Institut für Humangenetik und
Medizinisches Versorgungszentrum der UMG
Bereich Humangenetik
Heinrich-Düker-Weg 12
37073 Göttingen

UNIVERSITÄTSMEDIZIN GÖTTINGEN UMG

Tagesaktuelle Liste FB-533

Untersuchungsverfahren

Gültig ab: 15.11.2021

Vorlage Version: FB_1.0-0421

Untersuchungsgebiet: Humangenetik

Untersuchungsart: Chromosomenanalyse *

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Angeborener Chromosomensatz	Heparin-Blut, Fibroblastenkultur, Fruchtwasser	Chromosomenbänderungsanalyse	AA-201 / 1.5; AA-206 / 1.5; AA-216 / 1.1	Metasystems Ikaros		x
Angeborener Chromosomensatz	Fruchtwasser	FISH (Fluoreszenz-in-situ-Hybridisierung) mit spezifischen Sonden	AA-205 / 1.6	Zeiss Axio Imager.Z2, Metasystems Isis	x	
Angeborener Chromosomensatz	EDTA-Blut, DNA, kultivierte Amnionzellen	Molekulare Karyotypisierung (Array-based Comparative Genomic Hybridization, array-CGH)	AA-209 / 1.2; AA-114 / 1.5	Agilent Scanner G2539A		x

Untersuchungsart: Molekularbiologische Untersuchungen (Amplifikationsverfahren) **

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Schwerhörigkeit, sensorineural: *GJB2, GJB6*	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR)	AA-045 / 1.4; AA-046 / 1.4; AA-098 / 1.6; AA-099 / 1.8; AA-100 / 1.5; AA-101 / 1.5; AA-110 / 1.6; AA-134 / 1.5; AA-209 / 1.3	3500xl Genetic Analyzer, 7900HT Real-Time PCR System		x
Molybdän-Cofaktor Defizienz (MOCS1, MOCS2)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	AA-059 / 1.3; AA-060 / 1.3; AA-098 / 1.6; AA-099 / 1.8; AA-110 / 1.6; AA-209 / 1.3	3500xl Genetic Analyzer		x
Thrombophilie (F5-Gen Leiden Variante)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	AA-081 / 1.3; AA-098 / 1.6; AA-099 / 1.8; AA-110 / 1.6; AA-209 / 1.3	3500xl Genetic Analyzer		x
Thrombophilie (Faktor II-Gen-3' Polymorphismus)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	AA-076 / 1.3; AA-098 / 1.6; AA-099 / 1.8; AA-110 / 1.6; AA-209 / 1.3	3500xl Genetic Analyzer		x
Homozysteinämie (MTHFR)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung	AA-086 / 1.4; AA-087 / 1.4; AA-098 / 1.6; AA-099 / 1.8; AA-110 / 1.6; AA-209 / 1.3	3500xl Genetic Analyzer		x

