

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

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

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

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| Antineutrophile cytoplasmatische Antikörper (ANCA)    | Vollblut, Serum,<br>EDTA-Plasma        | Indirekte<br>Immunfluoreszenz<br>mikroskopie  | AA-1351-V003          | IFT, Euroimmun | flexibel               |
| Autoantikörper gegen Zellkerne (ANA)                  | Vollblut, Serum,<br>EDTA-Plasma        | Indirekte<br>Immunfluoreszenz<br>mikroskopie  | AA-1351-V003          | IFT, Euroimmun | flexibel               |
| Immunglobulin (Ig) A                                  | Serum                                  | Turbidimetrie   | AA-1480-V006          | Cobas pro      | flexibel               |
| Immunglobulin (Ig) G                                  | Serum                                  | Turbidimetrie   | AA-1480-V006          | Cobas pro      | flexibel               |
| Immunglobulin (Ig) M                                  | Serum                                  | Turbidimetrie   | AA-1480-V006          | Cobas pro      | flexibel               |
| Rheumafaktor  | Serum                                  | Turbidimetrie   | AA-1480-V006          | Cobas pro      | flexibel               |
| Retikulozyten   | EDTA-Blut                              | Bestimmung<br>zytochemisch-<br>zytometrischer<br>Merkmale   | AA-0178-V011          | Sysmex XN1000  | flexibel               |
| Großes Blutbild                                       | EDTA-Blut                              | Partikelzählung,<br>Partikelgrößenbes-<br>timmung,<br>Bestimmung<br>zytochemisch-<br>zytometrischer<br>Merkmale | AA-0178-V011          | Sysmex XN1000  | flexibel               |
| Kleines Blutbild                                      | EDTA-Blut                              | Partikelzählung,<br>Partikelgrößenbes-<br>timmung,<br>Bestimmung<br>zytochemisch-<br>zytometrischer<br>Merkmale | AA-0178-V011          | Sysmex XN1000  | flexibel               |
| Thrombozyten  | EDTA-Blut                              | Partikelzählung,<br>optisch-<br>elektronisch  | AA-0178-V011          | Sysmex XN1000  | flexibel               |
| Chlorid   | Serum, Urin                            | Ionenselektive<br>Elektrode (ISE)   | AA-1480-V007          | Cobas pro      | flexibel               |
| Kalium  | Serum, Urin                            | Ionenselektive<br>Elektrode (ISE)   | AA-1480-V007          | Cobas pro      | flexibel               |

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| Natrium   | Serum, Urin                            | Ionenselektive<br>Elektrode (ISE)    | AA-1480-V007          | Cobas pro                  | flexibel               |
| Monoklonale Gammopathie                               | Serum                                  | Kapillarelektropho-<br>rese          | AA-1530-V003          | Sebia Capillarys 3<br>Octa | flexibel               |
| Serumeiweiß   | Serum                                  | Kapillarelektropho-<br>rese          | AA-1486-V006          | Sebia Capillarys 3<br>Octa | flexibel               |
| Antithrombin III                                      | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| D-Dimer   | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Fibrinogen  | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| INR   | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Partielle Thromboplastinzeit                          | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Plasmathrombinzeit                                    | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Faktor 9  | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Anti-Xa   | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |
| Faktor 13   | Citratplasma                           | Turbidimetrischer<br>Immunoassay     | AA-1528-V006          | ACLTop 350                 | flexibel               |
| freies Protein S                                      | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLTop 350                 | flexibel               |

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| von Willebrand-Faktor Antigen                         | Citratplasma                           | Turbidimetrischer<br>Immunoassay     | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| von Willebrand-Faktor Aktivität                       | Citratplasma                           | Turbidimetrischer<br>Immunoassay     | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| Protein C   | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| APC-Resistenz, FV Leiden                              | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| Faktor 8  | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| Faktor 12   | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLS Top 350                 | flexibel               |
| Thromboplastinzeit                                    | Citratplasma                           | optische<br>Detektionsverfahr-<br>en | AA-1528-V006          | ACLS Top 350 (Quick<br>Test) | flexibel               |
| AFP   | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas e411 / Roche           | flexibel               |
| Anti-Müller-Hormon                                    | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |
| Beta-HCG  | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas e411 / Roche           | flexibel               |
| CA 125  | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas e411 / Roche           | flexibel               |
| CA 15-3   | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas e411 / Roche           | flexibel               |
| CA 19-9   | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas e411 / Roche           | flexibel               |
| Carcinogenic embryonic antigen (CEA)                  | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |
| Cortisol  | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |
| C-Peptid  | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |
| cyclischen Citrullin Peptid-Antikörper (Anti-CCP)     | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |
| DHEA-Sulfat   | Serum                                  | ECLIA                                | AA-1487-V006          | Cobas pro                    | flexibel               |

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| Folsäure  | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Follikelstimulierendes Hormon (FSH)                     | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| FT3   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| FT4   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Holotranscobalamin (Active B12)                         | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Humanes Threoglobulin (HTG)                             | Serum                                  | ECLIA                     | AA-1487-V007          | Cobas pro                       | flexibel               |
| Humanes Wachstumshormon (hGH)                           | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Insulin   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| insulinähnlicher Wachstumsfaktor-1 (IGF-1)              | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Luteinisierendes Hormon (LH)                            | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| N-terminales pro-B-Typ natriuretisches Peptid (NPROBNP) | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas e411 / Roche              | flexibel               |
| Osteocalcin   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Östradiol   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Parathormon   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Progesteron   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Prokollagen Typ 1 N-terminales Propeptid (P1NP)         | Serum                                  | ECLIA                     | AA-1487-V007          | Cobas pro                       | flexibel               |
| Prolaktin   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Totales prostataspezifisches Antigen (PSA)              | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Sexualhormonbindendes Globulin (SHBG)                   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| β-CrossLaps   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Testosteron   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Troponin T  | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas e411 / Roche              | flexibel               |
| Thyroidea stimulierendes Hormon (TSH)                   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas e411                      | flexibel               |
| Vitamin B12   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Vitamin D   | Serum                                  | ECLIA                     | AA-1487-V006          | Cobas pro                       | flexibel               |
| Inhibin B   | Serum                                  | ELISA                     | AA-1703-V001          | Dynex DSX                       | flexibel               |
| AFP   | Fruchtwasser                           | Immunometrie<br>(CLIA)    | AA-1517-V005          | Kryptor compact<br>plus, Brahms | flexibel               |
| Chromogranin A (CGA)                                    | Serum                                  | Immunometrie<br>(CLIA)    | AA-1517-V005          | Kryptor compact<br>plus, Brahms | flexibel               |
| Freies Beta-HCG   | Serum                                  | Immunometrie<br>(CLIA)    | AA-1517-V005          | Kryptor compact<br>plus, Brahms | flexibel               |

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| PAPP-A  | Serum                                  | Immunometrie<br>(CLIA)   | AA-1517-V005          | Kryptor compact<br>plus, Brahms | flexibel               |
| Procalcitonin   | Serum                                  | Immunometrie<br>(CLIA)   | AA-1517-V005          | Kryptor compact<br>plus, Brahms | flexibel               |
| Erythrozyten  | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Erythrozytenzylinder                                  | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Granulierte Zylinder                                  | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Hyaline Zylinder                                      | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Leukozyten  | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Leukozytenzylinder                                    | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Platteneithelien                                      | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Rundepithelien  | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Übergangsepithelien                                   | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| Wachszylinder   | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                                       | AA-1467-V004          | Mikroskop                       | flexibel               |
| atypische Lymphozten                                  | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop                       | flexibel               |
| Basophile Granulozyten                                | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop                       | flexibel               |



