



Genpanel Knochen-/Gefäss- und Bindegewebserkrankungen

Zerebelläre autosomal-dominante Arteriopathie mit subkortikalen Infarkten – CADASIL und DD

13 Gene: *ABCC6, APP, ATP1A2, CACNA1A, COL3A1, COL4A1, COL4A2, COLGALT1, FOXC1, GLA, HTRA1, NOTCH3, TREX1*

Referenz: Familial cerebral small vessel disease v1.17), Genomics England PanelApp (panelapp.genomicsengland.co.uk, access date: 28.01.2025)

Familiäres thorakales Aortenaneurysma und Aortendissektion (FTAAD)

33 Gene: *ABL1, ACTA2, ADAMTSL4, ARIH1, BGN, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FLCN, FLNA, IPO8, LOX, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2*

Referenz: Thoracic aortic aneurysm or dissection (Version 1.127), Genomics England PanelApp (panelapp.genomicsengland.co.uk, access date: 28.01.2025)

Ehlers-Danlos-Syndrome

49 Gene: *ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, EFEMP1, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, GORAB, LOX, LTBP2, LTBP4, PLOD1, PRDM5, PYCR1, RIN2, ROBO3, SKI, SLC39A13, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469*

Referenz: Ehlers-Danlos syndrome with a likely monogenic cause v3.21, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 28.01.2025)

Loeys-Dietz-Syndrom

6 Gene: *SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*

Referenz: Ehlers-Danlos syndromes v2.66, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 07.11.2022)

Osteogenesis Imperfecta

35 Gene: *ALPL, B3GALT6, B4GALT7, BMP1, CASR, COL1A1, COL1A2, COPB2, CREB3L1, CRTAP, FAM46A, FKBP10, GORAB, IFITM5, KDELR2, LRP5, MESD, NBAS, NOTCH2, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, TRPV6, UNC45A, WNT1, WNT11*

Referenz: Osteogenesis imperfecta v4.9, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 28.01.2025)

Skelettdysplasien: schwere/letale Formen

39 Gene: *ALPL, CEP120, CFAP410, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, FGFR1, FGFR2, FGFR3, FLNB, IFT140, IFT172, IFT52, IFT80, IFT81, KIAA0586, KIAA0753, NEK1, P3H1, SLC26A2, SLC26A2, SOX9, TRAF3IP1, TRIP11, TTC21B, WDR19, WDR34, WDR35, WDR60*

Referenz: u.a. Stembalska, Dudarewicz, Smigiel, Adv Clin Exp Med, 2021, PMID: 34019743

Skelettdysplasien: allgemein

423 Gene: *ABCC9, 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, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, AXIN1, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CBF, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, 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, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ERI1, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FBXW11, 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, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HHAT, 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, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS1, MEGF8, MEOX1, MESD, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MNX1, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEPRO, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1,*

ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSMC3, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RINT1, ROR2, RPGRI1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SETD5, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SMOC1, SNRNPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB3, TMC01, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UBA2, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6

Referenz: Skeletal Dysplasia v7.16, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 28.01.2025)

Die aufgeführten Gene entsprechen den der jeweiligen Referenz zugrundeliegenden Empfehlungen. Die gemäss Genomics England PanelApp aufgeführten Gene entsprechen den als «diagnostic-grade», also diagnostisch gewerteten Genen (s.a. <https://panelapp.genomicsengland.co.uk/#!/Guidelines>).

Die Auswertung weiterer Gene ist nach Rücksprache mit unserem Labor möglich.