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Yq11.21-23 Mikrodeletionen	EDTA-Blut, DNA	PCR	AA-089 / 1.3; AA-116 / 1.3; AA-098 / 1.6; AA-110 / 1.6; AA-209 / 1.3	BioRad C1000; ABI TC2720		x
Fragiles X-Syndrom: *FMR1*	EDTA-Blut, DNA	PCR, Fragmentanalyse	AA-098 / 1.6; AA-103 / 1.5; AA-122 / 1.6; AA-209 / 1.3; AA-248 / 1.3; AA-249 / 1.3; AA-250 / 1.3	BioRad C1000; 3130 Genetic Analyzer		x
Lynch-Syndrom / Hereditäres Nicht-Polypöses Kolorektales Karzinom (HNPCC): *EPCAM , MLH1, MSH2, MSH6, PMS2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Familiärer Brust- und Eierstockkrebs: *BRCA1, BRCA2, ATM, CHD1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, BARD1, BRIP1, STK11, TP53*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Polyposis-Syndrome (Familiäre adenomatöse Polyposis (FAP), MUTYH-assoziierte Polyposis (MAP), Cowden-Syndrom, Peutz-Jeghers-Syndrom, Hyperplastisches Polyposis-Syndrom (HPS), Serratiertes Polyposis-Syndrom (SPS), Hereditäres Mixed Polyposis-Syndrom	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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(HMPS)): *APC, MUTYH, PIK3CA, PTEN, STK11, RNF43, GREM1*			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Juvenile Polyposis: *BMPR1A, ENG, PTEN, SMAD4, STK11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Polyposis, unbestimmt: *APC, MSH3, MUTYH, NTHL1, POLD1, POLE, BMPR1A, ENG, GREM1, MLH1, MSH2, MSH6, PMS2, PTEN, RNF43, SMAD4, STK11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Darmkrebs (CRC), unbestimmt: *APC, BMPR1A, ENG, EPCAM, FAN1, FLCN, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SETD6, SMAD4, STK11, TP53*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Gastrointestinale Stromatumore: *KIT, MAX, MEN1, NF1, PDGFRA, SDHAF2, SDHB, SDHC, SDHD, TMEM127*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4;	Genetic Analyzer 3130, Genetic Ana-		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Li-Fraumeni-Syndrom: *TP53*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Magenkarzinom: *CDH1, ATM, BMPR1A, CHEK2, MLH1, MSH2, MSH6, PMS2, STK11, TP53, CTNNA1, EPCAM, PDGFRA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Malignes Melanom: *CDKN2A, BAP1, BRCA2, POT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Nierenzellkarzinom: *BAP1, DICER1, EPCAM, FH, FLCN, MET, PALB2, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TSC1, TSC2, VHL*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Familiäres Pankreaskarzinom: *BRCA1, BRCA2, CDKN2A, CHEK2, CFTR, PALB2, STK11, APC, ATM, CFTR, MLH1, MSH2, MSH6, PALLD, PMS1, PMS2, PRSS1, PTEN, RABL3, SPINK1, VHL, TP53*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Fanconi Anämie: *BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, UBE2T, XRCC2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Phäochromozytome und Paragangliome / Multiple endokrine Neoplasien: *CDC73, CDKN1B , DLST, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Prostatakarzinom: *BRCA1, BRCA2, CHEK2, MSR1, RNASEL, HOXB13, PALB2, RAD51D*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Retinoblastom: *RB1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Schilddrüsenkarzinom: *PTEN, RET, SDHB, SDHC, SDHD, APC, ATM, CHEK2, MEN1, MUTYH, SDHAF2, STK11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Von-Hippel-Lindau-Syndrom: *VHL*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Makrozephalie (Basisdiagnostik): *CHD8, DNMT3A, EZH2, NSD1, NFIX, PTEN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Weaver-Syndrom: *EZH2, EED, SUZ12, HACE1, NSD1, NFIX*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Sotos-Syndrom: *NSD1, NFIX, EZH2, EED, SUZ12, HACE1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Beckwith-Wiedemann-Syndrom: *H19, KCNQ1OT1, CDKN1C, NSD1, ICR1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Alexander-Krankheit: *GFAP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Bannayan-Riley-Ruvalcaba-Syndrom, Lhermitte-Duclos-Syndrom, Cowden syndrome 1, Macrozephalie/Autismus-Syndrom: *PTEN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Brittle Cornea-Syndrom (BCS): *ZNF469*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Dysplasie, kranio-metaphysäre (CMD): *ANKH, GJA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Greig-Zephalopolysyndaktylie-Syndrom: *GLI3, KIF7*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Joubert-Syndrom 12, Akrokallosales Syndrom: *KIF7*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Hydrozephalus mit Morbus Hirschsprung: *L1CAM*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kosaki-Syndrom (KOGS): *PDGFRB*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Krämpfe-Skoliose-Makrozephalie –Syndrom (SSMS): *EXT2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Luscan-Lumish-Syndrom (LLS): *SETD2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
MACS-Syndrom / RIN2-Syndrom: *RIN2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Makrozephalie mit Makrosomie und Gesichtsdysmorphien (MMFD): *RNF135*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Makrozephalie/Megalenzephalie-Syndrom (MGCPH): TBC1D7	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Megalenzephalie-Polymikrogyrie-Polydaktylie-Hydrozephalus-Syndrom (MPPH1-3): *PIK3R2, AKT3, CCND2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Megalenzephalen Leukoenzephalopathie mit subkortikalen Zysten (MLC): *HEPACAM, MLC1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Megalenzephalie-Kapillar-Malformation-Polymikrogyrie-Syndrom (MCAP): *PIK3CA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Mentale Retardierung-Makrozephalie, autosomal rezessiv (MRT41): *KPTN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Mentale Retardierung-Makrozephalie, Suszeptibilität für Autismus (AUTS18): *CHD8*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Mentale Retardierung-Makrozephalie, X-chromosomal (Cabezas; MRX93; MRX72; Turner type): *CUL4B, BRWD3, RAB39B, HUWE1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Opitz-Kaveggia-Syndrom (OKS): *MED12*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Perlman-Syndrom (PRLNMS): *DIS3L2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Phelan-McDermid-Syndrom: *SHANK3*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Polyhydramnion, Megalenzephalie und Symptomatische Epilepsie: *STRADA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Proteus-Syndrom: *AKT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Simpson-Golabi-Behmel-Syndrom (SGBS): *GPC3, OFD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Smith-Kingsmore-Syndrom (SKS): *MTOR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Illumina MiSeq, Illumina Nextseq550		x
Tatton-Brown-Rahman-Syndrom (TBRS): *DNMT3A*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Tenorio-Syndrom (TNORS): *RNF125*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Makrozephalie-Gesamt-Diagnostik: *AKT1, AKT3, ANKH, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, EED, EXT2, EZH2, GFAP, GJA1, GLI3, GPC3, H19, HACE1, HEPACAM, HUWE1, KCNQ1OT1, KIF7, KPTN, L1CAM, MED12, MLC1, MTOR, NFIX, NSD1, OFD1, PDGFRB, PIK3CA, PIK3R2, PTEN, RAB39B, RIN2, RNF125, RNF135, SETD2, SHANK3, STRADA, SUZ12, TBC1D7, ZNF469, HIST1H1E, ASPA, KIDINS220, MSL3, BMP4, BRAF, DEPDC5, FIBP, FOXP1, HRAS, KRAS, MAP2K1, MAP2K2, MYCN, NFIA, NFIB, NOTCH2NL, NRAS, ODC1, PTCH1, PTPN11, RAF1, RHEB, ROR2, SHOC2, SOS1, SPRED1, SUFU, SZT2, TSC1, TSC2, ZBTB20*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Primäre Mikrozephalie (MCPH): *ASPM, CDK5RAP2, CEP152, MCPH1, ANKLE2, ARX, BRAT1, CASC5, CASK, CDC6, CDK5, CDK6, CDT1, CENPE, CENPJ, CEP135, CEP63, CKAP2L, DCX, DDX11, DHCR7, DNA2,*						

