Tagesaktuelle Liste der Verfahren im flexiblen Akkreditierungsbereich nach DIN EN ISO 15189:2023

D-ML-21458-02 **gültig ab 24.02.2025**

MVZ der Universitätsmedizin Mainz GmbH
Fachgebiet Humangenetik, Molekulargenetisches Labor
Langenbeckstraße 1, Gebäude 706N
55131 Mainz

Untersuchungen im Bereich:
Medizinische Laboratoriumsdiagnostik

Untersuchungsgebiete:
Humangenetik (Molekulare Humangenetik, molekulare Zytogenetik)

Untersuchungsarten:
molekulare Chromosomenanalyse
Molekularbiologische Untersuchungen (Hybridisierungsverfahren und Amplifikationsverfahren)

Innerhalb der mit ** gekennzeichneten Untersuchungsbereiche ist dem Laboratorium, ohne dass es einer vorherigen Information und Zustimmung der Deutschen Akkreditierungsstelle GmbH bedarf, die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet. Die aufgeführten Untersuchungsverfahren sind beispielhaft. Das Laboratorium verfügt über eine aktuelle Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich.

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart:
Chromosomenanalyse**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät
angeborener Chromosomensatz	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	SNP-HD-Array	SOP 87 , Version 08	Affymetrix Gene Chip Scanner; Gene Chip Fluidics Station 450
angeborener Chromosomensatz	EDTA-Blut, genomische DNA; genomische DNA	SNP-XON-Array	SOP 88, Version 04	Affymetrix Gene Chip Scanner; Gene Chip Fluidics Station 450

Untersuchungsgebiet: Humangenetik

Untersuchungsart:

Molekularbiologische Untersuchungen (Hybridisierungsverfahren)**

Beckwith-Wiedemann-Syndrom, Silver-Russell-Syndrom: *H19, KCNQ1OT1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Silver-Russell-Syndrom: *MEST*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Angelman-Syndrom: *UBE3A*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Prader-Willi-Syndrom: *SNRPN*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Temple-Syndrom (matUPD14); Kagami-Ogata-Syndrom (patUPD14): *MEG3*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Silver-Russell-Syndrom: *MEST*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MS-MLPA	SOP 14, Version 11 und SOP 15, Version 06	ABI SeqStudio 8 Flex Genetic Analyzer System
Brust- und Eierstockkrebs, hereditär: *BRCA1, BRCA2*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Muskeldystrophie Typ Duchenne/Becker: *DMD*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Marfan-Syndrom: *FBN1*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
CMT1A, HNPP: *PMP22*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Hereditäre spastische Paraplegie (HSP): *ATL1*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Spinale Muskelatrophie: *SMN1, SMN2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
autosomal-dominante Polyzystische Nierenerkrankung (ADPKD): *PKD1, PKD2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Idiopathischer Kleinwuchs: *SHOX*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Schwerhörigkeit, nicht-syndromal: *GJB6, STRC, OTOA*	EDTA-Blut, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Li-Fraumeni-Syndrom: *CHEK2, TP53*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Neurofibromatose: *NF1, NF2, SPRED1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System

Neuralrohrdefekte: *VANGL1, VANGL2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Subtelomerscreening	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Opitz-Syndrom: *MID1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Mikrodeletionsscreening	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Tuberöse Sklerose: *TSC1, TSC2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Rubinstein-Taybi-Syndrom: *CREBBP, EP300*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Nicht-obstruktive Azoospermie, schwere Oligozoospermie: *AZF*	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
MECP2 -Duplikationssyndrom	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Rett-Syndrom: *MECP2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Dravet-Syndrom und Differentialdiagnosen: *SCN1A*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Aicardi-Goutières-Syndrom: *RNASEH2A, RNASEH2B, RNASEH2C, TREX1, SAMHD1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Holoprosenzephalie: *SHH, ZIC2, SIX3, GLI2, TGIF1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Lissenzephalie: *PAFAH1B1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Sotos-Syndrom: *APC2, NFIX, NSD1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät
Alzheimer/Demenz: *APP*	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Parkinson: *ATP13A2, GCH, LRRK2, PARK7, PRKN, PINK1, SNCAM*	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Schwannomatose: *SMARCB1, LZTR1, NF2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Gorlin-Goltz-Syndrom: *PTCH1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
DiGeorge-Syndrom (22q11.2)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Komplementdefekte (Neisseria): *CFHR1, CFHR2, CFHR3, CFHR4, CFHR5*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	MLPA	SOP 14, Version 11	ABI SeqStudio 8 Flex Genetic Analyzer System
Achromatopsie: *ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP*	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Albinismus: *AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC38A8, SLC45A2, SNAI2, SOX10, TYR, TYRP1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Beckwith-Wiedemann-Syndrom: *H19, LIT1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	Bisulfit-Pyrosequenzierung, MS-MLPA	SOP 14, Version 11; SOP 15, Version 06 ; SOP 18, Version 04	Qiagen PyroMark Q96 ID; ABI SeqStudio 8 Flex Genetic Analyzer System
Silver-Russell-Syndrom: *H19, MEST*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	Bisulfit-Pyrosequenzierung, MS-MLPA	SOP 14, Version 11; SOP 15, Version 06 ; SOP 18, Version 06	Qiagen PyroMark Q96 ID; ABI SeqStudio 8 Flex Genetic Analyzer System
Temple-Syndrom (matUPD14), Kagami-Ogata-Syndrom (patUPD14): *MEG3*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	Fragmentanalyse, Bisulfit-Pyrosequenzierung	SOP 20, Version 04	Qiagen PyroMark Q96 ID; ABI SeqStudio 8 Flex Genetic Analyzer System
Prader-Willi-Syndrom: *SNRPN*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	Bisulfit-Pyrosequenzierung, MS-MLPA	SOP 19, Version 05, SOP 23, Version 05; SOP 14, Version 11	Qiagen PyroMark Q96 ID; ABI SeqStudio 8 Flex Genetic Analyzer System

Angelman-Syndrom: *UBE3A*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	Bisulfit-Pyrosequenzierung, MS-MLPA	SOP 19, Version 05, SOP 23, Version 05; SOP 14, Version 11	Qiagen PyroMark Q96 ID; ABI SeqStudio 8 Flex Genetic Analyzer System
Aniridie: *PAX6*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lebersche Optikusatrophie Hot-Spots: *MTND1P* (G3460A), *MTND4P* (G11778A) und *MTND6P* (T14484C)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/Pyrosequenzierung	SOP 71, Version 03	Qiagen PyroMark Q96 ID
Fragiles-X-assoziiertes-Tremor-Ataxie-Syndrom (FXTAS); Fragiles X-Synrom: *FMR1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/Fragmentanalytik	SOP 3, Version 09	ABI SeqStudio 8 Flex Genetic Analyzer System
Mütterliche Kontamination (STR)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/Fragmentanalytik	SOP 80, Version 03	ABI SeqStudio 8 Flex Genetic Analyzer System
Repeat-Analyse Chorea Huntington: *HTT*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/Fragmentanalytik	SOP 57, Version 08	ABI SeqStudio 8 Flex Genetic Analyzer System
Rett-Syndrom Sequenzierung: *MECP2*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Schwerhörigkeit, nicht-syndromal: *GJB2, GJB6*	EDTA-Blut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung	SOP 16, Version 05	ABI SeqStudio 8 Flex Genetic Analyzer System
Spingolipidosen Typ 1 (*HEXA*); Spingolipidosen Typ 2 (*HEXB*)	EDTA-Blut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Mukopolysaccharidosen TypI (*IDUA*); Typ III A (*SGSH*); TypIII B (*NAGLU*); Typ IV B (*GLB1*); Typ VII (*GUSB*)	EDTA-Blut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Idiopathischer Kleinwuchs: *SHOX*	EDTA-Blut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung	SOP 30, Version 05	ABI SeqStudio 8 Flex Genetic Analyzer System
X-Inaktivierungstatus (HUMARA)	EDTA-Blut, genomische DNA; genomische DNA	PCR/Fragmentanalytik	SOP 27, Version 07	ABI SeqStudio 8 Flex Genetic Analyzer System
Li-Fraumeni-Syndrom: *CHEK2, TP53*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung	SOP 25, Version 07; SOP 54, Version 04; SOP 81, Version 02	ABI SeqStudio 8 Flex Genetic Analyzer System

