2, SLC6A3, SPR, TH, TPH2)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TwistWE.v1	Illumina Series (NovaSeq)		x	
Leukoenzephalopathie mit Verlust der weißen Substanz (EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)		x	

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, ZFYVE26, ZFYVE27)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a, TwistWE.v1	Illumina Series (NovaSeq)		x	
Cystische Fibrose (Mukoviszidose, CF) (CFTR-Gen [OMIM #602421])	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
DCM1Y (TPM1-Gen [OMIM #191010])	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-0015-V005	LC480II, LC1.2 (Roche); CFX96, Biorad	x	-	
Hypophosphatämie (CLCN5, DMP1, ENPP1, FAM20C, FGF23, PHEX, SLC34A1, SLC34A3, SLC9A3R1)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen-Pipeline, VarSeq (Golden Helix)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a	Illumina Series (NovaSeq)		x	
Hypophosphatasie (ALPL)	EDTA-Blut, DNA; DNA	Sequence capture (TWIST), Sequencing-by synthesis, Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel: TPv3a	Illumina Series (NovaSeq)		x	
Arzneimittelunverträglichkeit von CYP2C9-Substraten (CYP2C9 (dbSNP rs1799853, rs1057910))	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	CFX96, Biorad	x	-	
Thrombophilie (Faktor V Leiden, APC-Resistenz, F5-Gen: dbSNP rs6025)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	CFX96, Biorad	-	x	
Thrombophilie (Prothrombin, F2, dbSNP rs1799963)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1727-V001	CFX96, Biorad	x		
HCV-Therapie (HCV-Therapie IL28B (dbSNP rs12979860))	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1727-V1	CFX96, Biorad	-	x	
Hämochromatose, hereditär (HFE-Gen: dbSNP rs1800562, rs1799945, rs1800730)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1727-V1	CFX96, Biorad	-	x	
Laktoseintoleranz (LCT-Gen: dbSNP rs182549)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1727-V1	CFX96, Biorad	x	-	
Methylentetrahydrofolatreduktase-(MTHFR-) Defizienz (MTHFR-Gen: dbSNP rs1801133, rs1801131)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1727-V1	CFX96, Biorad	x	-	
Alpha-1-Antitrypsin-Mangel (SERPINA1-Gen:dbSNP rs17580, rs28929474)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-MR-QM-1727-V1	CFX96, Biorad	x	-	
Fructoseintoleranz (hereditäre), hereditäre Fruktose-1,6-bisphosphatase-(FBP1-) Mangel (ALDOB [OMIM #612724], FBP1 [OMIM #611570])	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Adrenogenitales Syndrom (AGS) (AGS Typ 4, Adrenogenitales Syndrom durch 11ß-Hydroxylase-Mangel; AGS Typ 2, Adrenogenitales Syndrom durch 3ß-Hydroxysteroiddehydrogenase-Mangel; AGS Typ 5, Adrenogenitales Syndrom durch 17-alpha-Hydroxylase-Mangel) (CYP11B1 [OMIM #610613], CYP11B2-Gen [OMIM #124080], CYP17A1-Gen [OMIM #609300], HSD3B2-Gen [OMIM #613890])	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Alpha-Thalassämie, Beta-Thalassämie; Anomale Hämoglobine (HbS, HbC, HbE u.a.) (HBB, HBA1 [OMIM #141800], HBA2 [OMIM #141850], HBD-, HBG1, HBG2-Promoter)	EDTA-Blut, DNA; DNA	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 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Propionazidämie (PCCA [OMIM #232000], PCCB [OMIM #232050])	EDTA-Blut, DNA; DNA	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 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Basalzellnävus-Syndrom (Gorlin-Goltz-Syndrom) (PTCH1 [OMIM #601309], PTCH2 [OMIM # 603673], SUFU [OMIM #607035])	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
APOE-Genotypisierung (Alzheimer Erkrankung, Spätform; Dysbetalipoproteinämie, Typ III-Hyperlipidämie) (APOE-Gen [OMIM #107741], dbSNP rs429358, rs7412)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Muskelatrophie, spinobulbär (SBMA, Kennedy Krankheit) (AR-Gen [OMIM #313700])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1300-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Dentatorubrale Pallido-Luysische Atrophie (DRPLA) (ATN1-Gen [OMIM #607462])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1300-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Morbus Wilson Erkrankung (WND) (ATP7B-Gen [OMIM #606882])	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Ataxien, spinocerebelläre autosomal-dominante (SCA1, SCA2, SCA3, SCA6, SCA7, SCA17) (ATXN1-Gen [OMIM #601556], ATXN2, ATXN3, ATXN7, CACNA1A, TBP)	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1300-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Butyrylcholinesterase (BCHE)-Defizienz und postoperative Apnoe (BCHE-Gen [OMIM #177400])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Ahornsiruperkrankung (MSUD) (BCKDHA-Gen [OMIM #608348], BCKDHB, DBT)	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	