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GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MFSD2A, NBN, NHEJ1, NIN, ORC1, ORC4, ORC6, PAFAH1B1 (LIS1), PCNA, PCNT, PHC1, PPM1D/WIP1, RAD50, RBBP8, RELN, RMI1, RNU4atac, RTTN, SASS6, STIL, TRAIP, TUBA1A, WDR62, XRCC4, XRCC9, ZNF335, EFTUD2, OSGEP, TP53RK, TPRKB, LAGE3, PNKP, WDR73, NCAPH2, CIT, NSMCE2, NEK3, CEP350, FANCM, CSNK2B, PYCR2, WRN, NUP133, NUP107, WDR4, ZEB2, TRMT1, TRAPPC9, NMT3A, DYNC1I2, VPS51, NDE1, BUB1B, KIF5C, KIF2A, TUBG1, TUBB2B, TBCD, POC1A , CHAMP1, MRE11A, STAMBP, COPB2						
Seckel-Syndrom (SKS): *CEP63, CEP152, CENPJ, CDK5RAP2, DNA2, RBBP8, TRAIP, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK6, CDT1, CENPE, CEP135, CKAP2L, DCX, DDX11, DHCR7, ERCC4, ERCC6, FAAP95 (FANCB), FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NEHJ1, NBN, NIN, ORC1, ORC4,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RELN, RMI1, RNU4atac, RTTN, SASS6, SLX4, STIL, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335						
LIG4-Syndrom (LIG): LIG4, XRCC4, BLM, DDX11, FANCD2, NBN, NHEJ1, PCNA, PPM1D, RAD50, ANKLE2, ARX, ASPM, ATR, ATRIP, B9D1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, MCPH1, MFSD2A, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNT, PHC1, PHF6, PHF9, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC9, ZNF335	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Meckel-Syndrom (MKS): *CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TMEM216, B9D1, B9D2, KIF14, TCTN2, TMEM67, TMEM231*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
			AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Kleinwuchs, mikrozephaler osteodysplastischer primordialer, Typ I und Typ 2 (MOPD1 und MOPD2 (MOPD)): *PCNT, RNU4ATAC, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Meier-Gorlin-Syndrom (MGS): *CDT1, CDC6, GMNN, ORC1, ORC4, ORC6, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDK5, CDK5RAP2, CDK6, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RNU4ATAC RMI1, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Nijmegen-Breakage-Syndrom (NBS): *NBN, RAD50, DDX11, BLM, FANCD2, LIG4, NHEJ1, PCNA, PPM1D, XRCC4, ANKLE2, ARX, ASPM, ATR, ATRIP, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, MCPH1, MFSD2A, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNT, PHC1, PHF6, PHF9, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC9, ZNF335*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Fanconi-Anämie (FA): *BRCA2, FANCA, FANCC, FANCD2, FANCE, XRCC9 (FANCG), ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95 (FANCB), FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PCNT, PALB2, PCNA, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, ZNF335*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Bloom-Syndrom (BS): *BLM, DDX11, FANCA, FANCC, NBN, RAD50, RMI1, XRCC9 (FANCG), ANKLE2, ARX, ASPM, ATR, ATRIP, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCD2, FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD51C, RBBP8, RELN, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, ZNF335						
Zerebro-Okulo-Fazio-Skeletales-Syndrom (COFS): *ERCC1, ERCC2, ERCC5, ERCC6, ERCC8, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CDK5RAP2, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, CKAP2L, DDX11, DNA2, ERCC4, FAAP95, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCI, GMNN, KMT2B, LIG4, NHEJ1, NBN, ORC1, ORC4, ORC6, PALB2, PIEZO2, PCNA, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RMI1, RTTN, SLX4, STIL, TRAIP, WDR62, XRCC4, XRCC9*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Filippi-Syndrom (FS): *CKAP2L, ANKLE2, ARX, ASPM, ATR, ATRIP, BLM, BRAT1, BRCA2, BRIP1, CASC5, CASK, CDC6, CDK5, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP63, CEP135, CEP152, DDX11, DCX, DHCR7, DNA2, ERCC4, ERCC6, FAAP95, FANCA, FANCC, FANCD2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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FANCE, FANCF, FANCI, GMNN, HNRNPU, KATNB1, KIF11, KMT2B, LAMB1, LIG4, MCPH1, MFSD2A, NHEJ1, NIN, NBN, ORC1, ORC4, ORC6, PAFAH1B1(LIS1), PALB2, PCNA, PCNT, PHC1, PHF6, PHF9, PPM1D, RAD50, RAD51C, RBBP8, RELN, RMI1, RNU4ATAC, RTTN, SASS6, SLX4, STIL, TRAIP, TUBA1A, UBE2T, WDR62, XRCC4, XRCC9, ZNF335						
Galloway-Mowat-Syndrom (GMS): *LAGE3, OSGEP, TP53RK, TPRKB*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hutchinson-Gilford-Progerie-Syndrom (HGPS): *LMNA, BANF1, FBN1, NARF, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1 ,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
(XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, WRN, XPA, XPC						
Mandibuloakrale Dysplasie (MAD): *LMNA, POLD1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, WRN, XPA, XPC						
Nestor-Guillermo-Progerie-Syndrom (NGPS): *BANF1, LMNA, FBN1, NARF, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Wiedemann-Rautenstrauch-Syndrom (WRS): *ANO6, CAV1, FBN1, LMNA, BANF1, PYCR1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ATM, ATP6V0A2, ATP7A, B3GALT6, B4GALT7, BLM,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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BRCA2, BRIP1,BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Werner-Syndrom (WS): *WRN, LMNA, POLD1, SPRTN, ZMPST24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC						
Ruijs-Aalfs-Syndrom (RAS): *SPRTN, WRN, LMNA, POLD1, ZMPST24, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC						
Cockayne-Syndrom (CS): *ERCC6, ERCC8, ERCC4, ERCC5, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Xeroderma pigmentosum (XP): DDB2, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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DKC1, EFEMP2, ELN, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, ZMPSTE24			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Bloom-Syndrom mit progeroiden Symptomen (BSPS): *BLM, PRKDC, SPRTN, WRN, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC, ZMPSTE24						
Lipodystrophie (LD): *AGPAT2, BSCL2, CAV1, CIDEC, LIPE, PLIN1, PPARG, PTRF, FBN1, LMNA, ZMPSTE24, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, CHD6, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Marfan-Lipodystrophie-Syndrom (MLS): *FBN1, AGPAT2, B3GALT6, B4GALT7, BSCL2, CAV1, LIPE, LMNA, PLIN1, PIK3R1, PPARG, PTRF, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, BLM, BRCA2, BRIP1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Short-Syndrom (STS): *PIK3R1, AGPAT2, BSCL2, CAV1, CIDEC, FBN1, LIPE, LMNA, PLIN1, PPARG, PTRF, ACD, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1, CHD6, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24						
Keppen-Lubinsky-Syndrom (KLS): *KCNJ6, ANO6, CAV1, LMNA, PIK3R1, POLD1, PYCR1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, LIPE, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC						
Progerie Typ Penttinen (PTP): *PDGFRB, B3GALT6, B4GALT7, CAV1, FBN1, LIPE, LMNA, PLIN1, PIK3R1, PPARG, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, BLM, BRCA2, BRIP1, BSCL2, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBLN5, HELLS, KCNJ6, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PEX11B, POLD1, POLH, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Cutis laxa assoziierte Syndrome (CLS): *ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PEX11B,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4;	Genetic Analyzer 3130, Genetic Ana-		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
PYCR1, ACD, AGPAT2, ANO6, ATM, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, HELLS, KCNJ6, LIPE, LMNA, LMNB1, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Lenz-Majewski-Syndrom (LMS): *PTDSS1, ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PEX11B, PYCR1, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
(FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPATG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24						
Rothmund-Thomson-Syndrom (RTS): *RECQL4, ACD, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, MRE11A, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24						
Dyskeratosis Congenita (DKC): *ACD, DKC1, NOLA2, NOLA3, PARN, RTEL1, TERC, TERT, TINF2, WRAP53, AGPAT2, ALDH18A1, ANO6, ATM, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1, LTPB4, MRE11A, NAA10, NARF, NEHJ1, PALB2, PCNA, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, SLX4, SMC2, SMC4, SPRTN, TINF2, TFAP2A, UBE2T, WFS1, WRN, XPA, XPC, ZMPSTE24*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Ataxia-Teleangiektasia/Louis-Bar-Syndrom (LBS): *ATM,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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MRE11A, PCNA, ACD, AG-PAT2, ALDH18A1, ANO6, ATP6V0A2, ATP7A, BANF1, B3GALT6, B4GALT7, BLM, BRCA2, BRIP1,BSCL2, CAV1, CHD6, CIDEC, CISD2, DDB2, DKC1, EFEMP2, ELN, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FAM111A, FANCA, FANCB (FAAP95), FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCL (PHF9), FBN1, FBLN5, HELLS, KCNJ6, LIPE, LMNA, LMNB1,LTPB4, NAA10, NARF, NEHJ1, NOLA2, NOLA3, PALB2, PARN, PDGFRB, PEX11B, PIK3R1, PLIN1, POLD1, POLH, PPARG, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, RECQL4, RIN2, RTEL1, SLX4, SMC2, SMC4, SPRTN, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRN, WRAP53, XPA, XPC, ZMPSTE24		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Hereditäre spastische Paraplegie (HSP), autosomal-dominant: *ATL1 (SPG3A), BICD2, BSCL2, CPT1C, HSPD1, WASHC5 (SPG8), KIF5A, NIPA1, REEP1 (SPG31), RTN2, SLC33A1, SPAST (SPG4), VAMP1, ZFYVE27*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Hereditäre spastische Paraplegie (HSP), autosomal-rezessiv und X-chromosomal: *CYP7B1 (SPG5A), FA2H, GJC2, SPG11, SPG7, ZFYVE26, ABCD1, L1CAM, PLP1, SLC16A2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATP13A2, B4GALNT1, CAPN1, C12orf65, C19orf12, CCT5, CYP2U1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FAM134B, FARS2, FLRT1, GAD1, GBA2, GPT2, GRID2, HACE1, IBA57, KIF1A, KIF1C, KLC2, KLC4, LYST, MAG, MARS2, NT5C2, PGAP1, RAB3GAP2, REEP2, SACS, SETX, SOD1, SPG20, TECPR2, TFG, UCHL1, USP8, VPS37A, WDR48, ZFR, CYP7B1 (SPG5A), FA2H, GJC2, SPG11, SPG7, ZFYVE26, L1CAM, ABCD1, PLP1, SLC16A2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hereditäre spastische Paraplegie (HSP), unbestimmt: *ATL1 (SPG3A), CYP7B1 (SPG5A), SPG11, KIF1A, KIF5A, REEP1 (SPG31), SPAST (SPG4), SPG7, ABCD1, ACP33, ADAR, AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, B4GALNT1, BICD2, BSCL2, C12orf65,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