Noonan-Syndrom: *PTPN11*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 66, Version 05; SOP 67, Version 06; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Rubinstein-Taybi-Syndrom: *CREBBP, EP300*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture; Sequencing-by-synthesis	SOP 66, Version 05; SOP 67, Version 06; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Schwerhörigkeit, mitochondrial: *MTRNR1*	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; mitochondrielle DNA, genomische DNA	PCR/DNA-Sequenzierung	SOP 25, Version 07; SOP 81, Version 02	ABI SeqStudio 8 Flex Genetic Analyzer System
Angeborener Herzfehler Typ 6: *GDF1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung	SOP 25, Version 07; SOP 81, Version 02	ABI SeqStudio 8 Flex Genetic Analyzer System
Muskelatrophie Typ Kennedy Repeatanalyse: *AR*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/Fragmentanalytik	SOP 3, Version 09	ABI SeqStudio 8 Flex Genetic Analyzer System
Maternal vererbter Diabetes mellitus mit Schwerhörigkeit *(MTTL1)* / MELAS (Myopathie, Enzephalopathie;Laktatazidose, Schlaganfall-ähnliche [´stroke-like´) Episoden	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat; mitochondrielle DNA, genomische DNA	PCR/Pyrosequenzierung	SOP 6, Version 04	Qiagen PyroMark Q96 ID
Bardet-Biedl-Syndrom: *ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT172, LZTFL1, MKKS, MKS1, NPHP1, PNPLA6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Corneale Dystrophie: *AGBL1, CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, OVOL2, PIKFYVE, PITX2, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1, ZEB1, ZNF469* (29 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Ectopia Lentis: *AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL18A1, FBN1, LTBP2, P3H2, PORCN, SUOX, VSX2* (14 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Fleckige Retina: *ABCA4, CHM, CYP4V2, ELOVL4, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13B* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Glaukom: *ADAMTS10, ASB10, B3GLCT, BEST1, BMP4, CNTNAP2, COL4A1, COL8A2, CYP1B1, FBN1, FOXC1, FOXE3, GJA1, ISPD, LMX1B, LTBP2, MAF, MYOC, NTF4, OPA1, OPA3, OPTN, PAX6, PIK3R1, PITX2, POMT1, PRSS56, PXDN, RPS19, RRM2B, SBF2, SH3PXD2B, SIX6, TBK1, TEK, TMEM126A, TTR, WDR36* (38 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Joubert-Syndrom: *AHI1, ARL13B, ARMC9, B9D1, B9D2, C2CD3, C21ORF2, C5ORF42, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423* (39 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Katarakt: *ABCB6, ABHD12, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BFSP1, BFSP2, CHMP4B, CLPB, COL4A1, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXE3, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX3, PXDN, RAB3GAP1, RECQL4, SIL1, SIPA1L3, SLC16A12, SLC33A1, TDRD7, TFAP2A, TMEM70, VIM, VSX2, WFS1, WRN* (78 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Lebersche kongenitale Amaurose: *AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, CWC27, DTHD1, GDF6, GUCY2D, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MYO7A, NMNAT1, OTX2, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TULP1* (30 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Makuladystrophie: *ABCA4, BEST1, C1QTNF5, CDH3, CFH, CLN3, CERKL, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, IMPG1, IMPG2, IRX1, MFSD8, PRDM13, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RP1L1, RPGR, RS1, TIMP3* (29 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Mikrophtalmie, Anophthalmie, Nanophthalmie: *ABCB6, ADAMTS18, ALDH1A3, ATOH7, BCOR, BMP4, BMP7, CHD7, COL4A1, COX7B, CYP1B1, ERCC2, ERCC5, ERCC6, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GDF3, GDF6, GJA1, GRIP1, HCCS, HESX1, HMX1, MAB21L2, MFRP, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PIGL, PITX2, POMGNT1, PQBP1, PRSS56, PXDN, RAB3GAP1, RARB, RAX, RBP4, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TENM3, TMEM98, TFAP2A, VAX1, VPS13B, VSX2, ZIC2* (61 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Nachtblindheit: *CABP4, CACNA1F, CACNA2D4, CYP4V2, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, RBP4, PDE6B, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1* (20 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Optikusatrophie: *ACO2, AFG3L2, ANTXR1, C12ORF65, CISD2, DNM1L, FDXR, MFN2, MT-ND1, MT-ND4, MT-ND6, NDUFS1, NR2F1, OPA1, OPA3, POLG, PRPS1, PDXK, RTN4IP1, SLC25A46, SLC52A2, SNX10, SPG7, TIMM8A, TMEM126A, WFS1, YME1L1* (26 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Retinitis Pigmentosa: *ABCA4, ABHD12, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C1QTNF5, C21ORF2, C2ORF71, C8ORF37, CA4, CACNA1F, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, EYS* , FAM161A, FLVCR1, GNPTG, GUCY2D, HGSNAT, HK1, IDH3B, IFT140, IFT172, IFT43, IMPDH1, IMPG2, INPP5E, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, MVK, NEK2, NMNAT1, NR2E3, NRL, OAT, OFD1, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX7, PHYH, PITPNM3, PLA2G5, POMGNT1, PRCD, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RDH12, RDH5, REEP6, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RP1L1, RPGR, RPGRIP1, RS1, SAG, SAMD11, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, VPS13B, WDR19, ZNF408, ZNF513* (119 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Senior-Loken-Syndrom: *CEP164, CEP290, IFT81, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19, ZNF423* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Stickler-Syndrom: *COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN* (8 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Usher-Syndrom: *ABHD12, ADGRV1, CDH23, CEP78, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, PEX1, USH1C, USH1G, USH2A* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Vitreoretinopathie: *ATOH7, BEST1, CAPN5, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CTC1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Zapfen- und Zapfen-Stäbchen Dystrophien: *ABCA4, ADAM9, ADAMTS18, AIPL1, ALMS1, ARHGEF18, ATF6, BEST1, C21ORF2, C2ORF71, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP78, CERKL, CLN3, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, GNAT2, GUCA1A, GUCY2D, KCNV2, MERTK, NMNAT1, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH12, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5* (48 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Augenerkrankungen: *AASS, ABCA4, ABCB6, ABHD12, ACBD5, ACKR3, ACO2, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRV1 (GPR98), ADIPOR1, AFG3L2, AGBL1, AGBL5, AGK, AHI1, AHR, AIPL1, ALDH18A1, ALDH1A3, ALMS1, ANTXR1, AP3B1, APTX, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ARSG, ASB10, ASPH, ATF6, ATOH7, B3GLCT, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S5, BLOC1S6, BMP4, BMP7, C10ORF11, C10ORF2, C12ORF65, C1QTNF5, C21ORF2, C2CD3, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CBS, CC2D2A, CCDC111, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP290, CEP41, CEP78, CERKL, CFH, CHD7, CHM, CHMP4B, CHN1, CHRDL1, CHST6, CIB2, CISD2, CLEC3B, CLN3, CLPB, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, CNTNAP2, COL11A1, COL11A2, COL17A1, COL18A1, COL2A1, COL4A1, COL5A1, COL8A2, COL9A1, COL9A2, COL9A3, COX7B, CPAMD8, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CSPP1, CTC1,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Augenerkrankungen: *CTDP1, CTNNA1, CTNNB1, CWC27, CYP1B1, CYP27A1, CYP4V2, CYP51A1, DCN, DFNB31, DHDDS, DHS6S1, DHX38, DNAJC30, DNM1L, DRAM2, DTHD1, DTNBP1, EDN3, EDNRB, EFEMP1, ELOVL4, ELP4, EMC1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, EYS, FAM126A, FAM161A, FBN1, FDXR, FGFR1, FLVCR1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FRMD7, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GCNT2, GDF3, GDF6, GJA1, GJA3, GJA8, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRIP1, GRK1, GRM6, GSN, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HESX1, HGSNAT, HK1, HKDC1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, IRX1, IRX5, IRX6, ISPD, JAG1, KCNJ13, KCNV2, KERA, KIAA0556, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF21A, KIF7, KIT, KIZ, KLHL7, KRT12, KRT3, LCA5, LCAT, LEMD2, LIM2, LMX1B, LOXHD1, LOXL3, LRAT, LRIT3, LRP2, LRP5, LRPAP1, LSS, LTBP2, LYST, LZTFL1,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Erweitertes Gen-Set Augenerkrankungen: MAB21L2, MAF, *MAK, MC1R, MERTK, MFN2, MFRP, MFSD8, MIP, MIR184, MITF, MKKS, MKS1, MLPH, MMACHC, MT-ND1, MT-ND4, MT-ND6, MTTP, MVK, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NDUFS1, NEK2, NF2, NHS, NMNAT1, NOTCH2NLC, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NTF4, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPTN, OTX2, OVOL2, P3H2 (LEPREL1), P4HA2, PANK2, PANK4, PAX2, PAX3, PAX6, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDXK, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PEX7, PHOX2A, PHYH, PIBF1, PIGL, PIK3R1, PIKFYVE, PITPNM3, PITX2, PITX3, PLA2G5, PNPLA6, POC1B, POLG, POMGNT1, POMT1, PORCN, PQBP1, PRCD, PRDM13, PRDM5, PRDX3, PRICKLE3, PRKCG, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PXDN, RAB27A, RAB28, RAB3GAP1,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Augenerkrankungen: *RARB, RAX, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROBO3, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPS19, RRM2B, RS1, RTN4IP1, SAG, SALL4, SAMD11, SBF2, SCO2, SDCCAG8, SEMA4A, SETX, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A4, SLC25A46, SLC33A1, SLC38A8, SLC39A5, SLC45A2, SLC4A11, SLC52A2, SLC6A6, SLC7A14, SMCHD1, SMOC1, SNAI2, SNRNP200, SNX10, SOX10, SOX2, SOX3, SPATA7, SPG7, SPP2, STRA6, STX3, SUFU, SUOX, TACSTD2, TBK1, TCF4, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TGFBI, TIMM8A, TIMP3, TK2, TLCD3B, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TMEM70, TMEM98, TOPORS, TOGARAM1, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TTR, TUB, TUBB3, TULP1, TYMP, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2, WASF1, WDPCP, WDR19, WDR36, WFS1, WRN, YME1L1, YAP1, ZEB1, ZIC2, ZNF408, ZNF423, ZNF469, ZNF513, ZNF644, ALPK1, Nys1, RILPL1, USP45, YAP1* (506 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Autismus-Spektrum-Störung: *ADNP, ANK2, ANKRD11, ARID1B, ASH1L, ASXL3, BAZ2B, BCKDK, BCL11A, CACNA1D, CACNA1H, CACNA2D3, CEP41, CHD2, CHD8, CIC, CNOT3, CNTN4, CNTNAP2, CTNND2, CUL3, CUX1, DDX3X, DEAF1, DIP2C, DSCAM, DYRK1A, EIF4E, ERBB2IP, FOXP1, GABRB3, GIGYF2, GRIA1, GRIN2B, GRIP1, ILF2, INTS6, IRF2BPL, KAT2B, KATNAL2, KDM5B, KDM6A, KMT2A, KMT2B, KMT2C, LEO1, MACROD2, MAGEL2, MBOAT7, MECP2, MED13, MED13L, MET, MYT1L, NAA15, NCKAP1, NCOR1, NLGN3, NLGN4X, NRXN1, PHF3, POGZ, PTCHD1, PTEN, RANBP17, RELN, RIMS1, RPL10, SCN2A, SCN9A, SET, SETD5, SHANK2, SHANK3, SLC6A1, SLC9A9, SMARCC2, SPAST, SRCAP, SRSF11, SYNGAP1, TAOK2, TBL1XR1, TBR1, TCF20, TMLHE, TNRC6B, TRIO, TRIP12, UBN2, UPF3B, USP15, USP7, WAC, WDFY3* (95 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Ehlers-Danlos-Syndrom: *COL5A1, COL5A2, COL1A1, COL1A2, COL3A1, PLOD1, ADAMTS2, B3GALT6, B4GALT7, CHST14, DSE, SLC39A13, TNXB, FKBP14, B3GAT3, ZNF469, CHST3* (17 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Thorakale Aortenaneurysmen und Aortendissektion (TAAD): *ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2, TGFB3, PRKG1, MFAP5* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Marfan-Syndrom und Typ 1 Fibrillinopathien: *FBN1, TGFBR2, TGFBR1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Genset Bindegewebserkrankungen: *ABCC6, ACTA2, ADAMTS10, ADAMTS2, ADAMTSL4, ALDH18A1, ATP6V0A2, ATP7A, B3GAT3, B4GALT7, CBS, CHST14, CHST3, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNB, GORAB, LTBP2, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469* (57 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Amyotrophe Lateralsklerose: *ALS2, ANG, ANXA11, ATL1, ATXN2, BSCL2, C9orf72, CHCHD10, CHMP2B, DCTN1, ERBB4, FIG4, FUS, GBE1, GRN, HEXA, HNRNPA1, HSPD1, KIAA0196, KIF5A, MATR3, NEFH, NEK1, OPTN, PFN1, PRF1, PRPH, REEP1, SETX, SIGMAR1, SLC52A2, SLC52A3, SOD1, SPAST, SPG11, SPG20, SQSTM1, TARDBP, TBK1, TRPM7, TUBA4A, UBQLN2, VAPB, VCP* (44 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Arthrogrypose: *ACAT1, ADGRG6, AGRN, BIN1, CACNA1E, CASK, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHUK, CNTNAP1, COL6A2, COLQ, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERBB3, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKBP10, FKTN, FLVCR2, GBA, GBE1, GFPT1, GLDN, GLE1, KAT6B, KIAA1109, KLHL40, LGI4, LMNA, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH8, NALCN, NEB, PIEZO2, PLOD2, PMM2, PPP3CA, RAPSN, RARS2, RIPK4, SCO2, SELENON, SMN1, SMN2, TGFB3, TK2, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN2, TSEN54, UBA1, VIPAS39, VPS33B, VRK1, ZBTB42, ZC4H2* (78 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Erweitertes Genset Ataxie: *ABCB7, ABHD12, ACO2, ADCK3 (COQ8A), ADGRG1, AFG3L2, AHI1, ALDH5A1, AMACR, ANO10, APTX, ARL13B, ARL6, ARSA, ATCAY, ATM, ATP1A3, ATP2B3, ATP8A2, ATXN10, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, BTD, C10ORF2, C5ORF42, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHP1, CLCN2, CLN5, CLN6, CLPP, COASY, COX20, CP, CSPP1, CSTB, CWF19L1, CYP27A1, CYP2U1, DARS2, DLAT, DNAJC19, DNAJC5, DNMT1, EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, FA2H, FAT2, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FOLR1, FXN, GALC, GBA, GBA2, GCLC, GFAP, GJB1, GLRB, GOSR2, GRID2, GRM1, GSS, HARS2, HEXA, HEXB, HIBCH, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0586, KIF1C, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, MARS2, MKKS, MKS1, MME, MRE11 (MRE11A), MTFMT, MTPAP, MTTP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEU1, NKX2-1, NKX2-5, NOL3, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPA3, OPHN1, PANK2, PAX6, PDE10A, PDE6D, PDHX, PDYN, PEX10, PEX2, PEX7, PHYH, PIK3R5, PLA2G6, PLD3, PMPCA, PNKD, PNKP, PNPLA6, POC1B, POLG, POLR3A, PPP2R2B, PRICKLE1, PRKCG, PRNP, PRRT2, RNF170, RNF216, RPGRIP1L, RUBCN, SACS, SAMD9L, SCN2A, SCYL1, SERAC1, SETX, SIL1, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A2, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TRPC3, TTBK2, TTC19, TTC21B, TTC8, TTPA, TUBB4A, UBA5, VAMP1, VLDLR, VRK1, WDPCP, WDR81, WFS1, WWOX, ZFYVE26, ZNF423* (213 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Choreatiforme Bewegungsstörungen: *ADCY5, ARSA, ATM, FRRS1L, FTL, GM2A, GNAO1, KCNA1, NKX2-1, PDE10A, PRNP, RNF216, SETX, VPS13A, XK* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Dystonie: *ADAR, ADCY5, ANO3, ATM, ATP1A2, ATP1A3, ATP7B, BCAP31, CACNA1A, CACNA1B, CIZ1, COL6A3, COX20, DCAF17, DCC, DNAJC12, FA2H, FTL, GCDH, GCH1, GNAL, HPCA, KCNA1, KCNMA1, KIF1C, KMT2B, MECR, NOTCH3, PANK2, PDGFB, PDGFRB, PLA2G6, PNKD, PRKRA, PRRT2, SCN8A, SGCE, SLC19A3, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A* (50 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hereditäre Spastische Paraplegie: *ABCD1, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARG1, ATAD3A, ATL1, ATP13A2, B4GALNT1, BSCL2, BTD, C12ORF65, C19ORF12, CAPN1, COASY, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS, DCTN1, DCTN2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FXN, GALC, GBA2, GBE1, GCH1, GJC2, HACE1, HSPD1, IBA57, IFIH1, KDM5C, KIAA0196, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, L2HGDH, MAG, MARS2, NIPA1, NKX6-2, NT5C2, PAH, PCYT2, PLA2G6, PLP1, PNPLA6, POLR3A, RARS, REEP1, REEP2, RNASEH2B, RTN2, SACS, SELENOI, SETX, SLC16A2, SLC25A15, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG7, SPR, TECPR2, TFG, TH, TUBB4A, UBAP1, UCHL1, VPS37A, ZFYVE26, ZFYVE27* (93 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Kongenitale myasthene Syndrome: *AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, FLAD1, GFPT1, GMPPB, LAMA5, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, STIM1, SYT2, VAMP1* (32 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Muskelatrophie: *AARS, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, REEP1, SCO2, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TBCE, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS* 1 (35 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Muskeldystrophien und Myopathien: *ACTA1, ACVR1, ADSS1, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, C10ORF2, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHCHD10, CHKB, CNTN1, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, CRYAB, DAG1, DES, DMD, DNA2, DNAJB5, DNAJB6, DNM2, DPM1, DPM2, DPM3, DYSF, EMD, FDX2, FHL1, FHL2, FKBP14, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, GOLGA2, HACD1, HNRNPA1, HNRNPA2B1, INPP5K, ISCU, ISPD, ITGA7, KBTBD13, KLHL40, KLHL41, KLHL9, LAMA2, LAMP2, LARGE, LARGE1, LDB3, LIMS2, LMNA, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MME, MSTN, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTM1, MTMR14, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYF6, MYH14, MYH2, MYH7, MYO18B, MYOT, MYPN, NEB, OPA1, ORAI1, PABPN1, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PUS1, PYROXD1, RBCK1, RRM2B, RXYLT1, RYR1, SELENON, SEPT9, SGCA, SGCB, SGCD, SGCG, SIL1, SMCHD1, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, SYNE1, SYNE2, TCAP, TIA1, TK2, TMEM126B, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIM54, TRIM63, TRIP4, TTN, VCP, VMA21, VPS13A, YARS2* (178 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Neuropathien: *SEPT9, AARS, ABHD12, AIFM1, AMACR, ARHGEF10, ATAD3A, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BSCL2, C12ORF65, CCT5, CHCHD10, COA7, COX10, COX6A1, CTDP1, DCAF8, DCTN1, DCTN2, DGAT2, DHTKD1, DNAJB2, DNAJB5, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, FXN, GAN, GLA, GARS, GDAP1, GJB1, GJB3, GNB4, GNE, HADHA, HADHB, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, MYOT, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PLEKHG5, PMP2, PMP22, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SETX, SGPL1, SH3TC2, SLC12A6, SLC25A46, SMAD3, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TECPR2, TFG, TRIM2, TRPA1, TRPV4, TTR, C10ORF2, TYMP, VCP, WNK1, YARS, ZFYVE26* (123 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Paroxysomale Bewegungsstörungen: *ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, NOTCH3, PNKD, POLG, PRKN, PRRT2, SCN1A, SCN8A, SLC2A1* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Periodische Paralysen: *CACNA1S, CLCN1, KCNE3, KCNJ18, KCNJ2, KCNJ5, SCN4A* (7 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Rhabdomyolyse: *ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ADCK3, AGL, AHCY, ALDOA, AMPD1, ANO5, C10ORF2, CAV1, COQ2, CPT2, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FKRP, FKTN, FLAD1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, HADHA, HADHB, ISCU, LAMA2, LAMP2, LDHA, LPIN1, MGME1, MPV17, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH3, OPA1, OPA3, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SLC16A1, SLC22A5, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TANGO2, TAZ, TK2, TYMP, YARS2* (110 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Spinale Muskelatrophie: *AARS, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, REEP1, SCO2, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TBCE, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1* (35 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Erweitertes Gen-Set Bewegungsstörungen: *SEPT9, AARS, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACAT1, ACO2, ACTA1, ACVR1, ADAR, ADCK3, ADCY5, ADGRG1, ADGRG6, ADSS1, AFG3L2, AGL, AGRN, AHCY, AHI1, AIFM1, AIMP1, ALDH18A1, ALDH5A1, ALDOA, ALG14, ALG2, ALS2, AMACR, AMPD1, AMPD2, ANG, ANO10, ANO3, ANO5, ANXA11, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARHGEF10, ARL13B, ARL6, ARL6IP1, ARSA, ASAH1, ASCC1, ATAD3A, ATCAY, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP1A3, ATP2A1, ATP2B3, ATP7A, ATP7B, ATP8A2, ATXN10, ATXN2, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BEAN1, BICD2, BIN1, BSCL2, BSCL2, BTD, BVES, C10ORF2, C12ORF65, C19ORF12, C5ORF42, C9orf72, CA8, CACNA1A, CACNA1A, CACNA1B, CACNA1E, CACNA1G, CACNA1S, CACNB4, CAMTA1, CAPN1, CAPN3, CASK, CASQ1, CAV1, CAV3, CAVIN1, CC2D2A, CCDC78, CCDC88C, CCT5, CEP290, CEP41, CFL2, CHAT, CHCHD10, CHKB, CHMP2B, CHP1, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHUK, CIZ1, CLCN1, CLCN2, CLN5, CLN6, CLPP, CNTN1, CNTNAP1, COA7, COASY, COL12A1, COL13A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ8A, COX10, COX20, COX6A1, CP, CPT1C, CPT2, CRYAB, CSPP1, CSTB, CTDP1, CTNNB1, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS, DARS2, DCAF17, DCAF8, DCC, DCTN1, DCTN2, DES, DGAT2, DGUOK,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Bewegungsstörungen: *DHCR24, DHTKD1, DLAT, DMD, DNA2, DNAJB2, DNAJB5, DNAJB6, DNAJC12, DNAJC19, DNAJC5, DNM2, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DSTYK, DYNC1H1, DYSF, EBF3, ECEL1, EEF2, EGR2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EMD, ENO3, ENTPD1, ERBB3, ERBB4, ERCC5, ERCC6, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FA2H, FAM134B, FARS2, FAT2, FBLN5, FBN2, FBXL4, FBXO38, FDX2, FDXR, FGD4, FGF14, FHL1, FHL2, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNC, FLVCR1, FLVCR2, FMR1, FOLR1, FRRS1L, FTL, FUS, FXN, G6PC, GAA, GALC, GAN, GARS, GBA, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GFAP, GFPT1, GJB1, GJB3, GJC2, GLA, GLDN, GLE1, GLRB, GM2A, GMPPB, GNAL, GNAO1, GNB4, GNE, GOLGA2, GOSR2, GRID2, GRM1, GRN, GSS, GYG1, GYS1, GYS2, HACD1, HACE1, HADHA, HADHB, HARS, HARS2, HEXA, HEXB, HIBCH, HINT1, HK1, HNRNPA1, HNRNPA2B1, HOXD10, HPCA, HSPB1, HSPB3, HSPB8, HSPD1, IBA57, IFIH1, IGHMBP2, IKBKAP, INF2, INPP5E, INPP5K, ISCU, ISPD, ITGA7, ITM2B, ITPR1, KARS, KAT6B, KBTBD13, KCNA1, KCNC3, KCND3, KCNE3, KCNJ10, KCNJ18, KCNJ2, KCNJ5, KCNMA1, KDM5C, KIAA0196, KIAA0586, KIAA1109, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, KIF7, KLHL40, KLHL41, KLHL9, KMT2B, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA5, LAMB2, LAMP2, LARGE,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; Beckman Coulter;

