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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 v1.14,), Genomics England PanelApp (panelapp.genomicsengland.co.uk, access date: 07.11.2022); https://panelapp.genomicsengland.co.uk/panels/50/

Familiäres thorakales Aortenaneurysma und Aortendissektion (FTAAD)

68 Gene: ABL1, ACTA2, BGN, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LOX, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, ARIH1, ASPH, FOXE3, IPO8, LTBP3, THSD4, ABCC6, ACVR1, ADAMTS2, ALDH18A1, ATP6V0A2, ATP7A, B4GALT7, CHST14, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, EMILIN1, FLCN, HEY2, HNRNPK, KCNN1, LTBP2, MAT2A, MED12, MYLK2, PKD1, PKD2, SLC39A13, TNXB, ZNF469

Referenz: Thoracic aortic aneurysm and dissection (Version 1.129), Genomics England PanelApp (panelapp.genomicsengland.co.uk, access date: 07.11.2022; https://panelapp.genomicsengland.co.uk/panels/700/)

Ehlers-Danlos-Syndrome

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

Referenz: Ehlers-Danlos syndromes v2.66, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 07.11.2022; https://panelapp.genomicsengland.co.uk/panels/53/)

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; https://panelapp.genomicsengland.co.uk/panels/53/)

Kraniosynostose-Syndrome

54 Gene: ALPL, ALX4, ARSB, ASXL1, BRAF, CDC45, COLEC11, CTSK, CYP26B1, EFNB1, ERF, FAM20C, FGFR1, FGFR2, FGFR3, FLNA, GLI3, GNAS, GNPTAB, HUWE1, IDS, IDUA, IFT122, IHH, IL11RA, JAG1, KAT6A, KMT2D, KRAS, MEGF8, MSX2, P4HB, PHEX, POR, PTPN11, RAB23, RECQL4, RUNX2, SKI, SLC25A24, SMAD6, SMO, SPECC1L, STAT3, TCF12, TFAP2B, TGFBR1, TGFBR2, TLK2, TMCO1, TWIST1, WDR35, ZEB2, ZIC1

Referenz: Craniosynostosis v.2.67, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 07.11.2022; https://panelapp.genomicsengland.co.uk/panels/168/)

Osteogenesis Imperfecta

31 Gene: ALPL, B3GALT6, B4GALT7, BMP1, CASR, COL1A1, COL1A2, CREB3L1, CRTAP, DSPP, FAM46A, FKBP10, GORAB, IFITM5, LRP5, NBAS, NOTCH2, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, TRPV6, WNT1

Referenz: Osteogenesis imperfecta v2.48, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 07.11.2022; https://panelapp.genomicsengland.co.uk/panels/196/)

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

373 Gene: ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARHGAP31, ARL6, ARSB, ARSE, ASXL1, ASXL2, ATP6V0A2, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, C21orf2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CHST14, CHST3, CHSY1, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DSPP, DVL1, DVL3, DYM, DYNC2LI1, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FAM46A, FAM58A, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, 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, GSC, GUSB, HDAC8, HES7, HGSNAT, HOXA13, HOXD13, HPGD, HSPG2, ICK, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, INPPL1, KAT6B, KIAA0753, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MEGF8, MEOX1, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MNX1, MPDU1, MSX2, MYCN, NAGLU, NANS, NBAS, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NSD1, NSDHL, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PITX1, PLOD2, PLS3, POC1A, POLR1A, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCAL1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTEX1D2, TCTN2, TCTN3, TERT, TGFB1, TGFB2, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC8, TWIST1, TYROBP, VDR, WDPCP, WDR34, WDR60, WISP3, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZSWIM6

Referenz: Skeletal Dysplasia v2.222, Genomics England PanelApp (panelapp.genomicsengland.co.uk; access date: 07.11.2022; https://panelapp.genomicsengland.co.uk/panels/309/)

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.