Deutsche Akkreditierungsstelle GmbH

Annex to the Accreditation Certificate D-ML-13206-01-00 according to DIN EN ISO 15189:2014

Period of validity: 17.08.2016 to 16.08.2021 Date of issue: 17.08.2016

Holder of certificate:

CeGaT GmbH
Paul-Ehrlich-Straße 23, 72076 Tübingen

Examinations in the field:

Medical Laboratory Diagnostics

Test area
Human genetics (molecular human genetics, cytogenetics)

Types of tests:
Molecular biological tests (amplification method)
Chromosome analysis

Within the given examination procedures marked with */**, the medical laboratory is permitted, without being required to inform and obtain prior approval from DAkkS, the following:
* select standard test procedures or equivalent procedures
** the modification, development and refinement of examination procedures.
The listed examination procedures are exemplary. The medical laboratory maintains a current list of all examination procedures within the flexible scope of accreditation.

Test area: Human Genetics (molecular human genetics)

Type of test:
Molecular biological analysis (amplification method) **

Analyte (measurement parameter)	Test material (matrix)	Test technique
POLG	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
HFE	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
SPR	genomic DNA	PCR with subsequent sequencing
TOR1A	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
OPA3	genomic DNA	PCR with subsequent sequencing
OPA1	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
GCH1	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
GJB2	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
GLA	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
FTL	genomic DNA	PCR with subsequent sequencing
ABCA3	genomic DNA	PCR with subsequent sequencing
SCN1A	genomic DNA	PCR with subsequent sequencing and deletion-/ duplication analysis
Neurodegenerative Diseases: Amyotrophic lateral sclerosis: (Gene panel: C9orf72, ALS2, ANG, ATXN1, ATXN2, ARHGEF28, CHCHD10, CHMP2B, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, HNRNPA2B1, MATR3, NEFH, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG20, SQSTM1, TARDBP, UBQLN2, VAPB, VCP, VEGFA, VPS54)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Neurodegenerative Diseases: Leukodystrophy / Leukoencephalopathy and differential diagnoses: (Gene panel: AARS2, ABCD1, ADAR, AIMP1, ALS2, ARSA, ASPA, CSF1R, CYP27A1, CYP7B1, DARS, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FAM126A, FIG4, FOLR1, FUS, GALC, GBA, GFAP, GJC2, GLB1, HEPACAM, HEXA, HSPD1, L2HGDH, LMNB1, MLC1, NDUFV1, NOTCH3. PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SETX, SOD1, SOX10, SPG11, SPG20, SPG21, SUMF1, TARDBP, TREX1, TUBB4A, VAPB, ZFYVE26)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)
Epilepsy: Idiopathic generalized and focal Epilepsy: (Gene panel: ALDH7A1, AMACR, CACNA1A, CACNA1H, CACNB4, CASR, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTN2, CPA6, DEPDC5, EFHC1, GABRA1, GABRB3, GABRD, GABRG2, GRIN2A, KCNA1, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, ME2, NIPA2, PRRT2, RBFOX1, RBFOX3, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC1A3, SLC2A1, STX1B, TBC1D24, UBR5	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Epilepsy: Epileptic Encephalopathies: (Gene panel: ACY1, ADAR, ADSL, ALDH7A1, ALG13, AMT, ARHGEF9, ARX, BRAT1, CACNA1A, CASK, CDKL5, CHD2, CNTNAP2, CPT2, DCX, DNM1, FLNA, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GCSH, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, HDAC4, HNRNPU, KCNA2, KCNB1, KCNQ2, KCNT1, MBD5, MECP2, MEF2C, MOCS1, MOCS2, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC9A6, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TREX1, TSC1, TSC2, UBE3A, WDR45, ZEB2)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)