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| Blasten   | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Eosinophile Granulozyten                              | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Kernschatten  | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Lymphozyten   | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Metamyelozyten  | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Monozyten   | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Morphologie Erythrozyten                              | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik                                   | Anweisung/<br>Version | Gerät     | Akkreditierungs-status |
|---|--|--|-----------------------|-----------|------------------------|
| Morphologie Leukozyten                                | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Morphologie Thrombozyten                              | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Myelozyten  | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Normoblasten  | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Plasmazellen  | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Promyelozyten   | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |
| Segmentkernige Granulozyten                           | Vollblut                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-0167-V010          | Mikroskop | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik                                      | Anweisung/<br>Version | Gerät        | Akkreditierungs-status |
|---|--|--|-----------------------|--------------|------------------------|
| Stabkernige Granulozyten                              | Vollblut                               | Hellfeldmikroskopi-<br>e nach Anfärbung<br>mittels Farbstoffen | AA-0167-V010          | Mikroskop    | flexibel               |
| Bilirubin   | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Glucose   | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Hämoglobin/Erythrozyten                               | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Ketone  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Leukozyten  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Nitrit  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| pH  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Proteine  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Urobilinogen  | Urin                                   | Teststreifen   | AA-1467-V004          | Teststreifen | flexibel               |
| Antistreptolysin O (ASLO)                             | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Complement C3c (C3)                                   | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Complement C4 (C4)                                    | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Albumin   | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Albumin   | Urin                                   | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Alkalische Phosphatase                                | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Amylase   | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Bilirubin, direkt                                     | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Bilirubin, gesamt                                     | Serum                                  | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |
| Calcium   | Serum, Plasma,<br>Urin                 | UV-/VIS-<br>Photometrie  | AA-1480-V006          | Cobas pro    | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik | Anweisung/<br>Version | Gerät     | Akkreditierungs-status |
|---|--|---------------------------|-----------------------|-----------|------------------------|
| Cholinesterase (CHE)                                  | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Cholesterin   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Kreatinkinase (CK)                                    | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| CK-MB   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Cystatin C (CYSC)                                     | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Eisen   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Eiweiß  | Urin                                   | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Eiweiß, gesamt  | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Gamma-GT (GGT)  | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Glucose   | Serum, NaF-<br>Plasma                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| GOT (AST)   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| GPT (ALT)   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Harnsäure   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Harnstoff   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| HDL Cholesterin                                       | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Homocystein   | Serum                                  | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |
| Kreatinin   | Serum,Urin                             | UV-/VIS-<br>Photometrie   | AA-1480-V006          | Cobas pro | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik                              | Anweisung/<br>Version | Gerät           | Akkreditierungs-status |
|---|--|--|-----------------------|-----------------|------------------------|
| Laktatdehydrogenase (LDH)                             | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Low densitiy lipoprotein (LDL)                        | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Lipase  | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Magnesium   | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Phosphat  | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Triglyceride  | Serum                                  | UV-/VIS-<br>Photometrie                                | AA-1480-V006          | Cobas pro       | flexibel               |
| Blutsenkung (in mm/h)                                 | EDTA-Blut                              | VIS-Photometrie  | AA-1595-V001          | Roller 20LC     | flexibel               |
| Lipoprotein (Lp) a                                    | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| TG  | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| alpha 1 Antitrypsin                                   | Serum                                  | Turbidimetrie<br>Turbidimetrie/Im-<br>munturbidimetrie | AA-1480-V006          | Cobas pro       | flexibel               |
| Coeruloplasmin  | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| C-reaktives Protein (Globaltest und high sensitive)   | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| Ferritin  | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| Haptoglobin   | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| HbA1c   | EDTA-Blut                              | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| Transferrin   | Serum                                  | Turbidimetrie  | AA-1480-V006          | Cobas pro       | flexibel               |
| 17-OH-Progesteron                                     | Serum                                  | ELISA  | AA-1703-V001          | Dynex DSX       | flexibel               |
| 5alpha-Dihydrotestosteron (DHT)                       | Serum                                  | ELISA  | AA-1703-V001          | Dynex DSX       | flexibel               |
| DHEA  | Serum                                  | ELISA  | AA-1703-V001          | Dynex DSX       | flexibel               |
| Osmolalität   | Serum, Urin                            | Kyroskopie   | AA-1678-V001          | Osmometer 3000D | flexibel               |
| Bakterien   | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                               | AA-1467-V004          | Mikroskop       | flexibel               |
| Pilze   | Urin,<br>Urinsediment                  | Hellfeldmikroskopi-<br>e                               | AA-1467-V004          | Mikroskop       | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version  | Gerät                                       | Akkreditierungs-status |
|---|--|---|--|---|------------------------|
| HLA-Crossmatch  | CPDA1-Blut,<br>Serum                   | Durchflusszytomet-<br>rie   | AA-0176-V009   | FACS Canto II, BD                           | flexibel               |
| 1,25 Dihydroxivitamin D   | Serum                                  | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| ACTH  | EDTA-Plasma                            | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| Aldosteron  | Serum                                  | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| Androstendion   | Serum                                  | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| BAP   | Serum                                  | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| Calcitonin  | Serum                                  | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| Renin   | EDTA-Plasma                            | CLIA  | AA-1624-V002   | LIAISON XL /<br>DiaSorin                    | flexibel               |
| Agammaglobulinämie hereditäre (BLNK, BTK, CD79A,<br>CD79B, IGHM, IGLL1, LRRC8A, PIK3R1)   | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V002,<br>AA-1729-V001,<br>AA-1617-V003,<br>AA-1709-V002,<br>AA-1733-V003 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Hereditäre periodische Fiebersyndrome (HPF) (ELANE,<br>IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3,<br>NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A) | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V002,<br>AA-1729-V001,<br>AA-1617-V003,<br>AA-1709-V002,<br>AA-1733-V003 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| Immundefekte im Kindesalter primäre (ADA, AK2, AP3B1, BLNK, BTK, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD79A, CD79B, CD8A, CIITA, CLPB, CORO1A, CSF3R, CXCR4, DCLRE1C, DOCK8, ELANE, FOXP1, G6PC3, GATA1, GATA2, GFI1, HAX1, IGHM, IGLL1, IKZF1, IL2RG, IL7R, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LRRC8A, LYST, MAGT1, NHEJ1, ORAI1, PIK3R1, PNP, PRKDC, PTPRC, RAB27A, RAG1, RAG2, RFX5, RFXANK], RFXAP, RHOH , RMRP, SBDS , SLC37A4, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP , TAZ, TRAC, UNC119 , USB1, VPS13B , VPS45, WAS, ZAP70) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Kombinierte T- und B-Zellimmundefekte ( ADA, AK2 , CD247, CD3D , CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DOCK8, FOXP1, IKZF1, L2RG, IL7R, ITK, JAK3, LCK, LIG4, MAGT1, NHEJ1, ORAI1 , PNP, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TRAC, UNC119, ZAP70.)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Neutropenie, kongenital (AP3B1, CLPB, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, JAGN1, LAMTOR2, LYST, RAB27A, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Omenn-Syndrom (OS) ( ADA, AK2, DCLRE1C, IL2RG, IL7R, JAK3, LIG4, RAG1, RAG2, RMRP)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| Schwere kombinierte Immundefekte T-B-) (ADA, AK2, DCLRE1C, LIG4, NHEJ1, PRKDC, RAG1, RAG2)           | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Schwere kombinierte Immundefekte T-B+) (CD247, CD3D, CD3E, CORO1A, FOXP1, IL2RG, IL7R, JAK3, PTPRC ) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V002, AA-1729-V001, AA-1617-V003, AA-1709-V002, AA-1733-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Agammaglobulinämie Bruton (XLA) (BTK-Gen)  | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0197-V007, AA-0269-V007, AA-1313-V007                             | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |
| Autoimmun-Polyendokrinopathie-Candidiasis-Ektodermaldystrophie-Syndrom Typ I (APECED) (AIRE-Gen)     | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0197-V007, AA-0269-V007, AA-1313-V007                             | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |
| CINCA-Syndrom (NLRP3-Gen)  | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0197-V007, AA-0269-V007, AA-1313-V007                             | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |
| Cryopyrin-assoziierte periodische Syndrome (CAPS) (NLRP3-Gen)  | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0197-V007, AA-0269-V007, AA-1313-V007                             | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |
| Familiäres Kälte-assoziiertes autoinflammatorisches Syndrom Typ I (NLRP3-Gen)                        | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0197-V007, AA-0269-V007, AA-1313-V007                             | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)     | Untersuchungs-<br>material<br>(Matrix) | Untersuchungste-<br>chnik | Anweisung/<br>Version                    | Gerät                              | Akkreditierungs-status |
|---|--|---------------------------|--|------------------------------------|------------------------|
| Hyper-IgD-und-periodisches-Fiebersyndrom (HIDS) (MVK-Gen) | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Hyper-IgM-Syndrom (AICDA-Gen)                             | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Hyper-IgM-Syndrom (CD40-Gen)                              | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Hyper-IgM-Syndrom (CD40LG-Gen)                            | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Hyper-IgM-Syndrom (UNG-Gen)                               | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Mevalonazidurie (MVK-Gen)                                 | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Mittelmeerfieber, familiäre Form (FMF) (MEFV-Gen)         | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Muckle-Wells-Syndrom (NLRP3-Gen)                          | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung      | AA-0197-V007, AA-0269-V007, AA-1313-V007 | ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik     | Anweisung/<br>Version                          | Gerät                                    | Akkreditierungs-status |
|--|--|--------------------------|--|--|------------------------|
| TNF-Rezeptor-1-assoziiertes periodisches Syndrom (TRAPS)<br>(TNFRSF1A-Gen)   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Wiskott-Aldrich-Syndrom (WAS) (WAS-Gen)  | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| X-gebundener schwerer kombinierter Immundefekt (X-<br>SCID) (IL2RG-Gen)  | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| X-gebundenes lymphoproliferatives Syndrom (XLP1)<br>(SH2D1A-Gen)   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Zyklische Neutropenie (CyN) / schwere kongenitale<br>Neutropenie (SCN) (ELANE)   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Rheumatoide Arthritis (IL4R (dbSNP rs1805010))   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer 3730                       | flexibel               |
| Rheumatoide Arthritis (TNF- $\alpha$ -Promotor (dbSNP rs361525,<br>rs1800629, rs1800750, rs1799724, rs1800630, rs1799964)) | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer 3730                       | flexibel               |
| Shwachman-Bodian-Diamond-Syndrom (SBDS-Gen )   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-0269-V007,<br>AA-1313-V007 | ABI Sequencer 3730                       | flexibel               |
| X-gekoppelte Agammaglobulinämie Typ Bruton, XLA (BTK-<br>Gen)  | EDTA-Blut, DNA<br>aus Blut             | (MS) MLPA                | AA-0103-V008                                   | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik               | Anweisung/<br>Version          | Gerät                         | Akkreditierungs-status |
|---|--|---|--------------------------------|-------------------------------|------------------------|
| ABO-Blutgruppenbestimmung                             | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| Antikörpersuchtest                                    | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| direkter Coombstest                                   | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| Kellsystem  | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| Rh-D-Bestimmung                                       | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| Rhesusformel  | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| Serumgegenprobe zur ABO-Bestimmung                    | EDTA-Blut /<br>Vollblut                | Hämagglutinations-<br>test / Geltechnik | AA-1475-V009                   | Banjo ID-Reader               | flexibel               |
| KIR   | Genomische<br>DNA                      | PCR-SSO                                 | AA-0207-V008                   | Luminex 200,<br>LabScan 3D    | flexibel               |
| KIR   | Genomische<br>DNA                      | PCR-SSP                                 | AA-0207-V008                   | Helmborg Score,<br>Genovision | flexibel               |
| HLA-Antikörper  | Serum, EDTA-<br>Plasma                 | Festphasenassay                         | AA-0394-V010                   | Luminex 200,<br>LabScan 3D    | flexibel               |
| HLA-Klasse I  | Genomische<br>DNA                      | Sanger-<br>Sequenzierung                | AA-0201-V0111,<br>AA-0206-V007 | ABI3730                       | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                                      | Untersuchungs-<br>material<br>(Matrix)                                 | Untersuchungstec-<br>hnik  | Anweisung/<br>Version         | Gerät   | Akkreditierungs-status |
|--|--|--|-------------------------------|---|------------------------|
| HLA-Klasse I, II   | Genomische<br>DNA  | Sequencing-by<br>synthesis<br>(Illumina),<br>MiSeq/NovaSeq<br>Illumina, IMGT<br>HLA-Datenbank  | AA-1391-V010                  | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                       | flexibel               |
| HLA-Klasse I, II   | Genomische<br>DNA (intern:<br>DNA aus Blut,<br>Schleimhauttupf-<br>er) | longrange-PCR,<br>Sequencing-by<br>synthesis (Illumina<br>bzw. Pacific<br>Biosciences of<br>California),<br>MiSeq/NovaSeq<br>Illumina bzw.<br>PacBio Sequel IIe,<br>IMGT HLA-<br>Datenbank | AA-1550-V006,<br>AA-1769-V001 | Illumina Series<br>(NovaSeq, Miseq,<br>etc), PacBio Sequel<br>IIe | flexibel               |
| HLA-Klasse I, II   | Genomische<br>DNA  | PCR-SSO  | AA-0211-V017                  | Luminex 200,<br>LABScan<br>OneLambda                              | flexibel               |
| HLA-Klasse I, II   | Genomische<br>DNA  | PCR-SSP  | AA-0192-V016                  | Helmsberg Secore,<br>Genovision                                   | flexibel               |
| HLA-Klasse I, II   | Genomische<br>DNA  | Sanger-<br>Sequenzierung   | AA-0215-V011                  | ABI3730   | flexibel               |
| Nachweis der Exone 5,7 und 10 des RHD Gens (Nachweis<br>der Exone 5,7 und 10 des RHD Gens) | fetale cfDNA aus<br>mütterlichem<br>Plasma (EDTA)                      | Real-time PCR  | AA-1721-V003                  | CFX96/384 Touch,<br>Bio-Rad                                       | flexibel               |