Biotinidase-Defizienz (BTD-Gen [OMIM #609019])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Congenitale bilaterale Aplasie des Vas deferens (CBAVD) (Azoospermie; Fertilitätsstörung) (CFTR-Gen [OMIM #602421])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis (Illumina); Dragen-Pipeline, Genoox (Franklin)	AA-MR-QM-1637-V3, AA-MR-QM-1391-V10, AA-MR-QM-1648-V2 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Pankreatitis, chronisch (CASR, CFTR-Gen [OMIM #602421], CPA1, CTRC, PRSS1, SPINK1)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis (Illumina); Dragen-Pipeline, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Metaphysäre Chondrodysplasie Typ Schmid (MCDS) (COL10A1-Gen [OMIM #120110])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Adrenogenitales Syndrom (AGS) (AGS Typ 3, Adrenogenitales Syndrom durch 21-Hydroxylase-Mangel) (CYP21A2-Gen [OMIM #613815])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005, AA-1599-V3	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Arzneimittelunverträglichkeit, Cytochrom P-450-bedingte (CYP2D6, CYP2C19, CYP1A2, CYP2B6, CYP2C8-Gen [OMIM #601129], CYP3A4, CYP3A5)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005, AA-MR-QM-0590	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Smith-Lemli-Opitz-Syndrom (DHCR7-Gen [OMIM #602858])	EDTA-Blut, DNA; DNA	Sequence Capture; Sequencing-by synthesis (Illumina); Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Muskeldystrophie Duchenne / Becker (DMD-Gen [OMIM #300377])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-0197-V007, AA-0269-V006, AA-1668-V001	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Myotone Dystrophie Typ 1 (Curschmann-Steinert-Syndrom) (DMPK-Gen [OMIM #605377])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1300-V3, AA-1299-V3	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Hämophilie A, Hämophilie B (F8-Gen [OMIM #300841], F9-Gen)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Tyrosinämie Typ I (FAH-Gen [OMIM #613871])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Achondroplasie (ACH) / Hypochondroplasie / Thanatophore Dysplasie (FGFR3-Gen [OMIM #134934])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Fragiles-X-Syndrom (FMR1-Gen [OMIM #309550])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-0277-V012	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Ataxie, Friedreichsche (FRDA1) (FXN-Gen [OMIM #606829])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-0313-V004, AA-1300-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Hämophilie A (Geninversionen int-22h/int-1h)	EDTA-Blut, DNA; DNA	PCR, Gelelektrophorese	AA-MR-QM-1413	Thermocycler, Agarosegelelektrophoresekammer		x	
Glucose-6-Phosphat-Dehydrogenase-Defizienz (Favismus) (G6PD-Gen [OMIM #305900])	EDTA-Blut, DNA; DNA	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)	-	x	