Neuromuscular diseases: Charcot-Marie-Tooth and sensoric Neuropathies (Gene panel: AARS, ABHD12, AIFM1, ARHGEF10, ATL1, ATL3, BSCL2, C10orf2, C12orf65, CTDP1, CTDP1, DCAF8, DHTKD1, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HADHA, HADHB, HARS, HINT1, HK1 (nur HMSNR Varianten), HOXD10, HSPB1, HSPB8, IFRD1, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR14, MTMR2, NDRG1, NEFL, NGF, NTRK1, OPA1, OPA3, PDHA1, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN9A, SEPT9, SH3TC2, SLC12A6, SOX10, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, TYMP, WNK1, YARS)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Neuromuscular diseases: Congenital und distal Myopathies (Gene panel: ACTA1, ACVR1, ANO5, BAG3, BIN1, C10orf2, CAV3, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, HNRNPA1, HNRNPA2B1, ISCU, KBTBD13, KLHL40, KLHL41, KLHL9, LAMP2, LDB3, MATR3, MEGF10, MSTN, MTM1, MYBPC1, MYBPC3, MYF6, MYH2, MYH7, MYH14, MYOT, NEB, OPA1, PABPN1, PLEC, POLG, POLG2, PTPLA, PUS1, RRM2B, RYR1, SEPN1, SIL1, STAC3, STIM1, SUCLA2, TIA1, TK2, TNNT1, TPM2, TPM3, TRIM32, TTN, VCP, VMA21, YARS2)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)
Hereditary eye diseases: Retinitis Pigmentosa, autosomal-recessive and X-linked (Gene panel: ABCA4, ARL2BP, BBS1, BEST1, C2orf71, C8orf37, CERKL, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, GNPTG, GPR125, IDH3B, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NMNAT1, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PRPF31, PROM1, RBP3, RBP4, RDH11, RDH12, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, SAG, SLC7A14, SPATA7, TTC8, TUB, TULP1, USH2A, ZNF513)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)
Hereditary eye diseases: Stargardt disease and Macular Dystrophies (Gene panel: ABCA4, BEST1, C1QTNF5, CDH3, CLN3, CNGB3, ELOVL4, FSCN2, IMPG1, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hereditary eye diseases: Cone Rod Dystrophies (Gene panel: ABCA4, ACBD5, ADAM9, AIPL1, C21orf2, C8orf37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CNNM4, CRX, CYP4V2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCYT1A, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5)	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)
Somatic tumor panel: ABCB1, ABCC2, ABCG2, ABL1, ABL2, ACD, ACVR1, ACVR1B, AJUBA, AKT1, AKT2, AKT3, ALK, AMER1, APC, AR, ARAF, ARFRP1, ARHGAP35, ARID1A, ARID1B, ARID2, ARID5B, ARNT, ASXL1, ATF1, ATM, ATP1A1, ATP5B, ATR, ATRX, AURKA, AURKB, AURKC, AXIN1, AXIN2, AXL, AZGP1, B2M, BAP1, BARD1, BCL10, BCL11A, BCL11B, BCL2, BCL2L1, BCL2L2, BCL3, BCL6, BCL9, BCOR, BCORL1, BCR, BIRC2, BIRC3, BIRC5, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRE, BRIP1, BTK, BTNL2, BUB1B, C11ORF30, CARD11, CASP8, CBFB, CBL, CCDC6, CCND1, CCND2, CCND3, CCNE1, CD1D, CD274, CD70, CD79A, CD79B, CD82, CDC27, CDC73, CDH1, CDH2, CDK12, CDK4, CDK6, CDK8, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CDX2, CEBPA, CEP57, CHD2, CHD4, CHEK1, CHEK2, CIC, CKS1B, COL1A1, CREB1, CREBBP, CRKL, CRTC1, CSF1R, CTCF, CTNNA1, CTNNB1, CUL3, CUL4B, CUX1, CYLD, CYP1A1, CYP1A2, CYP2A6, CYP2B6, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP2E1, CYP3A4, CYP3A5, DAXX, DCC, DDB2, DDIT3, DDR2, DDX3X, DEK, DIAPH1, DICER1, DIS3, DNMT1, DNMT3A, DOT1L, DPYD, DST, EGFR, EGR3,	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
