

Értelmi elmaradás (Intellectual disability) génpanel v:2023.10.12.

Panel tartalma 2620 gén:

A2ML1, AAAS, AARS, AASS, ABAT, ABCA2, ABCB11, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABCG5, ABHD12, ABHD16A, ABHD5, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAN, ACAT1, ACBD6, ACE2, ACER3, ACIN1, ACO2, ACOT9, ACOX1, ACOX2, ACP5, ACTA2, ACTB, ACTG1, ACTL6A, ACTL6B, ACVR1, ACY1, ACSF3, ACSL4, ADA, ADAM22, ADAMTS10, ADAR, ADARB1, ADAT3, ADCY5, ADD1, ADD3, ADGRG1, ADGRG4, ADGRG6, ADGRV1, ADK, ADNP, ADPRHL2, ADRA2B, ADSL, AFF2, AFF3, AFF4, AFG3L2, AFP, AGA, AGAP1, AGK, AGL, AGMO, AGO1, AGO2, AGPAT2, AGPS, AGT, AGTR2, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AIPL1, AIRE, AK1, AK2, AKAP17A, AKAP4, AKAP6, AKR1C2, AKR1D1, AKT1, AKT3, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALS2, ALX1, ALX3, ALX4, AMER1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKRD11, ANKRD17, ANO10, ANO3, ANO5, ANTXR1, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP2S1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, APTX, AQP7, AR, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGAP35, ARHGAP36, ARHGAP6, ARHGEF2, ARHGEF4, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARIH1, ARL13B, ARL14EP, ARL6, ARMC4, ARMC9, ARSA, ARSB, ARSE, ARSF, ARV1, ARX, ASAH1, ASB12, ASCC3, ASCL1, ASH1L, ASL, ASMT, ASMTL, ASNS, ASPA, ASPH, ASPM, ASS1, ASTN1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATM, ATN1, ATP11A, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP2B3, ATP2C2, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATP9A, ATR, ATRX, ATXN1, ATXN1_CAG, ATXN10, ATXN2, ATXN2L, ATXN3, ATXN3L, ATXN7, AUH, AUTS2, AVP, AVPR2, AWAT2, B3GALNT2, B3GALT6, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B9D1, B9D2, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BDP1, BEAN1, BFSP2, BGN, BHLHA9, BICD2, BICRA, BIN1, BLM, BLOC1S1, BMP15, BMP4, BMPER, BMPR1B, BOLA3, BPIFB6, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRIP1, BRPF1, BRSK2, BRWD3, BSCL2, BSND, BTBD, BTK, BUB1, BUB1B, C12orf4, C12orf57, C12orf65, C16orf62, C19orf12, C2CD3, C2orf71, C3orf58, C4orf26, C5orf42, C8orf37, C9orf72, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1H, CACNA1I, CACNA1S, CACNA2D1, CACNA2D2, CACNA2D3, CACNB4, CACNG2, CAD, CAMK2A, CAMK2B, CAMK2G, CAMK4, CAMTA1, CANT1, CAP1, CAPN10, CAPN15, CAPRIN1, CAPZA2, CARS, CARS2, CASK, CASP2, CASR, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC114, CCDC115, CCDC174, CCDC186, CCDC22, CCDC32, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC82, CCDC88A, CCDC88C, CCNA2, CCNB3, CCND2, CCNO, CCT5, CD96, CD99, CDC40, CDC42, CDC42BPB, CDC45, CDC6, CDH11, CDH15, CDH2, CDH23, CDH3, CDK10, CDK13, CDK16, CDK19, CDK5R1, CDK5RAP2, CDK8, CDK9, CDKL5, CDKN1C, CDON,

