Gen-Panels Entwicklungsstörungen & Fehlbildungen | LI

Hinweise:

- Gen-Panels mit mehr als 10 Genen dürfen gemäss Analysenliste des BAG nur durch Ärzte mit einem FMH-Titel Medizinische Genetik verordnet werden.
- Die Gen-Panels sind nicht endgültig und können je nach Fragestellung auf Wunsch angepasst werden.
- Die Gen-Panels können abhängig von der aktuellen Datenlage zum Zeitpunkt der Analysen von denjenigen auf dieser Liste abweichen.

Amelogenesis imperfecta / Dentinogenesis imperfecta v3 (37 Gene)

AMBN, ACP4, AMELX, AMTN, CLDN16, CLDN19, CNNM4, COL17A1, DLX3, DSPP, ENAM, FAM20A, FAM20C, FAM83H, GPR68, ITGB4, ITGB6, KLK4, LAMA3, LAMB3, LAMC2, LTBP3, MMP20, ODAPH, ORAI1, PEX1, PEX26, PEX6, RELT, ROGDI, SLC10A7, SLC13A5, SLC24A4, SMOC2, SP6, STIM1, WDR72

Arthrogrypose v8 (163 Gene)

ACTA1, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, ALG3, ANTXR2, ASCC1, ASXL1, ATAD1, ATP1A2, B3GALNT2, B4GAT1, BICD2, CACNA1E, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CNTN1, CNTNAP1, COASY, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CRLF1, CRPPA, DAG1, DHCR24, DNM2, DOK7, DPAGT1, DPM2, DYNC1H1, EBP, ECEL1, ERBB3, ERCC5, ERCC6, ERCC8, ERGIC1, EXOSC3, FAM20C, FBN2, FGFR2, FGFR3, FKBP10, FKRP, FKTN, FLNA, FLNB, FLNC, GBA, GBE1, GLDN, GLE1, GMPPB, HSPG2, IRF6, KAT6B, KCNK3, KIAA1109, KIDINS220, KIF21A, KLHL40, KLHL41, KLHL7, LAMA2, LARGE1, LGI4, LMOD3, LMX1B, MAGEL2, MED12, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYL1, MYLPF, MYMK, MYOD1, NALCN, NEB, NEK9, NUP88, ORAI1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PIEZO2, PIP5K1C, PLOD1, PLOD2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRG4, RAPSN, RIPK4, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCYL2, SELENON, SKI, SLC18A3, SLC29A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMN1, SMPD4, STAC3, STIM1, SYNE1, TGFB2, TGFB3, TGFBR1, TGFBR2, TNNI2, TNNT1, TNNT3, TOR1A, TOR1AIP1, TPM2, TPM3, TRIP4, TRPV4, TSEN54, TTN, UBA1, VAMP1, VIPAS39, VPS33B, ZC4H2, ZMPSTE24

Cavernomatose, familiäre zerebrale v3 (3 Gene)

CCM2, KRIT1, PDCD10

Cornelia de Lange-Syndrom & Differentialdiagnosen v4 (21 Gene)

AFF4, ANKRD11, ASXL1, BRD4, CREBBP, EBF3, EHMT1, EP300, HDAC8, KMT2A, MED13L, NAA10, NIPBL, NR2F1, PHIP, PUF60, RAD21, SET, SETD5, SMC1A, SMC3, TAF1, TAF6, TBL1XR1

Ektodermale Dysplasie v3 (53 Gene)

ANAPC1, APCDD1, AXIN2, C3orf52, CDH3, CDSN, DSG4, DSP, EDA, EDAR, EDARADD, ERCC2, GJB2, GJB6, GRHL2, HOXC13, HR, IKBKG, JUP, KDF1, KREMEN1, KRT14, KRT25, KRT71, KRT74, KRT81, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, RMRP, RSPO4, SDR9C7, SNRPE, SREBF1, ST14, TP63, TSPEAR, WDR35, WNT10A

Fallot-Tetralogie v1 (8 Gene)

FLT4, GATA4, GATA6, GDF1, JAG1, NKX2-5, TBX1, ZFPM2

Fehlbildungen der Extremitäten v5 (197 Gene)