Galaktosämie (GALT-Gen [OMIM #606999])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1391-V010, AA-1648-V002	Illumina Series (NovaSeq)	-	x	
Morbus Gaucher (GBA-Gen [OMIM #606463])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Glutarazidurie Typ 1 (GCDH-Gen [OMIM #608801])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1391-V010, AA-1648-V002	Illumina Series (NovaSeq)	-	x	
Taubheit, autosomal-rezessiv 1A; Hörverlust, autosomal-rezessiv, nicht-syndromal (GJB2-Gen [OMIM #121011])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Alopezie (Atrichia mit papulären Läsionen, APL) (HR-Gen [OMIM #602302])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Chorea Huntington (HTT-Gen [OMIM #613004])	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1300-V003, AA-0316-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Isovalerianazidämie (IVD-Gen [OMIM #607036])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005	Illumina Series (NovaSeq)	-	x	
Kongenitaler Laktasemangel (LCT-Gen [OMIM #603202])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005	Illumina Series (NovaSeq)	-	x	
Rett Syndrom (RTT) (MECP2-Gen [OMIM #300005])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Methylmalonazidurie, Vitamin B12-resistent (MUT-Gen [OMIM #609058])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005	Illumina Series (NovaSeq)	-	x	
Arzneimittelunverträglichkeit (NAT2-Gen OMIM #612182)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Phenylketonurie/Hyperphenylalaninämie (PKU/HPA) (PAH-Gen [OMIM #612349])	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)	-	x	
Hereditäre Neuropathie mit Neigung zu Drucklähmung (HNPP) (PMP22-Gen [OMIM #601097])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Creutzfeldt-Jakob Erkrankung, familiäre Form (CJD), Gerstmann-Sträussler-Scheinker-Syndrom (GSSS) (PRNP-Gen [OMIM #176640])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Pankreatitis, chronisch (PRSS1-Gen [OMIM #276000])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Alpha-1-Antitrypsin-Mangel (SERPINA1-Gen [OMIM #613490])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Léri-Weill Dyschondrosteose, Langer mesomele Dysplasie, idiopathischer Kleinwuchs (SHOX-Gen [OMIM #312865])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Muskelatrophie, spinale Typ I – III (IV) (SMA1,2,3,4) (SMN1-Gen [OMIM #600354], SMN2)	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Taubheit, autosomal-rezessiv 16, DFNB16 (STRC-Gen [OMIM #606440])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	

Ehlers-Danlos-Syndrom mit Tenascin-X-Defizienz, classic like Typ 1 (clEDS) (TNXB-Gen [OMIM #600985])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Azathioprin-Therapie (TPMT-Gen [OMIM #187680])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Amyloidose, familiäre Form (TTR-Gen [OMIM #176300])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Crigler-Najar-Syndrom Typ 1, 2 (Hyperbilirubinämie) (UGT1A1-Gen [OMIM #191740])	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Multi Drug resistance (ABCB1 (MDR1) (dbSNP rs1045642))	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Alkohol-Intoleranz (ADH1B, ALDH2-Gen)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Apolipoprotein B-Defizienz (FLDB) (APOB-Gen: dbSNP rs12713559)	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
HIV-1-Wirtsresistenz (CCR5-Gen: dbSNP rs333, CCR2-Gen: dbSNP rs1799864, SDF1-Gen: dbSNP rs1801157)	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Verträglichkeit catecholaminerger Neurotransmitter (Catechol-O-Methyltransferase) (COMT (dbSNP rs4680))	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Detoxifizierungsstörung (CYP1A1 (dbSNP rs4646903, rs1048943))	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V004	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Hörverlust, rezessiv (Hörverlust, autosomal-rezessiv, nicht-syndromal) (GJB6-Gen (Deletion D13S1830))	EDTA-Blut, DNA; DNA	PCR, Gelelektrophorese	AA-0272-V005	Thermocycler, Gelelektrophoresekammer	-	x	
Detoxifizierungsstörung (GSTM1, GSTP1, GSTT1) (Xenobiotika-Metabolisierung)	EDTA-Blut, DNA; DNA	Restriktionsanalyse	AA-MR-QM-0143-V4	Thermocycler, Agarosegelelektrophoresekammer	-	x	
HCV-Therapie (HCV-Therapie ITPA (dbSNP rs1127354, rs7270101))	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V1, AA-MR-QM-0269-V7, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Morbus Crohn, Inflammatory Bowel Disease (NOD2-Gen/CARD15: dbSNP rs2066844, rs2066845, rs2066847)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Statin-Unverträglichkeit, Myopathie unter Hochdosis-Therapie (SLCO1B1 (dbSNP rs4149056))	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V1, AA-MR-QM-0269-V7, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
SULT1A1-bedingter verzögerten Phase II-Metabolismus (SULT1A1 (dbSNP rs9282861))	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-0143-V4	Thermocycler, Agarosegelelektrophoresekammer	-	x	
Meulengracht- (Gilbert-) Syndrom (UGT1A1-Gen: dbSNP rs3064744)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Cumarin-und Cumarinderivat-Sensitivität (VKORC1 (dbSNP rs9934438, rs28527768))	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V1, AA-MR-QM-0269-V7, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Pseudoxanthoma Elasticum (ABCC6)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-MR-QM-1668-V1, AA-MR-QM-0269-V7, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730		x	