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

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|---|--|--|--|---------------------------------------|------------------------|
| <b>Cutis laxa</b> (ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, EFEMP2, ELN, FBLN5, LTBP4, PYCR1)                               | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Kollagen 4-assoziierte intrazerebrale Blutungen</b> (COL4A1, COL4A2)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Loeys-Dietz-Syndrom (LDS)</b> (SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Bikuspide Aortenklappe, mit Risiko für Aortenaneurysma und Aortenstenose/-dilatation</b> (GATA5, NOTCH1, ROBO4, SMAD6) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Thorakale Aortenerweiterung mit dem Risiko der Aortendissektion</b> (ACTA2, BGN, COL1A1, COL3A1, COL4A5, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FLNA, FOXE3, GATA5, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Marfan-ähnliche Erkrankungen</b> (ADAMTS10, ADAMTS17, ADAMTSL2, ADAMTSL4, EFEMP1, FBN1, FBN2, LTBP2, LTBP3, MED12, SKI, UPF3B, ZDHHC9)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Osteogenesis imperfecta (OI)</b> ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, MBTPS2, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Kraniosynostosen</b> (ALPL, ALX4, CDC45, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT140, IFT43, IL11RA, IMPAD1, MYH3, P4HB, POR, RAB23, RECQL4, SCARF2, SEC24D, SMAD6, TCF12, TWIST1)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |



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

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik   | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|---|--|--|--|---------------------------------------|------------------------|
| <b>Angeborene Herzfehler</b> (ACTC1, ACVR2B, ADAMTS10, ARHGAP31, BMPR2, BRAF, CBL, CFAP53, CHD7, CITED2, CREBBP, CRELD1, DNAH11, DNAH5, DNAI1, DOCK6, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HRAS, JAG1, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, LZTR1, MAP2K1, MAP2K2, MED12, MED13L, MGP, MMP21, MRAS, MYH11, MYH6, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NPHP4, NR2F2, NRAS, NSD1, PITX2, PKD1L1, PPP1CB, PTPN11, RAF1, RBM10, RBPJ, RIT1, RRAS, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SOS1, SOS2, SPRED1, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGFB1, TGFB2, TLL1, ZEB2, ZFPM2, ZIC3) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Arrhythmogene Erkrankungen</b> (ABCC9, ACTC1, ACTN2, AKAP9, ALPK3, ANK2, ANKRD1, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CRYAB, CSRP3, DES, DSC2, DSG2, DSP, FHL1, FHOD3, FLNC, GAA, GLA, GPD1L, HCN4, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PKP2, PLN, PRDM16, PRKAG2, RAF1, RANGRF, RBM20, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLC4A3, SNTA1, TAZ, TCAP, TECRL, TGFB3, TMEM43, TNNC, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V006 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Hämophilie A</b> (Geninversionen int-22h/int-1h)   | EDTA-Blut, DNA aus Blut                | Long Range PCR   | AA-1413-V003   | --                                    | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik                   | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|---|--|---|--|--|------------------------|
| <b>Hämochromatose</b> (HFE-Gen: dbSNP rs1800562, rs1799945, rs1800730)  | EDTA-Blut, DNA aus Blut                | Fluoreszenz-markierte Hybridisierungssonden | AA-1727-V001   | BioRad CFX96   | flexibel               |
| <b>Alpha-1-Antitrypsin-Mangel</b> (SERPINA1-Gen:dbSNP rs17580, rs28929474)  | EDTA-Blut, DNA aus Blut                | Fluoreszenz-markierte Hybridisierungssonden | AA-1727-V001   | BioRad CFX96   | flexibel               |
| <b>Arzneimittelunverträglichkeit von CYP2C9-Substraten</b> (CYP2C9, dbSNP rs1799853, rs1057910)   | EDTA-Blut, DNA aus Blut                | Fluoreszenz-markierte Hybridisierungssonden | AA-1727-V001; DB-0181-V008   | BioRad CFX96   | flexibel               |
| <b>Arzneimittelunverträglichkeit, Cytochrom P-450-bedingte</b> (CYP2D6, CYP2C19, CYP1A2, CYP2B6, CYP2C8, CYP3A4, CYP3A5)  | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung                        | AA-0590-V011; DB-0183-V007, AA-0272-V005; AA-0269-V007; AA-1668-V001 | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Laktoseintoleranz</b> (LCT-Gen: dbSNP rs182549)  | EDTA-Blut, DNA aus Blut                | Fluoreszenz-markierte Hybridisierungssonden | AA-1727-V001   | BioRad CFX96   | flexibel               |
| <b>5 Fu-Toxizität</b> (DPYD-Gen c.[1236G>A; 1129-5923G>C, 483 DPYD-Gen c.[1236G>A;1129-5923G>C 483+18G>A] (Haplotyp B) c.1679T>G c.1905+1G>A (Exon 14 Skipping Mutation) c.2846A>T) | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung                        | AA-1679-V001   | ABI Sequencer 3130XL   | flexibel               |
| <b>5 Fu-Toxizität, Morbus-Meulengracht, Irinotecan-Verträglichkeit</b> (DPYD, UGT1A1)   | EDTA-Blut, DNA; DNA                    | Fluoreszenz-markierte Hybridisierungssonden | AA-1771-V001   | Aria DX RealTime qPCR instrument                               | flexibel               |
| <b>HCV-Therapie</b> ( IL28B dbSNP rs12979860)   | EDTA-Blut, DNA aus Blut                | Fluoreszenz-markierte Hybridisierungssonden | AA-1727-V001   | BioRad CFX96   | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                            | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version                          | Gerät  | Akkreditierungs-status |
|--|--|--|--|--|------------------------|
| <b>Butyrylcholinesterase (BCHE)-Defizienz und postoperative Apnoe</b> (BCHE-Gen) | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001 | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Kongenitaler Laktasemangel</b> (LCT)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1391-V010, AA-1648-V002       | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Arzneimittelunverträglichkeit</b> (NAT2)                                      | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001 | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Azathioprin-Therapie</b> (TPMT)   | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001 | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Multi Drug resistance</b> (ABCB1 (MDR1), dbSNP rs1045642)                     | EDTA-Blut, DNA aus Blut                | Restriktionsanalyse  | AA-0143-V004                                   | Thermocycler   | flexibel               |
| <b>Verträglichkeit catecholaminerger Neurotransmitter</b> (COMT dbSNP rs4680)    | EDTA-Blut, DNA aus Blut                | Restriktionsanalyse  | AA-0143-V004                                   | Thermocycler   | flexibel               |
| <b>Detoxifizierungsstörung</b> (CYP1A1 , dbSNP rs4646903, rs1048943)             | EDTA-Blut, DNA aus Blut                | Restriktionsanalyse  | AA-0143-V004                                   | Thermocycler   | flexibel               |
| <b>Detoxifizierungsstörung</b> (GSTM1, GSTP1, GSTT1 )                            | EDTA-Blut, DNA aus Blut                | Restriktionsanalyse  | AA-0143-V004                                   | Thermocycler   | flexibel               |
| <b>HCV-Therapie</b> (HCV-Therapie ITPA, dbSNP rs1127354, rs7270101)              | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001 | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|--|--|--|--|--|------------------------|
| <b>Statin-Unverträglichkeit, Myopathie unter Hochdosis-Therapie</b> (SLCO1B1, dbSNP rs4149056) | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>SULT1A1-bedingter verzögerten Phase II-Metabolismus</b> (SULT1A1, dbSNP rs9282861)          | EDTA-Blut, DNA aus Blut                | Restriktionsanalyse  | AA-0143-V004   | Thermocycler   | flexibel               |
| <b>Cumarin-und Cumarinderivat-Sensitivität</b> (VKORC1, dbSNP rs9934438, rs28527768)           | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Hypophosphatämie</b> (CLCN5, DMP1, ENPP1, FAM20C, FGF23, PHEX, SLC34A1, SLC34A3, SLC9A3R1)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Hypophosphatasie</b> (ALPL)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Kongenitale Defekte der Glykosylierung (CDG-Syndrome)</b><br>(ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, CAD, CCDC115, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC2) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Mukopolysaccharidosen (MPS)</b> (ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Maligne Hyperthermie</b> (RYR1, CACNA1S)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Hyperoxalurie</b> (AGXT, GRHPR, HOGA1)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|---|--|--|--|---------------------------------------|------------------------|
| <b>Fettstoffwechselstörungen</b> (ABCA1, ANGPTL3, APOA1, APOA5, APOB, APOC2, APOE, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LMF1, LPL, MTP, PCSK9, SAR1B)    | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>MODY-Diabetes (Maturity-Onset Diabetes of the Young)</b> (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Porphyrien</b> (ALAD, CPOX, HMBS, PPOX, ALAS2, FECH, UROD, UROS)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Adipositas, monogene</b> (KSR2, LEP, LEPR, MC3R, MC4R, MRAP2, NTRK2, PCSK1, POMC, SH2B1, SIM1)   | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                                     | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik   | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|---|--|--|--|--|------------------------|
| <b>Sphingolipidosen</b> (GLA, GAA, GM2A, HEXA, HEXB, GALC, PSAP, NPC1, NPC2, SMPD1)       | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| Morbus Gaucher (GBA)  | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005; AA-0269-V007; AA-1668-V001   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| Morbus Gaucher (GBA)  | EDTA-Blut, DNA aus Blut                | (MS) MLPA  | AA-0103-V009   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Harnstoffzyklusdefekte</b> (ARG1, ASL, ASS1, CPS1, NAGS, OTC, SLC25A13, SLC25A15)      | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Störungen der Fettsäure-Oxidation</b> (ACADM, HADHA, HADHB, ACADVL, ETFA, ETFB, ETFDH) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                       | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version  | Gerät   | Akkreditierungs-status |
|---|--|---|--|---|------------------------|
| <b>Fruktose-Intoleranz, hereditäre</b> (ALDOB, FBP1)                        | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Hämochromatose, hereditäre</b> (BMP6, HAMP, HFE, HJV,<br>SLC40A1, TFR2 ) | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Adrenogenitales Syndrom (AGS)</b> (CYP11B1, CYP11B2,<br>CYP17A1, HSD3B2) | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Adrenogenitales Syndrom (AGS)</b> (CYP21A2)                              | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung  | AA-1599-V003;<br>AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001  | ABI Sequencer<br>3730XL; ABI<br>Sequencer 3130XL;<br>ABI Sequencer 3730 | flexibel               |
| <b>Adrenogenitales Syndrom (AGS)</b> (CYP21A2)                              | EDTA-Blut, DNA<br>aus Blut             | (MS) MLPA   | AA-1599-V003,<br>AA-0103-V009  | ABI Sequencer<br>3730XL; ABI<br>Sequencer 3130XL;<br>ABI Sequencer 3730 | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version  | Gerät   | Akkreditierungs-status |
|---|--|---|--|---|------------------------|
| <b>Ahornsirupkrankheit (MSUD)</b> (BCKDHA, BCKDHB, DBT) | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Alkoholintoleranz</b> (ADH1B, ALDH2)                 | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung  | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001   | ABI Sequencer<br>3730XL; ABI<br>Sequencer 3130XL;<br>ABI Sequencer 3730 | flexibel               |
| <b>Alpha-1-Antitrypsin-Mangel</b> (SERPINA1)            | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung  | AA-0272-V005;<br>AA-0269-V007;<br>AA-1668-V001   | ABI Sequencer<br>3730XL; ABI<br>Sequencer 3130XL;<br>ABI Sequencer 3730 | flexibel               |
| <b>Biotinidasemangel</b> (BTD)                          | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Carnitinzyklusdefekte</b> (CPT1A, CPT2, SLC25A20)    | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |

