Untersuchungsverfahren: Humangenetik - Molekulargenetik

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Lynch-Syndrom / Hereditäres Nicht-Polypöses Kolorektales Karzinom (HNPCC): EPCAM, MLH1, MSH2, MSH3, MSH6, PMS2	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Familiärer Brust- und Eierstockkrebs: ATM, BARD1, BRCA1, BRCA2, BRIP1, CHD1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Polyposis-Syndrome (Familiäre adenomatöse Polyposis (FAP), MUTYH-assoziierte Polyposis (MAP), juvenile Polypose (JP), Peutz-Jeghers-Syndrom, Serratiertes Polyposis-Syndrom (SPS), Hereditäres Mixed Polyposis-Syndrom (HMPS)): APC, BMPR1A, GREM1, MUTYH, NTHL1, POLD1, POLE, RNF43, SMAD4, STK11	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Pankreas-Ca assoziierte Syndrome: APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PMS2, PALB2, STK11, TP53	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Magen-Ca, unbestimmt: CDH1, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PMS2	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Prostata-Ca, unbestimmt: ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MAH3, MSH6, PALB2, PMS2	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Darmkrebs (CRC), unbestimmt: AKT1, APC, AXIN2, BMPR1A, BRAF, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, RNF43, RPS20, SMAD4, STK11	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
kongenitaler Hyperinsulinismus (CHI), Maturity-onset diabetes of the young (MODY): ABCC8, AKT2, APPL1, BLK, CEL, FOXA2, GCK, GLUD1, HADH, HK1, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KDM6A, KLF11, KMT2D, NEUROD1, NSD1, PAX4, PDX1, SLC16A1, TRMT10A	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Rasopathien, Noonan-Syndrom, LEOPARD-Syndrom, CFC-Syndrom, Mosaik-Rasopathien, fetales RASopathie-Syndrom: ARAF, BRAF, CBL, CDC42, CUL3, ERF, FBXW11, GRB2, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MAPK1, MAPK3, MRAS, NF1, NF2, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RIT2, RRAS, RRAS2, SETD5, SHOC2, SOS1, SOS2, SPRED1, SPRED2, YWHAZ	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, QIAcuity four, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Gefäßmalformationen, regionaler Überwuchs, Proteus-Syndrom, CLOVE-Syndrom, MCAP-Syndrom: AKT1, ANTXR1, ELMO2, EPHB4, GJA4, GLMN, GNA11, GNA14, GNAQ, IDH1, IDH2, MAP3K3, PIK3CA, PIK3R1, PTEN, RASA1, SMAD4, STAMBP, TEK, VEGFR2, VHL	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, QIAcuity four, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Megalenzephalie, Polymikrogyrie, postaxiale Polydaktylie und Hydrozephalus-Syndrom (MPPH): AKT3, CCND2, PIK3R2	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Morbus Osler: ACVRL1, ENG, GDF2	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Cerebral cavernous malformations (CAM): CCM2, KRIT1, PDCD10	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Lymphatische Malformationen (LM): ADAMTS3, ANGPT2, ARAF, CCBE1, CELSR1, EPHB4, FAT4, FLT4, FOXC2, GATA2, GJC2, KIF11, PIEZO1, PTPN14, SOX18, VEGFC	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Neurofibromatose (NF), Legius-Syndrom, Schwannomatose, McCune-Albright-Syndrom, juvenile myelomonozytäre Leukämie (JMML): GNAS, LZTR1, NF1, NF2, PRKAR1A, PTPN11, SMARCB1, SPRED1	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Tumorprädisposition (kleines Panel): AKT1, APC, ATM, AXIN2, BARD1, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, CTNNA1, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, PRKAR1A, PTEN, RAD51C, RAD51D, RNF43, RPS20, SMAD4, SMARCB1, SPRED1, STK11, TP53	EDTA-Blut, DNA, Speichel	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
hereditäre Lymphödeme, inkl. pränatale Lymphödeme AKT1, BRAF, CBL, CCBE1, FAT4, FLT4, FOXC2, GATA2, GJC2, LZTR1, MAP2K1, MAP2K2, NRAS, PIK3CA, PTEN, PTPN14, RASA1, SHOC2, SOX18, SPRED1, VEGFC	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	Enrichment (Illumina, Nextera Flex for Enrichment), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32716-001, 32888-003, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Whole exome sequencing	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
individuelle Exome-basierte Multigen-Panel-Analyse	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
HPO-basierte Exome Analyse	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Amyotrophe Lateralsklerose(ALS): ANG, CHCHD10, FUS, SOD1, TARDBP, UBQLN2, VAPB, VCP	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Alzheimer'sche Erkrankung: APOE, APP, PSEN1, PSEN2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Adams-Oliver-Syndrom (AOS): DLL4, DOCK6, EOGT, NOTCH1, RBPJ, ARHAGP40, ARHGAP31	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

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Hämophilie A: F8	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Marfan-Syndrom und Typ 1 Fibrillinopathien: FBN1, TGFBR1, TGFBR2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Ehlers-Danlos-Syndrom, klassischer Typ: COL1A1, COL5A1, COL5A2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Ehlers-Danlos-Syndrom, vaskulärer Typ: COL3A1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
thorakale Aortenerweiterung: ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