C19orf12, CCT5, CPT1C, CYP2U1, DDHD1, DDHD2, DNM2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAM134B, FARS2, FLRT1, FUS, GAD1, GBA2, GJC2, GRID2, IBA57, IFIH1, WASHC5 (SPG8), KIF1C, KLC2, KLC4, L1CAM, LYST, MAG, MARS2, NIPA1, NT5C2, PGAP1, PLP1, PMCA4, PNPLA6, RAB3GAP2, REEP2, RNA-SEH2B, RTN2, SETX, SLC16A2, SLC33A1, SOD1, SOX10, SPG20, TARDBP, TECPR2, TFG, TUBB4A, USP8, VAMP1, VCP, VPS37A, WDR48, ZFR, ZFYVE26, ZFYVE27, ATP13A2, UCHL1, CAPN1, DSTYK, GPT2, HACE1, VCP, SACS, VAMP1, AFG3L2, MARS2, , ABCD1, SETX, OPTN, FUS, TARDBP, SOX10, NKX6-2, KCNA1, KI-DINS220, UBAP1, AGTPBP1, SLC2A1 , PLA2G6						
Spastische Ataxien: *AFG3L2, KIF1C, MARS2, SACS, SOX10, SPG7, VAMP1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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ALS-ähnliche Hereditäre Spastische Paraplegie (HSP): *ALS2, SETX, SPG11, SOD1, TARDBP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hereditäre Motorisch-Sensible Neuropathien/Charcot-Marie-Tooth-Neuropathien (HMSN/CMT), Distale Hereditäre Motorische Neuropathie (dHMN), Hereditäre Sensible und Autonome Neuropathien (HSAN): *AARS, AIFM1, ARHGEF10, ATL1, ATL3, ATP7A, BSCL2, CCT5, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IGHMBP2, IKBKAP, INF2, JPH1, KARS, KIF1A, KIF1B, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRDM12, PRPS1, PRX, RAB7, REEP1, SBF1, SBF2/MTMR13, SCN11A,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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SCN9A, SETX, SH3TC2, SLC12A6, SLC5A7, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TDP1, TFG, TRIM2, TRPV4, VCP, WNK1, YARS						
Hereditäre Neuropathie mit Neigung zu Druckparesen (HNPP): *PMP22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kabuki-Syndrom: *KMT2D, KDM6A, RAP1A, RAP1B*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Rasopathien/Noonan-Syndrom: *PTPN11, A2ML1, BRAF, CBL, CDC42, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS,RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Costello-Syndrom (CSTLO): *HRAS*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Leopard-Syndrom (LPRD): *BRAF, PTPN11,RAF1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kardiofaziokutanes Syndrom (CFC): *BRAF, MAP2K1, MAP2K2, KRAS*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
CHARGE-Syndrom: *CHD7, SEMA3E, TBX1, TBX22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
DiGeorge-Syndrom (DGS): *TBX1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Abruzzo-Erickson-Syndrom (ABERS), CHARGE-like Syndrom: *TBX22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Neurofibromatose: *NF1, NF2, SPRED1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Legius-Syndrom: *SPRED1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Robinow-Syndrom: *WNT5A, DVL1, DVL3, ROR2, FGD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Aarskog-Syndrom: *FGD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Cornelia-de-Lange-Syndrom: *NIPBL, DXS423E / SMC1A, SMC3, RAD21, HDAC8, AFF4, SETD5, KMT2A, MAU2, BRD4*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
CHOPS-Syndrom: *AFF4*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
KBG-Syndrom (KBGS): *ANKRD11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Trichorhinophalangeales Syndrom 1 und 3 (TRPS1 und TRPS3): *TRPS1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Coffin-Siris-Syndrom: *ARID1B, ARID1A, ARID2, DPF2, SMARCB1, SMARCA4, SMARCE1, SMARCA2, SOX11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Nicolaides-Baraitser-Syndrom (NCBRS): *SMARCA2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Baraitser-Winter-Syndrom (BRWS1): *ACTB, ACTG1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Borjeson-Forssman-Lehmann-Syndrom (BFLS): *PHF6*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Cutis laxa (ADCL1) / Williams-Beuren-Syndrom (WBS): *ELN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Simpson-Golabi-Behmel-Syndrom (SGBS): *GPC3, GPC4, OFD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Townes-Brocks-Syndrom (TBS): *SALL1, DACT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Duane-Radial Ray-Syndrom (DRRS) / Okihiro-Syndrom: *SALL4*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Branchiooto(renales) Syndrom 1 und 2 (BOR1, BOR2): *EYA1, SIX1, SIX5*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Rubinstein-Taybi-Syndrom (RSTS): *CREBBP, EP300*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Floating-Harbor-Syndrom (FLHS): *SRCAP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Coffin-Lowry-Syndrom (CLS): *RPS6KA3*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Okulodentodigitale Dysplasie (ODDD): *GJA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Ritschner-Schintzel-Syndrom (RTSC): *WASHC5/KIAA0196, CCDC22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Lujan-Fryns-Syndrom (LFS), Opitz-Kaveggia-Syndrom (OKS): *MED12*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Cohen-Syndrom (COH): *VPS13B*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Mental retardation, autosomal dominant (MRD32): *KAT6A*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Glass-Syndrom: *SATB2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Schuurs-Hoeijmakers-Syndrom (SHMS): *PACS1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Kraniosynostosen (CRAN): *FGFR1, FGFR2, FGFR3, GLI3, MEGF8, RAB23, TWIST1, ADAMTS10, ALX1, ALX3, ALX4, CD96, EFNB1, ERF, IFT122, IFT43, IL11RA, LRP2, MN1, P4HB, POR, RECQL4, RUNX2, SEC24D, SKI, SMO, WDR19, WDR35, ZIC1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kardiomyopathie, dilatativ (DCM): *DMD, LMNA, MYBPC3, MYH7, TNNT2, TPM1, ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CASQ2, CRYAB, CSRP3, CFT1, DES, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKTN, GATAD1, ILK, LAMA2, LAMA4, LAMP2, LDB3, MURC, MYH6, MYL2, MYL3, MYPN, NEXN, NPPA, PDLIM3, PLN, PRDM16, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TBX5, TCAP, TMPO, TNNC1, TNNI3, TTR, VCL*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kardiomyopathie, hypertroph (HCM): *ACADVL, ACTC1, CSRP3, FHL1, MYBPC3, MYH7, MYL2, MYL3, MYPN, PLN, PRKAG2, TNNC1,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
TNNI3, TNNT2, TPM1, TRIM63, ACTN2, ANKRD1, CACNA1C, CALR3, CASQ2, CAV3, CBL, COX15 CRYAB, DES, FHL2, FXN, GLA, GAA, JPH2, KLF10, LDB3, MAP2K1, MAP2K2, MYH6, MYLK2, MYO6, MYOZ2, NEXN, PDLIM3, RAF1, RYR2, SLC25A4, TCAP, TTR, VCL			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Arrythmogene rechtsventrikuläre Kardiomyopathie / Dysplasie (ARVC/D): *DSC2, DSG2, DSP, JUP, PKP2, TGFB3, TMEM43, CASQ2, RYR2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Non-compaction Kardiomyopathie (NCCM): *ACTC1, HCN4, MIB1, MYBPC3, MYH7, NEXN, PRDM16, TAZ, TPM1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kardiomyopathie, restriktiv (RCM): *ACTC1, DES, MYH7, MYL2, MYPN, TNNI3, TNNT2, TPM1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Morbus Fabry: *GLA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): *CASQ2, CALM1, KCNE1, KCNJ2, RYR2, TRDN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Long QT-Syndrom (LQT): *ANK2, KCNE1, KCNE2, KCNH2, KCNQ1, SCN5A, AKAP9, CACNA1C, CALM1, CAV3, KCNE3, KCNJ2, KCNJ5, RYR2, SCN4B, SNTA1, TRDN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Short QT-Syndrom (SQT): *CACNA1C, KCNH2, KCNQ1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Brugada-Syndrom: *CACNA1C, CACNA2D1, CACNB2, PKP2, SCN1B, SCN5A, TRPM4, AKAP9, CALM1, CAV3, KCNE3, KCNJ2, KCNJ5, RYR2, SCN4B, SNTA1, TRDN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hypercholestrinämie (LDLR_Defizienz): *LDLR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hypercholesterinämie (primär): *APOB, PCSK9, LDLRAP1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Marfan-Syndrom: *FBN1, TGFBR1, TGFBR2, ACTA2, COL3A1, MYH11, MYLK, SMAD3, TGFB2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Ehlers-Danlos-Syndrom (vaskuläre Form): *COL3A1, ACTA2, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Thorakale Aortenerweiterung: *ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Bindegewebserkrankung mit Gefäßbeteiligung: *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, LTPB2, MYH11,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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MYLK, MYLK2, NOTCH1, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1, TGFBR2			AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Maturity-onset diabetes of the young (MODY): *ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Familiärer Hyperinsulinismus / Hyperinsulinämische Hypoglykämie (HHF): *ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1, UCP2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Familiäre Hypercholesterinämie (FH): *APOB, LDLR, LDLRAP1, PCSK9*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Familiäre hypokalzurische Hyperkalzämie (HHC): *CASR, AP2S1, GNA11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kallmann-Syndrom / Isolierter Hypogonadotroper Hypogonadismus (IHH): *ANOS1/KAL1, AXL, CCDC141, CHD7, DUSP6, FEZF1, FGF8, FGF17, FGFR1, FLRT3, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, NSMF, POLR3B, PROK2, PROKR2, TAC3, TACR3, SEMA3A, SOX10, SPRY4, SRA1, WDR11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Morbus Osler / Hereditäre Hämorrhagische Teleangiektasie (HHT): *ACVRL1, ENG, SMAD4, GDF2, RASA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Cystische Fibrose (CF): *CFTR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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			AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Familiäre Skeletterkrankungen: *ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS15, ADAMTS17, ADAMTS2, ADAMTSL2, APC2, AGPS, AIFM1, ALDH18A1, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR1, ARHGAP31, ARL6, ARSB, ARSE, ASXL1, ATP6V0A2, ATP6V1E1, ATP6V1A, ATP7A, ATR, AXIN1, B3GALT6, B3GALTL, B3GAT3, B4GALT7, B9D1, B9D2, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BGN, BHLHA9, BMP1, BMP4, BMPER, BMPR1B, C12ORF57, CA2, CANT1, CASR, CC2D2A, CCDC28B, CCDC8, CD96, CDC6, CDC45, CDC73, CDH3, CDKN1C, CDT1, CENPE, CENPJ, CEP152, CEP290, CEP63, CHST14, CHST3, CHSY1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL27A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CREB3L1, CREBBP, CRTAP, CSF1R, CSGALNACT1, CTSA, CTSC,*						