monogene Adipositas (KSR2, LEP, LEPR, MC3R, MC4R, MRAP2, NTRK2, PCSK1, POMC, SH2B1, SIM1)	EDTA-Blut, DNA; DNA	Sequence Capture (TWIST); Sequencing-by synthesis; Dragen, Genoox (Franklin)	AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V005 Panel TPv3a	Illumina Series (NovaSeq)		x	
5 Fu-Toxizität (DPYD-Gen c.[1236G>A; 1129-5923G>C, 483DPYD-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; DNA	Fragmentlängenanalyse	AA-1679-V001	ABI Sequencer 3130XL, 3730XL, 3730	x		
5 Fu-Toxizität, Morbus-Meulengracht, Irinotecan-Verträglichkeit (DPYD, UGT1A1)	EDTA-Blut, DNA; DNA	Fluoreszenz-markierte Hybridisierungssonden	AA-1771-V001	Aria DX RealTime qPCR instrument	x		
Short Tandem repeats-/Mikrosatelliten-Analyse	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-1730-V001	ABI Sequencer 3130XL	x	--	
Zieldiagnostik / Bestätigungsanalyse SNV	EDTA-Blut, DNA; DNA	Fragmentlängenanalyse	AA-0272-V005; AA-0269-V007; AA-1668-V001	ABI Sequencer 3730XL, 3130XL, 3730	--	--	
Zieldiagnostik / Bestätigungsanalyse CNV	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL, 3130XL, 3730	--	x	
Pseudoxanthoma Elasticum (ABCC6-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Adrenogenitales Syndrom, AGS (AGS Typ 3, Adrenogenitales Syndrom durch 21-Hydroxylase-Mangel) (CYP21A2-Gen [OMIM #613815])	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008, AA-1599-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Hörverlust, autosomal-rezessiv, nicht-syndromal (GJB2-, GJB6-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730, Liquid Handler	-	x	
MECP2-Duplikationsyndrom (MECP2-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Rett-Syndrom (MECP2-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Fechtner-Syndrom (MYH9-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Charcot-Marie-Tooth Neuropathie Typ 1 (PMP22-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL, 3130XL, 3730	x	*	
Hereditäre Neuropathie mit Neigung zu Drucklähmungen, HNPP (PMP22-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V009	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Pankreatitis, chronisch (PRSS1, SPINK1-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Bannayan-Riley-Ruvalcaba-Syndrom (PTEN-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Familiäres Mamma-/Ovarialkarzinom (HBOC) (SCN5A-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Taubheit, autosomal-rezessiv (STRC-, OTOA-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Ehlers-Danlos-Syndrom (EDS), classical like Typ 1, mit Tenascin-X-Defizienz (TNXB-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
TSC2/PKD1-Contiguous-Gene-Syndrom (TSC2-, PKD1-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	-	x	

Whole Exome Sequencing	EDTA-Blut, DNA; DNA	Sequence Capture, Sequencing-by synthesis, Bioinformatikpipeline, Genoox (Franklin)	AA-MR-QM-1654-V4, AA-MR-QM-1637-V3, AA-MR-QM-1617-V3, AA-MR-QM-1648-V3, AA-MR-QM-1662-V1, AA-MR-QM-1652-V1, AA-MR-QM-1504-V7, AA-MR-QM-1635-V5 Panel TWIST Exom v1	Illumina Series (NovaSeq)		x	
Whole Genome Sequencing	EDTA-Blut, ; DNA	Adapter ligation, Sequencing-by synthesis, Dragen, Varseq (Golden Helix)	AA-MR-QM-1781-V1	Illumina Series (NovaSeq)		x	
Bestätigungsanalysen (Sanger und MLPA)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V9	Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730		x	
Pulmonale alveoläre Mikrolithiasis (PAM) (SLC34A2)	EDTA-Blut, DNA; DNA	Sanger-Sequenzierung	AA-1668-V001, AA-0269-V007, AA-0272-V005	ABI Sequencer 3730XL, 3130XL, 3730		x	

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)*

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version_x000D_ Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung (siehe Zeile 3), sofern zutreffend
Muskeldystrophie Duchenne / Becker (DMD-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	x		
Leri-Weill-Dyschondrosteose (LWD), Langer mesomele Dysplasie (LMD); idiopathischer Kleinwuchs (SHOX-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	x		
Spinale Muskelatrophie, SMA (SMN1, SMN2-Gen)	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-MR-QM-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	x		
Morbus Gaucher (GBA-Gen [OMIM #606463])	EDTA-Blut, DNA; DNA	(MS) MLPA	AA-0103-V008	ABI Sequencer 3730XL, 3130XL, 3730	x		