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Bindegewebserkrankungen inkl. thorakales Aortenaneurysma: ABCC6, ACTA2, ACVR1, ADAMTS2, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, LOX, LTBP4, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PYCR1, SKI, SLC2A10, SLC39A13, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Kardiomyopathie: AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AKAP9, ALMS1, ALPK3, ANKRD1, BAG3, CALR3, CASQ2, CAV3, CHKB, COA5, COA6, COX15, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, DYSF, ELAC2, EMD, ETFA, ETFB, ETFDH, FHL1, FKRP, FKTN, FLNC, GAA, GATAD1, GBE1, GLA, GTPBP3, HADHA, HADHB, HCN4, HRAS, ILK, JPH2, JUP, KARS, LAMA2, LAMA4, LAMP2, LDB3, LIMS2, LMNA, MIB1, MRPL3, MRPL44, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOT, MYOZ2, MYPN, NDUFB11, NDUFV2, NEB, NEBL, NEXN, PDLIM3, PKP2, PLN, PNPLA2, POMT1, POMT2, PPA2, PRDM16, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SCO2, SDHA, SGCA, SGCB, SGCD, SGCG, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SYNE1, TAZ, TCAP, TK2, TMEM43, TMEM70, TNNC1, TNNI3, TNNT2, TOR1AIP1, TPM1, TSFM, TTN, TTR, VCL, VCP, ACADM, ACADS, CPTA1, GFM1, GNE, LARGE1, PLEC, POMGNT1, SEPN1, SYNE2, TGFB3	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Rhythmusstörungen: ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, GJA5, GNAI2, GPD1L, HCN4, JUP, KCNA5, KCNAB2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LMNA, MYL4, NPPA, PKP2, PLN, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Cholestase: ABCB11, ABCB4, ABCC12, ABCC2, ABCC4, ABCG5, ABCG8, AKR1D1, ATP7B, ATP8B1, BAAT, CFTR, CLDN1, CLDN14, CYP7B1, DCDC2, EPHX1, GCKR, GPBAR1, HFE, HSD17B13, IL12A, IL12RB2, IL2RA, JAG1, MBOAT7, MST1, MYO5B, NOTCH2, NR1H4, NR1I2, PKHD1, PNPLA3, SERPINA1, SLC10A1, SLC10A2, SLC25A13, SLC4A2, SLC51A, SLC51B, SLCO1B1, SLCO1B3, STAT4, TJP2, TM6SF2, UGT1A1, VIPAS39, VPS33B	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Epilepsie: AARS1, ABAT, ACY1, ADK, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, ARHGEF9, ARV1, ARX, ATIC, ATP6V1A, AUH, BCKDK, BRAT1, BTD, CACNA1A, CACNA1E, CAD, CAMK2A, CAMK2B, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CNNM2, CNPY3, COQ4, COQ8A, CPLX1, CPT1A, CPT2, CUX2, CYFIP2, D2HGDH, DDX3X, DENND5A, DEPDC5, DHDDS, DHFR, DNM1, DOCK7, DPYD, EEF1A2, ETFA, ETFB, ETFDH, ETHE1, FGF12, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GNAO1, GNB1, GPHN, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, HACE1, HCN1, HLCS, HNRNPU, HPD, IDH2, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, L2HGDH, LGI1, LIAS, LNPK, MBD5, MBOAT7, MDH2, MECP2, MEF2C, MLYCD, MMACHC, MOCS1, MOCS2, MOCS3, MTOR, NEXMIF, NPRL2, NPRL3, NTRK2, PACS2, PC, PCBD1, PCDH19, PDHA1, PDSS2, PHACTR1, PHGDH, PIGA, PLCB1, PLPBP, PNKP,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Alport- und Alport-like-Syndrom: CD151, COL4A3, COL4A4, COL4A5, FN1, LMX1B, MYH9, PXDN	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Schwerhörigkeit, nicht syndromal und syndromal: ABHD12, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ATP6V1B1, BCAP31, BCS1L, BDP1, BSND, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDH23, CDKN1C, CEACAM16, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DNMT1, EDN3, EDNRB, EPS8, EPS8L2, ESPN, ESRRB, EXOSC2, EYA1, EYA4, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOXB1, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LOXHD1, LRTOMT, MANBA, MARVELD2, MIR96, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PJVK, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, S1PR2, SALL1, SEMA3E, SERPINB6, SIX1, SIX5,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Makrozephalie, Großwuchs: AKT1, AKT2, AKT3, APC2, ASPA, ASXL2, ATP7A, BRAF, BRWD3, CCDC88C, CCND2, CHD3, CHD4, CHD8, CRADD, CUL4B, DNMT3A, EED, EZH2, FIBP, GALC, GCDH, GFAP, GPC3, H1-4, HEPACAM, HERC1, HEXA, HEXB, HRAS, HUWE1, IDUA, IGF2, KPTN, KRAS, L1CAM, L2HGDH, MAP2K1, MAP2K2, MED12, MLC1, MPDZ, MTOR, NF1, NFIB, NFIX, NPR2, NRAS, NSD1, PAK1, PHF6, PIGA, PIGN, PIGT, PIK3CA, PIK3R2, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RIN2, RNF125, RNF135, SETD2, SOS1, STRADA, TBC1D7, TSC1, TSC2, UPF3B	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Mikrozephalie: AMPD2, ANKLE2, ASNS, ASPM, ATR, CASK, CDK5RAP2, CDK6, CENPE, CENPJ, CEP135, CEP152, CEP63, CHMP1A, CIT, CLP1, COASY, DYRK1A, EXOSC3, EXOSC8, EXOSC9, FOXG1, IER3IP1, KAT6A, KIF11, KIF14, KNL1, MBD5, MCPH1, MECP2, MED17, MFSD2A, NCAPD2, NCAPD3, NCAPH, NIN, NSMCE2, NUP37, PCLO, PCNT, PHC1, PLK4, PNKP, PPP1R15B, RAB18, RARS2, RBBP8, SASS6, SEPSECS, SLC25A19, SLC25A46, SLC9A6, STAMBP, STIL, TBC1D23, TOE1, TOP3A, TRAIP, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TUBGCP4, TUBGCP6, VPS53, VRK1, WDR62, ZNF335	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Kleinwuchs: ALMS1, BTK, CCDC8, CDKN1C, CUL7, FGF8, FGFR1, FGFR3, FLNA, GH1, GHRHR, GHSR, GLI2, GLI3, HESX1, IFT172, IGF1, IGF1R, IGF2, IGFALS, IGSF1, LHX3, LHX4, NPR2, OBSL1, OTX2, PDE4D, PIK3R1, PITX2, POU1F1, PRKAR1A, PROKR2, PROP1, RNPC3, SHOX, SOX2, SOX3, SRCAP, STAT5B	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Ziliopathien: ACVR2B, AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP4, C2CD3, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CFAP298, CFAP300, CFAP418, CFAP53, CHD1L, CPLANE1, CRELD1, CSPP1, DCDC2, DDX59, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, EVC, EVC2, EXOC8, FAM149B1, FRAS1, GANAB, GAS8, GDF1, GLI3, GLIS2, HNF1B, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, INPP5E, INTU, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LEFTY2, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MMP21, MUC1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, PAX2, PDE6D, PIBF1, PKD1, PKD1L1, PKD2, PKHD1, POC1B,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Ataxie: ABCA1, ABCB7, ABHD12, AFG3L2, AMACR, ANO10, APTX, ARG1, ARSA, ASL, ASS1, ATCAY, ATM, ATP1A3, ATP7B, ATP8A2, BCKDHA, BCKDHB, MTRFR, CA8, CACNA1A, CACNA1C, CACNA1G, CACNB4, CAMTA1, CAPN1, CCDC88C, CLCN2, COA7, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPS1, CSF1R, CTBP1, CWF19L1, CYP27A1, DARS2, DBT, DDHD2, DGAT2, DNAJC12, DNAJC5, DNMT1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ETFA, ETFB, ETFDH, FGF14, FLVCR1, FRRS1L, FXN, GBA2, GFAP, GNAO1, GNB1, GOSR2, GPR88, GRID2, GRM1, HEXA, HEXB, HSD17B4, ITPR1, IVD, KCNA1, KCNA4, KCNC3, KCND3, KCNJ10, KIF1C, KMT2B, MARS2, MRE11, MTTP, NKX6-2, NPC1, NPC2, OTC, PDE2A, PDHA1, PDSS1, PDSS2, PDYN, PEX6, PEX7, PHYH, PIK3R5, PLA2G6, PMPCA, PNKP, PNPLA6, POLG, POLR3A, PPP2R2B, PRICKLE1, PRKCG, PRNP, PRRT2, PUM1, RNF170, RNF216, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC25A15, SLC2A1, SLC52A2, SLC52A3, SLC6A19, SNAP25, SNX14, SOD1, SPG7, SPTBN2, STUB1, SYNE1, SYT1, TACO1, TBCD, TDP1, TDP2,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Demenzerkrankungen: ADAR, ALS2, ANG, ANXA11, APOE, APP, ATXN2, ATXN2, BICD2, BSCL2, CA2, CCNF, CFAP410, CHCHD10, CHMP2B, COL4A1, COL4A2, COLGALT1, CSF1R, CTC1, CTSA, CYLD, DAO, DCTN1, ERBB4, ERCC6, ERCC8, ERLIN1, FIG4, FOXC1, FUS, GALC, GBA, GBE1, GLA, GRN, HEXA, HMBS, HNRNPA1, HNRNPA2B1, HTRA1, IFIH1, ITM2B, KIF5A, MAPT, MATR3, MYORG, NEFH, NEK1, NOP56, NOTCH3, NPC1, NPC2, OCLN, OPTN, PARK7, PDGFB, PDGFRB, PFN1, PRNP, PRPH, PON1, PON2, PON3, PPARGC1A, PSEN1, PSEN2, REEP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCFD1, SETX, SIGMAR1, SLC20A2, SLC52A2, SLC52A3, SNCA, SNCB, SMPD1, SNORD118, SOD1, SPART, SPG11, SQSTM1, TAF15, TARDBP, TBK1, TIA1, TREM2, TREX1, TRPM7, TTPAL, TUBA4A, TYROBP, UBQLN2, UNC13A, VAPB, VCP, XPR1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Phäochromozytom, Paragangliom: AIP, BAP1, CDC73, CDKN1B, DLST, EGLN1, EGLN2, EPAS1, FH, GDNF, KIF1B, MAX, MDH2, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, SRGAP1, TMEM127, VHL	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Prämature Ovarialinsuffizienz (POI): BMP15, CLPP, FIGLA, FSHR, HARS2, HFM1, LARS2, MCM8, NOBOX, NR5A1, STAG3, WT1, CYP17A1, CYP19A1, DIAPH2, FOXL2, FSHB, GDF9, HSD17B4, LHCGR, MCM9, POF1B, STAR, WNT4, BNC1, ERCC6, CPEB1, ESR1, ESR2, INHA, SOHLH1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Cerebrale Mikroangiopathien inkl. zerebrale autosomal-dominante Arteriopathie mit subkortikalen Infarkten und Leukenzephalopathie (CADASIL): APP, COL4A1, COL4A2, COLGALT1, CTSA, FOXC1, GLA, HTRA1, ITM2B, NOTCH3, SNORD118, TREX1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Hereditäre spastische Paraparesen (HSP), spastische Paraplegien (SPG), spastische Spinalparalysen und Differenzialdiagnosen: AAAS, ABCD1, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ARSA, ATL1, ATP13A2, ATP7B, B4GALNT1, BICD2, BSCL2, BTD, C19orf12, CAPN1, CCT5, CLCN2, CLN8, CPT1C, CYP27A1, CYP2U1, CYP7B1, DARS2, DDHD1, DDHD2, DNAJC12, DNM2, DSTYK, EIF2B5, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FARS2, FRRS1L, GAD1, GALC, GAN, GBA2, GBE1, GCH1, GJC2, GNAO1, GNB1, GPR88, GPT2, GRID2, HACE1, HEXA, HPDL, HSPD1, IBA57, IFIH1, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KLC2, KLC4, KMT2B, L1CAM, LYST, MAG, MARS1, MARS2, MMACHC, MTRFR, NIPA1, NKX6-2, NT5C2, OPA3, PANK2, PCYT2, PGAP1, SPG7, PLA2G6, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RETREG1, RNASEH2B, RTN2, SACS, SELENOI, SETX, SLC16A2, SLC33A1, SLC39A14, SOD1, SPART, SPAST, SPG11, SPG21, SPR, SYNE1, TBCD, TECPR2, TFG, TH, TTR, TUBB4A, UBAP1, UBQLN2, UBTF, UCHL1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Genetisch bedingte Adipositas: ADCY3, ADRB2, ADRB3, AFF4, AGRP, ALMS1, APC2, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, C2orf80, CARTPT, CCDC28B, CEP164, CEP19, CEP290, CFAP418, CHD2, CREBBP, CUL4B, DYRK1B, EHMT1, ENPP1, EP300, FMR1, FTO, GHR, GHRL, GNAS, IFT172, IFT27, IFT74, KCTD13, KDM6A, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKRN3, MKS1, MRAP2, NDN, NFIX, NIPBL, NPAP1, NR0B2, NSD1, NTRK2, PCSK1, PHF6, POMC, PPARG, PPP1R3A, PTEN, PYY, RAB23, RAI1, RGMA, SDCCAG8, SDC3, SH2B1, SIM1, SNRPN, TMEM67, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Albinismus: AP3B1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SOX10, TYR, TYRP1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Anämie, megaloblastär: ABCD4, ALAS2, AMN, CBLIF, CDAN1, CDIN1, CUBN, DBA2, DHFR, GATA1, KCNN4, LMBRD1, MMAA, MTHFD1, MTR, MTRR, NDUFS1, RPL11, RPL15, RPL18, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, SFXN4, SLC19A2, SLC19A3, SLC25A19, SLC46A1, TCN2, TPK1, TSR2, UBA1, UMPS	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Entwicklungsstörungen - Angelmann-Syndrom und Rett-like-Syndrom: ARID1B, ARX, ASXL1, ASXL2, ASXL3, ATRX, CAMK2A, CAMK2B, CDKL5, CNTNAP2, DDX3X, DYRK1A, EHMT1, FOXG1, HERC2, KDM5C, KIAA2022, MBD5, MECP2, MEF2C, NRXN1, PDHA1, POGZ, SLC6A8, SLC9A6, SYNGAP1, TCF4, UBE3A, ZEB2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Entwicklungsstörungen - syndromale Formen: A2ML1, ABCC6, ABCC9, ACSL4, ACTB, ACTG1, AFF2, AGL, AHI1, AIFM1, AKT1, AKT3, ALMS1, ALX1, ALX3, ALX4, ANK3, ANKRD11, AP1S2, ARHGAP31, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARX, ASPA, ASXL1, ASXL2, ASXL3, ATP6AP2, ATP7A, ATRX, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS7, BBS9, BCL11A, BCOR, BMP1, BRAF, BRWD3, C12orf57, C2CD3, CPLANE1, CACNG2, CAMK2A, CAMK2B, CASK, CBL, CBS, CC2D1A, CC2D2A, CCDC22, CCDC8, CCND2, CDH15, CDKL5, CDKN1C, CEP290, CHD7, CHD8, CHRNA7, CLCN4, CLIC2, CNTNAP2, COL1A1, COL1A2, CRBN, CREBBP, CRTAP, CSPP1, CTC1, CTCF, CTNNB1, CUL4B, CUL7, DCHS1, DCX, DDX3X, DHCR7, DHODH, DIS3L2, DKC1, DLG3, DLL4, DMD, DNMT3A, DOCK6, DVL1, DYRK1A, EDN3, EDNRA, EDNRB, EFNB1, EFTUD2, EHMT1, EIF2B5, EIF2S3, EMD, EOGT, EP300, EPB41L1, EPG5, ERCC6, ERCC8, ESCO2, EZH2, CCNQ, FANCA, FANCB, FANCC, FANCG, FAT4, FGD1, FGFR1, FGFR2, FGFR3, FHL1, FKBP10, FLNA, FLNB, FLVCR2, FMR1, FOXG1, FOXP1, FOXP2, FTSJ1, FUCA1, G6PC1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Entwicklungsstörungen - X-gebundene Formen: ACSL4, AFF2, AGTR2, AIFM1, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CCDC22, CDKL5, CLCN4, CLIC2, CUL4B, DCX, DDX3X, DKC1, DLG3, DMD, EIF2S3, EMD, CCNQ, FANCB, FGD1, FHL1, FLNA, FMR1, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HCFC1, HDAC8, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IKBKG, IL1RAPL1, IQSEC2, KDM5C, KDM6A, NEXMIF, L1CAM, LAMP2, LAS1L, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NAA10, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PHKA1, PHKA2, PLP1, PORCN, PQBP1, PRPS1, PTCHD1, RAB39B, RAB40AL, RBM10, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TAF1, THOC2, TIMM8A, TSPAN7, UBE2A, UPF3B, USP9X, WDR45, ZDHHC15, ZDHHC9, ZNF711, ZNF81	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Migrationsstörungen: ACTB, ACTG1, ADGRG1, AKT3, ARF1, ARFGEF2, ARX, B3GALNT2, B4GAT1, CCND2, CDK5, COL4A1, COL4A2, CRADD, CRPPA, CSNK2A1, CTNNA2, CTU2, DAG1, DCHS1, DCX, DDX3X, DYNC1H1, EMX2, ERMARD, FAT4, FH, FKRP, FKTN, FLNA, GMPPB, GRIN1, GRIN2B, KATNB1, KIF2A, KIF5C, KIFBP, LAMB1, LAMC3, LARGE1, MEF2C, MTOR, NDE1, NEDD4L, OCLN, PAFAH1B1, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRUNE1, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RELN, RTTN, RXYLT1, SHH, SIX3, TBC1D20, TMTC3, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, WDR81	