DAkkS
Deutsche
Akkreditierungsstelle

Deutsche Akkreditierungsstelle

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00 nach DIN EN ISO 15189:2014

Gültig ab: 27.10.2022
Ausstellungsdatum: 27.10.2022

Inhaber der Akkreditierungsurkunde:

Universitätsmedizin Göttingen
Institut für Humangenetik und MVZ der UMG
Bereich Humangenetik
Heinrich-Düker-Weg 12, 37073 Göttingen

Das Medizinische Laboratorium erfüllt die Mindestanforderungen gemäß DIN EN ISO 15189:2014 und gegebenenfalls zusätzliche gesetzliche und normative Anforderungen, einschließlich solcher in relevanten sektoralen Programmen, um die nachfolgend aufgeführten Konformitätsbewertungstätigkeiten durchzuführen.

Die Anforderungen an das Managementsystem in der DIN EN ISO 15189 sind in einer für Medizinische Laboratorien relevanten Sprache verfasst und stehen insgesamt in Übereinstimmung mit den Prinzipien der DIN EN ISO 9001.

Untersuchungen im Bereich:

Medizinische Laboratoriumsdiagnostik

Untersuchungsgebiete:
Humangenetik (Molekulare Humangenetik)
Humangenetik (Zytogenetik)

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

Diese Urkundenanlage gilt nur zusammen mit der schriftlich erteilten Urkunde und gibt den Stand zum Zeitpunkt des Ausstellungsdatums wieder. Der jeweils aktuelle Stand der gültigen und überwachten Akkreditierung ist der Datenbank akkreditierter Stellen der Deutschen Akkreditierungsstelle zu entnehmen (www.dakks.de)

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart: Chromosomenanalyse **

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Angeborener Chromosomensatz	peripheres Blut, Fibroblasten, Fruchtwasser	Chromosomenbänderungs-analyse
Angeborener Chromosomensatz	Fruchtwasser	FISH (Fluoreszenz-in-situ-Hybridisierung) mit spezifischen Sonden
Angeborener Chromosomensatz	kultivierte Amnionzellen, peripheres Blut; genomische DNA	Molekulare Karyotypisierung (Array-based Comparative Genomic Hybridization (array-CGH)