ABL1, ARHGAP31, ARL6, ARSL, ASXL1, ATR, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BRCA2, BRIP1, BTRC, CCND2, CCNQ, CDH3, CENPF, CEP290, CEP55, CHSY1, CIBAR1, CKAP2L, COL2A1, CYP26B1, DDX59, DHODH, DLL4, DLX5, DOCK6, DVL1, DVL3, DYM, DYNC2I1, EBP, EFNB1, EFTUD2, EIF4A3, EOGT, EPHA4, ERCC4, ESCO2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FBLN1, FGD1, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FRAS1, FREM2, FZD2, GATA1, GDF5, GJA1, GLI1, GLI2, GLI3, GNAS, GPC3, GRIP1, HDAC4, HDAC8, HEATR3, HNRNPK, HOXA13, HOXD13, IFT27, IFT43, IHH, IQCE, KCNH1, KCNN3, KIAA0825, KIF7, KYNU, LBR, LMBR1, LMX1B, LRP4, LTBP1, LZTFL1, MAPKAPK5, MECOM, MEGF8, MGP, MIR17HG, MKKS, MKS1, MYCN, NCAPG2, NEK1, NIPBL, NOG, NOTCH1, NSDHL, NXN, OFD1, ORC1, PALB2, PAX3, PCNT, PDE3A, PDE6D, PIGV, PIK3CA, PIK3R2, PITX1, POLR1A, PORCN, PRKACB, PRMT7, PTHLH, RAB23, RAD51, RAD51C, RBM8A, RBPJ, RECQL4, RNU4ATAC, ROR2, RPGRIP1L, RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS7, SALL1, SALL4, SCUBE3, SF3B4, SFRP4, SHOX, SLC26A2, SLX4, SMAD6, SMC1A, SMC3, SMO, SMOC1, SOX9, SPINT2, STKLD1, SUFU, TBX15, TBX3, TBX4, TBX5, TFAP2A, TFAP2B, TGDS, THPO, TMEM67, TP63, TRAF7, TRAPPC2, TRPS1, TRPV4, TTC8, TWIST1, TXNDC15, UBA2, UBE2T, UBE3B, USP9X, VPS35L, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, XRCC2, ZIC3, ZSWIM6

Geistige Behinderung / Entwicklungsverzögerung v11 (1641 Gene)

AARS1, ABCA2, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ACAD9, ACADS, ACBD6, ACO2, ACOX1, ACSF3, ACSL4, ACTB, ACTG1, ACTL6A, ACTL6B, ACVR1, ACY1, ADAM22, ADAR, ADARB1, ADAT3, ADD3, ADGRG1, ADGRL1, ADK, ADNP, ADPRS, ADSL, AFF2, AFF4, AFG2A, AFG2B, AGA, AGAP1, AGMO, AGO1, AGO2, AGPAT2, AGPS, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AK1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG9, ALKBH8, AMER1, AMFR, AMPD2, AMT, ANAPC1, ANK2, ANK3, ANKRD11, ANKRD17, ANKS1B, ANO10, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP32, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARMC9, ARPC4, ARSL, ARV1, ARX, ASH1L, ASL, ASNS, ASPA, ASPM, ASXL1, ASXL2, ASXL3, ATAD3A, ATG7, ATIC, ATN1, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP5F1E, ATP5PO, ATP6AP2, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATP9A, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B9D1, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BICRA, BLM, BLOC1S1, BLTP1, BOD1, BOLA3, BPTF, BRAF, BRAT1, BRCA1, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BUB1B, BUD13, C12orf4, C12orf57, C2orf69, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D1, CACNA2D2, CAD, CAMK2A, CAMK2B, CAMK4, CAMSAP1, CAMTA1, CAPN10, CAPN15, CAPRIN1, CARS1, CARS2, CASK, CBL, CBS, CBY1, CC2D1A, CC2D2A, CCBE1, CCDC115, CCDC174, CCDC22, CCDC32, CCDC47, CCDC82, CCDC88A, CCDC88C, CCND2, CDC42, CDC42BPB, CDH11, CDH2, CDK10, CDK13, CDK19, CDK5RAP2, CDK6, CDK8, CDK9, CDKL5, CDON, CELF2, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP19, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CERT1, CHAMP1, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHRNA7, CIC, CIT, CKAP2L, CLCN2, CLCN3, CLCN4, CLCN6, CLDN11, CLDN5, CLP1, CLPB, CLPP, CLTC, CNKSR1, CNKSR2, CNNM2, CNOT1, CNOT3, CNOT9, CNPY3, CNTNAP1, CNTNAP2, COA8, COASY, COG1, COG5, COG6, COG7, COG8, COL18A1, COL27A1, COL3A1, COL4A1, COL4A2, COLEC11, COLGALT1, COPB1, COPB2, COQ2, COQ4, COQ8A, COX10, COX15, CPE, CPLANE1, CPLX1, CPSF3, CRADD, CRBN, CREBBP, CRIPT, CRLS1, CRPPA, CSDE1, CSF1R, CSGALNACT1, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CTBP1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTR9, CTU2, CUL4B, CUX1, CUX2, CWC27, CWF19L1, CYB5R3, CYFIP2, CYP26B1, CYP27A1, CYP2U1, D2HGDH, DAG1, DAGLA, DARS1, DARS2, DBT, DCAF17, DCHS1, DCPS, DCX, DDB1, DDC, DDHD2, DDOST, DDX23, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DENND5A, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHTKD1, DHX30, DHX37, DHX9, DIAPH1, DIP2B, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DMD, DMPK, DMXL2, DNAH14, DNAJC12, DNAJC19, DNM1, DNM1L, DNMT3A, DNMT3B, DOCK3, DOCK6, DOCK7, DOHH, DONSON, DPAGT1, DPF2, DPH1, DPH5, DPM1, DPM3, DPP6, DPYD, DPYSL5, DSCAM, DTYMK, DYM, DYNC1H1, DYNC1I2, DYRK1A, EARS2, EBF3, EBP, ECHS1, EDEM3, EED, EEF1A2, EEF1B2, EEF1D, EEF2, EFL1, EFTUD2, EHMT1, EIF2AK2, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2, ELOVL4, ELP2, EMC1, EMC10, EMG1, EML1, EMX2, ENTPD1, EP300, EPG5, EPRS1, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERI1, ERLIN2, ESAM, ESCO2, ETHE1, EXOC7, EXOSC2, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT2, EXTL3, EZH1, EZH2, FAM149B1, FAM50A, FANCD2, FAR1, FARS2, FASTKD2, FAT1, FAT4, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXW11, FBXW7, FCSK, FDFT1, FEM1B, FGD1, FGF12, FGF13, FGFR1, FGFR2, FGFR3, FH, FIBP, FIG4, FILIP1, FITM2, FKRP, FKTN, FLNA, FMN2, FMR1, FOLR1, FOSL2, FOXG1, FOXP1, FOXP2, FOXP4, FOXRED1, FRA10AC1, FRAS1, FRMD4A, FRMD5, FRMPD4, FRRS1L, FRY, FTO, FTSJ1, FUCA1, FUT8, FZR1, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALNT2, GALT, GAMT, GAN, GATA6, GATAD2B, GATM, GBA2, GCDH, GCH1, GCSH, GDF1, GDF11, GDI1, GEMIN4, GEMIN5, GFAP, GFM1, GFM2, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLIS3, GLRA2, GLUL, GMPPA, GMPPB, GNAI1,