ELAC2, ELF3, EML4, ENG, EP300, EPCAM, EPHA2, EPHA3, EPHA5, EPHA7, EPHB1, EPHB4, EPHB6, EPHX1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERRFI1, ESR1, ETS1, ETV1, ETV4, ETV5, ETV6, EWSR1, EXT1, EXT2, EZH1, EZH2, FAM175A, FAM46C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FAT1, FBXW7, FES, FGF10, FGF14, FGF19, FGF23, FGF3, FGF4, FGF6, FGFBP1, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT4, FN1, FOXA1, FOXA2, FOXE1, FOXL2, FOXO1, FOXO3, FOXP1, FOXQ1, FRS2, FUBP1, FUS, G6PD, GABRA6, GALNT12, GATA1, GATA2, GATA3, GATA4, GATA6, GDNF, GID4, GLI1, GNA11, GNA13, GNAQ, GNAS, GOT1, GPC3, GPR124, GRIN2A, GRM3, GSK3B, GSTM1, GSTP1, GSTT1, GUSB, H3F3A, H3F3B, HGF, HIF1A, HIST1H3B, HLA-A, HLA-B, HLA-C, HLF, HMGA2, HNF1A, HNF1B, HOXA9, HOXB13, HRAS, HSD3B1, HSP90AA1, HSP90AB1, IDH1, IDH2, IGF1R, IGF2, IGF2R, IKBKB, IKBKE, IKZF1, IL2, IL21R, IL6ST, IL7R, ING1, ING4, INHBA, INPP4B, INPPL1, IRF2, IRF4, IRF6, IRS2, ITGB2, ITK, JAK1, JAK2, JAK3, JUN, KAT6A, KCNJ5, KDM5A, KDM5C, KDM6A, KDR, KEAP1, KEL, KIAA1549, KIT, KLF4, KLF6, KLHL6, KMT2A, KMT2B, KMT2C, KMT2D, KRAS, LAMP1, LATS1, LATS2, LCK, LGI1, LIFR, LIG4, LMO1, LPP, LRP1B, LRRK2, LTK, LYL1, LYN, LZTR1, MAF, MAFB, MAGEA1, MAGI2, MALT1, MAML1, MAP2K1, MAP2K2, MAP2K4, MAP3K1, MAP3K6, MAPK1, MAPK8, MAPK8IP1, MAX,	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
MBD1, MC1R, MCL1, MDM2, MDM4, MECOM, MED12, MEF2B, MEN1, MET, MGA, MITF, MLH1, MLH3, MLLT10, MLLT3, MMP2, MN1, MOB1A, MOB1B, MPL, MPO, MRE11A, MSH2, MSH3, MSH6, MSR1, MTHFR, MTOR, MTR, MTRR, MUC1, MUTYH, MXI1, MYB, MYC, MYCL, MYCN, MYD88, MYH11, MYH9, NAT1, NAT2, NBN, NCOA1, NCOA2, NCOA3, NCOR1, NF1, NF2, NFE2L2, NFKB1, NFKB2, NFKBIA, NIN, NKX2-1, NKX3-1, NOTCH1, NOTCH2, NOTCH3, NPM1, NRAS, NSD1, NTRK1, NTRK2, NTRK3, NUMA1, NUP93, NUP98, PAK3, PALB2, PALLD, PARK2, PAX3, PAX5, PAX7, PBRM1, PBX1, PCBP1, PDCD1LG2, PDGFB, PDGFRA, PDGFRB, PDK1, PER1, PHF6, PHOX2B, PIK3C2B, PIK3CA, PIK3CB, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PIM1, PKHD1, PLCG1, PLCG2, PML, PMS1, PMS2, POLD1, POLE, POLH, POLQ, POT1, POU2AF1, POU2F2, POU5F1, PPM1D, PPP2R1A, PRDM1, PRDM16, PREX2, PRF1, PRKACA, PRKAR1A, PRKCI, PRKD1, PRKDC, PRSS1, PRSS8, PRX, PSIP1, PSPH, PTCH1, PTEN, PTGS2, PTPN11,	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
PTPRC, PTPRD, PTPRT, QKI, RAC1, RAD21, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RALGDS, RARA, RASA1, RASAL1, RB1, RBM10, RBM15, RECQL, RECQL4, REL, RET, RHEB, RHOA, RHOH, RICTOR, RINT1, RNASEL, RNF2, RNF43, ROS1, RPL22, RPL5, RPTOR, RRM1, RUNX1, RUNX1T1, RXRA, RYR1, SACS, SAV1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SELP, SEMA4A, SEPT9, SETBP1, SETD2, SETDB1, SF3B1, SGK1, SH2D1A, SIN3A, SKP2, SLC15A2, SLC1A3, SLC22A1, SLC22A2, SLC22A6, SLC26A3, SLCO1B1, SLCO1B3, SLIT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMO, SMUG1, SNCAIP, SOCS1, SOS1, SOX10, SOX11, SOX17, SOX2, SOX9, SPEN, SPINK1, SPOP, SPRED1, SPTA1, SRC, SRD5A2, SRSF2, SSX1, STAG2, STAT3, STAT4, STAT5B, STK11, STK3, STK4, SUFU, SULT1A1, SUZ12, SYK, TAF1, TAF15, TAL1, TAP1, TBL1XR1, TBX3, TCF3, TCF7L1, TCF7L2, TCL1A, TERC, TERF2IP, TERT, TET1, TET2, TFE3, TGFBR2, THBS1, TIMP3, TLR4, TLX1, TLX3, TMEM127, TMPRSS2, TNF, TNFAIP3, TNFRSF14, TNK2, TOP1, TOP2A, TP53, TP53BP1, TPMT, TPX2, TRAF3, TRAF7, TRIM24, TRRAP, TSC1, TSC2, TSHR, TYMS, U2AF1, UBR5, UGT1A1, UGT2B15, UGT2B17, UGT2B7, UIMC1, USP9X, VEGFA, VHL, VKORC1, WASF3, WHSC1, WISP3, WRN, WT1, WWTR1, XPA, XPC, XPO1, XRCC1, XRCC2, YAP1, ZBTB2, ZFHX3, ZNF217, ZNF703	genomic DNA	Next-Generation-Sequencing (Illumina-Technology)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Additional detection of selected translocations in genes: ALK, BCL2, BCR, BRAF, BRD4, EGFR, ERG, ETV4, ETV6, EWSR1, FGFR1, FGFR2, FGFR3, FUS, MYB, MYC, NOTCH2, NTRK1, PAX3, PDGFB, RARA, RET, ROS1, SSX1TFE3, SUZ12, TAF15, TCF3, TMPRSS2		

Test area: Human Genetics (cytogenetics)

Type of test: chromosome analysis

Analyte (measurement parameter)	Test material (matrix)	Test technique
Duplication/Deletion	genomic DNA	comparative genome-hybridization of chromosomes (Array CGH Agilent SureScan DX)