

EXPLORATION DES ATAXIES CEREBELLEUSES HEREDITAIRES

Liste des gènes analysés en NGS au 1/09/2020 :

AAAS, AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ABCD5, ACO2, ACOX2, ADCY5, ADGRB3, ADK, ADPRHL2, AFG3L2, AGTPBP1, AIFM1, AIMP1, ALAS2, ALG6, AMACR, AMT, ANO10, AP4M1, AP5Z1, APOB, APTX, ARFGF2, ARL6IP1, ARSA, ASL, ASS1, ATAD3A, ATAD3B, ATCAY, ATG5, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2B2, ATP2B3, ATP7B, ATP8A2

B4GALNT1, BCAP31, BEAN1, BOLA3, BRAF, BRAT1, BTD

C12ORF65, C19ORF12, CA8, CACNA1A, CACNA1C, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CCDC88C, CHP1, CLCN2, CLN5, CLN6, CLN8, COA7, COG5, COL18A1, COQ2, COQ4, COQ5, COQ8A, COQ8B, COQ9, COX10, COX20, CP, CPS1, CRAT, CRBN, CRPPA, CSF1R, CTBP1, CTDP1, CTSA, CTSD, CUL4B, CWF19L1, CYP27A1, CYP7B1

DARS2, DBT, DCAF17, DKC1, DLAT, DNAJC3, DNAJC5, DNAJC19, DNMT1, DOCK3, DOCK6

EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELF2, ELOVL4, ELOVL5, EMC1, EP300, EPRS1, ERCC4, ERCC5, ERCC6, ERCC8, EXOSC9

FA2H, FAT1, FAT2, FDXR, FGF12, FGF14, FLVCR1, FOXRED1, FRMD4A, FTL, FXN

GALC, GBA, GBA2, GBE1, GCDH, GCSH, GDAP2, GFAP, GJB1, GJC2, GLB1, GLDC, GLRX5, GM2A, GMPPB, GNAO1, GOSR2, GPAA1, GPR88, GRID2, GRIK2, GRIN2B, GRM1, GSN

HARS1, HEXA, HEXB, HIBCH, HPRT1, HSD17B4, HSD17B10, HSPD1, HTRA1

IFRD1, ITM2B, ITPR1

KCNA1, KCNA2, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KIF1A, KIF1C, KIF26B, KIF5A, KIF7

L2HGDH, LAMA1, LITAF, LMNB1, LMNB2, LONP1, LRPPRC, LRSAM1, LYST

MAG, MARS2, MBIP, MECP2, MED17, MFSD8, MGME1, MLC1, MMACHC, MMADHC, MME, MORC2, MRE11A, MSTO1, MTCL1, MTPAP, MTPP, MYO5A

NALCN, NAXE, NDUFAF2, NDUFS7, NEU1, NFASC, NGLY1, NIPA1, NKX2-1, NKX6-2, NOL3, NPC1, NPC2, NRDC, NUBPL

OPA1, OPA3, OPHN1, OTC, OTUD4

PANK2, PAX6, PCDH12, PCNA, PDE2A, PDE10A, PDE8B, PDHA1, PDHX, PDSS1, PDSS2, PDYN, PEX2, PEX6, PEX7, PEX10, PEX16, PHYH, PIBF1, PIGN, PIGS, PIK3R5, PITRM1, PLA2G6, PLD3, PLEKHG4, PLP1, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, POLG, POLG2, POLR3A, POLR3B, POMGNT2, PRICKLE1, PRKCG, PRNP, PRPS1, PRRT2, PRUNE1, PTRH2, PUM1

QARS1

RARS1, RBM10, REPS1, RNF216, RPIA, RUBCN

SACS, SAMD9L, SCARB2, SCN1A, SCN2A, SCN8A, SCYL1, SDHA, SEPSECS, SETX, SIL1, SLC1A3, SLC2A1, SLC6A19, SLC9A1, SLC9A6, SLC17A5, SLC20A2, SLC25A1, SLC25A15, SLC25A46, SLC30A9, SLC30A10, SLC44A1, SLC52A2, SNX14, SPART, SPG7, SPG11, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, STUB1, STXBP1, SYNE1, SYT14

TARDBP, TBC1D23, TBC1D24, TDP1, TDP2, TGM6, THG1L, TINF2, TMEM92, TMEM240, TOR1AIP1, TPK1, TPP1, TRAPPC11, TRPC3, TTBK2, TTC19, TTC21B, TTPA, TUBB4A, TWNK

UBA5, UBR4, UBTF, UCHL1, UFM1

VAMP1, VCP, VPS13A, VPS13D, VWA3B

WDR73, WDR81, WFS1, WWOX

XK, XPA, XRCC1

ZFYVE26, ZFYVE27, ZNF592.