Gen-Panels Wachstumsstörungen | 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.

Kleinwuchs v7 (185 Gene)

ACAN, ACTB, ACTG1, ALMS1, AMMECR1, ANAPC1, ANKRD11, ARCN1, ATR, B3GAT3, BCS1L, BLM, BRAF, BRCA2, BRIP1, BTK, CBL, CCDC186, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP57, CEP63, CHD7, COG4, COL27A1, CREBBP, CRIPT, CUL7, DHCR7, DONSON, EP300, ERCC4, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGF8, FGFR1, FGFR3, FN1, FOXP4, GGPS1, GH1, GHR, GHRHR, GHSR, GLI2, GLI3, GMNN, GNAS, HDAC8, HESX1, HMGA2, HRAS, IDUA, IFT172, IGF1, IGF1R, IGF2, IGFALS, IGSF1, IHH, IKBKG, IL2RG, INSR, INTS1, IRS1, KANSL1, KDM3B, KDM6A, KMT2D, KRAS, LARP7, LFNG, LHX3, LHX4, LIG4, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, MSTO1, MTX2, NBAS, NBN, NF1, NHLRC2, NIPBL, NLRP2, NLRP5, NLRP7, NOTCH2, NPR2, NRAS, OBSL1, ORC1, ORC4, ORC6, OSGEP, OTX2, PADI6, PALB2, PAPPA2, PCNT, PIK3R1, PISD, PITX2, PLAG1, PLK4, PNPLA6, POC1A, POP1, POU1F1, PPP1CB, PPP3CA, PRMT7, PROKR2, PROP1, PTPN11, PUF60, RAD21, RAD51, RAF1, RALA, RAP1B, RASA2, RBBP8, RIT1, RNPC3, RNU4ATAC, ROR2, RPL10, RPS6KA3, RRAS, RRAS2, RTTN, SAMD9, SETD5, SGMS2, SHOC2, SHOX, SLX4, SMARCA2, SMARCE1, SMC1A, SMC3, SOS1, SOS2, SOX11, SOX2, SOX3, SPINK5, SPR, SPRED2, SRCAP, STAT5B, TALDO1, TBCE, TBX19, TBX2, TBX3, TOP3A, TRIM37, TRMT10A, UBE2T, VPS50, WRN, XRCC4, ZFP57, ZNF668

Kleinwuchs, idiopathisch nichtsyndromal v1 (2 Gene)

ACAN, SHOX

Makrozephalie / Überwuchssyndrome v8 (97 Gene)

ABCC9, AKT1, AKT2, AKT3, AMER1, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD3, CHD4, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, DVL1, EED, EIF2B5, EZH2, FGFR3, FIBP, GATAD2B, GCDH, GFAP, GLI3, GNAS, GPC3, GPC4, GPSM2, GRIA3, H1-4, HEPACAM, HERC1, HRAS, HUWE1, KCNH1, KDM1A, KIF7, KMT2E, KPTN, KRAS, L1CAM, LBR, MED12, MLC1, MPDZ, MTOR, NF1, NFIA, NFIB, NFIX, NONO, NPR2, NRAS, NSD1, ODC1, OFD1, PDGFRB, PHF21A, PHF6, PIGA, PIGM, PIK3CA, PIK3R2, PPP1CB, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RASA1, RIN2, RNF125, RNF135, SETD2, SHANK3, SOS1, SOST, STRADA, SUZ12, SYN1, SZT2, TBC1D7, TCF20, TMEM94, TRIO, TRIP12, TSC1, TSC2, UPF3B, WASHC5, WNT5A, ZBTB20, ZBTB7A

Segmentale Überwuchssyndrome v3 (15 Gene)

AKT1, AKT2, AKT3, ARAF, CCND2, CDKN1C, GJA4, NLRP2, PADI6, PIK3CA, PIK3R1, PIK3R2, PTEN, RASA1, SUZ12

Skeletterkrankungen v6 (458 Gene)

ABCC9, ABL1, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSK, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, ATR, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B9D1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP152, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRTAP, CSGALNACT1, CSNK1G1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DCC, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DROSHA, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFNB1, EFTUD2, EIF2AK3, ENPP1, EOGT, EP300, ERF, ESCO2, EVC, EVC2, EXOC6B, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBLN1, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GNAS, GNPAT, GNPNAT1, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HDAC4, HDAC8, HEATR3, HES7, HGSNAT, HHAT, HNRNPK, HOXA11, HOXA13, HOXD13, HPGD, HS2ST1, HSPG2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, INPPL1, KAT6B, KDELR2, KIAA0753, KIF22, KIF24, KIF5B, KIF7, KMT2D, LARP7, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, LYSET, MAFB, MAN2B1, MANBA, MAP3K7, MASP1, MATN3, MBTPS1, MBTPS2, MEGF8, MEOX1, MESD, MESP2, MGP, MIR17HG, MKKS, MKS1, MMP13, MMP2, MMP9, MNX1, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NRCAM, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PDIA6, PEX5, PEX7, PFN1, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLEKHM1, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SIK3, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35B2, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUCO, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFBR1, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TWIST2, TYROBP, UBA2, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZIC1, ZMPSTE24, ZNF687, ZSWIM6