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

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

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

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



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

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

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Pulmonale arterielle Hypertonie (PAH)</b> (ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNA5, KCNK3, SMAD1, SMAD4, SMAD9, TBX4)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Nierenerkrankungen</b> (ACE, ACTA2, ACTG2, ACTN4, AGT,AGTR1, AGXT, AHI1, ANKS6, ANLN, ANOS1, APOL1, ARHGAP24, ARHGDI1, BICC1, BMP4, BMP7, CC2D2A, CD2AP, CDC5L, CEP164, CEP290, CEP83, CFH, CHD1L, CHRM3, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DACH1, DCDC2, DGKE, DSTYK, DZIP1L, EMP2, ETV4, ETV5, EYA1, FGF20, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GANAB, GATA3, GDNF, GLA, GLIS2, REM1, GRHPR, GRIP1, HNF1B, HOGA1, HPSE2, IFT172, INF2, INVS, IQCB1, ITGA3, ITGA8, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, LRIG2, MAPKBP1, MUC1, MYH9, MYO1E, NEIL1, NEK8, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, PAX2, PAX8, PDSS2, PKD1, PKD2, PKHD1, PLCE1, PTPRO, REN, RET, ROBO2, RPGRIP1L, SALL1, SCARB2, SDCCAG8, SIX1, SIX2, SIX5, SLC41A1, SMARCAL1, SOX17, TMEM216, TMEM237, TMEM67, TRAP1, TRPC6, TTC21B, UMOD, UPK2, UPK3A, WDR19, WNT4, WT1, XPNPEP3, ZIC3, ZNF423) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Pankreatitis, chronisch (hereditäre)</b> (CASR, CFTR, CPA1, CTSC, PRSS1, SPINK1)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|---|--|--|--|--|------------------------|
| <b>Schwerhörigkeit/Taubheit</b> (ABCC1, ABHD12, ACTG1, ADCY1, ADGRV1, AIFM1, ATP6V1B1, BDP1, BSND, CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP250, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, CLRN2, COCH, COL11A1, COL11A2, COL4A6, COX1, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ERAL1, ESPN, ESRP1, ESRRB, EYA1, EYA4, FAM189A2, FOXI1, GAB1, GATA3, GIPC3, GJB2,GJB3, GJB6, GPRASP2, GPSM2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MIR182, MIR183, MIR96, MITF, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7, PJVK, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, RNR1, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLITRK6, SMPX, SNAI2, SOX10, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMEM132E, TMIE, TmprSS3, TNC, TPRN, TRIOBP, TRNE, TRNL1, TRNS1, TRNS2, TRRAP, TSPEAR, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Hörverlust, autosomal-rezessiv, nicht-syndromal</b> (GJB2)   | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005; AA-0269-V007; AA-1668-V001   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Hörverlust, autosomal-rezessiv, nicht-syndromal</b> (GJB2, GJB6)   | EDTA-Blut, DNA aus Blut                | (MS) MLPA  | AA-0103-V009   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|---|--|--|--|--|------------------------|
| <b>Ataxien</b> (ABCB7, ABHD12, ADGRG1, AFG3L2, AHI1, AMACR, ANO10, APTX, ARL13B, ARSA, ATCAY, ATG5, ATM, ATP13A2, ATP1A3, ATP8A2, ATXN10, B4GALNT1, BTD, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CC2D2A, CCDC88C, CEP290, CEP41, CHP1, CLCN2, CLN5, CLN6, COA7, COQ8A, CP, CPLANE1, CSPP1, CWF19L1, CYP27A1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, GALC, GBA, GBA2, GCLC, GDAP2, GJB1, GJC2, GOSR2, GRID2, GRM1, INPP5E, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0586, KIF1C, KIF26B, KIF7, MARS2, MICU1, MME, MRE11, MTPAP, NEU1, NKX6-2, NPC1, NPC2, NPHP1, OFD1, OPA1, OPA3, PANK2, PDE10A, PDE6D, PDHX, PDYN, PEX10, PEX2, PIK3R5, PLA2G6, PLD3, PLP1, PMPCA, PNKP, PNPLA6, POC1B, POLG, POLR3A, PRKCG, PRNP, PUM1, RNF216, RPGRIP1L, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC17A5, SLC1A3, SLC9A1, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, THG1L, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRPC3, TTBK2, TTC21B, TTPA, TUBB4A, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR81, WFS1, WWOX, XRCC1, ZNF423) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Ataxien, spinocerebelläre autosomal-dominante (SCA)</b><br>(ATXN 1 und/oder 2,3,7, CACNA1A, TBP)   | EDTA-Blut, DNA aus Blut                | Fragmentlängenanalyse, TP-PCR  | AA-1300-V003   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>Ataxie, Friedreich'sche (FRDA1) (FXN)</b>  | EDTA-Blut, DNA aus Blut                | Fragmentlängenanalyse, TP-PCR; Long-Range PCR                                    | AA-0313-V004, AA-1300-V003   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik   | Anweisung/<br>Version  | Gerät  | Akkreditierungs-status |
|---|--|--|--|--|------------------------|
| <b>Epilepsien</b> (AARS, ACTL6B, ADAM22, ADGRV1, ADRA2B, ALDH7A1, ALG13, AP3B2, ARHGEF15, ARHGEF9, ARV1, ARX, ATP1A2, BRAT1, CACNA1A, CACNA1E, CACNA1H, CACNB4, CAD, CASR, CDK19, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN2, CLCN4, CNPY3, CNTN2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DCX, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EPM2A, FGF12, FOXG1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GLUL, GNAO1, GOSR2, GOT2, GPHN, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HDAC4, HNRNPU, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH5, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MBD5, MDH2, MECP2, MEF2C, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PRDM8, PRICKLE1, PRICKLE2, PRRT2, RANBP2, RANGAP1, RELN, RHOBTB2, RNF13, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SMC1A, SPTAN1, SRPX2, ST3GAL3, STX1B, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc)                          | flexibel               |
| <b>Rett Syndrom (RTT)</b> (MECP2)   | EDTA-Blut, DNA aus Blut                | Sanger-Sequenzierung   | AA-0272-V005; AA-0269-V007; AA-1668-V001   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |
| <b>MECP2-Duplikationsyndrom</b> (MECP2)   | EDTA-Blut, DNA aus Blut                | (MS) MLPA  | AA-0103-V009   | ABI Sequencer 3730XL; ABI Sequencer 3130XL; ABI Sequencer 3730 | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Neuropathien, hereditäre</b> (AARS, ABCD1, ABHD12, AFG3L2, AIFM1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARHGEF10, ARL6IP1, ATL1, ATL3, ATP13A2, ATP1A1, ATP7A, B4GALNT1, BAG3, BICD2, BSCL2, C12orf65, C19orf12, CAPN1, CAPN3, CCT5, COX6A1, CPT1C, CTDP1, CYP2U1, CYP7B1, DCTN1, DDHD1, DDHD2, DES, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DPM3, DSTYK, DYNC1H1, EGR2, ELP1, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GBA2, GDAP1, GJB1, GJC2, GNB4, HARS, HINT1, HK1, HOXD10, HPDL, HSPB1, HSPB3, HSPB8, HSPD1, IBA57, IGHMBP2, INF2, JPH1, KARS, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, L1CAM, LAMA2, LITAF, LMNA, LRSAM1, MAG, MARS, MATR3, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NIPA1, NKX6-2, NT5C2, NTRK1, OPA1, PCYT2, PDK3, PLEKHG5, PLP1, PMP22, PNKP, PNPLA6, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, REEP2, RETREG1, RTN2, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SELENOI, SEPT9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC16A2, SLC25A46, SLC33A1, SLC5A7, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTLC1, SPTLC2, SURF1, SYT2, TDP1, TECPR2, TFG, TRIM2, TRPV4, TTR, TUBB4A, UBAP1, UCHL1, VCP, VPS37A, WASHC5, WNK1, VARS, ZFYV(F26, ZFYV(F27)) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| <b>Charcot-Marie-Tooth-Neuropathien, CMT</b> (ATL1, DNM2, GARS, GDAP1, GJB1, HINT1, IGHMBP2, MFN2, MPZ, NEFL, NGF, PMP22, SH3TC2)  | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