Untersuchungsart:
Molekularbiologische Untersuchungen (Hybridisierungsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version_x000D_ Pipeline/Kit/Panel+Version	Gerät	CE-Verfahren	in Haus-Verfahren	Erläuterung zu weiterer Bearbeitung (siehe Zeile 3), sofern zutreffend
Myotone Dystrophie Typ1, DM1 (Curschmann-Steinert-Syndrom) (DMPK-Gen)	EDTA-Blut, DNA; DNA	Southern-Blot-Hybridisierung	AA-MR-QM-1299-V003	ABI Sequencer 3730XL, 3130XL, 3730	-	x	
Fragiles X-Syndrom (FMR1-Gen)	EDTA-Blut, DNA; DNA	Southern-Blot-Hybridisierung	AA-MR-QM-0277-V13	Thermocycler, Hybridisierungsofen	-	x	

Untersuchungsgebiet: Mikrobiologie

Untersuchungsart:
Agglutinationsteste

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Staphylokokken (Koagulase, clumping factor, Protein A, Polysaccharide)	Bakterienkultur	Partikelagglutination	AA-1478-V002		x	
Streptokokken (Lancefield-Antigen)	Bakterienkultur	Partikelagglutination	AA-1452-V002		x	
Treponema pallidum Infektion assoziierte, nichtspezifische Lipoidantikörper (IgG, IgM)	Serum, EDTA-Plasma	Partikelagglutination	AA-1673-V001	Kartenschüttler, Biorad	x	

Untersuchungsart:
Empfindlichkeitstestungen von Bakterien, Parasiten, Pilzen

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Bakterien (Resistenztestung)	Keimkolonien in Reinkultur	Agardiffusionstest	AA-1473-V005	manuell	x	
Bakterien (Resistenztestung)	Keimkolonien in Reinkultur	Bouillondilutionsverfahren als minimale Hemmkonzentration (MHK)/Break-Point, vollmechanisiert	AA-1518-V006	Vitek2, Biomerieux	x	
Bakterien (Resistenztestung)	Keimkolonien in Reinkultur	trägergebundener Gradientendiffusionstest	AA-1473-V005	manuell	x	

Untersuchungsart:
Keimdifferenzierung/-identifizierung/-typisierung

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hefen	Pilzisolat	biochemisch aufwändig	AA-1518-V006	Vitek2, Biomerieux	x	
Anaerobier, Corynebakterien	Bakterienisolat	biochemisch aufwändig	AA-1518-V006	Vitek2, Biomerieux	x	
gram-negative aerobe Bakterien	Bakterienisolat	biochemisch aufwändig	AA-1518-V006	Vitek2, Biomerieux	x	
gram-positive aerobe Bakterien	Bakterienisolat	biochemisch aufwändig	AA-1518-V006	Vitek2, Biomerieux	x	
Neisseria sp., Haemophilus sp.	Bakterienisolat	biochemisch aufwändig	AA-1518-V006	Vitek2, Biomerieux	x	
Bakterien (Orientierungs-/Differenzierungsteste)	Keimkolonien in Reinkultur	biochemisch, orientierend (Katalase, Oxidase, Nitrocefin (Beta-Lactamase))	AA-1449-V003, AA-1455-V008		x	
Pneumokokken	Keimkolonien in Reinkultur	biochemisch, orientierend (Optochin)	AA-1455-V008		x	
Staphylococcus saprophyticus	Keimkolonien in Reinkultur	biochemisch, orientierend (Novobiocin)	AA-1472-V008		x	

Untersuchungsart:
Kulturelle Untersuchungen

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Bakterien, Pilze (Anlage und Ablesen)	Abstrich (urogenital, HNO), Blut, Haut, Wunde, Punktat, Stuhl,	in CO2-angereicherter Atmosphäre, mikroaerobe/anaerobe Atmosphäre, spezifisch, unspezifisch	AA-1482-V006, AA-1490-V005, AA-1531-V004, AA-1717-V002, AA-1537-V004, AA-1565-V002		x	
Bakterien, Pilze	Urin	spezifisch, unspezifisch, Keimzahlbestimmung	AA-1472-V008		x	
gram-negative multiresistente Bakterien	Abstrich (rektal), Wunde, Stuhl, Urin	spezifisch (selektiv)	AA-1765-V002		x	