GNAO1, GNAS, GNB1, GNB2, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GOT2, GPAA1, GPC3, GPC4, GPHN, GPT2, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM1, GRM7, GSS, GSX2, GTF2E2, GTF2H5, GTPBP2, GTPBP3, GUSB, H1-4, H3-3A, H3-3B, H4C3, H4C5, H4C6, H4C9, HACE1, HAX1, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HEATR3, HECTD4, HECW2, HEPACAM, HERC1, HERC2, HESX1, HGSNAT, HIBCH, HID1, HIKESHI, HIVEP2, HK1, HLCS, HMGB1, HNMT, HNRNPD, HNRNPH1, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HNRNPUL2, HOXA1, HPCA, HPD, HPDL, HPRT1, HRAS, HS2ST1, HSD17B10, HSD17B4, HTRA2, HTT, HUWE1, HYCC1, IARS1, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT172, IFT74, IGF1, IGF1R, IKBKG, IL1RAPL1, IMPA1, IMPDH2, INPP4A, INPP5E, INPP5K, INTS1, INTS11, INTS8, IPO8, IPP, IQSEC1, IQSEC2, IRF2BPL, IRX5, ISCA2, ITCH, ITGAV, ITPA, ITPR1, ITSN1, IVD, JAM2, JAM3, JARID2, KANSL1, KARS1, KAT5, KAT6A, KAT6B, KAT8, KATNB1, KATNIP, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNJ6, KCNK4, KCNK9, KCNMA1, KCNN2, KCNN3, KCNQ2, KCNQ5, KCNT1, KCNT2, KCTD7, KDM1A, KDM2B, KDM3B, KDM4B, KDM5A, KDM5B, KDM5C, KDM6A, KDM6B, KIAA0586, KIAA0753, KIDINS220, KIF11, KIF14, KIF1A, KIF21B, KIF2A, KIF4A, KIF5C, KIF7, KIFBP, KLHL15, KLHL20, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KPNA7, KPTN, KRAS, L1CAM, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LAS1L, LEO1, LETM1, LHX2, LIAS, LIG4, LINGO1, LINS1, LIPT2, LMBRD1, LMBRD2, LMNB1, LMNB2, LNPK, LONP1, LRP2, LRPPRC, LSS, LTBP1, LYRM7, LZTFL1, LZTR1, MAB21L1, MAB21L2, MACF1, MADD, MAF, MAG, MAGEL2, MAN1B1, MAN2B1, MAN2C1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K7, MAP4K4, MAPK1, MAPK8IP3, MAPKAPK5, MASP1, MAST1, MAST3, MAT1A, MBD5, MBOAT7, MBTPS2, MCCC2, MCM3AP, MCM6, MCOLN1, MCPH1, MDH2, MECP2, MED11, MED12, MED12L, MED13, MED13L, MED17, MED23, MED25, MED27, MEF2C, MEGF8, MEIS2, MESD, METTL23, METTL5, MFF, MFSD2A, MGAT2, MGME1, MICOS13, MICU1, MID1, MINPP1, MIR17HG, MKKS, MLC1, MLYCD, MMAA, MMACHC, MMADHC, MMUT, MN1, MOCS1, MOCS2, MOGS, MORC2, MPC1, MPDU1, MPDZ, MPLKIP, MRAS, MRE11, MRPS22, MRPS34, MSL3, MSMO1, MSTO1, MT-ATP6, MT-CO1, MT-CO2, MT-CO3, MT-ND5, MT-TK, MT-TL1, MT-TS1, MT-TV, MTFMT, MTO1, MTOR, MTR, MTRFR, MTRR, MTSS2, MVK, MYCBP2, MYCN, MYH10, MYO5A, MYRF, MYT1L, NAA10, NAA15, NAA20, NACC1, NAE1, NAGA, NAGLU, NALCN, NANS, NAPB, NARS1, NARS2, NAXD, NAXE, NBEA, NBN, NCAPG2, NCKAP1, NCOR1, NDE1, NDP, NDST1, NDUFA1, NDUFA11, NDUFA2, NDUFA6, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB8, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NECTIN1, NEDD4L, NEMF, NEU1, NEURL4, NEUROD2, NEUROG1, NEXMIF, NF1, NFASC, NFIA, NFIB, NFIX, NFU1, NGLY1, NHLRC2, NHS, NIPA1, NIPBL, NKAP, NKX6-2, NLGN3, NLRP3, NONO, NOVA2, NPRL2, NR2F1, NR2F2, NR4A2, NRAS, NRCAM, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NSUN2, NT5C2, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP107, NUP188, NUP214, OCLN, OCRL, ODC1, OFD1, OGDH, OGDHL, OGT, OPHN1, ORC1, OSGEP, OTUD5, OTUD6B, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PAN2, PANK2, PARN, PARS2, PAX5, PAX6, PBX1, PC, PCDH12, PCDH19, PCDHGC4, PCGF2, PCLO, PCNT, PCYT2, PDE2A, PDE4D, PDE6D, PDHA1, PDHX, PDSS1, PDSS2, PDZD8, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX2, PEX26, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM2L1, PGM3, PHF21A, PHF5A, PHF6, PHF8, PHGDH, PHIP, PI4KA, PIBF1, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY, PIK3CA, PIK3R1, PIK3R2, PIP5K1C, PISD, PITRM1, PLA2G6, PLAA, PLCB1, PLCH1, PLEKHG2, PLK4, PLP1, PLPBP, PLXNA1, PLXNA3, PMM2, PMPCA, PMPCB, PNKP, PNP, PNPLA6, PNPO, PNPT1, POC1A, POGZ, POLA1, POLG, POLR1A, POLR1C, POLR2A, POLR3A, POLR3B, POLRMT, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PORCN, POU3F2, POU3F3, POU4F1, PPFIA3, PPFIBP1, PPIL1, PPM1D, PPOX, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PQBP1, PRDM13, PRDM15, PREPL, PRICKLE2, PRKACB, PRKAR1B, PRMT7, PRODH, PRORP, PROSER1, PRPF8, PRPS1, PRR12, PRRT2, PRSS12, PRUNE1, PSMC3, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTPN11, PTPN23, PTRH2, PTRHD1, PUF60, PUM1, PURA, PUS1, PUS3, PUS7, PYCR1, PYCR2, QARS1, QRICH1, RAB11A, RAB11B, RAB18, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RABGAP1, RABL6, RAC1, RAC3, RAD21, RAF1, RAI1, RALA, RALGAPA1, RAP1B, RAP1GDS1, RARS1, RARS2, RBBP8, RBL2, RBM10, RBM28, RBPJ, RCBTB1, RELN, RERE, RFT1, RFX3, RFX7, RGS6, RHOBTB2, RIC1, RIMS2, RIT1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNF220, RNFT2, RNH1, RNPC3, RNU4ATAC, ROBO1, ROGDI, RORA, RPGRIP1L, RPH3A, RPL10, RPS6KA3, RRAS2, RRM2B, RSPRY1, RSRC1, RTEL1, RTN4IP1, RTTN, RUSC2, SALL1, SAMD9, SAMHD1, SART3, SASS6, SATB1, SATB2, SBDS, SC5D, SCAF4, SCAMP5, SCAPER, SCN10A, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHB, SDHD, SEMA6B, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGPL1, SGSH, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHQ1, SHROOM4, SIAH1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SKIC3, SLC12A2, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A2, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A42, SLC2A1, SLC30A9, SLC32A1, SLC33A1, SLC35A2, SLC35A3, SLC35B2, SLC35C1, SLC38A3, SLC39A8, SLC44A1, SLC45A1, SLC46A1, SLC4A10, SLC4A4, SLC5A7, SLC6A1, SLC6A17, SLC6A3, SLC6A8, SLC6A9, SLC9A6, SLC9A7, SLF2, SLITRK2, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMG8, SMG9, SMOC1, SMPD1, SMPD4, SMS, SNAP25, SNAP29, SNAPC4, SNIP1, SNRPB, SNX14, SNX27, SOBP, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX4, SOX5, SOX6, SPART, SPECC1L, SPEN, SPG11, SPOP, SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SRCAP, SRD5A3, SRP54, SRRM2, SRSF1, ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STEEP1, STIL, STIM1, STRA6, STRADA, STT3A, STUB1, STX1A, STX1B, STXBP1, SUCLA2, SUFU, SUOX, SUPT16H, SURF1, SUZ12, SVBP, SYN1, SYNCRIP, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAB2, TACO1, TAF1, TAF13, TAF2, TAF4, TAF6, TAF8, TANC2, TANGO2, TAOK1, TARS2,

TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBC1D7, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX2, TCEAL1, TCF12, TCF20, TCF4, TCF7L2, TCTN2, TDP2, TECPR2, TECR, TEFM, TELO2, TET3, TFE3, TGIF1, TH, THG1L, THOC2, THOC6, THRA, THUMPD1, TIAM1, TIMM50, TIMM8A, TIMMDC1, TKFC, TKT, TLK2, TM4SF20, TMCO1, TMEM106B, TMEM107, TMEM147, TMEM165, TMEM216, TMEM218, TMEM222, TMEM231, TMEM237, TMEM240, TMEM63A, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNIK, TNPO2, TNR, TNRC6B, TOE1, TOGARAM1, TOP3A, TOR1A, TOR1AIP1, TP53RK, TP73, TPI1, TPK1, TPP2, TPRKB, TRA2B, TRAF7, TRAIP, TRAK1, TRAPPC10, TRAPPC11, TRAPPC12, TRAPPC2L, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM32, TRIM8, TRIO, TRIP12, TRIT1, TRMT1, TRMT10A, TRMT5, TRNT1, TRPM3, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSPAN7, TSPOAP1, TTC5, TTC8, TTI1, TTI2, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUSC3, TWNK, UBA2, UBA5, UBAP2L, UBE2A, UBE3A, UBE3B, UBE4A, UBR1, UBR7, UBTF, UFC1, UFM1, UFSP2, UGDH, UGP2, UNC45A, UNC79, UNC80, UPB1, UPF3B, UQCC2, USP27X, USP7, USP9X, VAC14, VAMP2, VARS1, VARS2, VLDLR, VPS11, VPS13B, VPS13D, VPS16, VPS35L, VPS37A, VPS41, VPS4A, VPS53, VRK1, WAC, WARS1, WARS2, WASF1, WASHC4, WASHC5, WBP4, WDFY3, WDR11, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR5, WDR62, WDR73, WDR81, WIPI2, WLS, WNK3, WNT1, WWOX, XPA, XRCC4, XYLT1, YAP1, YARS1, YIF1B, YWHAG, YY1, ZBTB11, ZBTB18, ZBTB20, ZBTB24, ZBTB7A, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFHX3, ZFHX4, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF148, ZNF292, ZNF335, ZNF407, ZNF462, ZNF526, ZNF699, ZNF711, ZNHIT3, ZSWIM6