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Autismus: AP1S2, ATRX, AUTS2, CACNA1C, CASK, CDKL5, CHD7, CNOT3, DHCR7, DPP6, EHMT1, FGD1, FOXP1, GNAI1, GRIN2B, HEXA, HPRT1, KDM5C, L1CAM, MBD5, MECP2, MID1, NHS, NLGN3, NLGN4X, OPHN1, PCDH19, PHF6, PNKP, PQBP1, PTCHD1, PTEN, RAI1, SCN1A, SHANK2, SHANK3, SLC9A5, TCF4, TSC1, TSC2, UBE3A, VPS13B, ZEB2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Arthrogrypose: ACTA1, ADCY6, ADGRG6, ALG3, BICD2, CHST14, CNTNAP1, DNM2, ECEL1, ERBB3, ERGIC1, FBN2, FKBP10, GLDN, GLE1, LGI4, MYBPC1, MYH3, MYH8, NALCN, NEK9, PIEZO2, PIP5K1C, PLOD2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, UNC50, VIPAS39, VPS33B, ZC4H2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Augenerkrankungen: ABCA4, ABCB6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTSL4, ADGRV1, AFG3L2, AGBL1, AGBL5, AGK, AHI1, AIPL1, ALDH1A3, ALMS1, ANTXR1, AP3B1, APOE, ARL13B, ARL2BP, ARL6, ARMC9, ARMS2, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, C1QTNF5, C2CD3, CFAP418, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP290, CEP41, CEP78, CERKL, CFAP410, CFB, CFH, CHD7, CHM, CHMP4B, CHST6, CIB2, CISD2, CLN3, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL17A1, COL18A1, COL2A1, COL4A1, COL4A2, COL8A2, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CSPP1, CTDP1, CTNNA1, CTNNB1, CYP1B1, CYP27A1, CYP4V2, CYP51A1, DCN, DHDDS, DNM1L, DRAM2, DTNBP1, EDN3, EDNRB, EFEMP1, ELOVL4, EPG5, EPHA2, ERCC1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Autoinflammatorische Syndrome: CARD14, CARD8, CD70, CECR1, ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A, WDR1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Autoimmunerkrankungen, Immundefekte: ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, ANKZF1, AP1S3, AP3B1, ARHGEF1, ARPC1B, ASAH1, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2orf69, C3, C5, C6, C7, C8A, C8B, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CCN6, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD48, CD55, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CHUK, CIB1, CIITA, CLEC7A, CLPB, COPA, COPG1, CR2, CSF3R, CTLA4, CTPS1, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1C, DDX58, DEF6, DKC1, DNAJC21, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, DUOX2, EFL1, EGFR, ELANE, EPCAM, EPG5, ERBIN, F12, FADD, FAS, FASLG, FCHO1, FCN3, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HSPA1L, HTR1A, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBKB,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Blutbildungsdefekte: ABCB6, ABCB7, ABCD4, ABCG5, ABCG8, ACD, ACTN1, ADA2, ADAMTS13, AK1, ALAS2, ALDOA, AMN, ANK1, ANKRD26, ANO6, ATP11C, ATR, BLM, BMP6, BPGM, BRCA1, BRCA2, BRIP1, CBLIF, CD36, CD55, CD59, CDAN1, CDIN1, COX4I2, CP, CTC1, CUBN, CYB5R3, CYCS, DHFR, DIAPH1, DKC1, DNAJC21, DUT, EFL1, EGLN1, EPAS1, EPB41, EPB42, EPO, EPOR, ERCC4, ERCC6L2, ETV6, EXOC3L2, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FERMT1, FGA, FGB, FGG, FLI1, FLNA, FTH1, FTL, FYB1, G6PD, GATA1, GATA2, GCLC, GFI1B, GGCX, GLRX5, GNE, GP1BA, GP1BB, GP6, GP9, GPI, GRHL2, GSR, GSS, HAMP, HFE, HJV, HK1, HRG, HSPA9, IKZF1, IKZF5, ITGA2B, ITGB3, JAK2, KCNN4, KIF23, KLF1, LIG4, LMAN1, LMBRD1, LPIN2, LYST, MAD2L2, MCFD2, MECOM, MMAA, MPIG6B, MPL, MT-ATP6, MTHFD1, MTR, MTRR, MYH9, MYSM1, NBEAL2, NBN, NDUFB11, NHP2, NOP10, NPM1, NT5C3A, P2RY12, PALB2, PARN, PFKM, PGK1, PIEZO1, PKLR, PRF1, PRKACG, PROC, PROS1, PUS1, RAD51, RAD51C,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Polyzystische Nierenerkrankung, Polyzystische Lebererkrankung, CAKUT: ACE, ACTA2, ACTG2, ADAMTS9, AGT, AGTR1, AHI1, ALDH1A2, ALG8, ALG9, ANKS6, ANOS1, ARL13B, ARL6, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BICC1, BMP4, BMP7, BNC2, BRCA2, C8ORF37, CC2D2A, CDC5L, CEP120, CEP164, CEP290, CEP83, CHD1L, CHD7, CHRM3, CHRNA3, COL4A1, CRB2, CRKL, CTU2, DACH1, DCDC2, DHCR7, DNAJB11, DSTYK, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DZIP1L, ETFA, ETV4, ETV5, EYA1, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAT4, FGF20, FGF8, FGFR2, FIBP, FOXC1, FOXC2, FOXF1, FOXP1, FRAS1, FREM1, FREM2, GANAB, GATA2, GATA3, GDNF, GLI2, GLI3, GLIS2, GLIS3, GREB1L, GREM1, GRIP1, HAAO, HNF1B, HOXD13, HPSE2, HSPA6, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, IFT80, INPP5E, INVS, IQCB1, ITGA8, JAG1, KIF14, KYNU, LIFR, LPP, LRIG2, LRP4, LRP5, LZTFL1, MAPKBP1, MKKS, MKS1, MNX1, MUC1, MYCN, NADSYN1, NEK1, NEK8, NOTCH2,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chromosomeninstabilitätssyndrome: APC, ATM, ATR, AURKA, AURKB, AXIN2, BLM, BRCA1, BRCA2, BRIP1, BUB1, BUB1B, BUB3, CAMLG, CCNE1, CDC20, CENPE, CENPF, CENPH, CENPJ, CEP152, CEP63, CKAP2, CLASRP, CTNNBIP1, DDB2, DDX11, DNA2, DNMT3B, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESPL1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBXW7, FERMT1, FOXM1, GTF2E2, GTF2H5, HAUS8, ID1, KIF11, KIF2A, KIF2B, KIF2C, KIF4A, KLF4, LIG1, LIG4, MAD1L1, MAD2L1, MAD2L2, MAPRE1, MCTS1, MDM2, MDM4, MPLKIP, MRE11, ANAPC1, NBN, NDC80, NHEJ1, NIN, NSMCE2, TP53, PALB2, POLH, PRKDC, PTTG1, RAD21, RAD50, RAD51, RAD51C, RAE1, RANBP1, RB1, RBBP8, RECQL4, REST, RFWD3, RMI1, RMI2, RNF168, SGO1, SGO2, SLX4, SMC1A, SMC3, SPINK7, TOP2A, TOP3A, TRAIP, TTK, UBE2T, USB1, UVSSA, VHL, WRN, XPA, XPC, XRCC2, ZBTB24	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Disorders of Sexual Development, Adrenogenitales-Syndrom (AGS): APC, ATM, ATR, AURKA, AURKB, AXIN2, BLM, BRCA1, BRCA2, BRIP1, BUB1, BUB1B, BUB3, CAMLG, CCNE1, CDC20, CENPE, CENPF, CENPH, CENPJ, CEP152, CEP63, CKAP2, CLASRP, CTNNBIP1, DDB2, DDX11, DNA2, DNMT3B, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESPL1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBXW7, FERMT1, FOXM1, GTF2E2, GTF2H5, HAUS8, ID1, KIF11, KIF2A, KIF2B, KIF2C, KIF4A, KLF4, LIG1, LIG4, MAD1L1, MAD2L1, MAD2L2, MAPRE1, MCTS1, MDM2, MDM4, MPLKIP, MRE11, ANAPC1, NBN, NDC80, NHEJ1, NIN, NSMCE2, TP53, PALB2, POLH, PRKDC, PTTG1, RAD21, RAD50, RAD51, RAD51C, RAE1, RANBP1, RB1, RBBP8, RECQL4, REST, RFWD3, RMI1, RMI2, RNF168, SGO1, SGO2, SLX4, SMC1A, SMC3, SPINK7, TOP2A, TOP3A, TRAIP, TTK, UBE2T, USB1, UVSSA, VHL, WRN, XPA, XPC, XRCC2, ZBTB24	