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

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Spastische Paraplegien</b> (AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12orf65, C19orf12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENOI, SLC16A2, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, TUBB4A, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27) | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                 | Akkreditierungs-status |
|--|--|--|--|---------------------------------------|------------------------|
| <b>Entwicklungsstörungen und Komorbiditäten,<br/>Wachstumsstörungen</b> (AARS, ABCC9, ABCD1, ACSL4, ACTB, ACTG1, ADAT3, ADNP, AFF2, AHDC1, AIFM1, AKT3, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMER1, AMPD2, ANK3, ANKLE2, ANKRD11, AP1S2, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASPA, ASPM, ATP6AP2, ATP7A, ATRX, AUTS2, B4GALT1, BCAP31, BCOR, BDNF, BRAF, BRD4, BRWD3, C12orf4, C12orf57, CA8, CACNA1C, CACNG2, CAD, CAMK2A, CAMK2B, CAMK2G, CASK, CBL, CC2D1A, CCDC115, CCDC22, CCND2, CDH15, CDK13, CDK5, CDK5RAP2, CDK6, CDKL5, CDKL5, CDKN1C, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP85L, CHAMP1, CHD4, CHD7, CHD8, CHMP1A, CIC, CIT, CLCN4, CLIC2, CLP1, CLTC, CNKSR2, CNOT3, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COL4A3BP, COLGALT1, COPB2, CRADD, CRBN, CREBBP, CSNK2A1, CTCF, CTNNB1, CUL4B, DBH, DCX, DDC, DDOST, DDX3X, DEAF1, DHCR24, DHCR7, DIS3L2, DKC1, DLG3, DLG4, DNAJC12, DNM1, DNMT3A, DOCK7, DOCK8, DOLK, DONSON, DPAGT1, DPF2, DPM1, DPM2, DPM3, DPP6, DVL1, DVL1, DVL3, DVL3, DYNC1H1, DYRK1A, EBP, EDC3, EED, EEF1A2, EHMT1, EIF2B5, EIF2S3, EIF3F, ELP2, EP300, EPB41L1, EXOSC3, EXOSC8, EXOSC9, EZH2, FANCB, FBXO31, FGD1, FLNA, FMN2, FMR1, FOXG1, FOXG1, FOXP1, FOXP2, FRMPD4, FTSJ1, GABRA1, GALT, GATAD2B, GCDH, GCH1, GDI1, GFAP, GIMAP1, GK, GLI3, GNAI1, GNAO1, GNB1, GPAA1, GPC3, GPKOW, GPSM2, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, HCCS, HCFC1, HCN1, HDAC4, HDAC6, HDAC8, HEPACAM, HERC1, HIST1H1E, | EDTA-Blut, DNA aus Blut                | Sequence Capture (TWIST), Sequencing-by-synthesis, Dragen, Varseq (Golden Helix) | AA-1637-V003, AA-1617-V003, AA-1648-V002, AA-1662-V001, AA-1652-V001, AA-1504-V007, AA-1635-V007 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