Erweitertes Gen-Set Bewegungsstörungen: *LARGE1, LARS2, LAS1L, LDB3, LDHA, LGI4, LIMS2, LITAF, LMNA, LMNB1, LMOD3, LPIN1, LRP4, LRPPRC, LRSAM1, MAG, MAP3K20, MARS, MARS2, MATR3, MCM3AP, MECR, MED25, MEGF10, MFN2, MGME1, MICU1, MKKS, MKS1, MME, MORC2, MPV17, MPZ, MRE11, MRE11A, MSTN, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTM1, MTMR14, MTMR2, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTPAP, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUSK, MYBPC1, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYO18B, MYO9A, MYOT, MYPN, NAGLU, NALCN, NDRG1, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEB, NEFH, NEFL, NEK1, NEU1, NGF, NIPA1, NKX2-1, NKX2-5, NKX6-2, NOL3, NPC1, NPC2, NPHP1, NT5C2, NTRK1, NUBPL, OFD1, OPA1, OPA3, OPHN1, OPTN, ORAI1, PABPN1, PAH, PANK2, PAX6, PCYT2, PDE10A, PDE6D, PDGFB, PDGFRB, PDHA1, PDHX, PDK3, PDYN, PEX10, PEX2, PEX7, PFKM, PFN1, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PHYH, PIEZO2, PIK3R5, PLA2G6, PLD3, PLEC, PLEKHG5, PLOD2, PLP1, PMM2, PMP2, PMP22, PMPCA, PNKD, PNKP, PNPLA2, PNPLA6, POC1B, POGLUT1, POLG, POLG2, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP2R2B* EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Bewegungsstörungen: *PPP3CA, PRDM12, PREPL, PRF1, PRICKLE1, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRPH, PRPS1, PRRT2, PRX, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RARS, RARS2, RBCK1, REEP1, REEP2, RETREG1, RIPK4, RNASEH2B, RNF170, RNF216, RPGRIP1L, RRM2B, RTN2, RUBCN, RXYLT1, RYR1, SACS, SAMD9L, SBF1, SBF2, SCN10A, SCN11A, SCN2A, SCN4A, SCN8A, SCN9A, SCO2, SCYL1, SELENOI, SELENON, SERAC1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC18A3, SLC19A3, SLC1A3, SLC20A2, SLC22A5, SLC25A1, SLC25A15, SLC25A20, SLC25A4, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC52A2, SLC52A3, SLC5A7, SLC6A3, SLC9A6, SMAD3, SMCHD1, SMN1, SMN2, SNAP25, SNX14, SOD1, SOX10, SPAST, SPEG, SPG11, SPG20, SPG20, SPG21, SPG7, SPG7, SPR, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, STAC3, STIM1, STUB1, SUCLA2, SUCLG1, SURF1, SYNE1, SYNE2, SYT14, SYT2, TAF1, TANGO2, TARDBP, TAZ, TBCE, TBK1, TCAP, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TFG, TGFB3, TGM6, TH, THAP1, TIA1, TK2, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM43, TMEM67, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TPP1, TRAPPC11, TRIM2, TRIM32, TRIM54, TRIM63, TRIP4, TRIP4, TRPA1, TRPC3, TRPM7, TRPV4, TSEN2, TSEN54, TTBK2, TTC19, TTC21B, TTC8, TTN, TTPA, TTR, TUBA4A, TUBB4A, TYMP, UBA1, UBA5, UBAP1, UBQLN2, UCHL1, VAC14, VAMP1, VAPB, VCP, VIPAS39, VLDLR, VMA21, VPS13A, VPS33B, VPS37A, VRK1, WARS1, WDPCP, WDR81, WFS1, WWOX, XK, YARS2, ZBTB42, ZC4H2, ZFYVE26, ZFYVE27, ZNF423 (734 Gene)* EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Adrenale Hyperplasie: *ARMC5, CYP11A1, CYP11B1, CYP17A1, CYP21A2, GNAS, HSD3B2, MC2R, MRAP, NNT, NR0B1, NR3C1, PDE11A, PDE8B, POMC, POR, PRKAR1A, STAR* (18 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Cholestase: *ABCB11, ABCB4, ABCC2, AKR1D1, ATP8B1, BAAT, CFTR, CREB3L3, CYP7B1, DCDC2, DGUOK, EPCAM, FAH, HSD3B7, JAG1, LCT, LMF1, MKS1, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, SCYL1, SERPINA1, SLC25A13, SLC26A3, SMPD1, SPINT2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, VIPAS39, VPS33B* (47 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
MODY: *ABCC8, BLK, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, RFX6 (* 13 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Monogener Diabetes: *ABCC8, BLK, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC16A1, SLC2A2, UCP2, WFS1, ZFP57* (28 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Fertilitätsstörung (männlich): *AK7, AMH, AMHR2, ANOS1, AR, ARMC2, AURKC, BNC2, BRDT, C7orf63, CATSPER2, CCDC108, CCDC39, CFAP43, CFAP44, CFTR, CYP11B1, CYP17A1, DMRT1, DNAH1, DNAH17, DNAH5, DPY19L2, FANCM, FGFR1, FSHB, FSHR, FSIP2, GNRHR, HSD3B2, INSL3, KISS1R, KLHL10, LRRC6, LRRC6, MAMLD1, MEIOB, NANOS1, NR0B1, NR5A1, PLCZ1, PMFBP1, PPP2R3C, PROK2, PROKR2, QRICH2, RSPO1, SEMA3A, SEPT12, SLC26A8, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPATA16, SPEF2, SPINK2, SRD5A2, SUN5, SYCE1, SYCP3, TACR3, TAF4B, TDRD9, TEX11, TEX14, TEX15, TRIM37, TSGA10, TTC18, TTC21A, TTC29, USP26, USP9Y, WDR11, WDR52, WDR66, WDR96, ZMYND15* (81 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Glukokortikoidmangel: *AAAS, AIRE, CYP11A1, MC1R, MC2R, MCM4, MRAP, NNT, NR3C1, POMC, STAR, TXNRD2* (9 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hyperlipidämie: *ABCA1, ABCG5, ABCG8, ALMS1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9* (18 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Hypomagnesiämie: *BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6* (19 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hyperparathyreoidismus: *AIRE, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GCM2, GNA11, MEN1, PTH, RET, TRPV6* (14 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hypothyreose: *CASR, DUOX1, DUOX2, DUOXA2, FOXE1, GCM2, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TTF1, UBR1, ZFAT1* (33 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Hypoglykämie und familiärer Hyperinsulinismus: *ABCC8, ACAT1, ACSF3, AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, MPV17, NHLRC1, OXCT1, PC, PCK1, PDX1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, UCP2* (50 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kallmann-Syndrom: *ARMC5, CYP11A1, CYP11B1, ANOS1, AXL, CDK9, CHD7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, PROK2, PROKR2, SEMA3A, SOX10, SOX2, SOX3, SRA1, SPRY4, TAC3, TACR3, WDR11, CYP21A2, GNAS, HSD3B2, MC2R, MRAP, NNT, NR0B1, NR3C1, PDE11A, PDE8B, POMC, POR, PRKAR1A, STAR* (31 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Monogene Adipositas: *ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R,* *MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP* (36 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Ovarialinsuffizienz: *AR, BMP15, BNC1, CFTR, CLPP, CPEB1, CYP17A1, CYP19A1, DIAPH2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4ENIF1, ERCC6, ESR1, ESR2, F2, F5, FANCM, FIGLA, FMR1, FOXL2, FSHR, GALT, GDF9, GNAS, HARS2, HFM1, HSD17B4, INHA, LARS2, LHCGR, LHX8, LMNA, MCM8, MCM9, MRPS22, MSH5, MTHFR, NANOS3, NLRP2, NLRP5, NOBOX, NR5A1, NUP107, PADI6, PANX1, PATL2, POLG, POR, PROC, PROS1, PSMC3IP, SERPINC1, SERPINE1, SMC1B, SOHLH1, SPIDR, STAG3, STAR, SYCE1, SYCE3, SYCP3, TACR3, THBD, TUBB8, WEE2, WT1, ZP1, ZP2, ZP3* (73 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Störung der Geschlechtsentwicklung: *AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARX, ATRX, BCOR, BMP15, CBX2, CDKN1C, CEP41, CFTR, CHD7, CREBBP, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DUSP6, DYNC2H1, ERCC3, FGF8, FGFR1, FIG4, FRAS1, FSHR, GATA4, GNRHR, HARS2, HSD17B3, HSD17B4, HSD3B2, IL17RD, IRF6, KISS1R, LHCGR, MAMLD1, MAP3K1, MCM9, MKRN3, MKS1, MRPS22, NR0B1, NR5A1, NUP107, POR, PROK2, PROKR2, PSMC3IP, RSPO1, SOHLH1, SOX3, SOX9, SRD5A2, SRY, STAR, TACR3, WNT4, WT1, ZFPM2* (65 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Endokrinologie Erweitertes Gen-Set: *AAAS, ABCA1, ABCB11, ABCB4, ABCC2, ABCC8, ABCG5, ABCG8, ACAT1, ACSF3, ADCY3, AGL, AIRE, AK7, AKR1C2, AKR1C4, AKR1D1, ALDOA, ALDOB, ALMS1, AMH, AMHR2, ANOS1, AP2S1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, AR, ARL6, ARMC2, ARMC5, ARX, ATP8B1, ATRX, AURKC, AXL, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BLK, BMP15, BMP4, BNC1, BNC2, BRDT, BSND, C7orf63, CASR, CATSPER2, CBX2, CCDC108, CCDC39, CDC73, CDK9, CDKN1A, CDKN1B, CDKN1C, CDKN2B, CDKN2C, CEP290, CEP41, CFTR, CHD7, CLCNKB, CLDN16, CLDN19, CLPP, CNNM2, CNNM4, CPEB1, CREB3L3, CREBBP, CTLA4, CUL4B, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, CYP7B1, DCDC2, DGUOK, DHCR7, DHH, DIAPH2, DMRT1, DNAH1, DNAH17, DNAH5, DPY19L2, DUOX1, DUOX2, DUOXA2, DUSP6, DYNC2H1, DYRK1B, EGF, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4ENIF1, ENO3, EPCAM, EPM2A, ERCC3, ERCC6, ESR1, ESR2, FAH, FAM111A, FANCM, FBP1, FEZF1, FGF17, FGF8, FGFR1, FIG4, FIGLA, FLRT3, FMR1, FOXE1, FOXL2, FOXP3, FRAS1, FSHB, FSHR, FSIP2, FXYD2, G6PC, GAA, GALT, GATA4, GATA6, GBE1, GCK, GCM2, GDF9, GHR, GHRHR, GLIS3, GLUD1, GNA11, GNAS, GNRH1, GNRHR, GPIHBP1, GYG1, GYS1, GYS2, HADH, HARS2, HESX1, HFM1, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HS6ST1, HSD17B3, HSD17B4, HSD3B2, HSD3B7, IGSF1, IL17RD, INHA, INS, INSL3, INSR, IRF6, IRS4, IYD, JAG1, KCNA1, KCNJ10, KCNJ11, KISS1, KISS1R, KLF11, KLHL10, KSR2, LAMP2, LARS2, LCT, LDHA, LDLR, LDLRAP1, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LHX8, LIPA, LMF1, LMNA, LPL, LRRC6, MAGEL2, MAGT1, MAMLD1, MAP3K1, MC2R, MC3R, MC4R, MCM4, MCM8, MCM9, MEIOB, MEN1, MKKS, MKRN3, MKS1, MRV17*...	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Endokrinologie Erweitertes Gen-Set: *MRAP, MRPS22, MSH5, MYO5B, NANOS1, NANOS3, NEUROD1, NEUROG3, NHLRC1, NIPA2, NKX2-1, NKX2-5, NNT, NOBOX, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR0B1, NR0B2, NR1H4, NR3C1, NR5A1, NSMF, NTRK2, NUP107, OTX2, OXCT1, PANX1, PATL2, PAX4, PAX8, PC, PCBD1, PCK1, PCSK1, PCSK9, PDE11A, PDE3A, PDE8B, PDX1, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, PFKM, PGAM2, PGK1, PGM1, PHF6, PHKA1, PHKA2, PHKB, PHKG2, PLCZ1, PMFBP1, POLG, POMC, POR, POU1F1, PPARG, PPP2R3C, PRKAG2, PRKAG3, PRKAR1A, PROK2, PROKR2, PROP1, PSMC3IP, PTF1A, PTH, PYGL, PYGM, QRICH2, RBCK1, RET, RFX6, RSPO1, SARS2, SCYL1, SDCCAG8, SECISBP2, SEMA3A, SEPT12, SERPINA1, SIM1, SLC12A3, SLC16A1, SLC16A2, SLC25A13, SLC26A3, SLC26A4, SLC26A8, SLC2A2, SLC37A4, SLC5A5, SMC1B, SMPD1, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPATA16, SPEF2, SPIDR, SPINK2, SPINT2, SPRY4, SRA1, SRD5A2, SRY, STAG3, STAR, SUN5, SYCE1, SYCP3, TAC3, TACR3, TAF4B, TBL1X, TDRD9, TEX11, TEX14, TEX15, TG, THRA, THRB, TJP2, TMEM216, TPO, TRH , TRHR, TRIM32, TRIM37, TRMU, TRPM6, TRPV6, TSGA10, TSHB, TSHR, TTC18, TTC21A, TTC29, TTC37, TTC8, TTF1, TUBB8, TXNRD2, UBR1, UCP2, UCP3, UGT1A1, USP26, USP9Y, VIPAS39, VPS13B, VPS33B, WDPCP, WDR11, WDR52, WDR66, WDR96, WEE2, WFS1, WNT4, WT1, ZFAT1, ZFP57, ZFPM2, ZMYND15, ZP1, ZP2, ZP3* (408 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Epileptische Enzephalopathie: *AARS, ABAT, ADAR, ADSL, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, APOPT1, ARHGEF9, ARV1, ARX, ASNS, ATP6V1A, BRAT1, CACNA1A, CASK, CDKL5, CHD2, CLCN4, CNTNAP2, COX6B1, CPT2, D2HGDH, DCX, DNM1, DNM1L, DOCK7, ECHS1, EEF1A2, ETHE1, FAR1, FARS2, FGF12, FLNA, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, GTPBP3, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HTT, IQSEC2, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, KIF1A, LRPPRC, LYRM7, MBD5, MECP2, MEF2C, MOCS1, MRPL44, MTFMT, MTHFR, NACC1, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NRXN1, NUBPL, PCDH19, PIGA, PLCB1, PNKP, PNPO, POLG, PURA, RMND1, RNASEH2A, RNASEH2B, RORB, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCO1, SDHAF1, SERAC1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TCF4, TREX1, TSC1, TSC2, TTC19, UBA5, UBE3A, UNC80, WDR45, WWOX, ZEB2, CACNA1E, CAD, CAMK2A, CAMK2B, CNNM2, CNPY3, CPLX1, CUX2, CYFIP2, DDX3X, DENND5A, DHDDS, FRRS1L, GABRB2, GABRB1, GNB1, GRIA4, GRIN2D, HACE1, ITPA, KCNQ5, KCNT2, LNPK, MBOAT7, MDH2, MOCS2, NEXMIF, NTRK2, PACS2, PHACTR1, PLPBP, PPP3CA, RHOBTB2, SCN3A, SLC1A2, SLC25A12, TBL1XR1, TRAK1, YWHAG (176 Gene)*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Idiopathisch generalisierte Epilepsie: *ALDH7A1, AMACR, CACNA1A, CACNA1H, CACNB4, CASR, CHRNA2, CHRNA4, CHRNB2, CLCN2, DEPDC5, EFHC1, GABRA1, GABRB3, GABRG2, GRIN2A, HCN1, KCNA1, KCNC1, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, MTOR, NIPA, NPRL2, NPRL3, PCDH19, POLG, PRRT2, RELN, RORB, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC1A3, SLC2A1, SLC6A1, STXBP1, TBC1D24* (43 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Metabolische Epilepsien: *ABAT, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ARG1, BTD, D2HGDH, DHFR, DNM1L, DPYD, DPYS, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GCDH, GCH1, GCSH, GLDC, GNE, GPAA1, GPHN, HIBCH, L2HGDH, LIAS, MOCS1, MTHFR, PGAP1, PGAP2, PGAP3, PGK1, PIGA, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGS, PIGT, PIGV, PIGW, PIGY, PLPBP, PNPO, POLG, PRODH, PTS, QDPR, SERAC1, SLC25A1, SLC25A15, SLC2A1, SLC39A8, SLC46A1, SLC6A9, SUOX* (64 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Myoklonische Epilepsien: *AFG3L2, ASAH1, ATP13A2, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAJC5, EPM2A, GOSR2, GRN, KCNC1, KCTD7, LMNB2, MFSD8, NEU1, NHLRC1, PPT1, PRDM8, PRICKLE1, SCARB2, SERPINI1, TPP1* (27 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Pädiatrische Epilepsie: *ABAT, AARS, ABCD1, ADAR, ADSL, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, AMACR, AMT, ANKRD11, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARG1, ARHGEF9, ARSA, ARV1, ARX, ASAH1, ASNS, ASPA, ATP13A2, ATP1A3, ATP6V1A, ATRX, BRAT1, BTD, CACNA1A, CACNA1H, CACNB4, CASK, CASR, CC2D1A, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL4A1, COX15, COX6B1, CPT2, CSF1R, CSTB, CTC1, CTSD, CTSF, CUL4B, CYP27A1, D2HGDH, DARS, DARS2, DCX, DDC, DEPDC5, DHFR, DNAJC5, DNM1, DNM1L, DOCK7, DPYD, DPYS, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FGF12, FH, FLNA, FOLR1, FOXG1, FOXRED1, GABRA1, GABRB2, GABRB3, GABRG2, GALC, GAMT, GCDH, GCH1, GFAP, GFM1, GJC2, GLB1, GLDC, GLRB, GNAO1, GNB1, GNE, GOSR2, GPAA1, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRN, GTPBP3, HACE1, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HSD17B10, HSPD1, HTRA1, HTT, IBA57, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF1A, L2HGDH, LGI1, LMNB1, LMNB2, LRPPRC, LYRM7, MAGI2, MARS2, MBD5, MECP2, MED12, MEF2C, MFSD8, MLC1, MOCS1, MRPL44, MTFMT, MTHFR, MTOR, NACC1, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEU1, NFU1, NHLRC1, NOTCH3, NRXN1, NUBPL, OFD1, OPHN1, PCDH19, PGK1, PHF6, PGAP1, PGAP2, PGAP3, PIGA, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGS, PIGT, PIGV,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Pädiatrische Epilepsie: *PLCB1, PLP1, PNKP, PNPO, POLG, POLR3A, POLR3B, PPT1, PRDM8, PRICKLE1, PRIMA1, PRODH, PRRT2, PSAP, PTS, PURA, PYCR2, QDPR, RAB39B, RARS, RELN, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A15, SLC25A22, SLC2A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMS, SNAP25, SNORD118, SOX10, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TPP1, TREX1, TSC1, TSC2, TTC19, TUBB4A, UBA5, UBE2A, UBE3A, UNC80, VPS13A, WDR26, WDR45, WWOX, YY1, ZEB2, ZFYVE26, CACNA1E, CAD, CAMK2A, CAMK2B, CNNM2, CNPY3, CPLX1, CUX2, CYFIP2, DDX3X, DENND5A, DHDDS, FRRS1L, GABRB1, GRIA4, GRIN2D, ITPA, KCNQ5, KCNT2, LNPK, MBOAT7, MDH2, MOCS2, NEXMIF, NTRK2, PACS2, PHACTR1, PLPBP, PPP3CA, RHOBTB2, RORB, SCN3A, SLC1A2, SLC25A12, TBL1XR1, TRAK1, YWHAG* (339 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Epilepsien Erweitertes Gen-Set: AARS, ABAT, ABCD1, ACVRL1, ADAR, ADSL, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, AMACR, AMT, ANKRD11, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARG1, ARHGEF9, ARSA, ARV1, ARX, ASAH1, ASNS, ASPA, ATP13A2, ATP1A3, ATP6V1A, ATRX, BRAT1, BTD, CACNA1A, CACNA1E, CACNA1H, CACNB4, CAD, CAMK2A, CAMK2B, CASK, CASR, CC2D1A, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNNM2, CNPY3, CNTNAP2, COL4A1, COX15, COX6B1, CPLX1, CPT2, CSF1R, CSTB, CTC1, CTSD, CTSF, CUL4B, CUX2, CYFIP2, CYP27A1, D2HGDH, DARS, DARS2, DCX, DDC, DDX3X, DENND5A, DEPDC5, DHDDS, DHFR, DNAJC5, DNM1, DNM1L, DOCK7, DPYD, DPYS, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FGF12, FH, FLNA, FOLR1, FOXG1, FOXRED1, FRRS1L, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GALC, GAMT, GCDH, GCH1, GCSH, GFAP, GFM1, GJC2, GLB1, GLDC, GLRB, GNAO1, GNB1, GNE, GOSR2, GPAA1, GPHN, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GTPBP3, HACE1, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HSD17B10, HSPD1, HTRA1, HTT, IBA57, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD7, KDM5C, KIF1A, L2HGDH, LGI1, LIAS, LMNB1, LMNB2, LNPK, LRPPRC, LYRM7, MAGI2, MARS2, MBD5, MBOAT7, MDH2, MECP2, MED12, MEF2C, MFSD8, MLC1, MOCS1, MOCS2, MRPL44, MTFMT, MTHFR, MTOR, NACC1, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4,	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Epilepsien Erweitertes Gen-Set: *NDUFS7, NDUFS8, NDUFV1, NECAP1, NEU1, NEXMIF, NFU1, NHLRC1, NIPA, NOTCH3, NPRL2, NPRL3, NRXN1, NTRK2, NUBPL, OFD1, OPHN1, PACS2, PCDH19, PGAP1, PGAP2, PGAP3, PGK1, PHACTR1, PHF6, PIGA, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGS, PIGT, PIGV, PIGW, PIGY, PLCB1, PLP1, PLPBP, PNKP, PNPO, POLG, POLR3A, POLR3B, PPP3CA, PPT1, PRDM8, PRICKLE1, PRIMA1, PRODH, PROSC, PRRT2, PSAP, PTS, PURA, PYCR2, QDPR, RAB39B, RARS, RELN, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, RORB, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC1A3, SLC25A1, SLC25A12, SLC25A15, SLC25A22, SLC2A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A8, SLC6A9, SLC9A6, SMC1A, SMS, SNAP25, SNORD118, SOX10, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TPP1, TRAK1, TREX1, TSC1, TSC2, TTC19, TUBB4A, UBA5, UBE2A, UBE3A, UNC80, VPS13A, WDR26, WDR45, WWOX, YWHAG, YY1, ZEB2, ZFYVE26* (350 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Anämien: *ADAMTS13, AK1, ALAS2, ALDOA, AMN, ANK1, ATM, ATR, ATRX, BLM, BPGM, BRCA2, BRIP1, C15ORF41, CDAN1, CLCN7, COX4I2, CUBN, CYB5R3, DHFR, DNAJC21, EGLN1, EPAS1, EPB41, EPB42, EPO, EPOR, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PD, GATA1, GCLC, GLRX5, GPI, GSR, GSS, HBA1 , HBA2, HBB, HFE, HK1, HSPA9, JAK2, KCNN4, KIF23, KLF1, LPIN2, MAD2L2, MTR, NBN, NDUFB11, NT5C3A, PALB2, PC, PDHA1, PDHX, PFKM, PGK1, PIEZO1, PKLR, PUS1, RAD51C, REN, RHAG, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, SEC23B, SH2B3, SLC11A2, SLC19A2, SLC25A38, SLC2A1, SLC30A10, SLC4A1, SLX4, SPTA1, SPTB, TCN2, TF, THBD, TMPRSS6, TPI1, TRNT1, TSR2, UROS, VHL, XRCC2, YARS2, GIF* (116 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kinderonkologisches Panel: *AP3B1, BRCA2, CSF3R, CXCR4, DKC1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, MPL, NHP2, NOP10, PALB2, RAB27A, RAC2, RAD51C, RBM8A, RMRP, RPL5, RPL11, RPL15, RPL26, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26, RTEL1, SBDS, SLC37A4, SLX4, SRP72, TAZ, TERC, TERT, TINF2, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53* (59 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Diamond-Blackfan Anämie: *GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7* (20 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Dyskeratosis Congenita: *ACD, AK2, CTC1, DKC1, FERMT1, LIG4, NHP2, NOP10, PARN, RECQL4, RTEL1, TERC, TERT, TINF2, USB1, WRAP53* (16 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Fanconi Anämie: *ATM, ATR, BLM, BRCA2, BRIP1, CXCR4, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, NBN, PALB2, RAD51C, SLX4, UBE2T, XRCC2* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hämophagozytische Lymphohistozytose: *AP3B1, AP3D1, CD27, CD48, FADD, FAS, FASLG, GATA2, ITK, LIPA, LYST, MAGT1, MYO5A, NLRC4, PRF1, RAB27A, RC3H1, RECQL4, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, XIAP* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Hereditäre Leukämie: *ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, CBL, CDKN2A, CEBPA, DDX41, DKC1, EPCAM, ETV6, FANCA, GATA2, HRAS, IKZF1, KRAS, MAP2K1, MAP2K2, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PAX5, PMS2, PTPN11, RIT1, RUNX1, SAMD9L, SBDS, SOS1, SRP72, TERC, TERT, TINF2, TP53* (39 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hermansky-Pudlak-Syndrom: *ABCA3, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1* (23 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Inherited Bone Marrow Failure Syndrom (IBMFS): *ACD, ACTB, AK2, ANKRD26, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, CBL, CDKN2A, CEBPA, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CXCR4, DDX41, DKC1, DNAJC21, DTNBP1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITK, JAGN1, KRAS, LAMTOR2, LYST, MAGT1, MAP2K1, MAP2K2, MKL1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PAX5, PGM3, PMS2, PRF1, PTPN11, RAB27A, RAC2, RAD51C, RBM8A, RECQL4, RIT1, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SH2D1A, SLC37A4, SLX4, SMARCD2, SOS1, SRP72, STX11, STXBP2, TERC, TERT, THPO, TINF2, TP53, UNC13D, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2* (128 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Neutropenie: *ACTB, CD40, CD40LG, CLPB, CSF2RA, CSF3R, CTSC, CXCR2, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, IFNGR2, JAGN1, LAMTOR2, LYST, MKL1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, SRP72, TAZ, USB1, VPS13B, VPS45, WAS, WDR1* (34 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Thrombozytenfunktionsstörungen: *AP3D1, ARPC1B, BLOC1S3, BLOC1S6, DTNBP1, GP1BA, GP1BB, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, NBEAL2, P2RY12, RUNX1, TBXA2R, THPO, WIPF1* (21 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Thrombozytopenie: *ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ANO6, ARPC1B, CD36, CYCS, ETV6, FLI1, STIM1, THPO, FLNA, FYB, GATA1, GFI1B, GGCX, GP1BA, GP1BB, GP9, HOXA11, HRG, ITGA2, ITGA2B, ITGB3, LMAN1, MASTL, MECOM, MPL, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PROC, PROS1, RASGRP2, RBM8A, RUNX1, SERPINC1, SERPIND1, SERPINF2, SLC19A2, SLFN14, SRC, THBD, TPM4, TTC7A, TUBB1, WAS, WIPF1* (52 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Hämatologie Erweitertes Gen-Set: *ABCA3, ABCB7, ABCG5, ABCG8, ACBD5, ACD, ACTB, ACTN1, ADAMTS13, AK1, AK2, ALAS2, ALDOA, AMN, ANK1, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BMP6, BPGM, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, CASP10, CBL, CD36, CD40, CD40LG, CDAN1, CDKN2A, CEBPA, CLCN7, CLPB, COX4I2, CP, CSF2RA, CSF3R, CTC1, CTSC, CUBN, CXCR2, CXCR4, CYB5R3, CYCS, DDX41, DHFR, DKC1, DNAJC21, DTNBP1, EGLN1, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPO, EPOR, ERCC4, ERCC6L2, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FLI1, FLNA, FTL, FTH1, FYB, G6PC3, G6PD,GATA1, GATA2, GCLC, GFI1, GFI1B, GFI1BA, GFI1BB, GGCX, GIF, GINS1, GLRX5, GP1BA, GP1BB, GP6, GP9, GPI, GPR143, GSR, GSS, HAMP, HAX1, HBA1 , HBA2, HBB, HFE, HJV, HK1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HRG, HSPA9, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KCNN4, KIF23, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, MAD2L2, MAGT1, MAP2K1, MAP2K2, MASTL, MCFD2, MECOM, MKL1, MLH1, MLPH, MPIG6B, MPL, MSH2, MSH6, MTHFD1, MTR, MYH9, MYO5A, NBEA, NBEAL2, NBN, NDUFB11, NF1, NHP2, NOP10,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hämatologie Erweitertes Gen-Set: *NRAS, NT5C3A, OCA2, ORAI1, P2RX1, P2RY12, P2RY12, PALB2, PARN, PAX5, PC, PDHA1, PDHX, PFKM, PGK1, PGM3, PIEZO1, PKLR, PLA2G4A, PLAU, PMS2, PRF1, PRKACG, PROC, PROS1, PTPN11, PUS1, RAB27A, RAC2, RAD51C, RASGRP2, RBM8A, RECQL4, REN, RHAG, RIT1, RMRP, RNF168, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SERPINC1, SERPIND1, SERPINF2, SFTPB, SFTPC, SH2B3, SH2D1A, SLC11A2, SLC19A2, SLC25A38, SLC2A1, SLC30A10, SLC37A4, SLC40A1, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCD2, SOS1, SPTA1, SPTB, SRC, SRP54, SRP72, STIM1, STX11, STXBP2, TAZ, TBXA2R, TBXAS1, TCN2, TERC, TERT, TF, TFR2, THBD, THPO, TINF2, TMPRSS6, TP53, TPI1, TPM4, TRNT1, TSR2, TTC7A, TUBB1, TYR, TYRP1, UBE2T, UNC13D, UROS, USB1, VHL, VIPAS39, VKORC1, VPS13B, VPS33B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, YARS2* (311 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Adams-Oliver-Syndrom: *ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1* (8 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Cutis Laxa: *ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E, ATP7A, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1, SLC2A10* (11 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Ektodermale Dysplasie: *ANTXR1, APCDD1, AXIN2, BCS1L, CDH3, CDSN, CTSC, DLX3, DSG4, DSP, DYNC2LI1, EDA, EDAR, EDARADD, ERCC2, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IFT43, IFT122, IFT52, JUP, KCTD1, KRT14, KRT71, KRT74, KRT81, KRT83, KRT85, KRT86, LIPH, LPAR6, LRP6, LSS, LTBP3, MPLKIP, MSX1, NFKBIA, OFD1, PAX9, PKP1, POC1A, PORCN, PRKD1, NECTIN1, NECTIN4, RMRP, RPL21, SNRPE, SOX18, ST14, TP63, TRPS1, UBR1, WDR19, WDR35, WNT10A, WNT10B* (68 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Epidermolysis bullosa: *ATP2C1, CDSN, COL17A1, COL7A1, CSTA, DSG1, DSG2, DSG4, DSP, DST, EXPH5, FERMT1, GRIP1, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5* (29 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Ichthyosis: *ABCA12, AAGAB, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ATP2A2, ATP2C1, CARD14, CDSN, CERS3, CSTA, CTSC, CYP4F22, DSG1, DSP, EBP, ELOVL4, ENPP1, ERCC2, ERCC3, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2E2, GTF2H5, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, MPLKIP, MVK, NIPAL4, NSDHL, PEX7, PHYH, PNPLA1, SDR9C7, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, ST14, STS, SULT2B1, SUMF1, TGM1, TGM5, TRPV3, WNT10A* (66 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Neurofibromatose: *LZTR1, NF1, NF2, SMARCB1, SPRED1* (5 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Pachyonychia congenita: *AAGAB, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3* (7 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Palmoplantare Keratodermie: *AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJB2, GJB4, GJB6, JUP, KRT1, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, PKP1, SERPINB7, SLURP1, TRPV3, WNT10A* (25 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Progerie und Progeroid Syndrom: *AGPAT2, ALDH18A1, ATP6V0A2, B4GALT7, BANF1, BLM, BSCL2, C1R, C1S, CAV1, COL3A1, CTC1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GTF2E2, GTF2H5, GORAB, KCNJ6, LMNA, MPLKIP, OBFC1, PIK3R1, PLIN1, PNPLA2, POLD1, PPARG, PSMB8, PTDSS1, CAVIN1, PYCR1, RECQL4, SLC25A24, WRN, ZMPSTE24* (40 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Xeroderma Pigmentosum: *BLM, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FERMT1, GTF2E2, GTF2H5, MPLKIP, POLH, RECQL4, USB1, UVSSA, XPA, XPC* (19 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Erweitertes Gen-Set Dermatologie: *AAGAB, ABCA12, ABCA3, ABHD5, ACD, AGPAT2, AK2, ALDH18A1, ALDH3A2, ALOX12B, ALOXE3, ANTXR1, AP1S1, AP3B1, AP3D1, APCDD1, AQP5, ARHGAP31, ATP2A2, ATP2C1, ATP6V0A2, ATP6V1A, ATP7A, AXIN2, B4GALT7, BANF1, BCS1L, BLM, BLOC1S3, BLOC1S6, BSCL2, C10ORF11, C1R, C1S, CARD14, CAV1, CAVIN1, CDH3, CDSN, CERS3, COL17A1, COL3A1, COL7A1, CSTA, CTC1, CTSC, CYP4F22, DDB2, DKC1, DLL4, DLX3, DOCK6, DSG1, DSG2, DSG4, DSP, DST, DTNBP1, DYNC2LI1, EBP, EDA, EDAR, EDARADD, EDN3, EDNRB, EFEMP2, ELN, ELOVL4, ENPP1, EOGT, EPG5, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, EVC, EVC2, EXPH5, FBLN5, FBN1, FERMT1, FGF10, FGFR2, FGFR3, FLG, FRMD7, GJA1, GJB2, GJB3, GJB4, GJB6, GORAB, GPR143, GRHL2, GRIP1, GTF2E2, GTF2H5, HOXC13, HPS1, HPS3, HPS4, HPS5, HPS6, HR, IFT122, IFT43, IFT52, ITGA3, ITGA6, ITGB4, JUP, KCNJ6, KCTD1, KDSR, KIT, KITLG, KLHL24, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT5, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT81, KRT83, KRT85, KRT86, KRT9, LAMA3, LAMB3, LAMC2, LIPH, LMNA, LOR, LPAR6, LRP6, LSS, LTBP3, LTBP4, LYST, LZTFL1, LZTR1, MAB21L2, MAF, MAK, MBTPS2, MC1R, MERTK, MFN2, MFRP, MFSD8, MIP, MIR184, MITF, MKKS, MKS1, MLPH, MMACHC, MPLKIP, MSX1, MT-ND1, MT-ND4, MT-ND6, MTTP, MVK, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NDUFS1, NECTIN1, NECTIN4, NEK2, NF1, NF2, NFKBIA, NHP2, NHS, NIPAL4* *NFKBIA, NHP2, NHS, NIPAL4, NMNAT1, NOP10, NOTCH1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NSDHL, NTF4, NYX, OAT, OBFC1, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPTN, OTX2, OVOL2, P3H2 (LEPREL1), PADI3, PANK2, PARN, PAX2, PAX3, PAX6, PAX9, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PERP, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHOX2A, PHYH, PIGL, PIK3R1, PIKFYVE, PITPNM3, PITX2, PITX3, PKP1, PLA2G5, PLEC, PLIN1, PNPLA1, PNPLA2, PNPLA6, POC1A, POC1B, POLD1, POLG, POLH, POMGNT1, POMT1, PORCN, PPARG, PQBP1, PRCD, PRDM13, PRDM5, PRKCG, PRKD1, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PSMB8, PTDSS1, PTPN11, PXDN, PYCR1, RAB27A, RAB28, RAB3GAP1, RAF1, RARB, RAX, RAX2, RBP3, RBP4, RBPJ, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, RMRP, ROBO3, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPL21, RPS19, RRM2B, RS1, RTEL1, RTN4IP1, SAG, SALL4, SAMD11, SBF2, SDCCAG8, SDR9C7, SEMA4A, SERPINB7, SETX, SFTPB, SFTPC, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A24, SLC25A4, SLC25A46, SLC27A4, SLC2A10, SLC33A1, SLC38A8, SLC45A2, SLC4A11, SLC52A2, SLC7A14, SLURP1, SMARCB1, SMCHD1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Erweitertes Gen-Set Dermatologie: *NFKBIA, NHP2, NHS, NIPAL4, NMNAT1, NOP10, NOTCH1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NSDHL, NTF4, NYX, OAT, OBFC1, OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW, OPN1MW, OPTN, OTX2, OVOL2, P3H2 (LEPREL1), PADI3, PANK2, PARN, PAX2, PAX3, PAX6, PAX9, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PERP, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHOX2A, PHYH, PIGL, PIK3R1, PIKFYVE, PITPNM3, PITX2, PITX3, PKP1, PLA2G5, PLEC, PLIN1, PNPLA1, PNPLA2, PNPLA6, POC1A, POC1B, POLD1, POLG, POLH, POMGNT1, POMT1, PORCN, PPARG, PQBP1, PRCD, PRDM13, PRDM5, PRKCG, PRKD1, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PSMB8, PTDSS1, PTPN11, PXDN, PYCR1, RAB27A, RAB28, RAB3GAP1, RAF1, RARB, RAX, RAX2, RBP3, RBP4, RBPJ, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, RMRP, ROBO3, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPL21, RPS19, RRM2B, RS1, RTEL1, RTN4IP1, SAG, SALL4, SAMD11, SBF2, SDCCAG8, SDR9C7, SEMA4A, SERPINB7, SETX, SFTPB, SFTPC, SH3PXD2B, SHH, SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A24, SLC25A4, SLC25A46, SLC27A4, SLC2A10, SLC33A1, SLC38A8, SLC45A2, SLC4A11, SLC52A2, SLC7A14, SLURP1, SMARCB1, SMCHD1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Erweitertes Gen-Set Dermatologie: *SNAP29, SNRNP200, SNRPE, SNX10, SOX10, SOX18, SOX2, SOX3, SPATA7, SPG7, SPINK5, SPP2, SPRED1, ST14, STRA6, STS, SUFU, SULT2B1, SUMF1, SUOX, TACSTD2, TBK1, TCF4, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TEK, TENM3, TERC, TERT, TFAP2A, TGFBI, TGM1, TGM5, TIMM8A, TIMP3, TINF2, TK2, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMEM98, TOPORS, TP63, TRAF3IP1, TREX1, TRIM32, TRPM1, TRPS1, TRPV3, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TTR, TUB, TUBB3, TULP1, TYMP, TYR, TYRP1, UBIAD1, UBR1, USB1, USH1C, USH1G, USH2A, UVSSA, VAX1, VCAN, VIM, VPS13B, VSX1, VSX2, WDPCP, WDR19, WDR35, WDR36, WFS1, WNT10A, WNT10B, WRAP53, WRN, XPA, XPC, YME1L1, ZEB1, ZIC2, ZMPSTE24, ZNF408, ZNF423, ZNF469, ZNF513* (466 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Aicardi-Goutières-Syndrom: *ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1* (7 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Coffin-Siris-Syndrom: *ARID1A, ARID1B, ARID2, DPF2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SOX11, SOX4* (10 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Cornelia-de-Lange-Syndrom: *ANKRD11, AFF4, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, UBE2A* (9 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Holoprosenzephalie: *CDON, CNOT1, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TDGF1, TGIF1, ZIC2* (14 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Joubert-Syndrom: *AHI1, ARL13B, ARMC9, B9D1, B9D2, C2CD3, C21ORF2, C5ORF42, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423* (39 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kabuki-Syndrom: *CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX5* (7 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Leukodystrophien / -enzephalopathien: *AARS, AARS2, ABCD1, ACOX1, ADAR, ADPRHL2, AIFM1, AIMP1, AIMP2, ALDH3A2, AMT, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARSA, ASPA, BCAP31, BOLA3, BOLA3, CARS2, CLCN2, COASY, COL4A1, COL4A2, COX10, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMC1, EPRS, ERCC6, ERCC8, FA2H, FAM126A, FARS2, FKRP, FKTN, FOLR1, FOXRED1, FUCA1, GALC, GAN, GBE1, GCDH, GCSH, GEMIN4, GFAP, GFM1, GJC2, GLA, GLB1, GLDC, GLRX5, GLRX5, GMPPB, HEPACAM, HEXA, HIBCH, HIKESHI, HSD17B4, HSPD1, HTRA1, IBA57, IDS, IFIH1, ISCA1, ISCA2, KARS, KCNT1, KIF5A, L2HGDH, LAMA2, LARGE1, LIAS, LIPT1, LIPT2, LMNB1, LRPPRC, LYRM7, MARS2, MARS2, MCOLN1, MLC1, MPV17, MRPL44, MTFMT, MTHFS, NACC1, NARS2, NAXE, NDUFAF5, NDUFS1, NDUFV1, NEU1, NFU1, 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, POMT2, PPT1, PSAP, PSAT1, PYCR2, QARS, RAB11B, RARS, RARS2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, SAMHD1, SCO1, SCP2, SDHA, SDHAF1, SERAC1, SLC16A2, SLC17A5, SLC1A4, SLC25A12, SLC33A1, SLC6A9, SNORD118, SOX10, SPG11, SPTAN1, STN1, SUMF1, SURF1, TACO1, TARS2, TBCD, TBCK, TGFBI, TMEM106B, TPP1, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TTC19, TUBB4A, TUFM, TYMP, TYROBP, UBTF, UFC1, UFM1, VARS, VARS2, VPS11, WARS2, WDR45, WDR45B, ZFYVE26, ZNHIT3 (205 Gene)*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lissenzepahlie / neuronale Migrationssstörungen: *ACTB, ACTG1, ADGRG1, AKT3, APAF1, ARF1, ARFGEF2, ARX, ATP6V0A2, B3GALNT2, B4GAT1, CCND2, CDK5, COL4A1, COL4A2, CRADD, CSNK2A1, CTNNA2, CTU2, DAG1, DCHS1, DCX, DDX3X, DYNC1H1, EMX2, ERMARD, FAT4, FH, FKRP, FKTN, FLNA, GMPPB, GRIN1, GRIN2B, ISPD, KATNB1, KIF1B, KIF2A, KIF5C, LAMB1, LAMC3, LARGE1, MACF1, 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, YWHAE* (80 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Makrozephalie / Overgrowth: *AKT1, AKT2, AKT3, APC2, ASPA, ASXL2, BRAF, BRWD3, CCDC88C, CCND2, CDKN1C, CHD3, CHD4, CHD8, CRADD, CUL4B, DHCR24, DIS3L2, DNMT3A, DVL1, DVL3, EED, EIF2B5, EZH2, FIBP, FOXP1, GCDH, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HERC1, HIST1H1E, HRAS, HUWE1, IGF2, KIAA0196, KIF7, KLLN, KPTN, KRAS, LBR, L1CAM, MAP2K1, MAP2K2, MED12, MLC1, MPDZ, MTOR, NFIB, NFIX, NRAS, NONO, NSD1, OFD1, PAK1, PHF6, PIGA, PIGN, PIGT, PIGV, PIK3CA, PIK3R2, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RAF1, RIT1, RIN2, RNF125, RNF135, SEC23B, SETD2, SHOC2, SOS1, SOS2, SUFU, STRADA, SYN1, TBC1D7, TMCO1, TSC1, TSC2, UPF3B, WASHC5, WNT5A* (92 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Mikrozephalie: *AKT3, AMPD2, ANKLE2, ASNS, ASPM, ATR, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP63, CHMP1A, CIT, CLP1, COASY, DONSON, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, EXOSC8, EXOSC9, FOXG1, GFM1, IER3IP1, KANSL1, KAT6A, KATNB1, KIF11, KIF14, KNL1, LIG4, MBD5, MCPH1, MECP2, MED17, MFSD2A, MRE11A, MYCN, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NSMCE3, NUP37, OPHN1, PAFAH1B1, PCLO, PCNT, PHC1, PHGDH, PLK4, PNKP, POMT1, PPP1R15B, PQBP1, RAB18, RARS2, RBBP8, RELN, RTTN, SASS6, SEPSECS, SLC25A19, SLC25A46, SLC9A6, STAMBP, STIL, TBC1D23, TOE1, TOP3A, TRAIP, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TUBB2B, TUBGCP4, TUBGCP6, VLDLR, VPS53, VRK1, WDR62, WDR73, WDR4, XRCC4, ZNF33* (94 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Rett- und Rett-like Syndrom: *AARS, ABAT, ADAR, ADSL, ALDH7A1, ALG13, AMT, ANKRD11, AP3B2, APOPT1, ARHGEF9, ARV1, ARX, ASNS, ATP6V1A, BRAT1, CACNA1A, CASK, CDKL5, CHD2, CLCN4, CNTNAP2, COX6B1, CPT2, D2HGDH, DCX, DNM1, DNM1L, DOCK7, ECHS1, EEF1A2, ETHE1, FAR1, FARS2, FGF12, FLNA, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, GTPBP3, HCN1, HECW2, HEPACAM, HIBCH, HNRNPU, HTT, IQSEC2, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, KIF1A, LRPPRC, LYRM7, MBD5, MECP2, MEF2C, MOCS1, MRPL44, MTFMT, MTHFR, NACC1, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NRXN1, NUBPL, PCDH19, PIGA, PLCB1, PNKP, PNPO, POLG, PURA, RMND1, RNASEH2A, RNASEH2B, RORB, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCO1, SDHAF1, SERAC1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TCF4, TREX1, TSC1, TSC2, TTC19, UBA5, UBE3A, UNC80, WDR45, WWOX, ZEB2, CACNA1E, CAD, CAMK2A, CAMK2B, CNNM2, CNPY3, CPLX1, CUX2, CYFIP2, DDX3X, DENND5A, DHDDS, FRRS1L, GABRB2, GABRB1, GNB1, GRIA4, GRIN2D, HACE1, ITPA, KCNQ5, KCNT2, LNPK, MBOAT7, MDH2, MOCS2, NEXMIF, NTRK2, PACS2, PHACTR1, PLPBP, PPP3CA, RHOBTB2, SCN3A, SLC1A2, SLC25A12, TBL1XR1, TRAK1, YWHAG* (176 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Zerebral Kavernöse Fehlbildungen: *CCM2, KRIT1, PDCD10, RASA1, EPHB4* (5 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Entwicklungsstörungen Erweitertes Gen-Set: *AARS, AARS2, ABCC8, ABCD1, ABCD3, ACOX1, ACSL4, ACTB, ACTG1, ACY1, ADAMTSL2, ADAR, ADCK3, ADGRG1, ADNP, ADPRHL2, ADSL, AFF2, AGA, AGPS, AGXT, AHI1, AIFM1, AIMP1, AIMP2, AKT1, AKT2, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMPD2, AMT, ANK2, ANKLE2, ANKRD11, ANO10, ANTXR2, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, APAF1, APC2, APOL2, APOL4, APOPT1, APTX, ARF1, ARFGEF2, ARG1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARMC9, ARSA, ARSB, ARX, ASAH1, ASH1L, ASNS, ASPA, ASPM, ASXL2, ASXL3, ATP13A2, ATP6AP2, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GLCT, B4GALT1, B4GAT1, B9D1, B9D2, BAZ2B, BCAP31, BCKDK, BCL11A, BCOR, BDNF, BOLA3, BRAF, BRD4, BRWD3, BTD, C21ORF2, C2CD3, C5ORF42, CACNA1D, CACNA1H, CACNA2D3, CAD, CARS2, CASK, CC2D2A, CCDC115, CCDC88C, CCM2, CCND2, CDK5, CDK5RAP2, CDK6, CDKL5, CDKN1C, CDON, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CHD2, CHD3, CHD4, CHD7, CHD8, CHI3L1, CHMP1A, CIC, CIT, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLP1, CNOT1, CNOT3, CNTN4, CNTNAP2, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COMT, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, COX10, COX15, COX6B1, CRADD, CSF1R, CSNK2A1, CSPP1, CTC1, CTNNA2, CTNND2, CTNS, CTSA, CTSC, CTSD, CTSK, CTU2, CUL3, CUL4B, CUX1, CYP27A1, D2HGDH, DAG1, DAOA, DARS, DARS2, DCHS1, DCX, DDC, DDOST, DDX3X, DEAF1, DHCR24, DHCR7, DHDDS, DIP2C, DIS3L2, DKC1, DLG3, DNMT3A, DOLK, DONSON, DPAGT1, DPF2, DPM1, DPM2,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Entwicklungsstörungen Erweitertes Gen-Set: *DPM3, DPYD, DRD3, DSCAM, DVL1, DVL3, DYM, DYNC1H1, DYRK1A, EARS2, EBP, EED, EFTUD2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4E, ELK1, EMC1, EMX2, EPHB4, EPRS, ERBB2IP, ERCC6, ERCC8, ERMARD, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, EXOSC9, EYA1, EZH2, FA2H, FAM126A, FANCB, FARS2, FAT4, FGD1, FGF8, FGFR1, FH, FIBP, FKRP, FKTN, FLNA, FLNB, FMR1, FOLR1, FOXG1, FOXH1, FOXP1, FOXP2, FOXRED1, FTSJ1, FUCA1, FUT8, GAA, GABRB3, GAD1, GALC, GALNS, GAMT, GAN, GBA, GBE1, GCDH, GDI1, GEMIN4, GFAP, GFM1, GIGYF2, GJC2, GK, GLA, GLB1, GLDC, GLI2, GLI3, GLRX5, GM2A, GMPPA, GMPPB, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPSM2, GRIA1, GRIA3, GRIN1, GRIN2B, GRIP1, GUSB, HCCS, HDAC8, HEPACAM, HERC1, HEXA, HEXB, HGSNAT, HIBCH, HIKESHI, HIST1H1E, HNRNPH2, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTR2A, HTRA1, HUWE1, HYAL1, IBA57, IDS, IDUA, IER3IP1, IFIH1, IGBP1, IGF2, IL1RAPL1, ILF2, INPP5E, INTS6, IQSEC2, IRF2BPL, IRF6, ISCA1, ISCA2, ISPD, KANSL1, KARS, KAT2B, KAT6A, KATNAL2, KATNB1, KCNA2, KCNQ2, KCNT1, KDM5B, KDM5C, KDM6A, KIAA0196, KIAA0556, KIAA0586, KIAA0753, KIAA2022, KIF11, KIF14, KIF1B, KIF2A, KIF5A, KIF5C, KIF7, KLF8, KLLN, KMT2A, KMT2B, KMT2C, KMT2D, KNL1, KPTN, KRAS, KRIT1, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LBR, LDB3, LEO1, LIG4, LIPA, LIPT2, LMNB1, LRPPRC, LYRM7, MACF1, MACROD2, MAGEL2, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MARS2, MBD5, MBOAT7, MBTPS2, MCOLN1, MCPH1, MECP2, MED12, MED13, MED13L, MED17, MEF2C, MET, MFSD2A, MFSD8, MGAT2, MID1, MKS1, MLC1, MOCS1, MOCS2, MOGS, MPDU1, MPDZ, MPI, MPV17, MRE11A, MRPL44, MTFMT* …	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Entwicklungsstörungen Erweitertes Gen-Set: *MTHFR, MTHFS, MTM1, MTOR, MYCN, MYOT, MYT1L, NAA15, NACC1, NAGA, NAGLU, NARS2, NAXE, NCAPD2, NCAPD3, NCAPH, NCKAP1, NCOR1, NDE1, NDP, NDUFA1, NDUFAF5, NDUFS1, NDUFV1, NEDD4L, NEU1, NFIB, NFIX, NFU1, NGLY1, NHEJ1, NHS, NIN, NIPBL, NKX6-2, NLGN3, NLGN4X, NODAL, NONO, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NRAS, NRXN1, NSD1, NSDHL, NSMCE3, NTNG1, NUBPL, NUP37, NUS1, NXF5, OCLN, OCRL, OFD1, OPHN1, OTC, PAFAH1B1, PAK1, PAK3, PC, PCBD1, PCDH19, PCLO, PCNT, PDCD10, PDE6D, PDHA1, PDSS1, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PGM1, PHC1, PHF3, PHF6, PHF8, PHGDH, PHYH, PI4KA, PIBF1, PIGA, PIGN, PIGT, PIGV, PIK3CA, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLK4, PLP1, PMM2, PMPCB, PNKP, POGZ, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PORCN, PPP1CB, PPP1R15B, PPP2R5B, PPP2R5C, PPP2R5D, PPT1, PQBP1, PRODH, PRPS1, PRUNE1, PSAP, PSAT1, PTCH1, PTCHD1, PTEN, PYCR2, QARS, QDPR, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAC3, RAD21, RAF1, RAI1, RANBP17, RARS, RARS2, RASA1, RBBP8, RBM10, RELN, RFT1, RIMS1, RIN2, RIT1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF125, RNF135, RNF216, RPGRIP1L, RPL10, RPS6KA3, RTN4R, RTTN, RXYLT1, SAMHD1, SASS6, SCN2A, SCN8A, SCN9A, SCO1, SCP2, SDHA, SDHAF1, SEC23B, SEPSECS, SERAC1, SET, SETD2, SETD5, SGSH, SHANK2, SHANK3, SHH, SHOC2, SHROOM4, SIX3, SIX5, SLC16A2, SLC17A5, SLC1A4, SLC25A12, SLC25A15, SLC25A19, SLC25A26, SLC25A46, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SLC46A1,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Entwicklungsstörungen Erweitertes Gen-Set: *SLC6A8, SLC9A6, SLC9A9, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMPD1, SMS, SNORD118, SOS1, SOS2, SOX10, SOX11, SOX3, SOX4, SPAST, SPG11, SPTAN1, SRCAP, SRD5A3, SRPX2, SRSF11, SSR4, STAMBP, STIL, STN1, STRADA, STT3A, STT3B, STXBP1, SUFU, SUGCT, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYP, TACO1, TAF1, TAOK2, TARS2, TBC1D20, TBC1D23, TBC1D7, TBCD, TBCK, TBL1XR1, TBR1, TCF20, TCF4, TCTN1, TCTN2, TCTN3, TDGF1, TECPR2, TGFBI, TGIF1, TIMM8A, TMCO1, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM67, TMLHE, TMTC3, TNRC6B, TOE1, TOP3A, TPH1, TPP1, TRAIP, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM37, TRIO, TRIP12, TRMT10A, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSPAN7, TTC19, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TUFM, TUSC3, TYMP, TYROBP, UBE2A, UBE3A, UBN2, UBTF, UFC1, UFM1, UPF3B, USP15, USP7, USP9X, VARS, VARS2, VLDLR, VPS11, VPS33A, VPS53, VRK1, WAC, WARS2, WASHC5, WDFY3, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WNT5A, XRCC4, YWHAE, ZC4H2, ZCCHC12, ZDHHC9, ZFYVE26, ZIC2, ZNF335, ZNF41, ZNF423, ZNF674, ZNF711, ZNF81, ZNHIT3* (809 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Hörstörungen: *ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANKH, ARSG, ATP2B2, ATP6V1B1, ATP6V1B2, BCAP31, BCS1L, BDP1, BRAF, BSND, BTD, C10ORF2, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC42, CDH23, CDK9, CDKN1C, CEACAM16, CEMIP, CEP250, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, CLRN3, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, ECE1, EDN3, EDNRA, EDNRB, EIF3F, ELMOD3, EPS8, EPS8L2, ERCC2, ERCC3, ESPN, ESRRB, EXOSC2, EYA1, EYA4, FAM136A, FAM65B, FAS, FDXR, FGF3, FGFR3, FITM2, FOXI1, GATA3, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB4, GJB6, GNPTAB, GNPTG, GPR98, GPRASP2, GPSM2, GRHL2, GRXCR1, GRXCR2, GSTP1, HAL, HARS, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, LARS2, LHFPL5, LHX3, LMX1A, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MET, MGP, MIR96, MITF, MPZL2, MSRB3, MTAP, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH14, MYH9, MYO15A, MYO1A, MYO1C, MYO1F, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, NR2F1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, OTOR, P2RX2, PAX3, PCDH15, PDE1C, PDZD7 , PEX1, PEX26, PEX26, PEX3, PEX5, PEX6, PISD, PJVK, PMP22, PNPT1, POLD1, POLR1C, POLR1D,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Hörstörungen: *POU3F4, POU4F3, PRPS1, PTPN11, PTPRQ, RAF1, RDX, RMND1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SOX2, SPATA5, SPINK5, STAG2, STRC, SUCLA2, SUCLG1, SYNE4, SYT2, TBC1D24, TBL1X, TCF21, TCOF1, TECTA, TFAP2A, TFCP2, TIMM8A, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TMPRSS5, TNC, TPRN, TRIOBP, TRMU, TSHZ1, TSPEAR, TUBB4B, TYR, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN, XYLT2* (285 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Hydrops Fetalis: *AHCY, AKT3, ALG1, ALG12, ALG8, ALG9, ALPK3, AP3B1, ARSA, ARSB, ASAH1, BLOC1S3, BRAF, BRIP1, BSND, CALCRL, CBL, CCBE1, CEP120, CHRNA1, CHRND, CHRNG, CLCNKA, CLCNKB, CLN3, CLN5, CLN6, CLN8, CNBP, CTNS, CTSA, CTSD, CTSK, DHCR7, DIS3L2, DMPK, DTNBP1, EBP, EPHB4, ERBB3, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT4, FH, FLT4, FOXC2, FOXP3, FUCA1, G6PC, G6PD, GAA, GALC, GALNS, GBA, GBE1, GLA, GLB1, GLE1, GLUL, GM2A, GNPTAB, GNPTG, GNS, , GUSB, HADH, HADHA, HADHB, HBA1, HBA2, HBZ, HDAC8, HEXA, HEXB, HFE, HGSNAT, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HYAL1, IDS, IDUA, ITGA9, KIAA0586, KIF11, KLF1, KRAS, LAMP2, LAMP3, LARS2, LBR, LIPA, LMOD3, LZTR1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MCOLN1, MFSD8, MVK, NAGA, NAGLU, NEB, NEK1, NEK9, NEU1, NF1, NIPBL, NPC1, NPC2, NR1H4, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PIGA, PIK3CA, PIK3R2, PKLR, PLD1, PMM2, PPT1, PSAP, PSAT1, PTH1R, PTPN11, RAD21, RAF1, RASA1, RIT1, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RYR1, SEC23B, SGPL1, SGSH, SHOC2, SLC17A5, SLC22A5, SLC26A2, SMC1A, SMC3, SMPD1, SOS1, SOS2, SOX18, SPRED1, SUMF1, TALDO1, TAPT1, TAZ, THSD1, TPP1, TRIP11, TSC1, TSC2, UROS, VEGFC, VSX1, WDR35* (192 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Antikörpermangel: *AICDA, ATP6AP1, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CXCR4, HELLS, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IL2RG, IRF2BP2, LRBA, LRRC8A, MS4A1, NFKB1, NFKB2, NFKBIA, PIK3CD, PIK3R1, SH3KBP1, STAT1, TCF3, TNFRSF13B, TNFRSF13C, UNG* (35 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ~~Beckman Coulter~~; ABI SeqStudio 8 Flex Genetic Analyzer System
Autoimmunes lymphoproliferatives Syndrom und Dysregulation des Immunsystems: *AP3B1, AP3D1, CASP10, CASP8, CD27, CD70, CTLA4, CYBA, CYBB, DKC1, FADD, FAS, FASLG, FOXP3, GATA2, IL10, IL10RA, IL10RB, IL21, IL21R, IL2RA, ITK, KRAS, LRBA, LYST, MAGT1, MYO5A, NCF1, NCF2, NCF4, PRF1, PRKCD, RAB27A, RECQL4, RTEL1, SH2D1A, STAT1, STAT3, STX11, STXBP2, TTC7A, UNC13D, XIAP, RIPK1* (44 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Autoinflammatorisches Syndrom: *ACP5, ADA, ADAM17, ADAR, BACH2, CARD14, CECR1, COPA, DDX58, ELANE, FOXP3, HSPA1L, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL36RN, ISG15, LPIN2, LRBA, MEFV, MVK, NFAT5, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSENEN, PSMB8, PSTPIP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, TRNT1, TTC7A, WDR1, XIAP, ZBTB24* (50 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Granulomatose-Phagozytosedefekte: *CD40, CD40LG, CFTR, CLPB, CSF3R, CXCR2, CXCR4, CYBA, CYBB, ELANE, FERMT3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, HAX1, IFNGR1, IFNGR2, IL12B, IL12RB1, IRF8, ISG15, ITGB2, JAGN1, MKL1, NCF1, NCF2, NCF4, NOD2, RAC2, RORC, SBDS, SLC35C1, SMARCD2, SRP54, STAT1, TAZ, TCIRG1, TYK2, USB1, VPS45, WAS, WDR1* (45 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Komplementdefekte (Neisseria): *ADIPOQ, ADIPOR1, ADIPOR2, ARMC4, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C3, C3AR1, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD46, CD55, CD59, CD93, CFB, CFD, CFH, CFI, CFP, CLU, COLEC11, CR2, CRP, DGKE, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, FCN1, FCN2, FCN3, HYDIN, LRRC6, MASP1, MASP2, MAT2A, NME8, OFD1, PIGA, PTX3, RSPH1, RSPH4A, RSPH9, SERPING1, SPAG1, THBD, VSIG4, VTN, ZMYND10* (75 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Schwerwiegende kombinierte Immundefekte: *ADA, AK2, ATM, BACH2, BCL11B, BLM, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CHD7, CIITA, CORO1A, CTPS1, DCLRE1C, DNMT3B, DOCK2, DOCK8, EPG5, FOXN1, IFNGR1, IKBKB, IL12RB1, IL21, IL21R, IL2RA, IL2RG, IL7R, IRF8, ITGB2, ITK, JAK3, LAT, LCK, LIG4, LRBA, MAGT1, MALT1, MAP3K14, MSN, MTHFD1, NFAT5, NHEJ1, NSMCE3, ORAI1, PARN, PGM3, PIK3CD, PMS2, PNP, POLE, POLE2, PRKDC, PTPRC, RAC2, RAG1, RAG2, RASGRP1, RFX5, RFXANK, RFXAP, RHOH, RLTPR, RMRP, RTEL1, SEMA3E, SH2D1A, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TFRC, TNFRSF4, TYK2, UNC119, WAS, ZAP70* (91 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Syndrome und intrinsische/angeborene Defekte der Immunität: *AIRE, C2, C3, CARD9, CLEC7A, CPT2, DNASE1, DNASE1L3, DNASE2, DOCK8, DSG1, ERBB2IP, FOXP3, IFIH1, IFNGR1, IFNGR2, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL6ST, IRF3, IRF8, ISG15, JAK1, MEFV, NUP214, POMP, PRKCD, PROCR, PSMA3, PSMB4, PSMB8, PSMB9, RANBP2, RNASEH2A, RNASEH2B, RNASEH2C, RORC, SAMHD1, SPINK5, STAT1, STAT2, STAT3, STAT5B, TBK1, TICAM1, TLR3, TLR5, TMEM173, TRAF3, TRAF3IP2, TREX1, TYK2, UNC93B1, USP18, ZNF341* (58 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Immunologie erweitertes Gen-Set: *ACD, ACP5, ACTB, ADA, ADAM17, ADAR, ADIPOQ, ADIPOR1, ADIPOR2, AICDA, AIRE, AK2, ANKRD26, AP3B1, AP3D1, ARMC4 , ARPC1B, ATM, ATP6AP1, ATR, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C21ORF59, C3, C3AR1, C4A, C4B, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CBL, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CD93, CDCA7, CDKN2A, CEBPA, CEBPE, CECR1, CENPF, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLEC7A, CLPB, CLU, COLEC11, COPA, CORO1A, CPT2, CR2, CRP, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, DCLRE1C, DDX41, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DRC1, DSG1, DTNBP1, DYX1C1, ELANE, EPCAM, EPG5, ERBB2IP, ERCC4, ERCC6L2, ETV6, EXTL3, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FCN1, FCN2, FCN3, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GAS8, GATA1, GATA2, GFI1, GINS1, HAX1, HELLS, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HSPA1L, HYDIN, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKG, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL6ST, IL7R, INVS, IRAK4, IRF2BP2, IRF3, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG4, LPIN2, LRBA, LRRC6,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Immunologie erweitertes Gen-Set: *LRRC8A, LYST, MAGT1, MALT1, MAP2K1, MAP2K2, MAP3K14, MASP1, MASP2, MAT2A, MEFV, MKL1, MLH1, MOGS, MPL, MRE11A, MS4A1, MSH2, MSH6, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NF1, NFAT5, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE3, NUP214, OFD1, ORAI1, OTULIN, PALB2, PARN, PAX5, PEPD, PGM3, PIGA, PIH1D3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, POLE2, POMP, PRF1, PRKCD, PRKDC, PROCR, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, PTPN11, PTPRC, PTX3, RAB27A, RAC2, RAD51C, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RBM8A, RECQL4, RFX5, RFXANK, RFXAP, RHOH, RIT1, RLTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPGR, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS29, RPS7, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SBDS, SEMA3E, SERPING1, SH2D1A, SH3KBP1, SKIV2L, SLC29A3, SLC35C1, SLC37A4, SLC46A1, SLC7A7, SLX4, SMARCAL1, SMARCD2, SOS1, SP110, SPAG1, SPINK5, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, THBD, THPO, TICAM1, TINF2, TLR3, TLR5, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TP53, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, VSIG4, VTN, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZMYND10, ZNF341, RIPK1* (438 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Coenzym-Q10-Mangel: *ADCK3, ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2, SLC25A26* (17 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Glykosilierungsstörungen: *ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, B4GALT1, CAD, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, GNE, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, NUS1, PGM1, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3, TMEM199* (54 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lysosomale Erkrankungen: *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, VPS33A* (103 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Peroxisomale Biogenese Störungen: *ABCD1, ABCD3, ACOX1, AGPS, AGXT, AMACR, DYM, EBP, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2, SUGCT, TRIM37* (28 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
X-gekoppelte Intelligenzminderung: *ABCD1, ACSL4, AFF2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CLCN4, CUL4B, DCX, DDX3X, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HNRNPH2, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, PTCHD1, RAB39B, RBM10, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TAF1, TIMM8A, TSPAN7, UBE2A, UPF3B, USP9X, ZC4H2, ZCCHC12, ZDHHC9, ZNF41, ZNF674, ZNF711, ZNF81* (100 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Aortopathien: *ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, ALDH18A1, ATP7A, B3GAT3, BGN, CBS, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A5, COL5A1, COL5A2, ELN, ENPP1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GATA5, HCN4, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, ZDHHC9* (52 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Arrhythmie: *ABCC9, AKAP9, ANK2, BAG3, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DBH, DES, DSC2, DSG2, DSP, FLNC, GATA6, HADHA, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LMNA, MYH6, MYH7, MYL4, NKX2-5, NOS1AP, NUP155, PKP2, PLN, PPA2, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTN* (58 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Arrhythmogene linksventrikuläre Dysplasie: *ABCC9, ACTC1, BAG3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FBXO32, FLNC, HCN4, JPH2, JUP, LAMP2, LDB3, LMNA, MIB1, MYBPC3, MYH6, MYH7, PKP2, PLEKHM2, PLN, PRDM16, RAF1, RBM20, RYR2, SCN5A, TAZ, TCAP, TNNT2, TPM1, TTN, VCL* (36 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Arrhythmogene rechtsventrikuläre Dysplasie (ARVC): *CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYH7, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN* (17 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Aterielle Fibrillation: *ABCC9, CACNA1C, CACNA2D1, CACNB2, GATA6, GJA5, HCN4, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LMNA, MYL4, NPPA, NUP155, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, TBX5, TNNI3* (27 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Dilatative Kardiomyopathien: *ABCC6, ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, APOA1, BAG3, CASQ2, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FKTN, FLNC, FOXD4, GATA6, GATAD1, GBE1, GLB1, HAND1, HCN4, ILK, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LRRC10, MLYCD, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYPN, NEBL, NEXN, PCCA, PCCB, PDLIM3, PKP2, PLEKHM2, PLN, PRDM16, RAF1, RBCK1, RBM20, RMND1, RYR2, SCN5A, SDHA, SGCD, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TTN, TTR, VCL, VPS13A* (88 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
hereditäre hämorrhagische Teleangiektasie: *ACVRL1, ENG, RASA1, SMAD4* (4 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Heterotaxie und Situs inversus: *ACVR2B, ANKS6, ARMC4, C21ORF59, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DYX1C1, FOXH1, GDF1, INVS, LEFTY2, LRRC6, MMP21, NODAL, PIH1D3, PKD1L1, SPAG1, TTC25, ZIC3, ZMYND10* (35 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Kardiomyopathien: *AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, AKAP9, ALMS1, ALPK3, ANKRD1, ANO5, APOA1, BAG3, BRAF, CALR3, CAPN3, CASQ2, CAV3, CBL, CDH2, COA5, COA6, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, DYSF, EEF1A2, ELAC2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FHL1, FKRP, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA6, GATAD1, GBE1, GFM1, GLA, GLB1, GMPPB, GTPBP3, GUSB, HADHA, HADHB, HAND1, HCN4, HFE, HRAS, ISPD, JPH2, JUP, KRAS , LAMA2, LAMP2, LARGE, LDB3, LMNA, LRRC10, LZTR1, MAP2K1, MAP2K2, MLYCD, MRPL3, MRPL44, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOT, MYOZ2, MYPN, NDUFAF2, NDUFB11, NDUFV2, NEXN, NF1, NRAS, PCCA, PCCB, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, PPA2, PPP1CB, PRDM16, PRKAG2, PTPN11, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SOS2, SPEG, SPRED1, TAB2, TAZ, TBX20, TBX5, TCAP, TGFB3, TK2, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRIM32, TSFM, TTN, TTR, VCL, VCP, VPS13A, XK* (168 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): *ANK2, CALM1, CALM2, CALM3, CASQ2, GNAI2, HCN4, KCNJ2, KCNQ1, LMNA, RYR2, SCN5A, TECRL, TRDN* (14 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Kongenitale Herzfehler: *ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, AFF4, B3GAT3, BCOR, BMPR2, CAD, CBL, CCDC11, CDK13, CELSR1, CELSR2, CELSR3, CFC1, CHD4, CHD7, CITED2, CREBBP, CRELD1, CTC1, DHCR7, DNAH11, DNAH5, DNAH6, DNAI1, DTNA, EFTUD2, EHMT1, EIF2AK4, ELN, ENG, FBN1, EVC2, FLNA, FOXC1, FOXH1, GANAB, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GPC3, HAND1, HAND2, HOXA1, HRAS, JAG1, KDM5B, KMT2D, LEFTY2, MED13L, MEIS2, MYH11, MYCN, MYH6, MYH7, NAA15, NF1, NIPBL, NKX2-5, NKX2-6, NME7, NODAL, NOTCH1, NOTCH2, NR2F2, NSD1, PITX2, PKD1L1, PLD1, POGZ, PPP1CB, PRDM6, PRKD1, RABGAP1L, RBFOX2, RBM10, SALL4, SMAD6, SOS2, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TLL1, TMEM260, TPM1, ZEB2, ZFPM2, ZIC3* (103 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Pulmonale arterielle Hypertonie: *ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, KLF2, NFU1, NOTCH3, RASA1, SMAD4, SMAD9, SOX17, TBX4* (21 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
QT-Syndrome und aterielle Fibrillation: *ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAV3, GATA6, GJA5, HCN4, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNK3, LDB3, LMNA, MYL4, NOS1AP, NPPA, NUP155, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TBX5, TECRL, TNNI3, TRDN* (38 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kardiologische Erkrankungen erweitertes Gen-Set: *AARS2, ABCA1, ABCC6, ABCC8, ABCC9, ABCG5, ABCG8, ABL1, ACAD9, ACADVL, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTN2, ACVR1, ACVR2B, ACVRL1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AFF4, AGK, AGL, AGPAT2, AKAP9, ALDH18A1, ALMS1, ALPK3, ANK2, ANKRD1, ANKS6, ANO5, APOA1, APOA5, APOB, APOC2, APOC3, APOE, AQP1, ARMC4, ATP13A3, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, ATPAF2, B3GALT6, B3GAT3, B4GALT7, BAG3, BCOR, BGN, BMPR1B, BMPR2, BRAF, C1R, C1S, C21ORF59, CACNA1C, CACNA2D1, CACNB2, CAD, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV1, CAV3, CBL, CBS, CCDC103, CCDC11, CCDC114, CCDC151, CCDC39, CCDC40, CDH2, CDK13, CELSR1, CELSR2, CELSR3, CFC1, CHD4, CHD7, CHRM2, CHST14, CITED2, COA5, COA6, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A1, COL4A5, COL5A1, COL5A2, COX15, CPT2, CREB3L3, CREBBP, CRELD1, CRYAB, CSRP3, CTC1, CTNNA3, DBH, DES, DHCR7, DMD, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAH6, DNAI1, DNAI2, DNAJC19, DNAL1, DOLK, DSC2, DSE, DSG2, DSP, DTNA, DYSF, DYX1C1, EEF1A2, EFEMP2, EFTUD2, EHMT1, EIF2AK4, ELAC2, ELN, EMD, ENG, ENPP1, EPG5, EPHB4, ETFA, ETFB, ETFDH, EVC, EVC2, FAH, FBLN5, FBN1, FBN2, FBXO32, FHL1, FHOD3, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXC1, FOXD4, FOXE3, FOXF1, FOXH1, FOXRED1, FXN, GAA, GANAB, GATA4, GATA5, GATA6, GATAD1, GBE1, GDF1, GDF2, GFM1, GJA1, GJA5, GLA, GLB1, GMPPB, GNAI2, GORAB, GPC3, GPIHBP1, GTPBP3, GUSB, HADHA, HADHB, HAND1, HAND2, HCN4, HFE, HOXA1, HRAS, ILK, INVS, ISPD, JAG1, JPH2, JUP, KAT6B, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNK3, KCNQ1, KDM5B, KLF2,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Kardiologische Erkrankungen erweitertes Gen-Set: *KMT2D, KRAS , LAMA2, LAMA4, LAMP2, LARGE, LDB3, LDLR, LDLRAP1, LEFTY2, LIPA, LMF1, LMNA, LOX, LPL, LRRC10, LRRC6, LTBP4, LZTR1, MAP2K1, MAP2K2, MAP3K8, MAT2A, MED12, MED13L, MEIS2, MFAP5, MIB1, MLYCD, MMP21, MRAS, MRPL3, MRPL44, MRPS22, MTO1, MYBPC3, MYBPHL, MYCN, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, MYLK2, MYOT, MYOZ2, MYPN, NAA15, NDUFAF2, NDUFB11, NDUFV2, NEBL, NEXN, NF1, NF2, NFU1, NIPBL, NKX2-5, NKX2-6, NME7, NODAL, NOS1AP, NOTCH1, NOTCH2, NOTCH3, NPPA, NR2F2, NRAS, NSD1, NSUN2, NUP155, PARS2, PCCA, PCCB, PCSK9, PDLIM3, PIH1D3, PITX2, PKD1L1, PKP2, PLD1, PLEC, PLEKHM2, PLN, PLOD1, PNPLA2, POGZ, POMT1, PPA2, PPP1CB, PRDM16, PRDM5, PRDM6, PRKAG2, PRKD1, PRKG1, PTPN11, PYCR1, RABGAP1L, RAF1, RASA1, RASA2, RBCK1, RBFOX2, RBM10, RBM20, RIT1, RMND1, RRAS, RYR2, SALL4, SASH1, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SEMA3D, SEMA3E, SGCA, SGCB, SGCD, SGCG, SHOC2, SKI, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SMAD9, SMARCB1, SMCHD1, SNTA1, SOS1, SOS2, SOX17, SPAG1, SPEG, SPRED1, STAMBP, TAB2, TAZ, TBX1, TBX20, TBX4, TBX5, TCAP, TECRL, TFAP2B, TGDS, TGFB2, TGFB3, TGFBR1, TGFBR2, TK2, TLL1, TMEM260, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TNXB, TOR1AIP1, TPM1, TRDN, TRIM32, TRPM4, TSFM, TTC25, TTN, TTR, UPF3B, VARS2, VCAN, VCL, VCP, VPS13A, XK, ZDHHC9, ZEB2, ZFPM2, ZIC3, ZMYND10, ZNF469 (419 Gene)*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ~~Beckman Coulter~~; ABI SeqStudio 8 Flex Genetic Analyzer System