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart: Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Schwerhörigkeit, sensorineural: *GJB2, GJB6*	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung, Fluoreszenz-markierte Hybridisierungssonden (Real-time PCR)
Molybdän-Cofaktor Defizienz (MOCS1, MOCS2)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung
Thrombophilie (F5-Gen Leiden Variante)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung
Trombophilie (Faktor II-Gen-3' Polymorphismus)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung
Homozysteinämie (MTHFR)	EDTA-Blut, DNA	PCR, Sanger-Sequenzierung
Yq11.21-23 Mikrodeletionen	EDTA-Blut, DNA	PCR
Fragiles X-Syndrom: *FMR1*	EDTA-Blut, DNA	Short Tandem Repeat typing
Lynch-Syndrom / Hereditäres Nicht-polyposes Kolorektalkarzinom: *MLH1, MSH2, MSH6, PMS2, EPCAM*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Polyposis-Syndrome (Familiäre adenomatöse Polyposis, MUTYH-assoziierte Polyposis, Cowden-Syndrom, Peutz-Jeghers-Syndrom, Hyperplastisches Polyposis-Syndrom (HPS), Serratiertes Polyposis-Syndrom (SPS), Hereditäres Mixed Polyposis-Syndrom (HMPS)): *APC, MUTYH, PIK3CA, PTEN, STK11, RNF43, GREM1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Gastrointestinale Stromatumore: *KIT, MAX, MEN1, NF1, PDGFRA, SDHAF2, SDHB, SDHC, SDHD, TMEM127*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Li-Fraumeni-Syndrom: *TP53*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Malignes Melanom: *CDKN2A, BAP1, BRCA2, POT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Prostatakarzinom: *BRCA1, BRCA2, CHEK2, MSR1, RNASEL, HOXB13, PALB2, RAD51D*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Retinoblastom: *RB1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Von-Hippel-Lindau-Syndrom: *VHL*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Makrozephalie (Basisdiagnostik): *CHD8, DNMT3A, EZH2, NSD1, NFIX, PTEN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Weaver-Syndrom: *EZH2, EED, SUZ12, HACE1, NSD1, NFIX*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Sotos-Syndrom: *NSD1, NFIX, EZH2, EED, SUZ12, HACE1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Beckwith-Wiedemann-Syndrom: *H19, KCNQ1OT1, CDKN1C, NSD1, ICR1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Alexander-Krankheit: *GFAP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Brittle Cornea-Syndrom: *ZNF469*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Dysplasie, kranio-metaphysäre (CMD): *ANKH, GJA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Greig-Zephalopolysyndaktylie-Syndrom: *GLI3, KIF7*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Joubert-Syndrom 12, Akrokallosales Syndrom: *KIF7*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Hydrozephalus mit Morbus Hirschsprung: *L1CAM*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Kosaki-Syndrom: *PDGFRB*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Krämpfe-Skoliose-Makrozephalie –Syndrom (SSMS): *EXT2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Luscan-Lumish-Syndrom (LLS): *SETD2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
MACS-Syndrom / RIN2-Syndrom: *RIN2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Makrozephalie mit Makrosomie und Gesichtsdysmorphien (MMFD): *RNF135*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Megalenzephalie-Polymikrogyrie-Polydaktylie-Hydrozephalus-Syndrom (MPPH1-3): *PIK3R2, AKT3, CCND2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Megalenzephalen Leukoenzephalopathie mit subkortikalen Zysten (MLC): *HEPACAM, MLC1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Megalenzephalie-Kapillar-Malformation-Polymikrogyrie-Syndrom (MCAP): *PIC3CA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Mentale Retardierung-Makrozephalie, autosomal rezessiv (MRT41): *KPTN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Mentale Retardierung-Makrozephalie, Suszeptibilität für Autismus (AUTS18): *CHD8*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Opitz-Kaveggia-Syndrom (OKS): *MED12*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Perlman-Syndrom (PRLNMS): *DIS3L2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Phelan-McDermid-Syndrom: *SHANK3*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Polyhydramnion, Megalenzephalie und Symptomatische Epilepsie: *STRADA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Proteus-Syndrom: *AKT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Simpson-Golabi-Behmel-Syndrom (SGBS): *GPC3, OFD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Smith-Kingsmore-Syndrom (SKS): *MTOR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Tatton-Brown-Rahman-Syndrom (TBRS): *DNMT3A*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Tenorio-Syndrom (TNORS): *RNF125*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
LIG4-Syndrom: *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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Galloway-Mowat-Syndrom (GMS): *LAGE3, OSGEP, TP53RK, TPRKB*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, RTEL1, SLX4, SMC2, SMC4, TERC, TERT, TINF2, TFAP2A, UBE2T, WFS1, WRAP53, WRN, XPA, XPC*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Wiedemann-Rautenstrauch-Syndrom (WRS): *ANO6, CAV1, FBN1, LMNA, BANF1, PYCR1, SPRTN, ZMPSTE24, ACD, AGPAT2, ALDH18A1, ATM, ATP6V0A2, ATP7A, 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), 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, TFAP2A, UBE2T, WFS1, WRAP53, XPA, XPC*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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, 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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, PLIN1, POLH, PPARG, PRKDC, PTDSS1, PTRF, 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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Cutis laxa assoziierte Syndrome (CLS): *ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PEX11B, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, PRKDC, PTDSS1, PTRF, PYCR1, RAD51C, 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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Ataxia-Teleangiektasia/Louis-Bar-Syndrom (LBS): *ATM, MRE11A, PCNA, ACD, AGPAT2, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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, RNASEH2B, 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, KIDINS220, UBAP1, AGTPBP1, SLC2A1 , PLA2G6*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Spastische Ataxien: *AFG3L2, KIF1C, MARS2, SACS, SOX10, SPG7, VAMP1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