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Dystonie: ACY1, ADCY5, ANO3, ATM, ATP13A2, ATP1A3, CACNA1B, CIZ1, COL6A3, DLAT, DNAJC12, FRRS1L, FTL, GCDH, GCH1, GNAL, GNAO1, GNB1, GPR88, HEXA, HPCA, KCNMA1, KCTD17, KMT2B, NKX2-1, PARK7, PDE2A, PDHA1, PDHX, PINK1, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, RELN, SCN8A, SGCE, SLC16A2, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, SUCLA2, SYT1, TAF1, TBCD, TH, THAP1, TOR1A, TTPA, TUBB4A, UBTF, UNC13A, VAC14, VPS13D	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Extremitätenfehlbildungen: ARHGAP31, ARHGAP40, ARID1A, ARID1B, ARID2, BHLHA9, BMP2, BMPR1B, CCNQ, CDH3, CHSY1, CREBBP, DHODH, DLL4, DLX5, DOCK6, DPF2, EFNB1, EFTUD2, EOGT, EP300, ESCO2, EVC2, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, GDF5, GDF6, GJA1, GLI1, GLI2, GLI3, HDAC8, HOXA13, HOXD13, IHH, IQCE, KIAA0825, KIF7, LMBR1, LRP4, MECOM, MYCN, NAA10, NIPBL, NOG, NOTCH1, PAX3, PDE3A, PDE4D, PRKAR1A, PTHLH, RAD21, RBM8A, RBPJ, RECQL4, ROR2, SALL1, SALL4, SF3B4, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOX11, TBX15, TBX3, TBX4, TBX5, TP63, TRPV4, WNT10B, WNT7A, YY1AP1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Fiebersyndrome, periodisch: ADA2, AP1S3, APOA1, APOA2, APOC2, CARD14, ELANE, FGA, GSN, HTR1A, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, LYZ, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NLRP7, NOD2, NTRK1, OSMR, OTULIN, PLCG2, PSMB8, PSTPIP1, RBCK1, RIPK1, SH3BP2, SLC29A3, STING1, TNFRSF11A, TNFRSF1A, TRNT1, TTR, WDR1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Glykosylphosphatidylinositol (GPI)-Ankerstörungen: GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGS, PIGT, PIGV, PIGW, PIGY	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Herzfehler, genetisch: AARS2, ABCA1, ABCC9, ABCG5, ABCG8, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, ACVRL1, AGK, AGL, AKAP9, AKT3, ALMS1, ALPK3, ANK2, ANKRD1, APOA5, APOB, APOE, BAG3, BMPR1B, BMPR2, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV1, CAV3, CBL, CCND2, CHKB, COA5, COA6, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, DYSF, EIF2AK4, ELAC2, EMD, ENG, EPHB4, ETFA, ETFB, ETFDH, FHL1, FKRP, FKTN, FLNC, GAA, GATAD1, GBE1, GJA5, GLA, GNAI2, GPD1L, GTPBP3, HADHA, HADHB, HCN4, HRAS, ILK, JPH2, JUP, KARS, KCNA5, KCNAB2, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LIMS2, LIPA, LMNA, LPL, LZTR1, MAP2K1, MAP2K2, MIB1, MRAS, MRPL3, MRPL44, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOT, MYOZ2, MYPN, NDUFB11, NDUFV2, NEB, NEBL, NEXN, NF1, NF2, NPPA, NRAS, PCSK9, PDLIM3, PIK3CA, PIK3R2, PKP2, PLN, PNPLA2,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hereditäre Sensomotorische Neuropathien (HMSN): AARS1, ABHD12, AIFM1, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CCT5, COA7, COX6A1, CTDP1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GJB3, GLA, GNB4, HARS1, HEXA, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, PDK3, PDXK, PHYH, PLEKHG5, PMP2, PMP22, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN9A, SCN11A, SEPTIN9, SETX, SH3TC2, SLC5A7, SLC12A6, SLC25A46, SORD, SPG11, SPTLC1, SPTLC2, SURF1, TDP1, TFG, TRIM2, TRPV4, TTR, VCP, WARS1, WNK1, YARS1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Hypogonadotroper Hypogonadismus: ANOS1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, OTX2, POLR3B, PROK2, PROKR2, SEMA3A, SEMA3E, SOX10, SOX2, SOX3, SPRY4, SRA1, TAC3, TACR3, WDR11	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Ichtyosis, Palmoplantarkeratose, Keratinisierungsstörungen: AAGAB, ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, ATP2C1, CARD14, CASP14, CAST, CDSN, CERS3, CHST8, CLDN1, CSTA, CTSC, CYP4F22, DSG1, DSP, ELOVL4, ENPP1, ERCC2, ERCC3, FLG, FLG2, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2E2, GTF2H5, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT83, KRT9, LIPN, LORICRIN, MBTPS2, MPLKIP, MVK, NIPAL4, NSDHL, PIGL, PNPLA1, POMP, PSAT1, RIN2, SDR9C7, SERPINB7, SERPINB8, SHOC2, SLC27A4, SLURP1, SNAP29, SPINK5, SRD5A3, ST14, STIM1, STS, SULT2B1, SUMF1, TGM1, TGM5, TRPV3, VIPAS39, WNT10A	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Joubert-Syndrom: AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, C2CD3, CC2D2A, CELSR2, CEP104, CEP120, CEP164, CEP290, CEP41, CPLANE1, CSPP1, EXOC8, FAM149B1, HYLS1, IFT172, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, OFD1, PDE6D, PIBF1, POC1B, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Klippel-Feil-Syndrom: DLL3, GDF3, GDF6, HAAO, HES7, KYNU, LFNG, MEOX1, MESP2, MYH3, MYO18B, NADSYN1, RIPPLY2, TBX2, TBX6, WBP11, ZIC3	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
kombinierte B- und T-Zellimmundefekte: CD3G, CD40, CD40LG, CD8A, CIITA, DOCK8, IKZF1, ITK, LCK, MAGT1, ORAI1, PNP, RFX5, RFXANK, RFXAP, RHOH, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, UNC119, ZAP70	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Muskeldystrophie, kongenitale: ACTA1, ACVR1, ADSS1, ANO5, BAG3, BIN1, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHCHD10, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNA2, DNAJB5, DNAJB6, DNM2, DYSF, FHL1, FHL2, FKBP14, FLNC, GNE, HACD1, HNRNPA1, HNRNPA2B1, ISCU, KBTBD13, KLHL40, KLHL41, KLHL9, KY, LAMP2, LDB3, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MSTN, MTM1, MTMR14, MYF6, MYH14, MYH2, MYH7, MYOT, MYPN, NEB, OPA1, ORAI1, PABPN1, PLEC, POLG, POLG2, PUS1, PYROXD1, RRM2B, RYR1, SELENON, SIL1, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, TIA1, TK2, TNNT1, TPM2, TPM3, TRIM32, TRIM54, TRIM63, TTN, TWNK, VCP, VMA21, YARS2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Kraniosynostosen: ALPL, ALX1, ALX3, ALX4, ASXL1, BMP4, BPNT2, CDC45, COLEC11, CYP26B1, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FREM1, FLNA, GLI3, IFT122, IFT140, IFT43, IL11RA, KAT6A, MASP1, MEGF8, MSX2, P4HB, POR, RAB23, RECQL4, RUNX2, SCARF2, SEC24D, SKI, SMAD6, TCF12, TMCO1, TWIST1, WDR19, WDR35, ZIC1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Leukodystrophien, Leukoenzephalopathien: AARS1, AARS2, ABCD1, ACOX1, ADAR, ADPRS, AIMP1, AIMP2, ALDH3A2, AMT, ARSA, ASPA, BCAP31, BOLA3, CARS2, CLCN2, COA8, COASY, COL4A1, COL4A2, COX10, COX15, COX6B1, CSF1R, CTC1, CTSA, CYP27A1, DARS1, DARS2, EARS2 , EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMC1, EPRS1, ERCC6, ERCC8, FA2H, FAM126A, FARS2, FKRP, FKTN, FOLR1, FOXRED1, FUCA1, GALC, GAN, GBE1, GCDH, GCSH, GEMIN4, GFAP, GFM1, GJC2, GLA, GLB1, GLDC, GLRX5, GMPPB, HEPACAM, HEXA, HIKESHI, HSD17B4, HSPD1, HTRA1, IBA57, IDS, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, KIF5A, L2HGDH, LAMA2, LARGE1, LIAS, LIPT1, LIPT2, LMNB1, LRPPRC, LYRM7, MARS2, MCOLN1, MLC1, MPV17, MTFMT, MTHFS, NACC1, NARS2, NAXE, NDUFS1, NDUFV1, NEU1, NFU1, NKX6-2, NOTCH3, NPC1, NPC2, NUBPL, OCLN, PC, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PLA2G6, PLAA, PLEKHG2, PLP1, PMPCB, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMT1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Gliedergürtel-Muskeldystrophien (LGMD) und Differentialdiagnosen: ABHD5, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AGRN, ALDOA, ALG13, ALG14, ALG2, ANK2, ANKRD1, ANO5, APTX, ASAH1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, CACNA1A, CACNA1S, CACNB4, CAPN3, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, COL12A1, COL4A1, COL4A2, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COLQ, COQ8A, COX15, CRPPA, CPT2, CRYAB, CSRP3, DAG1, DES, DGUOK, DMD, DNA2, DNAJB3, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FAM111B, FHL1, FKBP14, FKRP, FKTN, FLNC, G6PC1, GAA, GBA2, GBE1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADH, HADHA, HADHB, HEXA, HINT1, HNRNPDL, HSPG2, IGHMBP2, ISCU, ITGA7, JPH2, KBTBD13, KCNA1, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, KIF21A, KLHL40, KLHL41, KLHL9, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LDHA,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Lipodystrophie: AGPAT2, AKT2, BANF1, BSCL2, CAV1, CAVIN1, CIDEC, FBN1, KCNJ6, LEMD2, LEP, LIPE, LMNA, LMNB2, OTULIN, PIK3R1, PLIN1, POLD1, PPARG, PSMB8, SPRTN, WRN, ZMPSTE24	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Lysomale Speichererkrankungen, Mukopolysaccharidose: ABCC8, ACY1, ADAMTSL2, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ANTXR2, ARG1, ARSA, ARSB, ASAH1, ASPA, ATP13A2, BTD, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSK, DHCR7, DPYD, DYM, ETFA, ETFB, ETFDH, FH, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HRAS, HYAL1, IDS, IDUA, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MOCS1, MOCS2, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX16, PEX26, PEX3, PEX5, PEX6, PGK1, PHYH, PPT1, PRODH, PSAP, QDPR, RAI1, SGSH, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX, TCF4, TPP1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Medulloblastom: APC, BRCA2, CTNNB1, ELP1, GPR161, MLH1, MSH2, MSH6, PALB2, PMS2, PTCH1, PTCH2, SMARCB1, SMARCA4, SUFU, TP53	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Nephrotisches Syndrom: ACE, ACTN4, COQ8B, ANLN, APOL1, ARHGDIA, CD2AP, COQ2, COQ6, COQ9, CRB2, DGKE, EMP2, INF2, ITGA3, LAMB2, LMX1B, MYO1E, NPHS1, NPHS2, PDSS2, PLCE1, PTPRO, SMARCAL1, TRPC6, WT1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Neurodegenrative Erkrankungen: AARS1, AARS2, ABCB7, ABCD1, ABHD12, ACOX1, ADAR, ADCY5, ADPRS, AFG3L2, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALS2, ANG, ANO10, ANO3, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOE, APP, APTX, ARL6IP1, ARSA, ASPA, ATCAY, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP7B, ATP8A2, B4GALNT1, BCAP31, BOLA3, BSCL2, C19orf12, CA2, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CCDC88C, CHCHD10, CHCHD2, CHMP2B, CIZ1, CLCN2, CLN3, CLN5, CLN6, CLN8, COA7, COA8, COASY, COL4A1, COL4A2, COL6A3, COLGALT1, COQ8A, COX10, COX15, COX20, COX6B1, CP, CPT1C, CSF1R, CTBP1, CTC1, CTSA, CTSD, CTSF, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DARS1, DARS2, DCAF17, DCTN1, DDHD1, DDHD2, DNAJC12, DNAJC5, DNAJC6, DNMT1, DSTYK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EMC1, EPRS1, ERCC6, ERCC8, ERLIN2, FA2H, FAM126A, FARS2, FBXO7, FGF14, FIG4, FKRP, FKTN, FLVCR1, FOLR1, FOXC1, FOXRED1, FRRS1L, FTL, FUCA1, FUS, FXN, GALC,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Neuromuskuläre Erkrankungen: AAAS, AARS1, ABCC9, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADCY6, ADGRG6, ADSS1, AGL, AGRN, AIFM1, ALDOA, ALG14, ALG2, ALG3, AMPD1, ANO5, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATP1A1, ATP1A2, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, BVES, CACNA1E, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCT5, CFAP276, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, COX6A2, CPT2, CRPPA, CRYAB, CTDP1, DAG1, DCAF8, DCTN1, DCTN2, DES, DGAT2, DHTKD1, DMD, DNA2, DNAJB2, DNAJB5, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, EMILIN1, ENO3, ERBB3, ERGIC1, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBLN5, FBN2, FBXO38, FGD4, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNB, FLNC, FXR1, G6PC1, GAA, GAN, GARS1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Neuropathien, Motoneuropathien: AAAS, AARS1, ABCA1, ABHD12, ACTA1, ADAR, AGRN, AIFM1, ALG14, ALG2, ALS2, AMACR, ANG, ANO5, ANXA11, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATM, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, MTRFR, CAPN3, CASQ1, CCDC78, CCT5, CD59, CFL2, CHAT, CHCHD10, CHKB, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CLCF1, CLP1, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COX10, COX6A1, CRLF1, CTDP1, CYP27A1, DAG1, DCAF8, DCTN1, DES, DGAT2, DHTKD1, DMD, DNAJB2, DNAJB5, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DYNC1H1, DYSF, EGR2, EMD, EPG5, EXOSC3, EXOSC8, FAM126A, RETREG1, FBLN5, FBXO38, FGD4, FHL1, FIG4, FKBP14, FKRP, FKTN, FUS, FXN, GAA, GALC, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GJB1, GLA, GMPPB, GNB4, GSN, GYG1, HADHA, HADHB, HARS1, HEXA, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB1, HSPB3, HSPB8, IFRD1, IGHMBP2, ELP1, INF2, CRPPA, ITGA7, KARS1, KBTBD13, KIF1A, KIF5A, KLHL13,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Nebennierenrindeninsuffizien (NNR-Insuffizienz):AAAS, AIRE, CDKN1C, CYP11A1, CYP21A2, DHCR7, LIPA, MC2R, MCM4, MRAP, NNT, NR0B1, NR5A1, PCSK1, POLE, POMC, PROP1, SAMD9, SGPL1, STAR, TBX19, TXNRD2, WNT4	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Optikusatrophie: ACO2, AFG3L2, C12orf65, CISD2, DNM1L, MFN2, MTPAP, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, TIMM8A, TMEM126A, WFS1, YME1L1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Osteogenesis imperfecta (OI): ALPL, ANO5, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, BMP1, COL1A1, COL1A2, CRTAP, FKBP10, GORAB, IFIH1, IFITM5, LRP5, P3H1, PLOD2, PLS3, PPIB, PYCR1, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TNFRSF11B, WNT1	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Periventrikuläre noduläre Heterotopie (PVNH): ARF1 , ARFGEF2 , DCX, ERMARD , FLNA, MAP1B , NEDD4L, TMTC3	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Mukoviszidose: CFTR	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Retinitis pigmentosa (RP): ABCA4, ADGRA3, ADIPOR1, AGBL5, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C8orf37, CA4, CDH16, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, GNAT1, GUCA1B, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, KIAA1549, KIZ, KLHL7, LRAT, MAK, MERTK, MVK, NAALADL1, NEK2, NEUROD1, NR2E3, NRL, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RCBTB1, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SAMD11, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRNT1, TTC8, TULP1, USH2A, ZNF408, ZNF513	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Small fiber neuropathy (SFN): ATL1, ATL3, CAV3, CRYAB, DES, DNAJB6, DNMT1, FLNC, GLA, LDB3, MATR3, MYH7, MYOT, SCN9A, SCN10A, SCN11A, SEPTIN9, SPTLC1, SPTLC2, TIA1, TRPA1, TTN, TTR, GBA	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x
Stoffwechselstörungen inkl. Mitochondropathien: AARS2, ABCC8, ABCD1, ABCD4, ABHD5, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACOX1, ACSF3, ADAR, ADPRS, AGA, AGK, AGL, AIFM1, AIMP1, AIMP2, ALDH3A2, ALDH6A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMPD1, AMT, ANO10, APPL1, APTX, ARG1, ARS1, ARSA, ARSB, ASL, ASPA, ASS1, ATP5F1A, ATP5F1D, ATP5F1E, ATPAF2, AUH, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BLK, BOLA3, C1QBP, CAD, CARS2, CCDC115, CD320, CEL, CHKB, CLCN2, CLPB, COA3, COA5, COA6, COA7, COA8, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CPS1, CPT1A, CPT2, CRAT, CSF1R, CTC1, CTNS, CTSA, CYC1, CYP27A1, DARS1, DARS2, DBT, DDOST, DGUOK, DHDDS, DLAT, DLD, DNA2, DNAJC12, DNAJC19, DNM1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, EARS2, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, EMC1, ENO3, EPRS1,	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Tumorprädisposition (großes Panel): ABRAXAS1, AIP, AKT1, ALK, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CCND1, CDC73, CDH1, CDK4, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CHEK2, CYLD, DDB2, DICER1, DIS3L2, EGFR, EGLN1, EGLN2, EPAS1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, IL1B, IL1RN, KIF1B, KIT, LZTR1, MAD2L2, MAX, MC1R, MEN1, MET, MITF, MLH1, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PALLD, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTCH2, PTEN, RAD50, RAD51, RAD51C, RAD51D, RASAL1, RB1, RECQL4, RET, RFWD3, RINT1, RNF43, RPS20, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPRED1, SRGAP1, STK11, SUFU, TERT, TMEM127, TP53, TSC1, TSC2, UBE2T, VHL, WRN, WT1, XPA, XPC, XRCC2	EDTA-Blut, DNA, Speichel	Enrichment (Twist BioScience, Twist Eome 2.0 plus Comp Exome spike-in plus MITO spike-in), Sequencing-by-synthesis, varvis (Limbus Medical Technologies GmbH), PCR, Sanger-Sequenzierung, MLPA	32863-002, 32884-004, 11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 33217-002, 32854-001	ABI 2700, ABI 9700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, Illumina MiSeq, Illumina NextSeq 550Dx, Illumina NovaSeq X Plus		x

Untersuchungsart:
Chromosomenanalyse**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Angeborener Chromosomensatz	EDTA-Blut, DNA, Amnionzellen, Chorionzotten, Speichel	Molekulare Karyotypisierung (SNP-Array)	14268-005, 32545-001	GCS3000DX V.2 Instrumenten System		x

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hämochromatose: HFE-Gen (C282Y und H63D)	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
CYP2C9-Metabolisierungsstatus: CYP2C9*1 (WT), CYP2C9*2 (R144C), CYP2C9*3 (I359L)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
CYP2C19-Metabolisierungsstatus: CYP2C19*2 (Pro227=), CYP2C19*2 (Trp212*), CYP2C19*17 (c.-806C>T)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Dihydropyrimidin-Dehydrogenase (DPD)-Metabolisierungsstatus (DPD-Defizienz): rs3918290, rs55886062, rs56038477, rs67376798, rs75017182	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Mukoviszidose: CFTR-Gen Exon 2-Deletion "sardische Mutation"(c.54-5811_164+2187del8108ins182)	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Mukoviszidose: CFTR-Gen nur Hauptmutationen (N1303K, F508del, R553*, G542*, G551D, R347P, 3849+10kb C>T, 1717-1G>A, CFTRdele2,3, W1282*, 2789+5G>A, 2183AA>G, R1162*, M1101K, 2143delT, 2184delA, 3272-26A>G, delI507, G85E, 621+1G>T, 3659delC, R334W, 1677delTA, 1078delT, E92*, 3905insT, E60*, I336K, 2184insA, A455E, Y1092*)	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Alzheimer'sche Erkrankung: APOE (APOE*4-Allel), APP (Exon 16 und 17)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Mastozytose: KIT (Exon 17)	EDTA-Blut, DNA	Anreicherung mittels ESMA, PCR, Sanger-Sequenzierung	19057-001, 11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Fragiles X-Syndrom (FraX), Prämature Ovarialinsuffizienz (FXPOI), FraX-Tremor-Ataxie-Syndrom (FXTAS): FMR1	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	