Neurodegenration erweitertes Gen-Set: A2M, AARS, AARS2, ABCA7, ABCD1, ACOX1, ADAR, ADGRG1, ADPRHL2, AIFM1, AIMP1, AIMP2, AKT3, ALDH3A2, ALS2, ANG, ANO3, ANXA11, AP4B1, AP4E1, AP4M1, AP4S1, APOE, APOPT1, APP, ARSA, ASPA, ATL1, ATP13A2, ATP1A3, ATP6AP2, ATXN2, ATXN3, BCAP31, BOLA3, BSCL2, C19ORF12, C9orf72, CARS2, CHCHD10, CHCHD2, CHMP2B, CLCN2, COASY, COL4A1, COL4A2, COLGALT1, COX10, COX15, COX6B1, CP, CRAT, CSF1R, CSTB, CTC1, CTSA, CTSD, CTSF, CYP27A1, D2HGDH, DARS, DARS2, DCAF17, DCTN1, DNAJC12, DNAJC5, DNAJC6, DNMT1, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, EMC1, EPM2A, EPRS, ERBB4, ERCC6, ERCC8, FA2H, FAM126A, FARS2, FBXO7, FH, FIG4, FKRP, FKTN, FOLR1, FOXC1, FOXRED1, FTL, FUCA1, FUS, GALC, GAN, GBA, GBE1, GCDH, GCH1, GEMIN4, GFAP, GFM1, GIGYF2, GJC2, GLA, GLB1, GLRX5, GMPPB, GNAL, GOSR2, GPSM2, GRN, HEPACAM, HEXA, HFE, HIBCH, HIKESHI, HNRNPA1, HSD17B4, HSPD1, HTRA1, HTRA2, HTT, IBA57, IDS, IFIH1, ISCA1, ISCA2, ITM2B, JPH3, JPH3, KARS, KCNC1, KCNT1, KCTD7, KIAA0196, KIF5A, KIFBP, L2HGDH, LAMA2, LAMC3, LARGE, LIPT2, LMNB1, LMNB2, LRPPRC, LRRK2, LYRM7, LYST, MAPT, MARS2, MATR3, MCOLN1, MFSD8, MLC1, MPO, MPV17, MRPL44, MTFMT, MTHFS, NACC1, NARS2, NAXE, NDE1, NDUFAF5, NDUFS1, NDUFV1, NEFH, NEK1, NEU1, NFU1, NHLRC1, NKX2-1, NKX6-2, NOS3, NOTCH3, NPC1, NPC2, NR4A2, NSDHL, NUBPL, OCLN, OPA3, OPTN, PANK2, PARK7, PC, PDGFB, PDGFRB, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PHGDH, PINK1, PLA2G6, PLAA, PLAU, PLEKHG2, PLP1, PMPCB, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMT1, POMT2, PPT1, PRDM8, PRF1, PRICKLE1, PRKN, PRKRA,	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Neurodegenration erweitertes Gen-Set: *PRNP, PRPH, PSAP, PSAT1, PSEN1, PSEN2, PYCR2, QARS, RAB11B, RAB18, RAB39B, RARS, RARS2, REEP1, REPS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, ROGDI, SAMHD1, SCARB2, SCO1, SCP2, SDHA, SDHAF1, SERAC1, SERPINI1, SETX, SGCE, SIGMAR1, SIGMAR1, SLC16A2, SLC17A5, SLC1A4, SLC20A2, SLC25A12, SLC30A10, SLC33A1, SLC39A14, SLC41A1, SLC52A2, SLC52A3, SLC6A3, SNCA, SNCAIP, SNCB, SNORD118, SOD1, SORL1, SOX10, SPAST, SPG11, SPG20, SPR, SPTAN1, SQSTM1, SRPX2, STN1, SUMF1, SURF1, SYNJ1, TACO1, TAF1, TARDBP, TARS2, TBCD, TBCK, TBK1, TBP, TGFB1, TH, THAP1, TMEM106B, TOR1A, TPP1, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRPM7, TTC19, TUBA1A, TUBA4A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUFM, TYMP, TYROBP, UBE3A, UBQLN2, UBTF, UCHL1, UFC1, UFM1, VAPB, VARS, VARS2, VCP, VPS11, VPS13A, VPS13C, VPS35, WAC, WARS2, WDFY3, WDR45, WDR45B, WDR62, ZFYVE26, ZNHIT3 (345 Gene)*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Bronchiektasien: *CCDC39, CCDC40, CFTR, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, NME8, RSPH4A, RSPH9, SCNN1A, SCNN1B* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Interstitielle Lungenerkrankungen: *ABCA3, CSF2RA, CSF2RB, DKC1, ELMOD2, HPS1, HPS4, ITGA3, NAF1, NF1, NKX2-1, PARN, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2* (26 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Primäre Ziliardyskenesie: *ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYX1C1, GAS8, HEATR2, HYDIN, INVS, LRRC6, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TTC25, ZMYND10* (42 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Zentrales Hyperventilations-Syndrom: *CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MECP2, PHOX2B, RAPSN, RET, SCN4A, SLC6A5, ZEB2* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Zystische Lungenerkrankungen: *EFEMP2, ELN, FBLN5, FLCN, LTBP4, SERPINA1, TSC1, TSC2* (8 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lungenerkrankungen erweitertes Gen-Set: *ABCA3, ABCC8, ACVRL1, AP3B1, AP3D1, AQP1, ARMC4, ATP13A3, BLOC1S3, BLOC1S6, BMPR1B, BMPR2, C21ORF59, CAV1, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DTNBP1, DYX1C1, EDN3, EFEMP2, EIF2AK4, ELMOD2, ELN, ENG, FBLN5, FLCN, FOXF1, GAS8, GDF2, GLRA1, GPR143, HEATR2, HPS1, HPS3, HPS4, HPS5, HPS6, HYDIN, INVS, ITGA3, KCNA5, KCNK3, KLF2, LRRC6, LTBP4, LYST, MECP2, NAF1, NF1, NFU1, NKX2-1, NME8, NOTCH3, OCA2, OFD1, PARN, PHOX2B, PIH1D3, RAPSN, RASA1, RET, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SCN4A, SCNN1A, SCNN1B, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC45A2, SLC6A5, SLC7A7, SMAD4, SMAD9, SMPD1, SOX17, SPAG1, STAT3, STK36, TBX4, TERC, TERT, TINF2, TSC1, TSC2, TTC25, TYR, TYRP1, ZEB2, ZMYND10* (127 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Maligne Hyperthermie: *CACNA1S, RYR1* (2 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Stoffwechselmyopathien / Rhabdomyolyse: *ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ADCK3, AGL, AHCY, ALDOA, AMPD1, ANO5, C10ORF2, CAV1, COQ2, CPT2, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FKRP, FKTN, FLAD1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, HADHA, HADHB, ISCU, LAMA2, LAMP2, LDHA, LPIN1, MGME1, MPV17, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH3, OPA1, OPA3, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SLC16A1, SLC22A5, SLC25A20, SLC25A4, SUCLA2, SUCLG1, TANGO2, TAZ, TK2, TYMP, YARS2* (110 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Neuralrohrdefekte: *SEPT6, ABCB6, ABL1, ABL2, GPR125, AHCY, ALDH1A2, ALDH1L1, ALX1, AMBRA1, AMD1, AMT, APAF1, APEX1, APOB, ARHGAP35, ARID1A, AXIN1, AZI2, B9D1, B9D2, BBS4, BCL10, BHMT, BHMT2, BMP4, BRCA1, BRD2, CASP3, CASP9, CAT, CBS, CC2D2A, CCL2, CCL3, CDH1, CDON, CELSR1, CELSR2, CELSR3, CEP290, CFL1, CHKA, CHUK, CITED2, COBL, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, COQ3, CRABP1, CRABP2, CTH, CTHRC1, CTNNBIP1, CUBN, CXCL6, CYCS, CYP26A1, CYP26B1, DAAM1, DACT1, DCHS1, DEAF1, DHFR, DLC1, DLG1, DLL3, DLX5, DNAAF1, DNMT1, DNMT3B, DVL1, DVL2, DVL3, EFNA5, ENAH, EP300, EPCAM, EPHA7, EPOR, ERCC2, FAT4, FGFR1, FGFR2, FKBP8, FOLH1, FOLR1, FOLR2, FOLR3, FOXE1, FOXH1, FOXN3, FPGS, FTCD, FUZ, FZD1, FZD2, FZD3, FZD4, FZD5, FZD6, GAMT, GAPDH, GART, GATA3, GCSH, GDF3, GDF6, GGH, GLDC, GLI2, GPC5, GRHL1, GRHL2, GRHL3, GSTO1, HDAC4, HES1, HES3, HES7, HIC1, HK1, HK2, HSPG2, HYLS1, ICMT, INADL, INPP5E, INS, INSR, INTU, INVS, IRF6, ITGA3, ITGA6, ITGB1, ITPK1, IZUMO1R(FOLR4), JARID2, KCNJ10, KDM6A, KIF14, KMT2D, LAMA5, LAMC1, LEP, LEPR, LFNG, LMNB1, LMO4, LPAR1, LRP6, MAP3K4, MAP3K8, MAPK9, MARCKS, MARCKSL1, MAT1A, MAT2A, MED12, MEN1, MEOX1, MESP2, MGMT, MKS1, MSGN1, MSX1, MSX2, MTHFD1, MTHFD2, MTHFD2L, MTHFR, MTHFS, MTR, MTRR, MTTP, MUT, MYT1L, NAP1L4, NAT1, NAT2, NCAM1, NDST1, NECTIN1, NEFL, NEUROG2, NNMT, NODAL, NOG, NOS1, NOS2, NOS3, NPHP3, NUMB, NUP50, PALLD, PAX2, PAX3, PAX7, PCMT1, PCYT1A, PDGFRA, PEMT, PFN1, PIGA, PIK3CA, PITX2, POR, POU3F1, PRICKLE1, PRICKLE2, PRICKLE3, PRICKLE4, PRKACA, PRKACB, PRKCB, PRKCG, PRMT1, PRMT2, PTCH1, PTK7, PTPN11, PTPN9* (321 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Neuralrohrdefekte: *PVR(NECL5), PVRL2(NECTIN2), RAB23, RAC1, RARA, RARG, RFC1, RGMA, RNMT, ROR2, RPGRIP1L, RYBP, SALL1, SALL2, SALL4, SARDH, SATB2, SCRIB, SDC4, SEC24B, SEMA4C, SESTD1, SETD2, SFRP1, SFRP2, SFRP3(FRZB), SHH, SHMT1, SHROOM1, SHROOM2, SHROOM3, SHROOM4, SIRT1, SKI, SLC19A1, SLC2A1, SLC2A4, SMARCA4, SMURF1, SMURF2, SNX1, SNX13, SNX2, SOD1, SOD2, SOX3, SP8, SPINT2, ST14, SUMO1, SUPT20H, T, TBX22, TBX6, TCFAP2A, TCN2, TCOF1, TECR, TFAP2A, TGDS, TGIF1, TMEM216, TMEM231, TMEM5, TMEM67, TP53, TP63, TRAF4, TRDMT1, TRPM6, TULP3, TWIST1, TXN2, TYMS, UCP2, VANGL1, VANGL2, VASP, VCL, WNT11, WNT3A, WNT5A, WNT7A, WNT9A, XNP(ATRX), XRCC1, XRCC3, YBX1, YYL, ZDBF2, ZIC1, ZIC2, ZIC3, ZNRF3* (321 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Alzheimer/Demenz: *ABCA7, A2M, APOE, APP, C9orf72, CHCHD10, CHMP2B, CSF1R, DCTN1, DNAJC5, DNMT1, EPM2A, FUS, GBA, GRN, HFE, HNRNPA1, HTRA1, ITM2B, JPH3, MAPT, MPO, NHLRC1, NKX2-1, NOTCH3, NOS3, PLAU, PRNP, PSEN1, PSEN2, RNF216, ROGDI, SIGMAR1, SNCA, SNCB, SORL1, SQSTM1, TARDBP, TBK1, TREM2, TRPM7, TUBA4A, TYROBP, UBE3A, UBQLN2, VCP, WAC, WDFY3* (48 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Arteriopathie (CADASIL): *ABCD1, APP, COL4A1, COL4A2, COLGALT1, CTSA, FOXC1, GLA, HTRA1, NOTCH3, SNORD118, TREX1* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Hereditäre zerebrale Angiopathien: *APP, COL4A1, COL4A2, COLGALT1, CTSA, FOXC1, GLA, HTRA1, NOTCH3, SNORD118, TREX1* (11 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Neurodegeneration mit Eisenablagerungen: *ATP13A2, C19ORF12, COASY, CP, CRAT, DCAF17, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Neuronale Ceroidlipofuszinose: *AFG3L2, ASAH1, ATP13A2, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAJC5, EPM2A, GOSR2, GRN, KCNC1, KCTD7, LMNB2, MFSD8, NEU1, NHLRC1, PPT1, PRDM8, PRICKLE1, SCARB2, SERPINI1, TPP1* (27 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Parkinson: *ATP13A2, ANO3, ATP1A3, ATP6AP2, ATXN2, ATXN3, C19ORF12, C9orf72, CHCHD2, CSF1R, DCTN1, DNAJC12, DNAJC6, EIF4G1, FBXO7, FTL, GBA, GCH1, GIGYF2, GLUD2, GNAL, GRN, HTRA2, HTT, JPH3, LRRK2, LYST, MAPT, NR4A2, OPA3, PANK2, PRKN, PARK7, PDGFB, PDGFRB, PINK1, PLA2G6, PRKRA, RAB39B, SGCE, SLC20A2, SLC30A10, SLC39A14, SLC41A1, SLC6A3, SNCA, SNCAIP, SPG11, SPR, SYNJ1, TAF1, TBP, TH, THAP1, TOR1A, TUBB4A, UCHL1 M, VPS13A, VPS13C, VPS35, WDR45* (60 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Gorlin-Goltz-Syndrom: *PTCH1, PTCH2, SUFU* (3 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Alport-Syndrom: *CD151, COL4A3, COL4A4, COL4A5, COL4A6, FN1, LMX1B, MYH9, PXDN* (9 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Cystische Nierenerkrankungen: *ANKS6, BICC1, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, ETFA, EYA1, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, RPGRIP1L, SDCCAG8, SEC61A1, SIX5, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423* (45 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Diabetes Insipidus: *AQP2, AVP, AVPR2, SLC12A1* (4 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hämolytisch Urämisches Syndrom: *ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MMUT, PIGA, PLG, THBD* (19 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Hypophasphatämische Rachitis: *ALPL, ANKH, CASR, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, GNA11, KL, OCRL, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR* (18 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Meckel-Syndrom: *AHI1, B9D1, B9D2, CC2D2A, CEP120, CEP290, CEP55, CSPP1, KIAA0586, KIAA0753, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN1, TCTN2, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Nephrolithiases: *ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GNA11, GPHN, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, OCRL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, VDR, XDH* (35 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Nephronophthisen: *ADAMTS9, ANKS6, ATXN10, CEP164, CEP290, CEP83, DCDC2, FAN1, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TRAF3IP1, TTC21B, WDR19, WDR35, XPNPEP3, ZNF423* (27 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Nephrotisches Syndrom: *ACTN4, ADCK4, ANKFY1, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAN1, FAT1, FBXW7, FN1, GAPVD1, INF2, ITGA3, ITGB4, ITSN1, ITSN2, KANK1, KANK2, KANK4, LAMA5, LAMB2, LMNA, LMX1B, MAFB, MAGI2, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH9, MYO1E, NPHP1, NPHP4, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, SYNPO, TBC1D8B, TNS2, TRPC6, TTC21B, WDR73, WT1, XPO5* (104 Gene)	EDTA-Blut, Mundschleimhaut,Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Pseudohyperaldosteronismus: *CUL3, HSD11B2, KCNJ5, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4* (10 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Renale Malformationen: *ACE, ACTG2, AGT, AGTR1, ALDH1A2, BICC1, BMP4, BMP7, CDC5L, CHD1L, DACH1, DSTYK, EYA1, FAM58A, FANCB, FGF20, FIBP, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GATA3, GDNF, GREB1L, GREM1, GRIP1, HNF1B, ITGA8, KIF14, KYNU, NEK8, NPHP3, OSR1, PAX2, PBX1, PUF60, REN, RET, ROBO2, SALL1, SDCCAG8, SIX1, SIX5, SLIT2, SOX17, SPRY1, SRGAP1, TBC1D1, TBX18, TFAP2A, TNS3, TRAP1, UMOD, UPF3B, UPK3A, WNT4, WNT5A, WT1* (59 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Renaltubuläre Azidose: *ATP6V0A4, ATP6V1B1, BCS1L, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B* (9 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Nierenerkrankungen erweitertes Gen-Set: *ACE, ACTG2, ACTN4, ACVR2B, ADAMTS13, ADAMTS9, ADCK4, ADCY10, AGT, AGTR1, AGXT, AHI1, ALDH1A2, ALMS1, ALPL, ALPL, ANKFY1, ANKS6, ANLN, AP2S1, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL3, ARL6, ARMC4, ARMC9, ATP1A1, ATP6V0A4, ATP6V1B1, ATXN10, AVP, AVPR2, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BMP7, BSND, C11ORF70, C21ORF2, C21ORF59, C2CD3, C3, C5ORF42, C8ORF37, CA2, CASR, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CD151, CD2AP, CD46, CDC5L, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFTR, CHD1L, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CLU, CNNM2, CNNM4, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPE, CPLANE1, CRB2, CSPP1, CUBN, CUL3, CYP24A1, CYP27B1, CYP2R1, DACH1, DCDC2, DDX59, DGKE, DHCR7, DLC1, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH17, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DSTYK, DYNC2H1, DYNC2LI1, DYX1C1, DZIP1L, EGF, EMP2, ENPP1, ETFA, EVC, EVC2, EXOC8, EYA1, FAH, FAM111A, FAM149B1, FAM20A, FAM58A, FAN1, FAN1, FANCB, FAT1, FBXW7, FGF20, FGF23, FIBP, FN1, FOXC1, FOXC2, FOXI1, FRAS1, FREM1, FREM2, FXYD2, GANAB, GAPVD1, GAS2L2, GAS8, GATA3, GDNF, GLI2, GLI3, GLIS2, GNA11, GPHN, GREB1L, GREM1, GRHPR, GRIP1, HEATR2, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, IFT81, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA8, ITGB4* ...	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Nierenerkrankungen erweitertes Gen-Set: *ITSN1, ITSN2, JAG1, KANK1, KANK2, KANK4, KCNA1, KCNJ1, KCNJ10, KCNJ5, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, KL, KLHL3, KYNU, LAMA5, LAMB2, LEFTY2, LMNA, LMX1B, LRP5, LRRC6, LZTFL1, MAFB, MAGED2, MAGI2, MAGT1, MAPKBP1, MCIDAS, MKKS, MKS1, MMACHC, MMADHC, MMUT, MOCOS, MOCS1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUC1, MYH9, MYO1E, NEK1, NEK8, NEK8, NIPA2, NME8, NODAL, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OSGEP, OSR1, PAX2, PBX1, PCBD1, PDE6D, PDPR, PDSS2, PHEX, PIBF1, PIGA, PIH1D3, PKD1, PKD2, PKHD1, PLCE1, PLG, PMM2, PNPLA6, POC1B, PTPRO, PUF60, PXDN, REN, RET, ROBO2, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SALL1, SARS2, SCAPER, SCARB2, SCLT1, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC7A9, SLC9A3R1, SLIT2, SMARCAL1, SOX17, SPAG1, SPRY1, SRGAP1, STK36, SUFU, SYNPO, TBC1D1, TBC1D8B, TBX18, TCTEX1D2, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TNS3, TRAF3IP1, TRAP1, TRAPPC3, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC25, TTC8, TXNDC15, UMOD, UPF3B, UPK3A, USP9X, VDR, VHL, VIPAS39, VPS33B, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR73, WNK1, WNK4* (441 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Nierenerkrankungen erweitertes Gen-Set: *WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, ZIC3, ZMYND10, ZNF423* (441 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
3-M-Syndrom: *ATR, BCS1L, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, CHD7, CREBBP, CUL7, DNA2, EP300, EYA1, FLNA, FLNB, HNRNPK, IRF6, KDM1A, KDM6A, KMT2D, LARP7, MAP3K7, NIN, NOTCH2, NSMCE2, OBSL1, ORC1, ORC4, ORC6, PCNT, PLK4, POC1A, RBBP8, RNU4ATAC, RTTN, SIX5, SH3PXD2B, SRCAP, TAB2, TRAIP, TRIM37, XRCC4* (44 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Amelogenesis Imperfecta: *AMELX, C4orf26, DLX3, DSPP, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, LTBP3, MMP20, SLC24A4, WDR72, WNT10B* (16 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Chondrodysplasie Punctata: *AGPS, ARSE, ASS, EBP, GDF5, GNPAT, LBR, MGP, NSDHL, PEX14, PEX19, PEX7* (10 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Dysostosen: *ALPL, ALX1, ALX3, ALX4, BMPER, CDC45, CDC6, CREBBP, CDT1, DHODH, DLL3, EFNB1, EFTUD2, EHMT1, EP300, EVC, EVC2, GDF3, GDF6, HDAC8, HES7, HSPG2, KAT6B, LIFR, LMX1B, MEOX1, MESP2, MNX1, MYCN, MYO18B, NIPBL, ORC1, ORC4, ORC6, POLR1A, POLR1C, POLR1D, RIPPLY2, SF3B4, SMC1A, SMC3, SNRPB, SRCAP, TBX4, TBX6, TCOF1, TWIST1, UBE2A* (48 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Brachydaktylie: *BMP2, BMPR1B, CHSY1, DHCR7, DLX5, ESCO2, FAM58A, GDF5, GNAS, HOXA13, HOXD13, IHH, MYCN, NOG, PDE3A, PDE4D, PTDSS1, PTHLH, RECQL4, ROR2, SOX9, TP63, WNT10B* (23 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Extremitätenfehlbildungen erweitertes Genset: *AFF4, ANKRD11, ARHGAP31, ARID1A, ARID1B, ATR, BHLHA9, BMP2, BMPR1B, BRCA2, BRD4, BRIP1, CDH3, CHSY1, DHCR7, DHODH, DLL4, DLX5, DOCK6, EOGT, ERCC4, ESCO2, FAM58A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FGF10, FGF16, FGF9, FGFR1, GDF5, GJA1, GLI3, GNAS, HDAC8, HOXA13, HOXD13, IHH, KCTD1, KYNU, LMBR1, LRP4, MYCN, NIPBL, NOG, NOTCH1, NSDHL, PALB2, PDE3A, PDE4D, PITX1, PTDSS1, PTHLH, PUF60, RAD21, RAD51C, RBM8A, RBPJ, RECQL4, ROR2, SALL1, SALL4, SF3B4, SLX4, SMC1A, SMC3, SOX9, TBX15, TBX3, TBX5, TP63, UBR1, WNT10B, WNT7A, XRCC2* (82 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Gastrointestinale Atresie: *CDK9, CHD7, CLMP, DHCR7, EFTUD2, FANCB, FANCC, GLI3, MID1, MNX1, MYCN, PTF1A, RFX6, SOX2, TTC7A* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Morbus-Hirschsprung: *BDNF, CELSR3, EDN3, EDNRB, KIFBP, L1CAM, MITF, NRG1, NRTN, PAX3, PHOX2B, RET, RMRP, SOX10, ZEB2* (15 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kleinwuchs erweitertes Gen-Set: *ACTB, ACTG1, AMMECR1, ARCN1, ATR, B3GAT3, BCS1L, BRAF, CBL, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, COL27A1, CREBBP, CUL7, DHCR7, DONSON, EP300, FGD1, FGFR3, FN1, GH1, GHR, GHRHR, GHSR, GLI2, GNAS, HDAC8, HESX1, HRAS, IDUA, IGF1, IGF1R, IGFALS, INSR, IRS1, KRAS, LARP7, LFNG, LHX3, LHX4, LZTR1, MAP2K1, MAP2K2, NIPBL, NOTCH2, NRAS, OBSL1, ORC1, ORC4, ORC6, OSGEP, OTX2, PCNT, PISD, PITX2, POC1A, POP1, POU1F1, PPP3CA, PRMT7, PROP1, PTPN11, PUF60, RAD21, RAF1, RALA, RASA2, RBBP8, RIT1, RNU4ATAC, RRAS, RTTN, SGMS2, SHOC2, SHOX, SMARCA2, SMARCE1, SMC1A, SMC3, SOS1, SOX11, SOX2, SOX3, SRCAP, STAT5B, TALDO1, TBX19, TBX2, TBX3, TOP3A, TRIM37, TRMT10A, XRCC4* (98 Gene)	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kraniosynostose: *ALPL, ALX3, ALX4, ASXL1, BMP4, CCBE1, CDC45, CEP120, COLEC11, CYP26B1, EDNRB, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, IFT43, IFT52, IL11RA, IMPAD1, MASP1, MEGF8, MSX2, NOG, PAX3, POR, RAB23, RECQL4, SCARF2, SEC24D, SIK1, SMAD6, SOX10, SPECC1L, TCF12, TGFBR1, TGFBR2, TWIST1, TWIST2, WDR19, WDR35, ZIC1* (49 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Lippen-Kiefer-Gaumenspalte: *COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, CTNND1, FOXE1, GRHL3, IRF6, KDM6A, KMT2D, MSX1, SATB2, SPECC1L, TBX2, TBX22, TGDS, TP63, TXNL4A, ZSWIM6* (21 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lymphatische Fehlbildungen: *ADAMTS3, CCBE1, FAT4, FLT4, FOXC2, GATA2, GJC2, KIF11, PIEZO1, PIK3CA, RASA1, SOX18* (12 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Metaphysäre Dysplasie: *ANKH, CDKN1C, COL10A1, FGFR3, FLNA, MMP13, MMP9, PISD, POP1, PTH1R, RMRP, RUNX2, SBDS* (13 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Mikromele Dysplasien: *ADAMTS10, ADAMTS17, ADAMTSL2, BMPR1B, DVL1, DVL3, EXT1, FBN1, FGFR3, GDF5, GNAS, GPC6, IFT122, IFT140, IFT43, IHH, INPPL1, LIFR, LRP4, LTBP2, MAB21L2, NPR2, PDE4D, PRKAR1A, ROR2, SHOX, SLC35D1, SMAD4, SOX9, TRIP11, TRPS1, WDR19, WDR35, WNT5A* (34 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Mineralisierungsstörungen: *ALPL, ANKH, B4GALT7, CASR, CLCN5, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, CYP27B1, ENPP1, FBN1, FGF23, FKBP10, GALNT3, MGP, P3H1, PHEX, PLOD2, PLS3, PPIB, PTDSS1, SERPINF1, SGMS2, SLC34A3, SLC39A13, SNX10, SOX9, TNFRSF11A, TNFRSF11B, VDR, XYLT2* (34 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Osteogenesis imperfecta: *ALPL, ANO5, ARCN1, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, BMP1, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, FAM46A, FGF23, FKBP10, GORAB, IFIH1, IFITM5, LEPRE1, LRP5, MBTPS2, MESD, P3H1, PHEX, PLOD2, PLS3, PPIB, PYCR1, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC34A3, SP7, SPARC, TENT5A, TMEM38B, TNFRSF11B, WNT1* (40 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Osteopetrose und weitere Dysplasien mit erhöhter Knochendichte: *AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DHCR24, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRP5, OSTM1, PLEKHM1, PNPLA6, PTDSS1, PTH1R, SDCCAG8, SFRP4, SLC29A3, SLCO2A1, SNX10, SOST, SQSTM1, TBXAS1, TCIRG1, TGFB1, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TRAPPC3, TRIM32, TTC21B, TTC8, TYROBP, WDPCP* (24 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Kurzrippendysplasie: *CEP120, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, GLI2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT,57, IFT80, IFT81, INTU, KIAA0586, KIAA0753, NEK1, TCTEX1D2, TCTN3, TMEM107, TTC21B, WDR19, WDR34, WDR35, WDR60* (28 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Spondylometaphysäre Dysplasie: *ACAN, ACP5, B3GALT6, B3GAT3, BGN, C21ORF2, CANT1, CHST3, COL11A1, COL11A2, COL2A1, COL9A1, DDR2, DDRGK1, DYM, EIF2AK3, EXTL3, FLNB, HSPG2, IMPAD1, INPPL1, KIF22, LONP1, MATN3, MMP13, NANS, NKX3-2, PAPSS2, PCYT1A, PISD, POP1, RAB33B, RMRP, RSPRY1, SLC39A13, SMARCAL1, TRAPPC2, TRPV4, WISP3, XYLT1* (40 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Vaskuläre Fehlbildungen: *ACVRL1, CCM2, ELMO2, ENG, GLMN, KRIT1, PDCD10, PIK3CA, PTEN, RASA1, SMAD4, SOX18, STAMBP, TEK* (14 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Skeletterkrankungen und Wachstumsstörungen erweitertes Gen-Set: *ACAN, ACP5, ACTB, ACTG1, ACVR2B, ACVRL1, ADAMTS10, ADAMTS17, ADAMTS3, ADAMTSL2, AFF4, AGA, AGPS, ALPL, ALX1, ALX3, ALX4, AMELX, AMER1, AMMECR1, ANKH, ANKRD11, ANKS6, ANO5, ARCN1, ARHGAP31, ARID1A, ARID1B, ARMC4, ARSB, ARSE, ASXL1, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BDNF, BGN, BHLHA9, BMP1, BMP2, BMP4, BMPER, BMPR1B, BRAF, BRCA2, BRD4, BRIP1, C21ORF2, C21ORF59, C4orf26, CA2, CANT1, CASR, CBL, CCBE1, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC8, CCM2, CDC45, CDC6, CDH3, CDK9, CDKN1C, CDT1, CELSR3, CENPJ, CEP120, CEP152, CEP63, CFAP53, CHD7, CHRNG, CHST3, CHSY1, CLCN5, CLCN7, CLMP, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTNND1, CTSA, CTSK, CUL7, CYP26B1, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNA2, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK6, DONSON, DSPP, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, DYX1C1, EBP, EDN3, EDNRB, EFNB1, EFTUD2, EHMT1, EIF2AK3, ELMO2, ENAM, ENG, ENPP1, EOGT, EP300, ERCC4, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EYA1, FAH, FAM111A, FAM20A, FAM20C, FAM46A, FAM58A, FAM83H, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAT4, FBN1, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FLT4, FN1, FOXC2, FOXE1, FOXH1, FREM1, FUCA1, GALNS, GALNT3, GATA2, GDF1, GDF3, GDF5, GDF6, GH1, GHR*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Skeletterkrankungen und Wachstumsstörungen erweitertes Gen-Set: *GHRHR, GHSR, GJA1, GJC2, GLB1, GLI2, GLI3, GLMN, GNA11, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPR68, GRHL3, GUSB, HDAC8, HES7, HESX1, HGSNAT, HNRNPK, HOXA13, HOXD13, HPGD, HRAS, HSPG2, HYALI, ICK, IDS, IDUA, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGFALS, IHH, IL11RA, IMPAD1, INPPL1, INSR, INVS, IRF6, IRS1, ITGB6, KAT6B, KCTD1, KDM1A, KDM6A, KIAA0586, KIF11, KIF22, KIFBP, KIT, KITLG, KL, KLK4, KMT2D, KRAS, KRIT1, KYNU, L1CAM, LAMB3, LARP7, LBR, LEFTY2, LEMD3, LEPRE1, LFNG, LHX3, LHX4, LIFR, LMBR1, LMX1B, LONP1, LRP4, LRP4, LRP5, LRRC6, LTBP2, LTBP3, LZTR1, LZTR1, MAB21L2, MAN2B1, MANBA, MAP2K1, MAP2K2, MAP3K7, MASP1, MATN3, MBTPS2, MEGF8, MEOX1, MESD, MESP2, MGP, MGP, MID1, MITF, MMP13, MMP13, MMP20, MMP21, MMP9, MMP9, MNX1, MSX1, MSX2, MYCN, MYO18B, NAGLU, NANS, NEK1, NEU1, NF1, NF2, NIN, NIPBL, NKX3-2, NODAL, NOG, NOTCH1, NOTCH2, NPR2, NRAS, NRG1, NRTN, NSDHL, NSMCE2, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, PALB2, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDCD10, PDE3A, PDE4D, PEX14, PEX19, PEX7, PHEX, PHOX2B, PIEZO1, PIH1D3, PIK3CA, PISD, PITRM1, PITX1, PITX2, PKD1L1, PLEKHM1, PLK4, PLOD2, PLS3, PNPLA6, POC1A, POLR1A, POLR1C, POLR1D, POP1, POP1, POP1, POR, POU1F1, PPIB, PPP3CA, PRKAR1A, PRMT7, PROP1, PTDSS1, PTEN, PTF1A, PTH1R, PTH1R, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RAD51C, RAF1, RALA, RASA1, RASA2, RBBP8, RBM8A, RBPJ, RECQL4, RET, RFX6, RIPPLY2, RIT1, RMRP, RNU4ATAC, ROR2, RRAS, RSPRY1, RTTN, RUNX2, RUNX2, SALL1, SALL4*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Skeletterkrankungen und Wachstumsstörungen erweitertes Gen-Set: *SATB2, SBDS, SBDS, SCARF2, SDCCAG8, SEC24D, SERPINF1, SERPINH1, SF3B4, SFRP4, SGMS2, SGSH, SH3PXD2B, SHOC2, SHOX, SIK1, SIX5, SKI, SLC17A5, SLC24A4, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLC9A3R1, SLCO2A1, SLX4, SMAD4, SMAD6, SMARCA2, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SNRPB, SNX10, SOS1, SOST, SOX10, SOX11, SOX18, SOX2, SOX3, SOX9, SP7, SPAG1, SPARC, SPECC1L, SPRED1, SQSTM1, SRCAP, STAMBP, STAT5B, SUMF1, TAB2, TALDO1, TBX15, TBX19, TBX2, TBX22, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTEX1D2, TCTN3, TEK, TENT5A, TGDS, TGFB1, TGFBR1, TGFBR2, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TOP3A, TP63, TRAIP, TRAPPC2, TRAPPC3, TRIM32, TRIM37, TRIP11, TRMT10A, TRPS1, TRPV4, TTC21B, TTC25, TTC7A, TTC8, TWIST1, TWIST2, TXNL4A, TYROBP, UBE2A, UBR1, VDR, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR72, WISP3, WNT1, WNT10B, WNT5A, WNT7A, XDH, XRCC2, XRCC4, XYLT1, XYLT2, ZEB2, ZIC1, ZIC3, ZMYND10, ZSWIM6* (566 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ~~Beckman Coulter~~; ABI SeqStudio 8 Flex Genetic Analyzer System