Untersuchungsart:
Ligandenassays

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chlamydia trachomatis IgG/IgA	Serum	ELISA	AA-1437-V013	Euroimmun Analyzer I (EuroImmun)/manuel (Euroimmun)	x	
Toxoplasma gondii, IgG, IgM	Serum	CLIA	AA-1401-V011, AA-MR-QM-1624-V5	Liaison XL, DiaSorin	x	
Treponema pallidum Antikörper (IgG + IgM quantitativ)	Serum, EDTA-Plasma	ELISA	AA-1437-V013	Euroimmun Analyzer I (Euroimmun)/manuell	x	
Treponema pallidum, IgG + IgM Suchtest	Serum	CLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Borrelia burgdorferi sensu lato IgG, IgM	Serum, EDTA-Plasma	CLIA	AA-1624-V005	LIAISON XL, DiaSorin	x	
Diphtherie-Toxoid, IgG	Serum, EDTA-Plasma	ELISA	AA-1437-V013	Euroimmun Analyzer I (Euroimmun)/manuell	x	
Yersinien Antikörper IgA/IgG/IgM	Serum	Immunoblot	AA-1583-V004	manuell (Mikrogen)/EUROBlotMaster (außer IgM)	x	
Borrelia burgdorferi sensu lato IgG, IgM	Serum, EDTA-Plasma	Immunoblot	AA-1583-V004	manuell (Mikrogen)/EUROBlotMaster	x	
Cryptosporidien Antigennachweis	Stuhl	ELISA	AA-1563-V007	Dynex DSX, R-Biopharm	x	
Entamoeba histolytica/dispar Antigennachweis	Stuhl	ELISA	AA-1563-V007	Dynex DSX, R-Biopharm	x	
Giardia lamblia Antigennachweis	Stuhl	ELISA	AA-1563-V007	Dynex DSX, R-Biopharm	x	
Treponema pallidum IgM	Serum, EDTA-Plasma	Immunoblot	AA-1583-V004	manuell (Mikrogen) + EUROBlotMaster (neu)	x	
Chlamydia trachomatis, psittaci, pneumoniae IgA, IgG, IgM	Serum, EDTA-Plasma	Immunoblot	AA-1583-V004	manuell (Mikrogen)/EUROBlotMaster	x	
Helicobacter pylori Antigennachweis	Stuhl	ELISA	AA-1563-V007	Dynex DSX, R-Biopharm	x	
Tetanus-Toxoid IgG	Serum, EDTA-Plasma	ELISA	AA-1437-V013	Euroimmun Analyzer I (Euroimmun)/manuell	x	
Clostridium difficile Toxin A/B-Antig.	Stuhl	ELISA	AA-1558-V002	Dynex DSX, R-Biopharm	x	
Clostridium difficile GDH	Stuhl	ELISA	AA-1558-V002	Dynex DSX, R-Biopharm	x	

Untersuchungsart:
Mikroskopie

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Bakterien	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Pilze	Urin, Urinsediment	Hellfeldmikroskopie	AA-1467-V004	Mikroskop	x	
Bakterien, Pilze	Abstrich	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-1444-V005, AA-1508-V004	Mikroskop	x	
Schimmelpilze	Pilzkultur	Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen	AA-1467-V004	Mikroskop	x	

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Bordetalla pertussis, Bordetella parapertussis, DNA	Abstrich, Sputum	Real-Time PCR	AA-1511-V004	CFX Opus, Biorad	x	

Chlamydia pneumoniae, Mycoplasma pneumoniae, DNA	Abstrich, Sputum	Real-Time PCR	AA-1523-V005	CFX Opus, Biorad	x	
Chlamydia trachomatis, Mycolpasma genitalium, Neisseria gonorrhoeae, Trichomonas vaginalis (STIs, sexually transmitted infections), DNA	Abstrich, Urin, Ejakulat	Real-Time PCR	AA-1659-V004	Alinity m, Abbott	x	
Chlamydia trachomatis, Mycoplasma hominis, genitalium, Neisseria gonorrhoeae, Trichomonas vaginalis, Ureaplasma urealyticum, parvum (STIs, sexually transmitted infections), DNA	Abstrich, Urin, Ejakulat	Real-Time PCR	AA-1458-V008	CFX Opus, Biorad	x	