Heterotaxie v4 (56 Gene)

ACTC1, ACTG2, ACVR2B, ANKS6, CCDC103, CCDC32, CCDC39, CCDC40, CCDC65, CFAP298, CFAP300, CFAP45, CFAP52, CFAP53, CFC1, CRELD1, DAW1, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAH9, DNAI1, DNAI2, DNAL1, FANCB, FOXH1, FOXJ1, GDF1, INVS, LEFTY2, LRRC56, LZTFL1, MMP21, MNS1, NKX2-5, NME8, NODAL, NPHP4, ODAD1, ODAD2, ODAD3, ODAD4, PITRM1, PKD1L1, SPAG1, ZIC3, ZMYND10

Hirnfehlbildungen v6 (400 Gene)

AARS1, AARS2, ABAT, ABCD1, ACOX1, ACTB, ACTG1, ADAR, ADGRG1, ADSL, AGA, AIFM1, AIMP1, AKT1, AKT3, ALDH3A2, ALDH5A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARF1, ARFGEF2, ARSA, ARX, ASNS, ASPA, ASPM, ATP1A2, ATP1A3, ATP6V0A2, ATP7A, ATP7B, ATR, AUH, B3GALNT2, B3GNT2, B4GAT1, BCAP31, BOLA3, BRWD3, BUB1B, CASK, CCDC88C, CCND2, CDH2, CDK13, CDK5, CDK5RAP2, CDKN1C, CDON, CENPJ, CEP135, CEP152, CEP63, CEP85L, CHD8, CHMP1A, CLCN2, CLN6, CLP1, CLPP, CNOT1, COA8, COL4A1, COL4A2, COQ2, COQ9, COX10, COX14, COX15, COX6B1, CPS1, CRADD, CRPPA, CSF1R, CSNK2A1, CTC1, CTNNA2, CUL4B, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DCAF17, DCHS1, DCX, DEPDC5, DGUOK, DHCR24, DHCR7, DIS3L2, DISP1, DLL1, DNM1L, DNMT3A, DPYSL5, DYNC1H1, DYRK1A, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EML1, EMX2, ERCC6, ERCC8, ERMARD, ETFA, ETFB, ETFDH, EXOSC3, EZH2, FA2H, FARS2, FASTKD2, FAT4, FBXL4, FGF8, FGFR1, FH, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXH1, FOXRED1, FUCA1, GALC, GALT, GAN, GBE1, GCDH, GFAP, GFM1, GJA1, GJB1, GJC2, GLA, GLB1, GLI2, GLI3, GLRX5, GMPPB, GPC3, GPSM2, GRIA3, GRIN1, GRIN2B, H3-3A, HEPACAM, HEXA, HIBCH, HMGCL, HS2ST1, HSD17B4, HSPD1, HTRA1, HTRA2, HUWE1, HYCC1, IBA57, IDH2, IDS, IKBKG, ISCA2, JAM3, KATNB1, KIF14, KIF2A, KIF5C, KIF7, KIFBP, KMT2D, KNL1, KPTN, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LARGE1, LIAS, LMNB1, LYRM7, MACF1, MAP1B, MAPK8IP3, MARS2, MBD5, MCOLN1, MCPH1, MED12, MED17, MEF2C, MLC1, MN1, MOCS1, MPDZ, MPV17, MRPL44, MRPS22, MTFMT, MTOR, MTR, NADK2, NDE1, NDUFA1, NDUFA11, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS8, NDUFV1, NDUFV2, NEDD4L, NF1, NFIX, NFU1, NODAL, NOTCH3, NPC1, NPC2, NPRL2, NPRL3, NSD1, NSDHL, NSRP1, NUBPL, OCLN, OCRL, OFD1, OPHN1, OSGEP, OTC, PAFAH1B1, PAH, PC, PCLO, PCNT, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PI4KA, PIDD1, PIGA, PIK3CA, PIK3R2, PLCH1, PLP1, PNKP, POLG, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PPT1, PQBP1, PSAP, PTCH1, PTEN, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAD21, RARS1, RARS2, RBBP8, RELN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, RNF216, RPIA, RRM2B, RTTN, RXYLT1, SAMHD1, SCN3A, SCO1, SCP2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETD2, SHH, SIX3, SLC16A2, SLC17A5, SLC19A3, SLC25A1, SLC25A12, SLC25A4, SLC35A2, SMC1A, SMO, SNAP29, SOX10, SOX11, SPART, SPG11, SPG7, SPRED1, SRD5A3, SRPX2, STAG2, STIL, SUCLA2, SUFU, SUMF1, SURF1, SYN1, TACO1, TBC1D20, TBC1D23, TBC1D32, TBCK, TCIRG1, TGIF1, TMEM70, TMTC3, TMX2, TP73, TREM2, TREX1, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TTC19, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TWNK, TYMP, TYROBP, UGT1A1, UPF3B, VLDLR, VPS50, VPS53, VRK1, WASHC5, WDR62, WDR81, WNK3, ZFYVE26, ZIC2, ZNF335

Joubert-Syndrom v8 (38 Gene)

AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, C2CD3, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT172, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOGARAM1, TXNDC15, ZNF423

Kabuki-Syndrom v1 (2 Gene)

KDM6A, KMT2D

Kraniosynostose v5 (77 Gene)

ACTB, ACTG1, ALPL, ALX4, ARSB, ASXL1, B3GAT3, BRAF, CDC45, CHD5, CHD7, COLEC11, CTSK, CYP26B1, EFNB1, ERF, ESCO2, FAM20C, FGF10, FGFR1, FGFR2, FGFR3, FLNA, GLI3, GNAS, GNPTAB, HNRNPK, HUWE1, IDS, IDUA, IFT122, IHH, IL11RA, JAG1, KAT6A, KMT2D, KRAS, LTBP1, MASP1, MEGF8, MSX2, NFIA, P4HB, PHEX, PJA1, POR, PPP1CB, PRRX1, PTCH1, PTPN11, RAB23, RECQL4, RNU12, RUNX2, SEC24D, SHOC2, SIX1, SKI, SLC25A24, SMAD3, SMAD6, SMO, SOX6, SPECC1L, STAT3, TCF12, TFAP2B, TGFBR1, TGFBR2, TLK2, TMCO1, TRAF7, TWIST1, WDR35, ZEB2, ZIC1, ZNF462