Azidämie / Azidurie: *ABCD4, ACADSB, ACAT1, ACSF3, ADK, AHCY, ALDH6A1, AMN, BCKDHA, BCKDHB, BCS1L, CBS, CD320, CLPB, CTH, CUBN, D2HGDH, DBT, DLD, ETFA, ETFB, ETFDH, FLAD1, GCDH, GIF, GNMT, HCFC1, HIBCH, HMGCL, IDH2, IVD, L2HGDH, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, PCCA, PCCB, PEPD, PPM1K, SERAC1, SLC25A1, SUCLA2, SUCLG1, SUGCT, TCN2, UMPS* (56 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Fettsäureoxidationstörung: *ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ALDH5A1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ* (26 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Glykogenspeicherkrankheit: *AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4* (29 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Harnstoffzyklusdefekte: *ACADM, ACADS, ACADVL, ARG1, ASL, ASS1, BCKDHA, BCKDHB, CA5A, CPS1, CPT1A, CPT2, DBT, DLD, ETFA, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, HMGCS2, IVD, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, NBAS, OAT, OTC, PC, PCCA, PCCB, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, SUCLA2, SUCLG1, TMEM70, UMPS* (49 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lipodystrophie: *AGPAT2, AKT2, BANF1, BSCL2, CAV1, CIDEC, FAM105B, FBN1, KCNJ6, LEMD2, LIPE, LMNA, LMNB2, PIK3LR, PLIN1, PPARG, PSMB8, PTRF, SPRTN, TBC1D4, ZMPSTE24* (21 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Metabolische Lebererkrankungen: *AGL, ALDOB, ATP7B, BCS1L, DGUOK, DNA2, FAH, FBP1, G6PC, GALE, GALK1, GALT, GBE1, GYS2, HAMP, HFE, HFE2, HPD, LIPA, MGME1, MPI, NPC1, NPC2, OPA1, PHKA2, PHKB, PHKG2, POLG, POLG2, PYGL, RNASEH1, RRM2B, SERPINA1, SLC25A4, SLC2A2, SLC37A4, SLC40A1, SMPD1, TAT, TFR2, TK2, TOP3A, TWNK* (43 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Mitochondriopathien erweitertes Gen-Set: *AARS2, ABCC2, ABHD5, ACAD9, ACADM, ACADS, ACADSB, ACADVL, AGK , AGL, AIFM1, ALDOA, AMPD1, AMT, ANO10, APOPT1, APTX, ATP5F1A, ATP5F1D, ATP5F1E, ATPAF2, AUH, BCS1L, BOLA3, C10ORF2, C12ORF65, C19orf70, C1QBP, CARS2, CHKB, CLPB, COA3, COA5, COA6, COA7, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CPS1, CPT1A, CPT2, CYC1, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ENO3, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FLAD1, FOXRED1, G6PC, GAA, GBE1, GFER, GFM1, GLDC, GTPBP3, GYG1, GYS1, HADH, HADHA, HADHB, HIBCH, HMGCL, HMGCS2, HSD17B10, IBA57, ISCA1, ISCA2, ISCU, KIF5A, LAMP2, LDHA, LIAS, LIPT1, LIPT2, LPIN1, LRPPRC, LYRM4, LYRM7, MARS2, MDH2, MFF, MFN2, MGME1, MIPEP, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, NARS2, MT-ATP6, MTFMT, MTO1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OGDH, OPA1, OPA3, OTC, PC, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PMPCA, PMPCB, PNPLA2, PNPT1, POLG, POLG2, PPA2, PRKAG2, PUS1, PYGM, RMND1, RNASEH1, RRM2B, SCO1, SCO2, SDHA, SDHAF1, SDHD, SERAC1, SFXN4, SLC16A1, SLC19A2, SLC19A3, SLC22A5, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLCO1B3, SLCO1B1, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM,*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Mitochondriopathien erweitertes Gen-Set: *TIMM8A, TIMMDC1, TK2 , TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WFS1, YARS2* (231 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Mitochondriopathien mitochondriale DNA: *MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY* (37 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Stoffwechselstörungen und Mitochondriopathien erweitertes Gen-Set: *AARS2, AARS2, ABCC2, ABCC8, ABCD1, ABCD3, ABCD4, ABHD5, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADAMTSL2, ADAR, ADCK3, ADK, ADPRHL2, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AIMP1, AIMP2, AKT2, ALDH3A2, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMN, AMPD1, AMT, ANO10, ANO5, ANTXR2, APOPT1, APPL1, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATP13A2, ATP5F1A, ATP5F1D, ATP5F1E, ATP6V0A2, ATP7B, ATPAF2, AUH, B3GLCT, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BLK, BOLA3, BSCL2, BTD, C10ORF2, C12ORF65, C19orf70, C1QBP, CA5A, CAD, CARS2, CAV1, CBS, CCDC115, CD320, CEL, CHKB, CIDEC, CLCN2, CLN3, CLN5, CLN6, CLN8, CLPB, COA3, COA5, COA6, COA7, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CPS1, CPT1A, CPT2, CRAT, CSF1R, CTC1, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CYC1, CYP27A1, D2HGDH, DARS, DARS2, DBT, DDOST, DGUOK, DHCR7, DHDDS, DLAT, DLD, DNA2, DNAJC12, DNAJC19, DNM1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, DYSF, EARS2, EBP, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, EMC1, ENO3, EPM2A, EPRS, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAH, FAM126A, FARS2, FASTKD2, FBP1, FBXL4, FH, FKRP, FKTN, FLAD1, FOLR1, FOXRED1, FUCA1, FUK, FUT8, G6PC, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System