Untersuchungsgebiet: Virologie

Untersuchungsart:
Ligandenassays*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Cytomegalievirus, IgG, IgM	Serum, EDTA-Plasma	CLIA	AA-1401-V011, AA-1487-V006, AA-MR-QM-1624-V5	Liaison XL, DiaSorin	x	
Enteroviren, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	
Epstein-Barr-Virus, VCA, EA, EBNA, IgG, IgM	Serum, EDTA-Plasma	CLIA	AA-1624-V004	LIAISON XL, DiaSorin	x	
Epstein-Barr-Virus	Serum, EDTA-Plasma	Immunoblot	AA-1538-V004	RemcomScan, Mikrogen/EUROBlotMaster	x	
Hepatitis-A-Virus, Ig (Suchtest)	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas e411, Roche	x	
Hepatitis-A-Virus, IgM	Serum	ECLIA	AA-MR-QM-1624	LIAISON XL, DiaSorin	x	
Hepatitis-B-Virus, Anti-HBc Ig	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas e411, Roche	x	
Hepatitis-B-Virus, Anti-HBc IgM	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Hepatitis-B-Virus, Anti-HBe Ig	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Hepatitis-B-Virus, Anti-HBs Ig	Serum, EDTA-Plasma	ECLIA	AA-1401-V011	Cobas pro, Roche	x	
Hepatitis-B-Virus, HBs-Antigen, qualitativ	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Hepatitis-B-Virus, HBs-Antigen, quantitativ	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas e411, Roche	x	
Hepatitis-C-Virus, Ig	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Hepatitis-C-Virus, Ig	Serum, EDTA-Plasma	Immunoblot	AA-1399-V005	manuell (Mikrogen)/EUROBlotMaster	x	
Hepatitis-D-Virus, Ig	Serum, EDTA-Plasma	ELISA	AA-1398-V008	manuell (Kit DiaSorin)	x	
Hepatitis-E-Virus, IgG, IGM	Serum, EDTA-Plasma	ELISA	AA-1398-V008	Dynex DSX, R-Biopharm	x	
Hepatititis-B-Virus, HBe-Antigen	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Herpes-Simplex-Virus, IgG	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	
Humanes Immundefizienzvirus, Antigen + Antikörper	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Humanes Immundefizienzvirus, Antikörper	Serum, EDTA-Plasma	Immunoblot	AA-1399-V005	manuell (Mikrogen)/EUROBlotMaster	x	
Masern IgG/IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	
Mumps IgG/IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	
Parvovirus B19, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	

Rötelnvirus, IgG	Serum, EDTA-Plasma	ECLIA	AA-1401-V011, AA-1487-V006	Cobas pro, Roche	x	
Varizella Zoster-Virus, IgG, IgM	Serum, EDTA-Plasma	ELISA	AA-1437-V012	manuell (EuroImmun)	x	
SARS-CoV-2 IgG quantitativ	Serum, EDTA-Plasma	ECLIA	AA-1624-V004	LIAISON XL, DiaSorin	x	
Denguevirus, IgG, IgM	Serum	Immunoblot	AA-MR-QM-1583-V5	manuell (Mikrogen)	x	
Chikungunyavirus, IgG, IgM	Serum	Immunoblot	AA-MR-QM-1583-V5	manuell (Mikrogen)	x	
Zikavirus, IgG, IgM	Serum	Immunoblot	AA-MR-QM-1583-V5	manuell (Mikrogen)	x	