Leukodystrophie und Leukenzephalopathie v6 (224 Gene)

AARS1, AARS2, ABAT, ABCD1, ACBD5, ACER3, ACOX1, ADAR, ADGRG1, ADSL, AGA, AIFM1, AIMP1, AIMP2, ALDH3A2, ALDH5A1, AP4B1, AP4E1, AP4M1, AP4S1, APP, ARSA, ASPA, ATP7A, ATP7B, ATPAF2, AUH, B3GALNT2, BCAP31, BCS1L, BOLA3, CIC, CLCN2, CLDN11, CLN6, CLPP, CNP, COA8, COL4A1, COL4A2, COQ2, COQ8A, COQ9, COX10, COX14, COX15, COX6B1, CPS1, CSF1R, CST3, CTC1, CTSA, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DCAF17, DEGS1, DGUOK, DNM1L, DPYD, EARS2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, ERCC6, ERCC8, ETFA, ETFB, ETFDH, FA2H, FARS2, FASTKD2, FBXL4, FDX2, FH, FIG4, FLVCR2, FOLR1, FOXRED1, FUCA1, GALC, GALNT2, GALT, GAN, GBE1, GCDH, GFAP, GFM1, GJA1, GJB1, GJC2, GLA, GLB1, GLRX5, GSN, HEPACAM, HEXA, HIBCH, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IBA57, IDH2, IDS, IFIH1, IKBKG, ISCA2, ITM2B, JAM3, KIF5A, L2HGDH, LAMB1, LARS2, LIAS, LIG3, LMNB1, LYRM7, MAL, MARS2, MCOLN1, MEF2C, MLC1, MOCS1, MPLKIP, MPV17, MRPL44, MRPS16, MRPS22, MTFMT, MTHFR, MTR, NADK2, NAXD, NAXE, NDUFA1, NDUFA11, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NKX6-2, NOTCH3, NPC1, NPC2, NUBPL, OCLN, OCRL, OTC, PAFAH1B1, PAH, PC, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLEKHG2, PLP1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POLR3K, PPT1, PRNP, PSAP, PSEN1, PSEN2, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RNF220, RPIA, RPS6KA3, RRM2B, SAMHD1, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SLC16A2, SLC17A5, SLC19A3, SLC25A1, SLC25A12, SLC25A4, SNORD118, SOX10, SPART, SPG11, SPG21, SPG7, SUCLA2, SUMF1, SURF1, TACO1, TBCK, TCIRG1, TMEM106B, TMEM63A, TMEM70, TPP2, TREM2, TREX1, TTC19, TUBB4A, TUFM, TWNK, TYMP, TYROBP, UFM1, UGT1A1, VPS11, ZFYVE26

Lymphödem (inkl. Rasopathien) v3 (49 Gene)

ADAMTS3, ANGPT2, ARAP3, BRAF, CBL, CCBE1, CELSR1, CHD7, DCHS1, EPHB4, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, HRAS, IKBKG, KIF11, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, NSD1, PIEZO1, PMM2, PPP1CB, PTPN11, PTPN14, RAF1, RASA1, RASA2, RIT1, RORC, RRAS, RRAS2, SHANK3, SHOC2, SOS1, SOS2, SOX18, SPRED1, TIE1, TSC1, TSC2, VEGFC

Mikrophthalmie/Anophthalmie v2 (49 Gene)

ABCB6, ALDH1A3, BCOR, BMP4, CAPN15, COL4A1, FAT1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, GDF3, GDF6, GRIP1, HCCS, HMGB3, KMT2D, MAB21L2, MFRP, MITF, MYRF, NAA10, OTX2, PAX2, PAX6, PITX2, PLK4, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SMCHD1, SMO, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TMEM98, TUBGCP4, VAX1, VPS35L, VSX2

Mikrozephalie und pontocerebelläre Hypoplasie v7 (245 Gene)