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Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
CTSK, CUL7, OFD1, DCHS1, DDR2, DDRGK1, DDX59, DHCR7, DHODH, GNPAT, DKK1, DLL3, DLL4, DLX3, DLX5, DLX6, DMP1, DNA2, DSE, DSPP, DVL1, DVL3, DYM, DYNC2H1, EBP, EF-EMP2, EFNB1, EFTUD2, EHMT1, EIF2AK3, ELN, EN1, ENPP1, EOGT, EP300, ERCC2, ERCC6, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, FAM111A, FAM20B, FAM20C, FAM58A, FAR1, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXW4, FERMT3, FGD1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FN1, FREM1, FZD2, GALNS, GALNT3, GBA, GCM2, GDF3, GDF5, GDF6, GHR, GJA1, GLA, GLB1, GLI3, GMNN, GNAI3, GNAS, GNPAT, GORAB, GPC3, GPC6, GPX4, GSC, H19, HAAO, HDAC4, HDAC6, HDAC8, HES7, HOXA11, HOXA13, HOXD13, HPGD, HRAS, HSPA9, HSPG2, I-ARS2, ICK, ID, IDH , IDS, I-DUA, IFITM5, IFT122, IFT43, IFT80, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, INPPL1, IRX5, ITGB3, KAT6B, KDELR2, KIAA0196/WASHC5, KIF22, KIF7, KL, KYNU, LBR,						