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



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

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

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix)                      | Untersuchungstec-<br>hnik   | Anweisung/<br>Version                          | Gerät   | Akkreditierungs-status |
|--|---|---|--|---|------------------------|
| <b>Zieldiagnostik / Bestätigungsanalyse CNV</b>  | EDTA-Blut, DNA<br>aus Blut                                  | (MS) MLPA   | AA-0103-V009                                   | ABI Sequencer<br>3730XL; ABI<br>Sequencer 3130XL;<br>ABI Sequencer 3730 | flexibel               |
| <b>Whole Exome Sequencing</b>  | EDTA-Blut, DNA<br>aus Blut,<br>Chorionzotten,<br>Amniozyten | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1654-0002,<br>AA-1637-V003,<br>AA-1617-V003 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>mTORopathien</b> (AKT1, AKT1S1, AKT3, CCND2, DEPDC5, DEPTOR, DOCK7, G3BP1, G3BP2, MLST8, MTOR, NPRL2, NPRL3, PAK2, PIK3CA, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PTEN, RICTOR, RPTOR, STK11, STRADA, TBC1D7, TSC1, TSC2 ) | EDTA-Blut, DNA<br>aus Blut                                  | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1637-V003,<br>AA-1391-V010,<br>AA-1648-V002 | Illumina Series<br>(NovaSeq, Miseq,<br>etc)                             | flexibel               |
| <b>Prader-Willi-Syndrom/Angelman-Syndrom</b> (Deletionen, Duplikationen, Methylierung in Chromosomenregion 15q11.2-q13)  | DNA aus Blut,<br>extrahierte DNA                            | (MS) MLPA   | AA-1756-V001                                   | Thermocycler; ABI<br>Sequencer 3730XL;<br>ABI Sequencer 3730            | flexibel               |
| Pankreas Elastase  | Stuhl   | ELISA   | AA-1590-V001                                   | Dynex DSX   | flexibel               |
| Calprotectin   | Stuhl   | ELISA   | AA-1598-V001                                   | Dynex DSX   | flexibel               |
| Staphylokokken<br>(Koagulase, clumping factor, Protein A, Polysaccharide)  | Bakterienkultur   | Partikelagglutination   | AA-1478-V002                                   |   | flexibel               |
| Streptokokken<br>(Lancefield-Antigen)  | Bakterienkultur   | Partikelagglutination   | AA-1452-V002                                   |   | flexibel               |
| Bakterien (Resistenztestung)   | Keimkolonien in<br>Reinkultur                               | Agardiffusionstest  | AA-1473-V005                                   | manuell   | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version      | Gerät              | Akkreditierungs-status |
|---|--|---|----------------------------|--------------------|------------------------|
| Bakterien (Resistenztestung)                          | Keimkolonien in Reinkultur             | Bouillondilutionsverfahren als minimale Hemmkonzentration (MHK)/Break-Point, vollmechanisiert | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| Bakterien (Resistenztestung)                          | Keimkolonien in Reinkultur             | trägergebundener Gradientendiffusionstest   | AA-1473-V005               | manuell            | flexibel               |
| Hefen   | Pilzisolat                             | biochemisch, aufwendig  | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| Anaerobier, Corynebakterien                           | Bakterienisolat                        | biochemisch, aufwendig  | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| gram-negative aerobe Bakterien                        | Bakterienisolat                        | biochemisch, aufwendig  | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| gram-positive aerobe Bakterien                        | Bakterienisolat                        | biochemisch, aufwendig  | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| Neisseria sp., Haemophilus sp.                        | Bakterienisolat                        | biochemisch, aufwendig  | AA-1518-V006               | Vitek2, Biomerieux | flexibel               |
| Bakterien (Orientierungs-/Differenzierungsteste)      | Keimkolonien in Reinkultur             | biochemisch, orientierend (Katalase, Oxidase, Nitrocefin (Beta-Lactamase))                    | AA-1449-V003, AA-1455-V008 |                    | flexibel               |
| Pneumokokken  | Keimkolonien in Reinkultur             | biochemisch, orientierend (Optochin)  | AA-1455-V008               |                    | flexibel               |
| Staphylococcus saprophyticus                          | Keimkolonien in Reinkultur             | biochemisch, orientierend (Novobiocin)  | AA-1472-V008               |                    | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                                     | Untersuchungs-<br>material<br>(Matrix)                                     | Untersuchungstec-<br>hnik  | Anweisung/<br>Version   | Gerät                                   | Akkreditierungs-status |
|---|--|--|---|---|------------------------|
| Bakterien, Pilze (Anlage und ablesen)   | Abstrich<br>(urogenital,<br>HNO), Blut,<br>Haut, Wunde,<br>Punktat, Stuhl, | in CO2-<br>Atmosphäre,<br>mikroaerobe/anae-<br>robe Atmosphäre,<br>spezifisch,<br>unspezifisch | AA-1482-V006,<br>AA-1490-V005,<br>AA-1531-V004,<br>AA-1717-V002,<br>AA-1537-V004,<br>AA-1565-V002 |   | flexibel               |
| Bakterien, Pilze  | Urin   | spezifisch,<br>unspezifisch,<br>Keimzahlbestimm-<br>ung  | AA-1472-V008  |   | flexibel               |
| Multiresistente gramnegative Bakterien  | Abstrich (rektal),<br>Wunde, Stuhl,<br>Urin                                | spezifisch   | AA-1765-V002  |   | flexibel               |
| Chlamydia trachomatis IgG/IgA   | Serum  | ELISA  | AA-1437-V013  | manuell<br>(EuroImmuno)                 | flexibel               |
| Chlamydia trachomatis IgG/IgA   | Serum  | ELISA  | AA-1437-V013  | Euroimmun<br>Analyzer I<br>(EuroImmuno) | flexibel               |
| Toxoplasma gondii, IgG, IgM   | Serum  | CLIA   | AA-1401-V0011   | Cobas e411                              | flexibel               |
| Treponema pallidum Infektion assoziierte, nichtspezifische<br>Lipoidantikörper (IgG, IgM) | Serum, EDTA-<br>Plasma   | Partikelagglutinati-<br>on   | AA-1673-V001  | Kartenschüttler,<br>Biorad              | flexibel               |
| Treponema pallidum Antikörper   | Serum, EDTA-<br>Plasma   | ELISA  | AA-1437-V013  | manuell<br>(EuroImmuno)                 | flexibel               |
| Treponema pallidum Antikörper   | Serum, EDTA-<br>Plasma   | ELISA  | AA-1437-V013  | Euroimmun<br>Analyzer I<br>(EuroImmuno) | flexibel               |
| Treponema pallidum, Ig (Lues-Suchtest)  | Serum  | ECLIA  | AA-1401-V011,<br>AA-1487-V006   | Cobas pro                               | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik                                   | Anweisung/<br>Version      | Gerät                 | Akkreditierungs-status |
|--|--|--|----------------------------|-----------------------|------------------------|
| Borrelia burgdorferi sensu lato IgG, IgM   | Serum, EDTA-Plasma                     | CLIA   | AA-1624-V005               | LIAISON XL / DiaSorin | flexibel               |
| Diphtherie-Toxoid, IgG   | Serum, EDTA-Plasma                     | ELISA  | AA-1437-V013               | manuell (EuroImmuno)  | flexibel               |
| Yersinien Antikörper IgA/IgG/IgM   | Serum                                  | Immunoblot   | AA-1538-V004               | manuell (Mikrogen)    | flexibel               |
| Borrelia burgdorferi sensu lato IgG, IgM   | Serum, EDTA-Plasma                     | Immunoblot   | AA-1538-V002               | manuell (Mikrogen)    | flexibel               |
| Bakterien, Pilze   | Abstrich                               | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-1444-V005, AA-1508-V004 | Mikroskop             | flexibel               |
| Schimmelpilze  | Pilzkultur                             | Hellfeldmikroskopie nach Anfärbung mittels Farbstoffen | AA-1467-V004               | Mikroskop             | flexibel               |
| Bordetella pertussis, Bordetella parapertussis, Bordetella holmesii, DNA   | Abstrich, Sputum                       | Real-Time PCR  | AA-1511-V004               | CFX Opus Biorad       | flexibel               |
| Chlamydia trachomatis, Neisseria gonorrhoeae, DNA  | Abstrich, Urin                         | Real-Time PCR  | AA-1458-V008               | CFX Opus Biorad       | flexibel               |
| Chlamydia pneumoniae, Mycoplasma pneumoniae, DNA   | Abstrich, Sputum                       | Real-time PCR  | AA-1523-V006               | CFX Opus Biorad       | flexibel               |
| Chlamydia trachomatis, Mycoplasma genitalium, Neisseria gonorrhoeae, Trichomonas vaginalis (STIs, sexually transmitted infections)                   | Abstrich, Urin, Ejakulat               | Real-time PCR  | AA-1659-V004               | Alinity m, Abbott     | flexibel               |
| Chlamydia trachomatis, Mycoplasma hominis, genitalium, Neisseria gonorrhoeae, Ureaplasma urealyticum, parvum (STIs, sexually transmitted infections) | Abstrich, Urin, Ejakulat               | Real-time PCR  | AA-1458-V008               | CFX Opus Biorad       | flexibel               |
| Cryptosporidien Antigennachweis  | Stuhl                                  | ELISA  | AA-1563-V007               | r-biopharm DSX        | flexibel               |
| Entamoeba histolytica/dispar Antigennachweis   | Stuhl                                  | ELISA  | AA-1563-V007               | r-biopharm DSX        | flexibel               |
| Giardia lamblia Antigennachweis  | Stuhl                                  | ELISA  | AA-1563-V007               | r-biopharm DSX        | flexibel               |
| Treponema pallidum IgM   | Serum, EDTA-Plasma                     | Immunoblot   | AA-1583-V004               | manuell (Mikrogen)    | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)     | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik | Anweisung/<br>Version         | Gerät                            | Akkreditierungs-status |
|---|--|---------------------------|-------------------------------|----------------------------------|------------------------|
| Chlamydia trachomatis, psittaci, pneumoniae IgA, IgG, IgM | Serum, EDTA-<br>Plasma                 | Immunoblot                | AA-1583-V004                  | manuell (Mikrogen)               | flexibel               |
| Helicobacter pylori Antigennachweis                       | Stuhl                                  | ELISA                     | AA-1563-V007                  | r-biopharm DSX                   | flexibel               |
| Tetanus-Toxoid IgG  | Serum, EDTA-<br>Plasma                 | ELISA                     | AA-1437-V013                  | manuell<br>(EuroImmuno)          | flexibel               |
| Interferon-Gamma Release-Assay                            | EDTA-Plasma                            | CLIA                      | AA-1624-V005                  | LIAISON XL /<br>DiaSorin         | flexibel               |
| Clostridium difficile Toxin A/B-Antig.                    | DHT                                    | ELISA                     | AA-1558-V002                  | Dynex DSX                        | flexibel               |
| Cytomegalievirus, IgG, IgM                                | Serum, EDTA-<br>Plasma                 | ELISA/CMIA                | AA-1401-V009,<br>AA-1437-V010 | Cobas e411 /<br>Roche, Dynex DSX | flexibel               |
| Epstein-Barr-Virus, VCA, EA, EBNA, IgG, IgM               | Serum, EDTA-<br>Plasma                 | ECLIA                     | AA-1624-V002                  | LIAISON XL /<br>DiaSorin         | flexibel               |
| Epstein-Barr-Virus  | Serum, EDTA-<br>Plasma                 | Immunoblot                | AA-1538-V004                  | RemcomScan                       | flexibel               |
| Hepatitis-A-Virus, Ig                                     | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 / Roche               | flexibel               |
| Hepatitis-A-Virus, IgM                                    | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 / Roche               | flexibel               |
| Hepatitis-B-Virus, Anti-HBc Ig                            | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-B-Virus, Anti-HBc IgM                           | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-B-Virus, Anti-HBe Ig                            | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-B-Virus, Anti-HBs Ig                            | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-B-Virus, HBs-Antigen, qualitativ                | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-B-Virus, HBs-Antigen, quantitativ               | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-C-Virus, Ig                                     | Serum, EDTA-<br>Plasma                 | CMIA                      | AA-1401-V009                  | Cobas e411 /<br>Roche; cobas pro | flexibel               |
| Hepatitis-C-Virus, Ig                                     | Serum, EDTA-<br>Plasma                 | Immunoblot                | AA-1399-V004                  | manuell (Mikrogen)               | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                 | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik          | Anweisung/<br>Version                    | Gerät                                | Akkreditierungs-status |
|---|--|-------------------------------|--|--------------------------------------|------------------------|