PCR, Fragmentanalyse, Repeat-primed PCR	11178-003, 23443-001, 19069-001, 53122-001, 19143-001, 53126-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Myotone Dystrophie Typ 1 (DM1): MD101, MD102 (DMPK-Gen)	EDTA-Blut, DNA	PCR, Fragmentanalyse, Repeat-primed PCR	11178-003, 23443-001, 19070-001, 65583-001, 19144-001, 66930-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Myotone Dystrophie Typ 2 (DM2), Proximale Myotone Myopathie (PROMM): CL3N58, CL3N59 (ZNF9-Gen)	EDTA-Blut, DNA	PCR, Fragmentanalyse	11178-003, 23443-001, 19076-001, 66967-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Spinozerebelläre Ataxie (SCA1, SCA2, SCA3, SCA6, SCA7, SCA11): ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP	EDTA-Blut, DNA	PCR, Fragmentanalyse	11178-003, 23443-001, 19078-001, 66975-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Chorea Huntington [OMIM#143100]: HTT-Gen	EDTA-Blut, DNA	PCR, Fragmentanalyse	11178-003, 23443-001, 19079-002, 52332-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Amyotrophe Lateralsklerose (ALS): C9ORF72	EDTA-Blut, DNA	PCR, Fragmentanalyse, Repeat-primed PCR	11178-003, 23443-001, 19080-001, 68276-001, 19152-001, 68279-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
X-Inaktivierung (XCI): FMR1, AR, RP2	EDTA-Blut, DNA, Speichel	PCR, Fragmentanalyse	11178-003, 23443-001, 19081-001, 63701-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Spinobulbäre Muskelatrophie (SBMA): AR	EDTA-Blut, DNA	PCR, Fragmentanalyse	11178-003, 23443-001, 19082-002, 63693-001	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
UPD6: 6q24	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	mMLPA	19086-002	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
UPD7: 7p12, 7q32	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	mMLPA	19086-002	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
UPD14: 14q32	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	mMLPA	19086-002	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
UPD15: Prader-Willi-Syndrom (PWS)WS/Angelman-Syndrom (AS)	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	mMLPA	19086-002	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Prader-Willi-Syndrom (PWS) / Angelman-Syndrom (AS): 15q11-13	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	PCR, Sanger-Sequenzierung, MLPA, mMLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 19086-002	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Silver-Russell-Syndrom (SRS) / Beckwith-Wiedemann-Syndrom (BWS): 11p15	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	PCR, Sanger-Sequenzierung, MLPA, mMLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001, 19086-002	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Multi-Locus-Imprinting Defekt (MLID): 6q24.2, 7p12.2, 7q32.2, 11p15.5, 14q32.2, 15q11.2, 19q13.43, 20q13.32	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	MLPA, mMLPA	12806-003, 32680-001, 19086-002	ABI 2700, ABI ProFlex System, ABI 3500xl Dx Genetic Analyzer		x
Hämochromatose: HFE, HFE2, HAMP, SLC40A1, TRF2	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Cystische Fibrose (CF): CFTR-Gen	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Pankreatitis: CFTR, PRSS1, SPINK1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Muskeldystrophie Duchenne/Becker (DMD/BMD): DMD-Gen	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Hereditäre Neuropathie mit Neigung zu Drucklähmungen (HNPP) / Charcot-Marie-Tooth Neuropathie Typ 1A (CMT1A, HMSN1A): 17p12	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Spinale Muskelatrophie (SMA): SMN1 (Exon 7 und 8), SMN2 (Exon 7 und 8)	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	MLPA	12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Magen-Karzinom, diffuses: CDH1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Sensorineutrale Schwerhörigkeit Typ 1: GJB2, GJB6	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Morbus Fabry: GLA	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Multiple endokrine Neoplasie Typ 1 (MEN1): MEN1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Multiple endokrine Neoplasie Typ 2 (MEN2): RET	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Thrombophilie Protein-C-Mangel (PROC): PROC	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Thrombophilie Proteil-S-Mangel (PROS): PROS1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Rett-Syndrom: MECP2, CDKL5, FOXG1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
SHOX-bedingter Kleinwuchs, Leri-Weill-Syndrom: SHOX	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Li-Fraumeni-Syndrom: TP53	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Tuberöse Sklerose Typ 1 und Typ 2: TSC1, TSC2	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Saethre-Chotzen-Syndrom, Brachyzephalie, isolierte, Skaphokephalie, isolierte: TWIST1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
von Hippel-Lindau Syndrom: VHL	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Wilms-Tumor, Denys-Drash-Syndrom: WT1	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Cowden-Syndrom: PTEN	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/ Version	Gerät	CE-Verfahren	in Haus-Verfahren
Familiäre Hypercholesterinämie: APOB100 (R3527H, R3558H), LPA, LDLR, PCSK9, LDLRAP1	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Achondroplasie, Hypochondroplasie, Thanatophore Dysplasie, Muenke-Syndrom: FGFR3	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
familiäre infantile Krämpfanfälle, Paroxysomale kinesiogene Dyskinesie: PRRT2	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Angelman-Syndrom (AS): UBE3A	EDTA-Blut, DNA, Speichel	PCR, Sanger-Sequenzierung	11178-003, 12842-001, 11226-002, 31495-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer		x
Untersuchung auf familiäre Varianten (SNV oder CNV), Validierung von Varianten	EDTA-Blut, DNA, Speichel, Amnionzellen, Chorionzotten	PCR, Sanger-Sequenzierung, MLPA	11178-003, 12842-001, 11226-002, 31495-001, 12806-003, 32680-001	ABI 2700, ABI ProFlex System, ABI Verity 96-well, Biomek NXP, ABI 3500xl Dx Genetic Analyzer, QIAcuity four		x