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
LEMD3, LEPRE1, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LPIN2, LRRK1, LRP4, LRP5, LRP6, LTBP2, LTBP3, LTBP4, LZTFL1, MAFB, MAN2B1, MAP2K1, MATN3, MED12, MEGF8, MEOX1, MESD, MESP2, MGP, MIR17HG, MIR140, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MTAP, MYCN, MYH11, MYLK, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NFIX, NIN, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRAS, NSDHL, NT5E, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, PAM16, PAPSS2, PAX1, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PHEX, PIK3R1, PISD, PITX1, PLEKHM1, PLCB3, PLOD1, PLOD2, PLOD3, PLS3, POC1A, POLD1, POLE, POLR1C, POLR1D, POLR3A, POP1, POR, PPIB, PRDM5, PRG4, PRKAR1A, PRKG1, PTCH1, PTCH2, PTDSS1, PTH1R, PTHLH, PTPN11, PVRL4, PYCR1, RAB23, RAB33B, RAD21, RAD51C, RASA1, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RIN2, RIPPLY2, RMRP, RNPC3, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RPS6KA3, RSPO2, RSPRY1						

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
, RUNX2, SALL1, SALL4, SAMD9, SBDS, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOX, SKI ,SLC17A5, SLC26A2, SLC29A3, SLC2A10, SLC34A1, SLC34A3, SLC35D1, ERC4, ERC5, ERC6, ERC7, SOX9, SP7, SPARC, STX16, SUFU, SUMF1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, THPO, TMCO1, TMEM165, TMEM38B, TMEM67, TMEM216, TNFRSF11A, TNFRSF11B, TNFSF11, TNXB, TONSL, TP63, TRAF6, TRAIP, TRAPPC2, TREM2, TRIM32, TRIP11, TRPS1, TRPV4, TTC8, TWIST1, TYROBP, USB1, VDR, WDPCP, WDR35, WDR60, WISP3, WNT1, WNT10B, WNT16, WNT3, NT5A, WNT7A, XYLT1, XYLT2, ZAK, ZBTB16, ZMPSTE24, ZNF469, ZNF521, ZSWIM6, SMAD6, MECOM, MBTPS2, ZIC1						
Microphthalmie (MCOP): *ABCB6, ALDH1A3, ALX1, ALX3, ALX4, BCOR, BMP4, BMP7, CENPF, CHD6,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4;	Genetic Analyzer 3130, Genetic Ana-		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
CHD7, CRIM1, FAM111A, FOXE3, FREM1, GDF3, GDF6, GLI2, HCCS, HMGB3, HIST1H2BJ, KDM6A, KMT2D, LRP5, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OTX2, PAX6, PORCN, PRSS56, RARB, RAX, RBP4, SALL2, SALL4, SHH, SIX6, SMC2, SMC4, SMCHD1, SMO, SMOC1, SOX2, STRA6, TCTN2, TENM3, TFAP2A, VAX1, VSX2		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Erbliche Schwerhörigkeit (DFN): *ACTG1, ADCY1, ADGRV1, AIFM1, ATP2B2, ATP6B1 (ATP6V1B1), CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, CLRN1-AS1, COCH, COL11A1, COL11A2, COL4A3, COL4A4, COL4A5, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GRHL2, GRXCR1, GRXCR2, GSDME (DFNA5), HARS, HGF, HOMER2, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MCM2, MET,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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MIR96, MITF, MSRB3, MTCO1, MTND1, MTRNR1, MTTH, MTTI, MTTL1, MTTS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PJVK, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, ROR1, S1PR2, SANS (USH1G), SERPINB6, SIX1, SIX5, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC44A4, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TECTA, TECTA, TIMM8A, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, USH2A, WBP2, WFS1, WHRN, WS2B, RIMBP2, ATP1A3, LRBA, ATP11A, PPIP5K2, SYNJ2, CEP78, MRPS2, TD, PHYH, PEX7, ARSG, ABCA4, ABHD12, ALMS1, ANKH, BCAP31, BCS1L, BDP1, BSND, C10ORF2, CATSPER2, CD151, CDKN1C, CHD7, CHSY1, COL2A1, COL9A1, COL9A2, DNMT1, GPRASP2, GPSM2, HARS2, HOXB1, HSD17B4, LARS2, MANBA, NDP, NLRP3, POLR1C, POLR1D, RPE65, SALL1, SEMA3E, SLC19A2, SPATA5, TCOF1,						

