

Full name:	Hereditary spastic paraplegia (188 genes) - ULB
Laboratory:	<u>Centre de Génétique Humaine - Erasme ULB</u>
Created:	15 Jul 2019 - 10:48
Changed:	03 May 2023 - 14:28

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABCD1</u>	97.56	1	
<u>ABHD12</u>	88.30	1	
<u>ADAR</u>	100.00	1	
<u>AFG3L2</u>	94.80	1	
<u>AIMP1</u>	99.97	1	
<u>ALDH18A1</u>	100.00	1	
<u>ALDH3A2</u>	99.94	1	
<u>ALS2</u>	100.00	1	
<u>AMPD2</u>	99.90	1	
<u>ANG</u>	100.00	1	
<u>AP4B1</u>	100.00	1	
<u>AP4E1</u>	99.99	1	
<u>AP4M1</u>	100.00	1	
<u>AP4S1</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AP5Z1</u>	99.25	1	
<u>ARG1</u>	100.00	1	
<u>ARL6IP1</u>	100.00	1	
<u>ARSI</u>	99.82	1	
<u>ASPA</u>	100.00	1	
<u>ATAD3A</u>	97.50	1	
<u>ATL1</u>	99.89	1	
<u>ATP13A2</u>	98.30	1	
<u>AUH</u>	99.24	1	
<u>B4GALNT1</u>	99.99	1	
<u>BICD2</u>	100.00	1	
<u>BSCL2</u>	100.00	1	
<u>BTD</u>	100.00	1	
<u>C19ORF12</u>	100.00	1	
<u>CAPN1</u>	99.97	1	
<u>CCT5</u>	99.92	1	
<u>CDK16</u>	99.96	1	
<u>CLN8</u>	100.00	1	
<u>COASY</u>	100.00	1	
<u>CPT1C</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CSF1R</u>	99.99	1	
<u>CTNNB1</u>	100.00	1	
<u>CYP27A1</u>	99.38	1	
<u>CYP2U1</u>	93.20	1	
<u>CYP7B1</u>	100.00	1	
<u>DARS1</u>	100.00	1	
<u>DARS2</u>	100.00	1	
<u>DDHD1</u>	99.58	1	
<u>DDHD2</u>	100.00	1	
<u>DSTYK</u>	100.00	1	
<u>ELOVL4</u>	100.00	1	
<u>ENTPD1</u>	100.00	1	
<u>ERCC2</u>	98.91	1	
<u>ERLIN1</u>	100.00	1	
<u>ERLIN2</u>	100.00	1	
<u>EXOSC3</u>	100.00	1	
<u>FA2H</u>	95.71	1	
<u>FARS2</u>	100.00	1	
<u>FBXO7</u>	99.78	1	
<u>FIG4</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FLRT1</u>	100.00	1	
<u>FRMD7</u>	99.98	1	
<u>FUS</u>	100.00	1	
<u>FXN</u>	100.00	1	
<u>GAD1</u>	100.00	1	
<u>GALC</u>	98.98	1	
<u>GAN</u>	98.52	1	
<u>GARS1</u>	97.89	1	
<u>GART</u>	99.99	1	
<u>GBA1</u>	100.00	1	
<u>GBA2</u>	100.00	1	
<u>GBE1</u>	100.00	1	
<u>GCH1</u>	99.71	1	
<u>GFAP</u>	100.00	1	
<u>GJA1</u>	100.00	1	
<u>GJC2</u>	96.72	1	
<u>GLB1</u>	100.00	1	
<u>GLRX5</u>	100.00	1	
<u>GLTP</u>	100.00	1	
<u>GPR143</u>	89.09	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>GSN</u>	97.80	1	
<u>HACE1</u>	98.41	1	
<u>HEPACAM</u>	99.85	1	
<u>HEXA</u>	100.00	1	
<u>HSD17B4</u>	100.00	1	
<u>HSPD1</u>	100.00	1	
<u>HYCC1</u>	100.00	1	
<u>IBA57</u>	92.01	1	
<u>IFIH1</u>	100.00	1	
<u>IRF2BPL</u>	97.56	1	
<u>KCNJ6</u>	100.00	1	
<u>KCNMA1</u>	99.93	1	
<u>KDM5C</u>	99.97	1	
<u>KIDINS220</u>	100.00	1	
<u>KIF1A</u>	98.74	1	
<u>KIF1C</u>	100.00	1	
