

## Spasztikus paraplegia (Spastic paraplegia) génpanel v:2025-05-23

Panel tartalma 163 gén:

ABCD1, ABHD16A, ACBD6, ACER3, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALK, ALS2, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BCAS3, BLOC1S1, BORCS8, BSCL2, BTBD, C19orf12, CACNA1A, CACNA1G, CAPN1, CCDC82, CHP1, CLDN11, COASY, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, DDX3X, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FICD, FXN, GAD1, GALC, GBA2, GBE1, GCH1, GJA1, GJC2, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HMBS, HPDL, HSPD1, IBA57, IFIH1, IRF2BPL, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KPNA3, L1CAM, L2HGDH, LETM1, LSM7, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NFU1, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA3, PAH, PCYT2, PI4KA, PLP1, PNPLA6, POLR3A, POLR3K, PPFIBP1, PPP2R2B, RAB3GAP2, RARS1, REEP1, REEP2, RETREG1, RHOB, RINT1, RNASEH2B, RNF170, RNU7-1, RTN2, SACS, SARS2, SELENOI, SERAC1, SETX, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SPART, SPAST, SPATA5L1, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTSSA, STN1, TAF8, TBP, TECPR2, TFG, TH, TMEM63C, TUBB4A, UBAP1, UCHL1, VAMP1, VPS37A, WASHC5, WDR45B, WDR48, ZFYVE26, ZFYVE27

Forrás:

Hereditary spastic paraplegia - childhood onset (Version 2.122)

<https://panelapp.genomicsengland.co.uk/panels/568/>

Spastic paraplegia - PS303350

<https://www.omim.org/phenotypicSeries/PS303350?sort=geneSymbols>

Spastic Paraplegia Panel

<https://blueprintgenetics.com/tests/panels/neurology/spastic-paraplegia-panel/>

(Megtekintve: 2025-05-23)