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
TFAP2A, TJP2, TMC2, TYR, KARS1, LARS2, TUBB4B						
Familiäre neurodegenerative Erkrankungen: *ACOX1, AARS2, ABCB7, ABCD1, ADAR, ADCK3, ADCY5, AFG3L2, ALDH3A2, ANG, ANO10, ANO3, ANXA11, APTX, ARSA, ASPA, ATCAY, ATM, ATN1, ATP13A2, ATP1A3, ATP2B3, ATP6AP2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, BEAN1, C21orf2, C9orf72, CA8, CACNA1A, CACNA1G, CACNB4, CASK, CCDC88C, CCNF, CHCHD10, CHCHD2, CHMP2B, CLN6, COL4A1, COMT, CP, CSF1R, CTSF, CWF19L1, CYP27A1, DAO, DAP12, DARS2, DCTN1, DJ1, DNAJC13, DNAJC3, DNAJC5, DNAJC6, DNMT1, EARS2, ECHS1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL4, ELOVL5, EPHA4, ERBB4, FAM126A, FBXO7, FGF12, FGF14, FIG4, FMR1, FRRS1L, FTL, FUCA1, FUS, FXN, GALC, GBA, GBE1, GCH1, GFAP, GJA1, GJC2, GLA, GLE1, GM2A, GNAL, GNAO1, GNE, GOSR2, GRID2, GRM1, GRN, HEPACAM, HEXA, HEXB, HNRNPA1, HNRNPA2B1, HSD17B4, HTRA1, HTRA2, ITPR1, KCNA1, KCNC3,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
KCND3, KIAA0226, KIF1C, KIF5A, KMT2B, L2HGDH, LMNB1, LRRK2, MAPT, MARS2, MATR3, MLC1, MRE11A, MTND1, MTND5, MTND6, MTPAP, MTTC, MTTF, MTTH, MTTK, MTTL1, MTTQ, MTTS1, MTTS2, NEFH, NEK1, NKX2-1, NOP56, NOTCH3, NPC1, NPC2, OPA3, OPHN1, OPTN, PANK2, PDE10A, PDE8B, PDGFB, PDGFRB, PDYN, PEX10, PFN1, PHYH, PIK3R5, PINK1, PLA2G6, PLP1, PMPCA, PNKD, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PON1, PON2, PON3, PPARGC1A, PPP2R2B, PPT1, PRICKLE1, PRKAR1B, PRKCG, PARK2, PRKRA, PRNP, PRPH, PRRT2, PSAP, PSEN1, PSEN2, PTRHD1, RAB29, RAB39B, RIC3, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, SACS, SAMHD1, SCA25, SCN2A, SCP2, SETX, SGCE, SLC17A5, SLC1A3, SLC20A2, SLC2A1, SLC30A10, SLC6A3, SLC9A1, SNCA, SNX14, SOD1, SOX10, SPAST, SPG7, SPR, SPTBN2, SQSTM1, STUB1, SUMF1, SYNE1, SYNJ1, SYT14, TAF1, TAF15, TARDBP, TBK1, TBP, TDP1, TGM6, TH, THAP1, TMEM106B,						