Stoffwechselstörungen und Mitochondriopathien erweitertes Gen-Set: *GEMIN4, GFAP, GFER, GFM1, GFM2, GIF, GJC2, GLA, GLB1, GLDC, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GMPPB, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GTPBP3, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEPACAM, HEXA, HEXB, HFE, HFE2, HGSNAT, HIBCH, HIKESHI, HLCS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HYAL1, IBA57, IDH2, IDS, IDUA, IFIH1, INS, INSR, ISCA1, ISCA2, ISCU, ITPA, IVD, KARS, KCNJ11, KCNT1, KIF5A, KLF11, L2HGDH, LAMA2, LAMP2, LARGE1, LCT, LDB3, LDHA, LIAS, LIPA, LIPE, LIPT1, LIPT2, LMBRD1, LMNA, LMNB1, LPIN1, LRPPRC, LYRM4, LYRM7, MAGT1, MAN1B1, MAN2B1, MANBA, MARS2, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MFF, MFN2, MFSD8, MGAT2, MGME1, MIPEP, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPI, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MRPS7, MT-ATP6, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, MTR, MT-RNR1, MT-RNR2, MTRR, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUT, MYH3, MYOT, NACC1, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Stoffwechselstörungen und Mitochondriopathien erweitertes Gen-Set: *NEUROD1, NFU1, NGLY1, NHLRC1, NKX6-2, NOTCH3, NPC1, NPC2, NUBPL, NUS1, OAT, OCLN, OGDH, OPA1, OPA3, OTC, PAH, PAX4, PC, PCBD1, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PLA2G6, PLAA, PLEKHG2, PLIN1, PLP1, PMM2, PMPCA, PMPCB, PNPLA2, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMT1, POMT2, PPA2, PPARG, PPM1K, PPT1, PRKAG2, PRKAG3, PRODH, PSAP, PSAT1, PTRF, PTS, PUS1, PYCR2, PYGL, PYGM, QARS, QDPR, RAB11B, RAI1, RARS, RARS2, RBCK1, REPS1, RFT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RRM2B, RYR1, SAMHD1, SCN4A, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHD, SEC23B, SERAC1, SERPINA1, SFXN4, SGSH, SI, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A4, SLC2A1, SLC2A2, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, SLC3A1, SLC40A1, SLC46A1, SLC5A1, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNORD118, SOX10, SPG11, SPG7, SPTAN1, SRD5A3, SSR4, STN1, STT3A, STT3B, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, TACO1, TANGO2, TARS2, TAT, TAZ, TBC1D4, TBCD, TBCK, TCF4, TCN2, TFAM, TFR2, TGFBI, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM106B, TMEM126A, TMEM126B, TMEM165, TMEM199, TMEM70, TOP3A, TPK1, TPP1, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM37*	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA, mitochondrielle DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Stoffwechselstörungen und Mitochondriopathien erweitertes Gen-Set: *TRIT1, TRMT10C, TRMT5, TSFM, TTC19, TUBB4A, TUFM, TUSC3, TWNK, TXN2, TYMP, TYROBP, UBTF, UFC1, UFM1, UGT1A1, UMPS, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS, VARS2, VPS11, VPS33A, WARS2, WDR45, WDR45B, WFS1, YARS2, ZFYVE26, ZMPSTE24, ZNHIT3* (672 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Gastrointestinale Neoplasien: *APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, BUB1B, CDC73, CDH1, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, EPCAM, FANCC, GALNT12, GREM1, IL1B, IL1RN, KIT, MAX, MEN1, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, PTEN, RET, RHBDF2, RPS20, SDHA, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, TMEM127, TP53, TSC1, TSC2, VHL* (52 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ~~Beckman Coulter~~; ABI SeqStudio 8 Flex Genetic Analyzer System
Gynäkologische Tumore: *APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, ERCC2, ERCC3, FAM175A, LZTR1, MEN1, MITF, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, SDHA, SMAD4, SMARCB1, SPRED1, STK11, SUFU, TP53, TSC1, TSC2, XRCC2* (46 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Hauttumore: *BAP1, BRCA1, BRCA2, CDK4, CDKN2A, DDB2, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, MC1R, MITF, MLH1, MSH2, MSH6, PMS2, POT1, PTCH1, PTCH2, PTEN, RB1, SUFU, TERT, TP53, WRN, XPA, XPC* (28 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Kolorektale Karzinome: *APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, ERCC2, ERCC3, FAM175A, LZTR1, MEN1, MITF, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, SDHA, SMAD4, SMARCB1, SPRED1, STK11, SUFU, TP53, TSC1, TSC2, XRCC2* (46 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Li-Fraumeni-Syndrom erweitertes Gen-Set: *APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, ERCC2, ERCC3, FAM175A, LZTR1, MEN1, MITF, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLE, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, SDHA, SMAD4, SMARCB1, SPRED1, STK11, SUFU, TP53, TSC1, TSC2, XRCC2* (46 Gene)	EDTA-Blut, Mundschleimhaut, Chorionzotten, Fruchtwasser, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System
Lungenkarzinom: *ATM, BRCA1, BRCA2, CDKN2A, CHEK2, EGFR, FAM111B, TP53* (8 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Nierenkarzinom und Harnwegstumore: *BAP1, CCND1, CDC73, CDKN1C, CHEK2, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, REST, RNF139, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TP53, TSC1, TSC2, VHL, WT1* (35 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Pädiatrische Tumore: *ALK, AIP, APC, AXIN2, BAP1, BLM, BMPR1A, BRAF, BUB1B, CBL, CDC73, CDKN1C, CEBPA, DICER1, DIS3L2, EPCAM, EZH2, FH, GATA2, GPC3, GPR161, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, NRAS, NSD1, NSUN2, PAX5, PHOX2B, PMS2, PRF1, PRKAR1A, PTCH1, PTEN, PTPN11, RAF1, RASA2, RECQL4, REST, RET, RIT1, RRAS, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1* (72 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Pankreaskarzinom: *APC, ATM, BMPR1A, BRCA1, BRCA2, BUB1B, CHEK2, CDKN2A, EPCAM, FANCC, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PALLD, PMS2, SMAD4, STK11, TP53, TSC1, TSC2, VHL* (24 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Paragangliom / Phärochromozytom: *BAP1, CDKN1B, EGLN1, EGLN2, EPAS1, FH, KIF1B, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SRGAP1, TMEM127, VHL* (20 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Prostatakarzinom: *ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, TP53* (12 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Tumore des ZNS: *AIP, ALK, APC, BRCA2, KIF1B, MLH1, MSH2, MSH6, NF1, NF2, PHOX2B, PMS2, PTEN, SDHA, SDHB, SDHD, SMARCA4, SMARCB1, SMARCE1, SPRED1, SUFU, TP53, VHL, LZTR1* (24 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System