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Influenzaviren, Respiratory-Syncytial-Virus, SARS-CoV-2, DNA	Abstrich	Real-time PCR	AA-1659-V004	Alinity m, Abbott	x	
Influenzaviren, Respiratory-Syncytial-Virus, DNA	Abstrich	Real-time PCR	AA-1523-V005	CFX Opus, Biorad		
Hepatitis-B-Virus, DNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1400-V007, AA-1659-V004	Alinity m, Abbott	x	
Hepatitis-B-Virus, DNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1602-V004	cobas 6800, Roche	x	
Hepatitis-C-Virus, RNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1400-V007, AA-1659-V004	Alinity m, Abbott	x	
Hepatitis-C-Virus, RNA, quantitativ	Serum, EDTA-Plasma	Real-time PCR	AA-1602-V004	cobas 6800, Roche	x	
Humane Papillomaviren, qualitativ	Abstrich	Real-time PCR	AA-1659-V004	Alinity m, Abbott	x	
Humans Immundefizienzvirus, RNA, quantitativ	EDTA-Plasma	Real-time PCR	AA-1400-V007, AA-1659-V004	Alinity m, Abbott	x	
Humans Immundefizienzvirus, RNA, quantitativ	EDTA-Plasma	Real-time PCR	AA-1602-V004	cobas 6800, Roche	x	
Parvovirus B19 DNA	Biopsat, EDTA-Plasma	Real-time PCR	AA-1570-V003	CFX Opus, Biorad	x	
HEV-RNA (HEV-RNA)	Serum, EDTA-Plasma	Real-time PCR	AA-1692-V003	CFX Opus, Biorad	x	
SARS-CoV-2	Abstrich, Rachenspülung	Real-time PCR	AA-1659-V004	Alinity m, Abbott	x	
SARS-CoV-2	Abstrich	Real-time PCR	AA-1602-V004	cobas 6800, Roche	x	
SARS-CoV-2	Abstrich (Nasenrachenraum)	Real-time PCR	AA-MR-QM-1523-V6	CFX Opus, Biorad	x	
Herpes-Simplex-Virus-1 und 2 DNA	Abstrich (urogenital)	Real-time PCR	AA-1458-V007	CFX Opus, Biorad	x	
Adenoviren, DNA; Enteroviren, Rhinoviren, RNA	Abstrich	Real-time PCR	AA-1523-V005	CFX Opus, Biorad	x	

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hepatitis-C-Virus, RNA, Genotyp Core	Serum, EDTA-Plasma	Real-time PCR	AA-1410-V003, AA-1428-V006	Thermocyler, Biometra Analytik		x
HDV-RNA (quantitativ) (HDV-RNA (quantitativ))	Serum, EDTA-Plasma	Real-time PCR	AA-1693-V005	CFX Opus, Biorad		x

Untersuchungsgebiet: Transfusionsmedizin

Untersuchungsart:
Agglutinationsteste*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
AB0-Blutgruppenbestimmung	EDTA-Blut, Blut	Hämagllutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	
Antikörpersuchtest	EDTA-Plasma	Hämagllutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	
direkter Coombstest	EDTA-Blut, Blut	Hämagllutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	

Kellsystem	EDTA-Blut, Blut	Hämaglutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	
Rh-D-Bestimmung	EDTA-Blut, Blut	Hämaglutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	
Rhesusformel	EDTA-Blut, Blut	Hämaglutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	
Serumgegenprobe zur AB0-Bestimmung	EDTA-Plasma	Hämaglutinationstest / Geltechnik	AA-MR-QM-1475-V10	Banjo-Reader, Biorad	x	

Untersuchungsart:
Durchflusszytometrie*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
HLA-Antikörper	Serum, EDTA-Plasma	Festphasenassay	AA-MR-QM-0394-V11	LABScan 3D, BMT	x	

Untersuchungsart:
Durchflusszytometrie**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
HLA-Crossmatch	CPDA1-Blut, Serum	Durchflusszytometrie	AA-MR-QM-0176-V10	FACS Canto II, BD		x

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
KIR	DNA	PCR-SSO	AA-MR-QM-0207-V9	LabScan 3D, BMT	x	
KIR	DNA	PCR-SSP	AA-MR-QM-0207-V9	Helmberg Score, Genovision	x	
HLA-Klasse I, II	DNA	PCR-SSO	AA-MR-QM-0211-V18	LabScan 3D, BMT	x	
HLA-Klasse I, II	DNA	PCR-SSP	AA-MR-QM-0192-V17	Helmberg Score, Genovision	x	
HLA-Klasse I, II	DNA	Sanger-Sequenzierung	AA-MR-QM-0215-V12	ABI3730, SeCore	x	

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
HLA-Klasse I, II	DNA	(PCR); Sequencing-by synthesis (Illumina); in-house pipeline	AA-MR-QM-1391-V11	MiSeq/NovaSeq, Illumina		x
HLA-Klasse I, II	DNA	long-read SMRT Sequencing (Pacific Biosciences of California)	AA-MR-QM-1769-V3	PacBio Sequel IIe, Pacific Biosciences of California		x
Nachweis der Exone 5, 7 und 10 des RHD Gens	fetale dfDNA aus mütterlichem Plasma (EDTA)	Real-time PCR	AA-MR-QM-1721-V4	CFX96/384 Touch, Bio-Rad		x
Rhesustypisierung (RHD/CE-Gen)	Genomische DNA	PacBio, IMGT HLA-Datenbank	AA-MR-QM-1769-V3	PacBio Sequel IIe, Pacific Biosciences of California		x