| Hepatitis-D-Virus, Ig   | Serum, EDTA-Plasma                     | ELISA                         | AA-1398-V007                             | manuell (Kit DiaSorin)               | flexibel               |
| Hepatitis-E-Virus, IgG, IGM   | Serum, EDTA-Plasma                     | ELISA                         | AA-1398-V007                             | r-biopharm DSX                       | flexibel               |
| Hepatitis-B-Virus, HBe-Antigen  | Serum, EDTA-Plasma                     | CMIA                          | AA-1401-V009                             | Cobas e411 / Roche; cobas pro        | flexibel               |
| Herpes-Simplex-Virus, IgG, IgM  | Serum, EDTA-Plasma                     | ELISA                         | AA-1437-V010                             | manuell (EuroImmuno)                 | flexibel               |
| Humanes Immundefizienzvirus, Antigen + Antikörper                     | Serum, EDTA-Plasma                     | CMIA                          | AA-1401-V009                             | Cobas e411 / Roche                   | flexibel               |
| Humanes Immundefizienzvirus, Antikörper                               | Serum, EDTA-Plasma                     | Immunoblot                    | AA-1399-V004                             | manuell (Mikrogen)                   | flexibel               |
| Masern IgG/IgM  | Serum, EDTA-Plasma                     | ELISA                         | AA-1437-V010                             | manuell (EuroImmuno)                 | flexibel               |
| Mumps IgG/IgM   | Serum, EDTA-Plasma                     | ELISA                         | AA-1437-V010                             | manuell (EuroImmuno)                 | flexibel               |
| Parvovirus B19, IgG, IgM  | Serum, EDTA-Plasma                     | ELISA                         | AA-1437-V010                             | manuell (EuroImmuno)                 | flexibel               |
| Rötelnvirus, IgG, IgM   | Serum, EDTA-Plasma                     | CMIA                          | AA-1401-V009                             | Cobas e411 / Roche                   | flexibel               |
| Varizella Zoster-Virus, IgG, IgM                                      | Serum, EDTA-Plasma                     | ELISA                         | AA-1437-V010                             | manuell (EuroImmuno)                 | flexibel               |
| Influenzaviren, Respiratory-Syncytial-Virus, SARS-CoV-2 (nur Alinity) | Abstrich                               | Real-time PCR (Oligoplex-PCR) | AA-1659-V004, AA-1523-V003,              | Alinity m, Abbott; CFX Opus, Biorad  | flexibel               |
| Hepatitis-B-Virus, DNA, quantitativ                                   | Serum, EDTA-Plasma                     | Real-time PCR                 | AA-1400-V006, AA-1602-V003, AA-1659-V001 | Alinity m, Abbott; cobas 6800, Roche | flexibel               |
| Hepatitis-C-Virus, RNA, Genotyp                                       | Serum, EDTA-Plasma                     | Real-time PCR                 | AA-1410-V002                             | Alinity m, Abbott                    | flexibel               |
| Hepatitis-C-Virus, RNA, Genotyp Core                                  | Serum, EDTA-Plasma                     | Real-time PCR                 | AA-1428-V006                             | Biometra-Thermocycler                | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix)   | Untersuchungstec-<br>hnik | Anweisung/<br>Version                          | Gerät  | Akkreditierungs-status |
|---|--|---------------------------|--|--|------------------------|
| Hepatitis-C-Virus, RNA, quantitativ                   | Serum, EDTA-<br>Plasma   | Real-time PCR             | AA-1400-V006,<br>AA-1602-V003,<br>AA-1659-V001 | Alinity m, Abbott;<br>cobas pro, Roche                             | flexibel               |
| Humane Papillomaviren, qualitativ                     | Abstrich   | Real-time PCR             | AA-1629-V001,<br>AA-1659-V001                  | Alinity m, Abbott  | flexibel               |
| Humans Immundefizienzvirus, RNA, quantitativ          | Serum, EDTA-<br>Plasma   | Real-time PCR             | AA-1400-V006,<br>AA-1602-V003,<br>AA-1659-V001 | Alinity m, Abbott;<br>cobas 6800, Roche                            | flexibel               |
| Parvovirus B19 DNA                                    | Biopsat, EDTA-<br>Plasma   | Real-time PCR             | AA-1570-V003                                   | Rotor-Gene,<br>Qiagen; CFX Opus,<br>Biorad                         | flexibel               |
| SARS-CoV-2 IgG quantitativ                            | Serum, EDTA-<br>Plasma   | CLIA                      | AA-1624-V005                                   | Liaison XL   | flexibel               |
| HDV-RNA (quantitativ) (HDV-RNA (quantitativ))         | Serum, EDTA-<br>Plasma   | Real-time PCR             | AA-1693-V002                                   | CFX Opus Biorad  | flexibel               |
| HEV-RNA (HEV-RNA)                                     | Serum, EDTA-<br>Plasma   | Real-time PCR             | AA-1692-V002                                   | CFX Opus Biorad  | flexibel               |
| SARS-CoV-2  | Abstrich (cobas<br>6800: nur<br>Nasopharyngeal-<br>Abstrich),<br>Rachenspülung | Real-time PCR             | AA-1659-V001,<br>AA-1602-V004                  | Alinity m, Abbott;<br>cobas 6800, Roche,<br>BioRad<br>CFX384,CFX96 | flexibel               |
| Herpes-Simplex-Virus-1 und 2 DNA                      | Abstrich<br>(urogenital)   | Real-time PCR             | AA-1458-V005                                   | CFX Opus Bio-Rad   | flexibel               |
| Adenoviren, DNA, Enteroviren, Rhinoviren, RNA         | Abstrich   | Real-time PCR             | AA-1523-V004                                   | CFX Opus Bio-Rad   | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix)  | Untersuchungstec-<br>hnik  | Anweisung/<br>Version                                   | Gerät                                 | Akkreditierungs-status |
|---|---|--|---|---------------------------------------|------------------------|
| ABL1  | EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark (cDNA wird analysiert) | Amplikon-basiertes NGS, Sequencing-by-synthesis, Dragen, JSI medical systems SeqNext                       | AA-1617-V003, AA-1729-V001, AA-1709-V002                | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Glioblastom (MGMT), MGMT-Promotormethylierung         | DNA aus FFPE-Gewebe, DNA aus Tumorgewebe  | Sanger-Sequenzierung; Amplikon-basiertes NGS, Sequencing-by-synthesis, Dragen, JSI medical systems SeqNext | AA-1461-V008, AA-1617-V003, AA-1729-V001, AA-1709-V002  | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| POLE  | DNA aus FFPE-Gewebe, DNA aus Tumorgewebe  | Sanger-Sequenzierung; Amplikon-basiertes NGS, Sequencing-by-synthesis, Dragen, JSI medical systems SeqNext | -AA-1461-V008, AA-1617-V003, AA-1729-V001, AA-1709-V002 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)         | Untersuchungstec-<br>hnik  | Anweisung/<br>Version                    | Gerät                                 | Akkreditierungs-status |
|---|--|--|--|---------------------------------------|------------------------|
| Fusionen bei soliden Tumoren (NTRK3-ETV6, EWSR1-NR4A3, EWSR1-PBX1, EWSR1-ZNF384, EWSR1-ATF1, EWSR1-PATZ1, EWSR1-DDIT3, EWSR1-SP3, EWSR1-FEV, EWSR1-CREB1, EWSR1-FLI1, EWSR1-ETV4, EWSR1-ETV1, EWSR1-ERG, YY1-EWSR1, EWSR1-ZNF444, EWSR1-SMARCA5, NFATC2-EWSR1, SS18-SSX1, SS18-SSX4, FUS-CREB3L2, FUS-CREB3L1, FUS-DDIT3, FUS-ERG, FUS-ATF1, FUS-FEV) | RNA aus FFPE-Gewebe, RNA aus Tumorgewebe, cDNA | Amplikon-basiertes NGS, Sequencing-by-synthesis, Dragen, JSI medical Systems Seqnext | AA-1463-V004, AA-1733-V002, AA-1617-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)   | Untersuchungstec-<br>hnik  | Anweisung/<br>Version                    | Gerät                                 | Akkreditierungs-status |
|---|--|--|--|---------------------------------------|------------------------|
| Fusion bei Leukämien/Lymphomen (ACTN4-MLL, BCR-ABL1, CBFβ-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-PDSSA, MLL-PICALM, MLL-SEPT11, MLL-SEPT2, MLL-SEPT5, MLL-SEPT6, MLL-SEPT9, MLL-SH3GL1, MLL-SORBS2, MLL-TET1, MLL-TOP3A, MLL-ZFYVE19, MN1-ETV6, NPM1-ALK, NPM1-MLF1, NPM1-RARA, NUP98-MLL, PAX5-PML, PML-RARA, RPN1-MECOM, RUNX1-MECOM, RUNX1-RUNX1T1, SET-NUP214, STIL-TAL1, TCF3-HLF, TCF3-PBX1) (Fusion AML-ALL- (BCR), CBFβ, DEK, ETV6, FUS, MLL, NPM1, PAX5, PML, MECOM, RUNX1, SET, SIL, TCF3) Fusion bei Leukämien/Lymphomen (ACTN4-MLL, BCR-ABL1, CBFβ-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- | EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA | Amplikon-basiertes NGS, Sequencing-by-synthesis, Dragen, JSI medical Systems Seqnext | AA-1463-V004, AA-1733-V002, AA-1617-V003 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)   | Untersuchungstechnik   | Anweisung/<br>Version                                  | Gerät  | Akkreditierungs-status |
|---|--|--|--|--|------------------------|
| Chimärismus (Chimärismusanalyse)                        | EDTA-Blut, EDTA-Knochenmark, Heparin-Blut, Heparin-Knochenmark, RNA aus Blut und Knochenmark, cDNA | Fluoreszenz-markierte Hybridisierungssonden (ddPCR), Fragmentlängenanalyse   | AA-1541-V003   | ABI Sequencer 3730XL, 3130XL, 3730, QX200 Droplet Digital PCR System (BioRad), QuantStudio 7 Pro | flexibel               |
| Phäochromozytom/Paragangliom (MAX)                      | EDTA-Blut, DNA aus Blut  | (MS) MLPA  | AA-0103-V008   | ABI Sequencer 3730XL, 3130XL, 3730   | flexibel               |
| Mikrosatelliteninstabilität (MSI-Analyse)               | DNA aus FFPE-Gewebe, DNA aus Tumorgewebe   | Fragmentlängenanalyse  | AA-0218, AA-0269                                       | Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730, Agarosegelelektrophoresenkammer                | flexibel               |
| Lynch-Syndrom (HNPCC), z.A. (MLH1-Promotormethylierung) | DNA aus FFPE-Gewebe  | Amplikon-basiertes NGS, Sequencing-by-synthesis, JSI medical systems SeqNext | AA-0229-V008, AA-1617-V003, AA-1615-V001, AA-1376-V004 | Illumina Series (NovaSeq, Miseq, etc)  | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version   | Gerät                                       | Akkreditierungs-status |
|---|--|---|---|---|------------------------|
| Familiäres Mamma-/Ovarialkarzinomsyndrom (HBOC)<br>(ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2,<br>PTEN, RAD51C, RAD51D, STK11, TP53)            | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Endokrinologische Tumorerkrankungen<br>(endokrinologische Neoplasien) (AIP, AP2S1, CASR, CDC73,<br>CDKN1B, GCM2, GNA11, GNAS, MEN1, PRKAR1A, PTH,<br>RET) | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version   | Gerät                                       | Akkreditierungs-status |
|--|--|---|---|---|------------------------|
| Familiäres Paragangliom-/Phäochromozytomsyndrom<br>(PCC/PGL) (MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD,<br>TMEM127, VHL) | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Hereditäre Nierenzellkarzinome (BAP1, FH, FLCN, MET,<br>PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, VHL)                       | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version   | Gerät                                 | Akkreditierungs-status |
|--|--|--|---|---------------------------------------|------------------------|
| Hämatologische Neoplasien mit Keimbahnprädisposition (ABCB7, ACD, ADH5, ALAS2, ALDH2, ANKRD26, ATG2B, ATM, BLM, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CSF3R, CTC1, DDX41, DKC1, DNAJC21, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, GLRX5, GSKIP, HAX1, HSPA9, JAGN1, KRAS, LIG4, MAD2L2, MBD4, MDM4, MECOM, MLH1, MSH2, MSH6, MYSM1, NBN, NF1, NHP2, NOP10, PALB2, PARN, PMS2, PTPN11, RAD50, RAD51, RAD51C, RFDWD3, RPL11, RPL15, RPL18, RPL23, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLC19A2, SLC25A38, SLX4, SRP54, SRP72, TERC, TERT, THPO, TINF2, TOP3A, TP53, TRNT1, TSR2, UBE2T, VPS45, WAS, WRAP53, WRN, XPC, XRCC2) | EDTA-Blut, DNA aus Blut                | Sequence capture (TWIST)<br>Sequencing-by-synthesis, Dragen, VarSeq (Golden Helix);<br><br>Einzelgensequenzierung, Amplikon-basiertes NGS, Sequencing-by-synthesis, JSI medical systems SeqNext) | AA-1476 AA-1637, AA-1617, AA-1648, AA-1662, AA-1652, AA-1504, AA-1635 | Illumina Series (NovaSeq, Miseq, etc) | flexibel               |
| Hereditäre Tumorsyndrome (AIP, APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDKN1B, CDKN2A, CDKN2B, CHEK1, CHEK2, DICER1, EPCAM, FH, FLCN, GATA2, GNAS, GREM1, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RB1, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, STK11, TMEM127, TP53, VHL)   | EDTA-Blut, DNA aus Blut                | (MS) MLPA  | AA-0103   | ABI Sequencer 3730XL, 3130XL, 3730    | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)    | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik   | Anweisung/<br>Version   | Gerät                                       | Akkreditierungs-status |
|--|--|---|---|---|------------------------|
| BRCA-Diagnostik, Therapie PARP-Inhibitor (BRCA1, BRCA2)  | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Lynch-Syndrom (HNPCC) (MLH1, MSH2, MSH6, PMS2,<br>EPCAM) | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST)<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix);<br><br>Einzelgensequenzi-<br>erung, Amplikon-<br>basiertes NGS,<br>Sequencing-by<br>synthesis, JSI<br>medical systems | AA-1476 AA-<br>1637, AA-1617,<br>AA-1648, AA-<br>1662, AA-1652,<br>AA-1504, AA-<br>1635 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |

