

Leukodisztrófia (Leukodystrophy) génpanel v:2022.01.28.

Panel tartalma 757 gén:

AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACTA2, ACTB, ACY1, ACSF3, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALDH7A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMN, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOA1BP, APOPT1, APP, ARCN1, ARFGEF2, ARHGAP31, ARHGEF10, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP11A, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BEST1, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTBD, C11ORF73, C12orf57, C12orf65, C19orf12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CHMP2B, CIC, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CP, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CTBP1, CTC1, CTD1P1, CTNS, CTSA, CTSD, CTSF, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, CSF1R, CSPP1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHH, DHX37, DLD, DLL4, DMXL2, DNAJC5, DNM1L, DNM2, DNMT1, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYRK1A, EARS2, EDNRB, EGR2, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX1L, FDX2, FGD4, FGFR1, FH, FIG4, FKR, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FTL, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALT, GALNT2, GALT, GAN, GATAD2B, GBE1, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GRN, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMBS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, HTRA1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIAA1161, KIDINS220, KIF1A, KIF1C, KIF5A, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE, LARGE1, LARS2, LETM1, LIAS, LIG3, LIPT1, LIPT2, LITAF, LMNB1, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MAPT, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFF, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDE1,

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Inherited white matter disorders (Version 1.150)

<https://panelapp.genomicsengland.co.uk/panels/42/> (Megtekintve: 2022.01.28.)