Tumorerkrankungen erweitertes Gen-Set: *ABCA3, ABCB7, ABCG5, ABCG8, ABRAXAS1, ACD, ACTB, ACTN1, ADAMTS13, AIP, AK1, AK2, AKT1, ALAS2, ALK, AMN, ANK1, ANKRD26, AP3B1, AP3D1, APC, ARPC1B, ATM, ATR, ATRX, AXIN2, BAP1, BARD1, BLM, BLOC1S3, BLOC1S6, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, C15ORF41, CBL, CCND1, CD70, CDAN1, CDC42, CDC73, CDH1, CDK4, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CEBPA, CEP57, CHEK2, CLCN7, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CUBN, CXCR4, CYB5R3, CYCS, CYLD, DDB2, DDX41, DHFR, DICER1, DIS3L2, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, EGFR, EGLN1, EGLN2, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPOR, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ETV6, EXO1, EXT1, EXT2, EZH2, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FAM111B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FH, FLCN, FLI1, FLNA, FYB, G6PC3, G6PD, GALNT12, GATA1, GATA2, GBA, GCLC, GFI1, GFI1B, GGCX, GINS1, GP1BA, GP1BB, GP9, GPC3, GPI, GPR143, GREM1, GSS, HAX1, HBA1, HBA2, HBB, HFE, HNF1A, HNF1B, HOXA11, HOXB13, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, IL1B, IL1RN, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KIF1B, KIF23, KIT, KITLG, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, LZTR1, MAD2L2, MAGT1, MAP2K1, MAP2K2, MASTL, MAX, MC1R, MCFD2, MECOM, MEN1, MET, MITF, MKL1, MLH1, MLH3, MPL, MRE11, MRE11A, MSH2, MSH3, MSH6, MTHFD1, MTR, MUTYH, MYH9, MYO5A, NBEAL2, NBN, NF1, NF2, NHP2, NOP10, NRAS, NSD1, NSUN2, NT5C3A, NTHL1, OCA2, P2RY12, PALB2, PALLD, PARN, PAX5, PC, PDGFRA, PDHA1, PDHX, PGM3, PHOX2B, PIEZO1, PIK3CA, PKLR, PMS1, PMS2*	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Tumorerkrankungen erweitertes Gen-Set: *POLD1, POLE, POLH, POT1, PPM1D, PRF1, PRKACG, PRKAR1A, PROC, PROS1, PTCH1, PTCH2, PTEN, PTPN11, PUS1, RAB27A, RAC2, RAD50, RAD51, RAD51C, RAD51D, RAF1, RASA2, RASAL1, RASGRP2, RB1, RBM8A, RECQL, RECQL4, REN, REST, RET, RFWD3, RHAG, RHBDF2, RINT1, RIT1, RNF139, RNF168, RNF43, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS20, RPS24, RPS26, RPS28, RPS29, RPS7, RRAS, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SCG5, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SHOC2, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCD2, SMARCE1, SOS1, SOS2, SPRED1, SPTA1, SPTB, SRC, SRGAP1, SRP54, SRP72, STK11, STX11, STXBP2, SUFU, TBXA2R, TCN2, TERC, TERT, TF, THBD, THPO, TINF2, TMEM127, TMPRSS6, TP53, TPI1, TRNT1, TSC1, TSC2, TUBB1, TYR, TYRP1, UBE2T, UNC13D, USB1, VHL, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, WRN, WT1, XIAP, XPA, XPC, XRCC2, YARS2* (373 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ~~Beckman Coulter~~; ABI SeqStudio 8 Flex Genetic Analyzer System