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
TMEM230, TMEM240, TOR1A, TPP1, TREM2, TREX1, TTBK2, TTPA, TUBB4A, UBA5, UBQLN2, UNC13A, VAMP1, VAPB, VCP, VLDLR, VPS13A, VPS13C, VPS35, WWOX, XK, XPR1, AFGL2, CTSF, ELOV4, HEXA, KCNA1, RNF216, TGTM6, PRKN, RAB38B, SCA17, APOE, APP, PDEN1, SPG4, PLP, EIF2B						
De novo: *ADNP, AHDC1, ALG13, ANKRD11, ARID1B, ASXL1, ASXL3, AUTS2, BCL11A, BRAF, CASK, CDK13, CDKL5, CHAMP1, CHD2, CHD3, CHD4, CHD8, CNKSR2, CNOT3, COL4A3BP, CREBBP, CSNK2A1, CTCF, CTNNB1, DDX3X, DNM1, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EP300, FOXG1, FOXP1, GABRB3, GATAD2B, GNAI1, GNAO1, GRIN2B, HDAC8, HNRNPU, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNH1, KCNQ2, KCNQ3, KDM5B, KIF1A, KMT2A, MECP2, MED13L, MEF2C, MSL3, MYT1L, NAA10, NFIX, NSD1, PACS1, PDHA1, POGZ, PPM1D, PPP2R1A, PPP2R5D, PTEN, PTPN11, PUF60, PURA, SATB2, SCN1A, SCN2A, SCN8A, SET, SETD5, SLC35A2, SLC6A1, SMAD4, SMARCA2,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
SMC1A, STXBP1, SUV420H1, SYNGAP1, TBL1XR1, TCF20, TCF4, TRIO, UPF3B, USP9X, WAC, WDR45, ZBTB18, ZC4H2						
Retinitis pigmentosa, autosomal-dominant und x-gekoppelt: *ARL3, BEST1, CA4, CACNA1F, CRX, FSCN2, GUCA1B, HK1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, OFD1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, SPP2, TOPORS*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Retinitis pigmentosa, autosomal-rezessiv und x-gekoppelt: *ABCA4, ADGRA3, ADIPOR1, AGBL5, AHI1, AIPL1, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CEP290, CERKL, CLCC1, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CWC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, GNAT1, GNPTG, GUCY2D, HGSNAT, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIF11, KIZ, LRAT, MAK, MERTK, MFRP, NEK2, NEUROD1, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF31, RBP3, RCBTB1, RDH11, RDH12,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
REEP6, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, SAG, SAMD11, SLC7A14, SPATA7, TRNT1, TTC8, TUB, TULP1, USH2A, WDR19, ZNF408, ZNF513						
Usher-Syndrom: *ABHD12, ADGRV1, ARSG, CDH23, CEP250, CIB2, CLRN1, GJB2, HARS, MYO7A, PCDH15, PDZD7, PRPH2, USH1C, USH1G, USH2A, WHRN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Refsum-Syndrom / Zellweger-Syndrom-Spektrum (Refsum / Zellweger / Neonatale Adrenoleukodystrophie): *PHYH, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Bardet-Biedl-Syndrom: *ARL6, BBIP, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CCDC28B, CEP290, IFT27, IFT74, IFT172, INPP5E, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TMEM67, TRIM32, TTC8, TTC21B, WDPCP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
Alagille-Syndrom: *JAG1, NOTCH2, BMP2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Leber'sche kongenitale Amaurose: *AGBL5, AIPL1, ALMS1, CABP4, CDH16, CEP290, CLUAP1, CNGA3, CRB1, CRX, DTHD1, GDF6, GUCY2D, IDH3A, IFT140, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, NMNAT1, OTX2, PDE6G, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kongenitale stationäre Nachtblindheit: *CABP4, CACNA1F, CACNA2D4, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Zapfendystrophie (Zapfen-Stäbchen-Dystrophie): *ABCA4, ADAM9, AIPL1, ALMS1, ATF6, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
CDHR1, CEP78, CEP250, CERKL, CFAP410, CNGA3, CNGB3, CNNM4, CRB1, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, NMNAT1, NR2E3, NRL, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RDH12, RGS9, RGS9BP, RIMS1, RPGR (inkl. ORF15), RPGRIP1, SEMA4A, TTLL5, TULP1, UNC119			AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Makuladystrophien / Morbus Stargardt: *ABCA4, BEST1, C1QTNF5, CDH3, CFH, CLN3, CNGB3, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, GUCA1A, GUCY2D, IMPG1, IMPG2, IRX1, MAP-KAP3, MFSD8, PPT1, PRDM13, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthe-sis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Ana-lyzer 3500xl, Illu-mina MiSeq, Illu-mina Nextseq550		x
Optikusatrophie und Le-ber´sche hereditäre Optikus-neuropathie (LHON): *MT-ND1, MT-ND4, MT-ND4L, MT-ND6, ACO2, AFG3L2, ANTXR1, ATP1A3, C12ORF65, CISD2, DNM1L, FDXR, MFN2, MTPAP, NBAS, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, SSBP1, TIMM8A, TMEM126A, WFS1, YME1L1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthe-sis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Ana-lyzer 3500xl, Illu-mina MiSeq, Illu-mina Nextseq550		x
Flecked Retina Erkrankungen: *ABCA4, CHM, CYP4V2, EF-EMP1, ELOVL4, KCNJ13,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
OAT, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RPE65, RS1, VPS13B		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Vitreoretinopathien (Familiäre Exsudative Vitreoretinopathie): *ATOH7, BEST1, CAPN5, COL18A1, COL2A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, TSPAN12, VCAN, ZNF408*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Senior-Loken-Syndrom: *CEP164, CEP290, INVS, IFT81, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TMEM67, TRAF3IP1, WDR19, ZNF423*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Achromatopsie: *ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
			AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Myopie und Differentialdiagnosen: *COL11A1, COL11A2, COL18A1, COL2A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, FBN1, FZD4, LRP2, LRP5, P3H2, VCAN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Katarakt: *ABCB6, ABHD12, ADAMTSL4, AGK, BFSP1, BFSP2, CHMP4B, CLPB, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP51A1, CYP27A1, DNMBP, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, GALK1, GALT, GCNT2, GEMIN4, GJA3, GJA8, GLA, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, LTBP2, MAF, MIP, MIR184, MYH9, NDP, NHS, OCRL, OPA3, P3H2, PAX6, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL2, RECQL4, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
TMEM114, UNC45B, VIM, VSX2, WFS1						
Mikrophthalmie-Anophthalmie-Kolobom-Komplex: *ABCB6, ACTB, ACTG1, ATOH7, C12ORF57, CRYBA4, CYP1B1, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM2, FZD4, GDF3, GDF6, GJA1, GRIP1, HESX1, HMX1, MFRP, NDUFB11, OCRL, OTX2, PAX2, PAX6, PIGL, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RAX, RBP4, SIX3, SIX6, SOX2, PXDN, RAB18, RAB3GAP1, RAB3GAP2, TMEM98, VPS13B, VSX2, ZIC2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Linsenluxation: *ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, CBS, COL11A1, COL18A1, COL2A1, FBN1, LTBP2, P3H2, VCAN, VSX2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Hornhautdystrophien/Keratoconus: *AGBL1, CHST6, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, MIR184, OVOL2, PAX6, PIKFYVE,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6;	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x

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PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469			AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0			
Septo-optische Dysplasie: *FGFR1, HESX1, OTX2, PROKR2, SOX2, SOX3*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Kongenitales Glaukom / Axenfeld-Rieger-Syndrom: *ASB10, COL4A1, CYP1B1, DDX58, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, SBF2, SH3PXD2B, TBK1, TEK, WDR36*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Vorderkammerdysgenesien / Axenfeld-Rieger-Syndrom: *ASPH, B3GLCT, COL4A1, COL4A2, CYP1B1, EYA1, FOXC1, FOXE3, GPATCH3, HMX1, LTBP2, MYOC, PAX6, PITX2, PXDN, SLC38A8*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8; AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		x
Albinismus (syndromal): *AP3B1, BLOC1S3, BLOC1S6, C10ORF11,*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT	AA-098 / 1.6; AA-099 / 1.8; AA-102 / 1.5; AA-103 / 1.5; AA-110 / 1.6; AA-112 / 1.8;	Genetic Analyzer 3130, Genetic Ana-		x

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Tagesaktuelle Liste FB-533

Untersuchungsverfahren

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Anweisung	Gerät	CE-Verfahren	In Haus-Verfahren
DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC38A8, SLC45A2, SOX10, TYR, TYRP1		HS), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-122 / 1.7; AA-209 / 1.4; AA-210/ 1.4; AA-212/ 1.4; AA-220 / 1.3; AA-222 / 1.1; AA-223 / 1.2; AA-245 / 1.6; AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-501 / 1.1; AA-502 / 1.0; AA-503 / 1.1; AA-504 / 1.2; AA-506 / 1.0	lyzer 3500xl, Illumina MiSeq, Illumina Nextseq550		
Whole exome sequencing	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect Human All Exon), Sequencing-by-synthesis, custom pipeline, Sanger-Sequenzierung, MLPA	AA-098 / 1.6; AA-099 / 1.8; AA-110 / 1.6; AA-112 / 1.3, AA-153 / 1.2, AA-209 / 1.3, AA-246 / 1.0; AA-247 / 1.0; AA-500 / 1.2; AA-504 / 1.2; AA-505 / 1.2	Genetic Analyzer 3130, Genetic Analyzer 3500xl, Illumina Nextseq550		x