AARS1, ADARB1, AFG2B, AGMO, AIMP2, AMPD2, ANKLE2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARF3, ASPM, ATP11A, ATP1A2, ATP6V0A1, ATP6V0C, ATP9A, ATR, ATRX, BLM, BPTF, BRCA2, BRD4, BRIP1, BUB1B, CAMK2B, CASK, CCDC88A, CCND2, CDC40, CDK5RAP2, CDK6, CDT1, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP55, CEP57, CEP63, CHAMP1, CHKA, CHMP1A, CIT, CKAP2L, CLP1, COASY, COPB1, COPB2, CPSF3, CREBBP, CRIPT, CSNK2A1, CTCF, CTNNB1, CTU2, DDX11, DHCR7, DIAPH1, DNA2, DNMT3A, DOHH, DONSON, DPM1, DPP6, DROSHA, DYNC1H1, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, EIF5A, ERCC4, ERCC5, ERCC6, ERCC8, EXOC7, EXOSC1, EXOSC3, EXOSC8, EXOSC9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FBRSL1, FOXG1, GMNN, GPT2, GTF2E2, H4C3, HDAC8, HHAT, HMGB1, HPDL, IARS1, IER3IP1, IGF1, IGF1R, KIF11, KIF14, KIFBP, KNL1, LAGE3, LARP7, LIG4, LMNB1, LMNB2, MBD5, MCM7, MCPH1, MED17, METTL5, MFSD2A, MINPP1, MORC2, MPLKIP, MRE11, MSMO1, MYCN, NAA20, NAPB, NARS1, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIPBL, NSD2, NSRP1, NUP107, NUP188, NUP214, NUP37, ORC1, ORC4, ORC6, OSGEP, PALB2, PCDH12, PCDHGC4, PCLO, PCNT, PDCD6IP, PDHA1, PHC1, PLK4, PNKP, POC1A, POGZ, PPIL1, PPP1R15B, PQBP1, PRDM13, PRIM1, PRUNE1, PTPN23, PUF60, PUS7, QARS1, RABGAP1, RAD21, RAD50, RAD51, RAD51C, RARS2, RBBP8, RMI1, RNU4ATAC, RPL10, RRP7A, RTTN, SARS1, SASS6, SEPSECS, SLC1A4, SLC25A19, SLC25A46, SLC38A3, SLC9A6, SLX4, SMARCA5, SMC1A, SMC3, SMG8, STAMBP, STIL, SVBP, TAF13, TBC1D23, TMX2, TNPO2, TOE1, TOP3A, TP53RK, TRAIP, TRAPPC12, TRAPPC14, TRAPPC6B, TRAPPC9, TRIO, TRIP13, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TTC5, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, UBA5, UBE3A, UFC1, UFM1, UGP2, UNC80, VPS50, VPS51, VPS53, VRK1, WDFY3, WDR11, WDR37, WDR4, WDR62, WDR73, XRCC4, YIF1B, YIPF5, ZEB2, ZNF335, ZNF526, ZNF668

Neuronale Migrationsstörungen v6 (110 Gene)

ACTB, ACTG1, ADGRG1, AKT3, APC2, ARF1, ARFGEF2, ARX, ASPM, ATP1A2, ATP1A3, B3GALNT2, B4GAT1, CASK, CCND2, CDH2, CDK13, CEP85L, CRADD, CRPPA, CSNK2A1, CTNNA2, DAG1, DCHS1, DCX, DEPDC5, DPYSL5, DYNC1H1, EML1, EMX2, FAT4, FIG4, FKRP, FKTN, FLNA, GPSM2, GRIN1, GRIN2B, H3-3A, KATNB1, KIF2A, KIF5C, LAMA2, LAMB1, LAMC3, LARGE1, MACF1, MAP1B, MAPK8IP3, MN1, MTOR, NDE1, NEDD4L, NPRL2, NPRL3, NSRP1, OCLN, OSGEP, PAFAH1B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PI4KA, PIDD1, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMT1, POMT2, PPP1R12A, PTEN, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, RXYLT1, SCN3A, SLC35A2, SMO, SNAP29, SOX11, TBC1D32, TMX2, TP73, TSC1, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, VLDLR, VPS50, WDR62, WNK3

Oro-fazio-digitales Syndrom (OFD) v5 (17 Gene)

C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, SCLT1, SCNM1, TBC1D32, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, WDPCP

Osteopetrose v3 (20 Gene)

AMER1, ANKH, CA2, CLCN7, CTSK, FAM20C, FERMT3, LEMD3, LRP5, OSTM1, PLEKHM1, PTH1R, RASGRP2, SNX10, SOST, TCIRG1, TGFB1, TNFRSF11A, TNFSF11, TYROBP

Rasopathien (inkl. Noonan-Syndrom) v9 (23 Gene)

BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2

Rett-Syndrom und ähnliche Erkrankungen v3 (17 Gene)

ALDH5A1, ARX, BDNF, CDKL5, FOXG1, FOXP2, KCNQ2, MECP2, MEF2C, NTNG1, PLP1, SCN2A, SHANK3, STXBP1, TCF4, UBE3A, ZEB2

Sotos-Syndrom v1 (3 Gene)

APC2, NSD1, NFIX

Ziliopathien v7 (122 Gene)

AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, CC2D2A, CCDC28B, CCDC32, CCNQ, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP418, CILK1, CPLANE1, CRB2, CSPP1, DCDC2, DDX59, DHCR7, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, FAM149B1, GLI2, GLI3, GLIS2, HNF1B, HYLS1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LAMA1, LBR, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIBF1, PIK3C2A, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, RPGRIP1L, SBDS, SCLT1, SDCCAG8, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, USP9X, VPS13B, WDPCP, WDR19, WDR35, XPNPEP3, ZNF423, ZSWIM6