ALS-ähnliche Hereditäre Spastische Paraplegie (HSP): *ALS2, SETX, SPG11, SOD1, TARDBP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, SCN9A, SETX, SH3TC2, SLC12A6, SLC5A7, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TDP1, TFG, TRIM2, TRPV4, VCP, WNK1, YARS	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Hereditäre Neuropathie mit Neigung zu Druckparesen (HNPP): *PMP22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Kabuki-Syndrom: *KMT2D, KDM6A, RAP1A, RAP1B*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Costello-Syndrom (CSTLO): *HRAS*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Leopard-Syndrom (LPRD): *BRAF, PTPN11,RAF1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Kardiofaziokutanes Syndrom (CFC): *BRAF, MAP2K1, MAP2K2, KRAS*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
CHARGE-Syndrom: *CHD7, SEMA3E, TBX1, TBX22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
DiGeorge-Syndrom (DGS): *TBX1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Abruzzo-Erickson-Syndrom (ABERS), CHARGE-like Syndrom: *TBX22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Neurofibromatose: *NF1, NF2, SPRED1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Legius-Syndrom: *SPRED1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Robinow-Syndrom: *WNT5A, DVL1, DVL3, ROR2, FGD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Aarskog-Syndrom: *FGD1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
CHOPS-Syndrom: *AFF4*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
KBG-Syndrom (KBGS): *ANKRD11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Trichorhinophalangeales Syndrom 1 und 3 (TRPS1 und TRPS3): *TRPS1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Nicolaides-Baraitser-Syndrom (NCBRS): *SMARCA2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Baraitser-Winter-Syndrom (BRWS1): *ACTB, ACTG1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Borjeson-Forssman-Lehmann-Syndrom (BFLS): *PHF6*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Cutis laxa (ADCL1) / Williams-Beuren-Syndrom (WBS): *ELN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Simpson-Golabi-Behmel-Syndrom (SGBS): GPC3, GPC4, OFD1	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Townes-Brocks-Syndrom (TBS): *SALL1, DACT1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Duane-Radial Ray-Syndrom (DRRS) / Okihiro-Syndrom: *SALL4*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Branchiooto(renales) Syndrom 1 und 2 (BOR1, BOR2): *EYA1, SIX1, SIX5*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Rubinstein-Taybi-Syndrom (RSTS): *CREBBP, EP300*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Floating-Harbor-Syndrom (FLHS): *SRCAP*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Coffin-Lowry-Syndrom (CLS): *RPS6KA3*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Okulodentodigitale Dysplasie (ODDD): *GJA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Ritschner-Schintzel-Syndrom (RTSC): *WASHC5/KIAA0196, CCDC22*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Lujan-Fryns-Syndrom (LFS), Opitz-Kaveggia-Syndrom (OKS): *MED12*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Cohen-Syndrom (COH): *VPS13B*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Mental retardation, autosomal dominant (MRD32): *KAT6A*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Glass-Syndrom: *SATB2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Schuurs-Hoeijmakers-Syndrom (SHMS): *PACS1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Kardiomyopathie, hypertroph (HCM): *ACADVL , ACTC1, CSRP3, FHL1, MYBPC3, MYH7, MYL2, MYL3, MYPN, PLN, PRKAG2, TNNC1, 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*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Kardiomyopathie, restriktiv (RCM): *ACTC1, DES, MYH7, MYL2, MYPN, TNNI3, TNNT2, TPM1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Morbus Fabry: *GLA*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): *CASQ2, CALM1, KCNE1, KCNJ2, RYR2, TRDN*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Short QT-Syndrom (SQT): *CACNA1C, KCNH2, KCNQ1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Hypercholestrinämie (LDLR_Defizienz): *LDLR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Hypercholesterinämie (primär): *APOB, PCSK9, LDLRAP1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Marfan-Syndrom: *FBN1, TGFBR1, TGFBR2, ACTA2, COL3A1, MYH11, MYLK, SMAD3, TGFB2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Thorakale Aortenerweiterung: *ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Bindegewebserkrankung mit Gefäßbeteiligung: *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBN1, FBN2, LTPB2, MYH11, MYLK, MYLK2, NOTCH1, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1, TGFBR2*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Familiäre Hypercholesterinämie (FH): *APOB, LDLR, LDLRAP1, PCSK9*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Familiäre hypokalzurische Hyperkalzämie (HHC): *CASR, AP2S1, GNA11*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
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 (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Morbus Osler / Hereditäre Hämorrhagische Teleangiektasie (HHT): ACVRL1, *ENG, SMAD4, GDF2, RASA1*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Cystische Fibrose (CF): *CFTR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Familiäre Skeletterkrankungen (SKELETT): 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, CTSK, CUL7, OFD1, DCHS1, DDR2, DDRGK1, DDX59, DHCR7, DHODH,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Fortsetzung siehe nächste Zeile