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

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)  | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstec-<br>hnik  | Anweisung/<br>Version  | Gerät                                       | Akkreditierungs-status |
|--|--|--|--|---|------------------------|
| Hereditäre Alpha-Tryptasämie (TPSAB1)  | EDTA-Blut                              | Fluoreszenz-<br>markierte<br>Hybridisierungsso-<br>nden;<br>Chimärismusanaly-<br>se;<br>Fragmentlängenana-<br>lyse | AA-1433-V006,<br>AA-1541-V003  | QX200 Droplet<br>Generator                  | flexibel               |
| Hypogonadotroper Hypogonadismus / Kallmann-Syndrom<br>(ANOS1, CHD7, DUSP6, FEZF1, FGF8, FGF17, FGFR1, FLRT3,<br>FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R,<br>LHB, NSMF, PROK2, PROKR2, SEMA3A, SOX10, SPRY4,<br>TAC3, TACR3, WDR11) | EDTA-Blut, DNA<br>aus Blut             | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix)                    | AA-1637-V003,<br>AA-1617-V003,<br>AA-1648-V002,<br>AA-1662-V001,<br>AA-1652-V001,<br>AA-1504-V007,<br>AA-1635-V007 | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Ovarialdysgenese (BMP15, FSHR, MCM9, NR5A1,<br>PSMC3IP)  | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix)                    | AA-1637-V002,<br>AA-1617-V003  | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Vorzeitige Ovarialinsuffizienz (BMP15, DIAPH2, ESR1,<br>FIGLA, FOXL2, FSHR, GDF9, INHA, LHCGR, NOBOX, NR5A1,<br>SOHLH1, SOHLH2, STAG3)   | EDTA-Blut, DNA<br>aus Blut             | Sequence capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>VarSeq (Golden<br>Helix)                    | AA-1637-V002,<br>AA-1617-V003  | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| Hypogonadotroper<br>Hypogonadismus 1 mit oder<br>ohne Anosmie (ANOS1-Gen)  | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung   | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005   | ABI Sequencer<br>3730XL, 3130XL,<br>3730    | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)                      | Untersuchungs-<br>material<br>(Matrix) | Untersuchungstechnik     | Anweisung/<br>Version  | Gerät                                    | Akkreditierungs-status |
|--|--|--------------------------|--|--|------------------------|
| Androgeninsensitivität (AIS) (AR-Gen)                                      | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                   | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Ovarialdysgenese Typ 2 /POI (BMP15-Gen)                                    | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                   | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Hypogonadotroper<br>Hypogonadismus 6 mit oder<br>ohne Anosmie (FGF8-Gen)   | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                   | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Hypogonadotroper Hypogonadismus 2 mit<br>oder ohne Anosmie (FGFR1-Gen)     | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                   | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| FISH-Rezeptor-Defizienz / Ovarialdysgenese Typ 1 /POI<br>(FSHR-Gen)        | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | AA-0197-V007,<br>AA-1313-V007,<br>AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005 | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Hypogonadotroper<br>Hypogonadismus 4 mit oder<br>ohne Anosmie (PROK2-Gen)  | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | -AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                  | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Hypogonadotroper<br>Hypogonadismus 3 mit oder<br>ohne Anosmie (PROKR2-Gen) | EDTA-Blut, DNA<br>aus Blut             | Sanger-<br>Sequenzierung | -AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005                                  | ABI Sequencer<br>3730XL, 3130XL,<br>3730 | flexibel               |
| Azoospermie (AZF-Mikrodeletionen)  | EDTA-Blut, DNA<br>aus Blut             | Fragmentlängenanalyse    | AA-0272-V005,<br>AA-0284-V008  | Thermocycler                             | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)  | Untersuchungstec-<br>hnik  | Anweisung/<br>Version                          | Gerät                                       | Akkreditierungs-status |
|---|---|--|--|---|------------------------|
| Ovariell<br>Hyperstimulationssyndrom (OHSS) (FSHR-Gen: dbSNP rs6166)  | EDTA-Blut, DNA<br>aus Blut  | Sanger-<br>Sequenzierung   | AA-1668-V001,<br>AA-0269-V007,<br>AA-0272-V005 | ABI Sequencer<br>3730XL, 3130XL,<br>3730    | flexibel               |
| V. a. Fertilitätsstörung, wiederholte Fehlgeburten (ANXA5-M2 Genotyp: dbSNP rs112782763, rs28717001, rs28651243, rs113588187)   | EDTA-Blut, DNA<br>aus Blut  | Sanger-<br>Sequenzierung   | AA-1668-V001,<br>AA-0272-V005,<br>AA-0269-V007 | ABI Sequencer<br>3730XL, 3130XL,<br>3730    | flexibel               |
| PGT-A (Aneuploidiediagnostik), Chromosomensatz (zur Abklärung einer de novo Chromosomenveränderung)                             | Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD, Genomische DNA aus Einzelzellen | NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA, Sureplex DNA Amplification System), Bluefuse Multi Software | AA-1585-V003,<br>AA-1700-V001                  | Illumina Series<br>(NovaSeq, Miseq,<br>etc) | flexibel               |
| PGT-SR (Translokationsdiagnostik), partieller Chromosomensatz (zur Abklärung einer bekannten familiären Chromosomenveränderung) | Trophektodermzellen im Rahmen einer PID und/oder Polkörper im Rahmen einer PKD, Genomische DNA aus Einzelzellen | NGS (Sequencing-by-synthesis) nach gesamtgenomischer Amplifikation (WGA, Sureplex DNA Amplification System), Bluefuse Multi Software | AA-1585-V003,<br>AA-1700-V001                  |   | flexibel               |



| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)  | Untersuchungstec-<br>hnik                                   | Anweisung/<br>Version   | Gerät  | Akkreditierungs-status |
|---|---|---|---|--|------------------------|
| Hohes Risiko einer schwerwiegenden Erbkrankheit für die Nachkommenschaft (PGT-M)  | Genomische DNA aus Einzelzellen, Polkörper, Trophektodermzellen                                   | Fragmentlängenanalyse                                       | AA-1378-V007  | Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Aneuploidiediagnostik (nicht invasiver Pränataltest): Trisomie 21, Trisomie 18, Trisomie 13, gonosomale Aberrationen, Mikrodeletionen | BCT-Blut (Streck), zellfreie (fetale&maternale) DNA aus Blut                                      | Gesamtgenomsequenzierung, Sequencing-by-synthesis, Dragen   | AA-1640-V001, AA-1641-V001, AA-1642-V001, AA-1643-V004, AA-1644-V001, | Illumina Series (NovaSeq, Miseq, etc)            | flexibel               |
| Kallmann-Syndrom (FGFR1-, GNRHR-, KISS1R-, GNRH1-, NELF-, PROK2-, PROKR2-Gen)   | EDTA-Blut, DNA aus Blut   | (MS) MLPA   | AA-0103-V008  | Thermocycler, ABI Sequencer 3730XL, 3130XL, 3730 | flexibel               |
| Segregationsanalyse, CNV-Bestätigung/-Ausschluss, STR-CNV-Analyse (Genomische Imbalancen)   | EDTA-Blut, Genomische DNA aus Blut, natives Abortgewebe / kultivierte Zellen aus Fruchtwasser und | Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR) | AA- 1420-V006   | LC480II, LC1.2 (Roche), CFX96/384Touch (BioRad)  | flexibel               |
| Uniparentale Disomie 14 (UPD 14)  | genomische DNA  | Mikrosatellitenanalyse (Fragmentanalyse STR)                | AA-1526-V002  | Thermocycler, ABI Sequencer 3730                 | flexibel               |
| Uniparentale Disomie 15 (UPD 15)  | genomische DNA  | Mikrosatellitenanalyse (Fragmentanalyse STR)                | AA-1526-V002  | Thermocycler, ABI Sequencer 3730                 | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n)   | Untersuchungs-<br>material<br>(Matrix)  | Untersuchungstechnik  | Anweisung/<br>Version                          | Gerät  | Akkreditierungs-status |
|---|---|---|--|--|------------------------|
| angeborener Chromosomensatz (Array-CGH)                 | Genomische DNA aus Blut sowie aus nativen Zellen und Zellkultur von Zellen aus Fruchtwasser, Chorionzotten oder | Array basierte CGH  | AA-0351-V014                                   | SureScan<br>Microarray Scanner<br>G2505C (Agilent) | flexibel               |
| angeborener Chromosomensatz (Chromosomenanalyse & FISH) | Peripheres Blut, Fruchtwasser, Chorionzotten, Abortgewebe, Nabelschnurblut, Haut, Knochenmark                   | Chromosomenbänderungsanalyse, Fluoreszenz in situ Hybridisierung (FISH): Pränataler Schnelltest, Mikrodeletionsdiagnostik, Chromosomenpainting, Subtelomeranalysen, Interphase-Untersuchungen, Vielfarbenkaryotypisierung | AA-0335-V013,<br>AA-0356-V010,<br>AA-1390-V005 |  | flexibel               |

| Analyt / Indikation<br>(Messgröße; Gen/e, Variante/n) | Untersuchungs-<br>material<br>(Matrix)   | Untersuchungstechnik  | Anweisung/<br>Version   | Gerät              | Akkreditierungs-status |
|---|--|---|---|--------------------|------------------------|
| angeborener Chromosomensatz                           | Genomische<br>DNA aus Blut<br>sowie aus<br>Zellkultur von<br>Zellen aus<br>Fruchtwasser<br>oder<br>Chorionzotten | Molekulare<br>Karyotypisierung  | AA-1651-V003  | Bionano Saphyr     | flexibel               |
| Fazioskapulohumerale Muskeldystrophie (FSHD1)         | EDTA-Blut; DNA   | Molekulare<br>Karyotypisierung  | AA-1651-V003  | Bionano Saphyr     | flexibel               |
| <b>Hämophilie</b> (F8, F9)                            | EDTA-Blut, DNA<br>aus Blut   | Sequence Capture<br>(TWIST),<br>Sequencing-by<br>synthesis, Dragen,<br>Varseq (Golden<br>Helix) | AA-1413-V003,<br>AA-1637-V003,<br>AA-1391-V010,<br>AA-1648-V002 | Illumina Plattform | flexibel               |