CDT1, CELF2, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CEP85L, CFAP47, CFP, CHAMP1, CHD1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHL1, CHM, CHMP1A, CHRDL1, CHRM1, CHRNA2, CHRNA4, CHRNB2, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CIC, CISD2, CIT, CKAP2L, CLCN2, CLCN3, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN19, CLIC2, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CMC4, CNKSR1, CNKSR2, CNNM2, CNOT1, CNOT2, CNOT3, CNPY3, CNTN3, CNTN4, CNTNAP1, CNTNAP2, COA3, COA5, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL18A1, COL1A1, COL1A2, COL25A1, COL2A1, COL4A1, COL4A2, COL4A3, COL4A3BP, COL4A4, COL4A6, COL6A1, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB1, COPB2, COQ2, COQ4, COQ5, COQ8A, COQ9, COX10, COX14, COX15, COX6B1, COX7B, CP, CPA6, CPD, CPE, CPLX1, CPS1, CPSF3, CPXCR1, CRADD, CRB1, CRB2, CRBN, CREBBP, CRLF2, CRX, CRYAA, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CTBP1, CTC1, CTCF, CTDP1, CTGF, CTNNA2, CTNNB1, CTNND1, CTNND2, CTNS, CTPS2, CTR9, CTSA, CTSD, CTSF, CTSK, CTTNBP2, CTU2, CUL3, CUL4B, CUL7, CUX1, CUX2, CWC27, CWF19L1, CXorf56, CXorf58, CYB5R3, CYC1, CYFIP2, CYP1B1, CYP27A1, CYP2U1, CYP7B1, CSDE1, CSF1R, CSF2RA, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CSTB, CSTF2, D2HGDH, DAB1, DACT1, DAG1, DALRD3, DARS, DARS2, DBT, DCAF17, DCC, DCHS1, DCHS2, DCPS, DCTN1, DCX, DDB1, DDB2, DDC, DDHD1, DDHD2, DDOST, DDR2, DDX11, DDX23, DDX3X, DDX53, DDX58, DDX59, DDX6, DEAF1, DECR1, DEGS1, DENND5A, DEPDC5, DGKH, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHPS, DHRSX, DHTKD1, DHX30, DHX32, DHX37, DIAPH1, DIAPH2, DIP2B, DIS3L2, DKC1, DLAT, DLD, DLG1, DLG2, DLG3, DLG4, DLGAP2, DLL1, DLL3, DLL4, DMD, DMP1, DMPK, DMXL2, DNA2, DNAAF3, DNAAF4, DNAH14, DNAJC12, DNAJC19, DNAJC3, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DNMT3B, DOCK11, DOCK3, DOCK6, DOCK7, DOCK8, DOHH, DOLK, DONSON, DPAGT1, DPF1, DPF2, DPF3, DPH1, DPH2, DPH5, DPM1, DPM2, DPM3, DPP6, DPYD, DPYS, DPYSL2, DPYSL5, DRD2, DROSHA, DSCAM, DSCR3, DSPP, DST, DSTYK, DTYMK, DVL1, DVL3, DYM, DYNC1H1, DYNC1I2, DYNC2H1, DYRK1A, EARS2, EBF3, EBP, ECEL1, EDA, EDEM3, EDNRA, EDNRB, EED, EEF1A2, EEF1B2, EFHC1, EFN1B1, EFTUD2, EGR2, EHMT1, EIF2A, EIF2AK1, EIF2AK2, EIF2AK3, EIF2S3, EIF3F, EIF4A3, EIF4G1, EIF5A, ELAC2, ELK1, ELN, ELOVL4, ELOVL5, ELP2, EMC1, EMC10, EMG1, EML1, EMX2, EN2, ENOX2, ENPP1, ENTPD1, EOGT, EOMES, EP300, EPB41L1, EPG5, EPHA7, EPM2A, EPPK1, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERGIC3, ERLIN2, ERMARD, ESCO2, ESX1, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC2, EXOC7, EXOSC3, EXOSC8, EXT1, EXT2, EXTL3, EYA1, EZH2, F5, FA2H, FAAH2, FAH, FAM111A, FAM111B, FAM120C, FAM126A, FAM160B1, FAM161A, FAM20A, FAM20C, FAM47B, FAM50A, FAM58A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FAR1, FARS2, FARSA, FARSB, FASN, FAT4, FBLN5, FBN1, FBN2, FBP1, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO25, FBXO28, FBXO31, FBXO7, FBXO8, FBXW11, FBXW4, FBXW7, FDFT1, FDXR, FEM1B, FGD1, FGD4, FGF10, FGF12, FGF13, FGF14, FGF3, FGFR1, FGFR2, FGFR3, FH, FHL1, FIBP, FICD, FIG4, FKBP14, FKBP6, FKBPL, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1,

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Forrás:

Intellectual disability - microarray and sequencing (Version 5.124)

<https://panelapp.genomicsengland.co.uk/panels/285/> (Megtekintve: 2023.05.16.)