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
GNPAT, DKK1, DLL3, DLL4, DLX3, DLX5, DLX6, DMP1, DNA2, DSE, DSPP, DVL1, DVL3, DYM, DYNC2H1, EBP, EFEMP2, 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, IARS2, ICK, ID, IDH , IDS, IDUA, IFITM5, IFT122, IFT43, IFT80, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, INPPL1, IRX5, ITGB3, KAT6B, KDELR2, KIAA0196/WASHC5, KIF22, KIF7, KL, KYNU, LBR, LEMD3, LEPRE1, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LPIN2, LRRK1, LRP4, LRP5, LRP6, LTBP2, LTBP3, LTBP4,		
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
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 , RUNX2, SALL1, SALL4, SAMD9, SBDS, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SFRP4, SGMS2, SGSH,		
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
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		
Cystische Fibrose (CF): *CFTR*	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Familiäre Skeletterkrankungen (SKELETT): 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, CTSK, CUL7, OFD1, DCHS1, DDR2, DDRGK1, DDX59, DHCR7, DHODH,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Fortsetzung siehe nächste Zeile		

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
GNPAT, DKK1, DLL3, DLL4, DLX3, DLX5, DLX6, DMP1, DNA2, DSE, DSPP, DVL1, DVL3, DYM, DYNC2H1, EBP, EFEMP2, 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, IARS2, ICK, ID, IDH , IDS, IDUA, IFITM5, IFT122, IFT43, IFT80, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, INPPL1, IRX5, ITGB3, KAT6B, KDELR2, KIAA0196/WASHC5, KIF22, KIF7, KL, KYNU, LBR, LEMD3, LEPRE1, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LPIN2, LRRK1, LRP4, LRP5, LRP6, LTBP2, LTBP3, LTBP4,		
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
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 , RUNX2, SALL1, SALL4, SAMD9, SBDS, SEC23A, SEC24D, SERPINF1, SERPINH1, SETBP1, SF3B4, SFRP4, SGMS2, SGSH,		
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
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, 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	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, 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,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis (Illumina), custom pipeline, Sanger-Sequenzierung, MLPA
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
	Fortsetzung der vorherigen Zeile	
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, TFAP2A, TJP2, TMC2, TYR, KARS1, LARS2, TUBB4B		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Familiäre neurodegenerative Erkrankungen (NEUROD): 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, KCND3, KIAA0226, KIF1C, KIF5A,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, SeqPilot (Modul: SeqNext), Varvis, Sanger-Sequenzierung, MLPA

Fortsetzung siehe nächste Zeile

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
	Fortsetzung der vorherigen Zeile	
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, 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		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
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, SMC1A, STXBP1, SUV420H1, SYNGAP1, TBL1XR1, TCF20, TCF4, TRIO, UPF3B, USP9X, WAC, WDR45, ZBTB18, ZC4H2	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, SeqPilot (Modul: SeqNext), Varvis, Sanger-Sequenzierung, MLPA

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Hereditäre Netzhautdystrophie: ABCA4, ABCB6, ABHD12, ACO2, ACTB, ACTG1, ADAM9, ADAMTS10, ADAMTS17, ADAMTSL4, ADGRA3, ADIPOR1, AFG3L2, AGBL1, AGBL5 ,AGK ,AHI1, AHR, AIPL1, ALMS1, ANTXR1, AP3B1, ARHGEF18, ARL2BP, ARL3, ARL6, ARSG, ASB10, ASPH, ATF6, ATOH7, ATP1A3, B3GLCT, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP2, C12orf57, C12orf65, C1QTNF5, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CBS, CCDC28B, CDH16, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP410, CFH, CHM, CHMP4B, CHST6, CIB2, CISD2, CLCC1, CLN3, CLPB, CLRN1, CLUAP1 ,CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL17A1, COL18A1, COL2A1, COL4A1, COL4A2, COL5A1, COL5A2, COL8A2, COL9A1, COL9A2, COL9A3, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4 ,CRYBB1, CRYBB2, CRYBB3 ,CRYGB, CRYGC ,CRYGD, CRYGS, CTDP1, CTNNA1, CTNNB1,	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, SureSelect XT HS), Sequencing-by-synthesis, custom pipeline, SeqPilot (Modul: SeqNext), Varvis, Sanger-Sequenzierung, MLPA