Primäre ciliäre Dyskinesie: *ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYX1C1, GAS8, HEATR2, HYDIN, INVS, LRRC6, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TTC25, ZMYND10* (42 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 4; SOP 68, Version 4; SOP 81, Version 1	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Ziliopathie erweitertes Gen-Set: *ACVR2B, AHI1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF2, C21ORF59, C2CD3, C5ORF42, C8ORF37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CPE, CRB2, CSPP1, DCDC2, DDX59, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH17, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DYX1C1, DYNC2H1, DYNC2LI1, EVC, EVC2, FAM58A, GAS2L2, GAS8, GLI2, GLI3, GLIS2, HEATR2, HYDIN, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT80, IFT81, INPP5E, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LEFTY2, LRRC6, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIH1D3, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCAPER, SCLT1, SDCCAG8, SPAG1, STK36, SUFU, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TTC21B, TTC25, TTC8, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, ZIC3, ZMYND10, ZNF423* (149 Gene)	EDTA-Blut, Mundschleimhaut, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Indikationsspezifisches NGS-Panel:	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 66, Version 05; SOP 67, Version 06 ; SOP 81, Version 02	NextSeq500; ABI SeqStudio 8 Flex Genetic Analyzer System
Whole Exome Sequencing	EDTA-Blut, Mundschleimhaut, Hautbiopsat, genomische DNA; genomische DNA	PCR/DNA-Sequenzierung, NGS Sequence capture, Sequencing-by-synthesis, Auswertesoftware: MaiVarView	SOP 68, Version 05; SOP 90, Version 01; SOP 81, Version 02	NextSeq2000; ABI SeqStudio 8 Flex Genetic Analyzer System