<u>KIF5A</u>	100.00	1	
<u>KLC2</u>	100.00	1	
<u>KLC4</u>	99.99	1	
<u>L1CAM</u>	99.98	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>L2HGDH</u>	99.75	1	
<u>LAMB1</u>	100.00	1	
<u>LYST</u>	100.00	1	
<u>MARS1</u>	100.00	1	
<u>MARS2</u>	100.00	1	
<u>MED12</u>	99.98	1	
<u>MFN2</u>	100.00	1	
<u>MLC1</u>	98.57	1	
<u>MTPAP</u>	99.98	1	
<u>MTRFR</u>	100.00	1	
<u>NANS</u>	100.00	1	
<u>NEFL</u>	100.00	1	
<u>NEXMIF</u>	100.00	1	
<u>NIPA1</u>	92.99	1	
<u>NKX6-2</u>	100.00	1	
<u>NT5C2</u>	100.00	1	
<u>OCLN</u>	100.00	1	
<u>OPA3</u>	100.00	1	
<u>OPTN</u>	100.00	1	
<u>PANK2</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PDYN</u>	100.00	1	
<u>PGAP1</u>	100.00	1	
<u>PLA2G6</u>	100.00	1	
<u>PLP1</u>	100.00	1	
<u>PNPLA6</u>	99.95	1	
<u>POLR3A</u>	100.00	1	
<u>POLR3B</u>	100.00	1	
<u>PPP2R2B</u>	100.00	1	
<u>PRUNE1</u>	100.00	1	
<u>PSEN1</u>	100.00	1	
<u>PUM1</u>	100.00	1	
<u>RAB18</u>	99.99	1	
<u>RAB3GAP1</u>	100.00	1	
<u>RAB3GAP2</u>	100.00	1	
<u>RARS1</u>	99.99	1	
<u>REEP1</u>	100.00	1	
<u>REEP2</u>	97.04	1	
<u>RETREG1</u>	100.00	1	
<u>RNASEH2A</u>	100.00	1	
<u>RNASEH2B</u>	94.58	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>RNASEH2C</u>	100.00	1	
<u>RNASET2</u>	100.00	1	
<u>RTN2</u>	99.44	1	
<u>SACS</u>	99.95	1	
<u>SAMD9L</u>	100.00	1	
<u>SAMHD1</u>	100.00	1	
<u>SERAC1</u>	100.00	1	
<u>SETX</u>	100.00	1	
<u>SIL1</u>	99.95	1	
<u>SLC16A2</u>	99.97	1	
<u>SLC25A15</u>	100.00	1	
<u>SLC25A46</u>	100.00	1	
<u>SLC2A1</u>	100.00	1	
<u>SLC33A1</u>	100.00	1	
<u>SLC39A14</u>	99.94	1	
<u>SMPD1</u>	100.00	1	
<u>SOD1</u>	100.00	1	
<u>SOX10</u>	100.00	1	
<u>SPART</u>	100.00	1	
<u>SPAST</u>	99.83	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SPG11</u>	100.00	1	
<u>SPG21</u>	100.00	1	
<u>SPG7</u>	95.78	1	
<u>SPR</u>	99.98	1	
<u>SUN1</u>	100.00	1	
<u>SYNE1</u>	100.00	1	
<u>TANGO2</u>	99.94	1	
<u>TARDBP</u>	99.43	1	
<u>TBC1D20</u>	95.06	1	
<u>TECPR2</u>	99.92	1	
<u>TFG</u>	99.98	1	
<u>TGM6</u>	100.00	1	
<u>TH</u>	98.79	1	
<u>TRPV4</u>	100.00	1	
<u>TTBK2</u>	100.00	1	
<u>TUBB4A</u>	100.00	1	
<u>TUBG1</u>	100.00	1	
<u>UBQLN2</u>	99.30	1	
<u>UCHL1</u>	99.66	1	
<u>USP8</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>VAMP1</u>	100.00	1	
<u>VAPB</u>	99.77	1	
<u>VCP</u>	97.10	1	
<u>VPS13B</u>	100.00	1	
<u>VPS37A</u>	99.28	1	
<u>WASHC5</u>	100.00	1	
<u>WDR45</u>	100.00	1	
<u>WDR45B</u>	99.94	1	
<u>WDR48</u>	100.00	1	
<u>ZC4H2</u>	100.00	1	
<u>ZFR</u>	100.00	1	
<u>ZFYVE26</u>	100.00	1	
<u>ZFYVE27</u>	100.00	1	