Fortsetzung siehe nächste Zeile

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
	Fortsetzung der vorherigen Zeile	
CWC27, CYP1B1, CYP27A1, CYP4V2, CYP51A1, DCN, DDX58, DHDDS, DHS6S1, DHX38, DNM1L, DNMBP, DRAM2, DTHD1, DTNBP1, EDN3 ,EDNRB, EFEMP1, ELOVL4, EMC1, EPG5, EPHA2, ERCC1, ERCC2, ERCC5, ERCC6, EYA1, EYS, FAM126A, FAM161A, FBN1, FDXR, FGFR1, FLVCR1, FOXC1, FOXE3 ,FOXL2, FRAS1, FREM2, FRMD7, FSCN2, FTL, FYCO1 ,FZD4, GALK1 ,GALT, GCNT2, GDF6, GEMIN4, GJA1, GJA3, GJA8, GJB2 ,GLA, GNAT1, GNAT2, GNB3, GNPTG, GPATCH3, GPR143, GPR179, GPR98, GRIP1, GRK1, GRM6, GSN, GUCA1A, GUCA1B, GUCY2D, HARS, HESX1, HGSNAT, HK1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, IRX1, JAG1, JAM3, KCNJ13, KCNV2, KIAA1549, KIF11, KIF3B ,KIZ, KLHL7, KRT12, KRT3, LCA5, LEMD2, LIM2, LMX1B, LONP1, LOXHD1, LOXL1, LRAT, LRIT3, LRMDA, LRP2, LRP5, LSS, LTBP2 ,LYST, LZTFL1, MAF, MAK, MAPKAPK3, MC1R, MERTK, MFN2, MFRP, MFSD8 ,MIP, MIR184, MITF ,		
	Fortsetzung siehe nächste Zeile	

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Fortsetzung der vorherigen Zeile		
MKKS ,MKS1, MLPH ,MT-ND1, MT-ND4, MT-ND4L, MT-ND6, MTPAP, MVK, MYH9, MYO5A, MYO7A, MYOC, NBAS ,NDP, NDUFB11 ,NEK2, NEUROD1 ,NHS, NMNAT1, NOTCH2, NPHP1 ,NPHP3, NPHP4 ,NR2E3, NR2F1, NRL, NTF4, NYX, OAT ,OCA2, OCRL, OFD1, OPA1, OPA3, OPN1LW ,OPN1MW, OPTN, OTX2, OVOL2, P3H2, PAX2, PAX3, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PIGL, PIKFYVE, ITPNM3, PITX2, PITX3, PLA2G5, POC1B, POMGNT1, PPT1, PRCD, PRDM13, PRDM5, PROKR2, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PXDN, RAB18, RAB27A, RAB28, RAB3GAP1, RAB3GAP2, RAX, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RS1, RTN4IP1, SAG, SAMD11, SBF2, SDCCAG8, SEMA4A, SH3PXD2B ,		
Fortsetzung siehe nächste Zeile		

Anlage zur Akkreditierungsurkunde D-ML-13161-02-00

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
	Fortsetzung der vorherigen Zeile	
SIL1, SIPA1L3, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A46, SLC33A1, SLC38A8, SLC45A2, SLC4A11, SLC7A14, SNRNP200, SOX10, SOX2, SOX3, SPATA7, SPG7, SPP2, SSBP1, TACSTD2, TBC1D20, TBK1, TCF4, , TDRD7, TEK, TGFBI, TGIF1, TIMM8A, TIMP3, TMEM114, TMEM126A, TMEM67, TMEM98, TOPORS, TRAF3IP1, TRIM32, TRNT1, TRPM1, TSPAN12, TST, TTC21B, TTC8, TTLL5, TUB, TUBA3D, TULP1, TYR, TYRP1, UBIAD1, UNC119, UNC45B, USH1C, USH1G, USH2A, USP45, VCAN, VIM, VPS13B, VSX1, VSX2 ,WDPCP, WDR19, WDR36, WFS1, WHRN , WRN, YME1L1, ZEB1, ZIC2, ZNF408, ZNF423, ZNF469, ZNF513, TFAP2A, PRR12, CEP19, ABCD1, ACOX1, HSD17B4, SCLT1, MCAT, KIT, SNAI2, ISCA2, TUBB4B, DNAJC30, BCOR, BMP4, BMP7, CHD7, FREM1, HCCS		
Whole exome sequencing	EDTA-Blut, DNA	Sequence Capture (Agilent Technologies, Human), Sequencing-by-synthesis, custom pipeline, SeqPilot (Modul: SeqNext), Varvis, Sanger-Sequenzierung, MLPA