

Full name:	Hereditary Spastic Paraplegia (94 genes) - KUL
Abbreviation:	HSP
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	17 Sep 2020 - 14:23
Changed:	29 Aug 2022 - 11:53

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABCD1</u>	95.00	0	interpretable range CS1>95%
<u>ADAR</u>	95.00	0	interpretable range CS1>95%
<u>AFG3L2</u>	95.00	0	interpretable range CS1>95%
<u>AIMP1</u>	95.00	0	interpretable range CS1>95%
<u>ALDH18A1</u>	95.00	0	interpretable range CS1>95%
<u>ALDH3A2</u>	95.00	0	interpretable range CS1>95%
<u>ALS2</u>	95.00	0	interpretable range CS1>95%
<u>AMPD2</u>	95.00	0	interpretable range CS1>95%
<u>AP4B1</u>	95.00	0	interpretable range CS1>95%
<u>AP4E1</u>	95.00	0	interpretable range CS1>95%
<u>AP4M1</u>	95.00	0	interpretable range CS1>95%
<u>AP4S1</u>	95.00	0	interpretable range CS1>95%
<u>AP5Z1</u>	95.00	0	interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ARL6IP1</u>	95.00	0	interpretable range CS1>95%
<u>ARSI</u>	95.00	0	interpretable range CS1>95%
<u>ATL1</u>	95.00	0	interpretable range CS1>95%
<u>ATP13A2</u>	95.00	0	interpretable range CS1>95%
<u>B4GALNT1</u>	95.00	0	interpretable range CS1>95%
<u>BICD2</u>	95.00	0	interpretable range CS1>95%
<u>BRAF</u>	95.00	0	interpretable range CS1>95%
<u>BSCL2</u>	95.00	0	interpretable range CS1>95%
<u>C19ORF12</u>	95.00	0	interpretable range CS1>95%
<u>CAPN1</u>	95.00	0	interpretable range CS1>95%
<u>CBL</u>	95.00	0	interpretable range CS1>95%
<u>CCT5</u>	95.00	0	interpretable range CS1>95%
<u>CPT1C</u>	95.00	0	interpretable range CS1>95%
<u>CYP27A1</u>	95.00	0	interpretable range CS1>95%
<u>CYP2U1</u>	95.00	0	interpretable range CS1>95%
<u>CYP7B1</u>	95.00	0	interpretable range CS1>95%
<u>DARS1</u>	95.00	0	interpretable range CS1>95%
<u>DDHD1</u>	95.00	0	interpretable range CS1>95%
<u>DDHD2</u>	95.00	0	interpretable range CS1>95%
<u>DSTYK</u>	95.00	0	interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ENTPD1</u>	95.00	0	interpretable range CS1>95%
<u>ERCC2</u>	95.00	0	interpretable range CS1>95%
<u>ERLIN1</u>	95.00	0	interpretable range CS1>95%
<u>ERLIN2</u>	95.00	0	interpretable range CS1>95%
<u>FA2H</u>	95.00	0	interpretable range CS1>95%
<u>FARS2</u>	95.00	0	interpretable range CS1>95%
<u>FLRT1</u>	95.00	0	interpretable range CS1>95%
<u>GBA2</u>	95.00	0	interpretable range CS1>95%
<u>GCH1</u>	95.00	0	interpretable range CS1>95%
<u>GJC2</u>	95.00	0	interpretable range CS1>95%
<u>HACE1</u>	95.00	0	interpretable range CS1>95%
<u>HRAS</u>	95.00	0	interpretable range CS1>95%
<u>HSPD1</u>	95.00	0	interpretable range CS1>95%
<u>IBA57</u>	95.00	0	interpretable range CS1>95%
<u>KCNA2</u>	95.00	0	interpretable range CS1>95%
<u>KDM5C</u>	95.00	0	interpretable range CS1>95%
<u>KIDINS220</u>	95.00	0	interpretable range CS1>95%
<u>KIF1A</u>	95.00	0	interpretable range CS1>95%
<u>KIF1C</u>	95.00	0	interpretable range CS1>95%
<u>KIF5A</u>	95.00	0	interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>KLC2</u>	95.00	0	interpretable range CS1>95%
<u>KRAS</u>	95.00	0	interpretable range CS1>95%
<u>L1CAM</u>	95.00	0	interpretable range CS1>95%
<u>MAP2K1</u>	95.00	0	interpretable range CS1>95%
<u>MAP2K2</u>	95.00	0	interpretable range CS1>95%
<u>MARS1</u>	95.00	0	interpretable range CS1>95%
<u>MARS2</u>	95.00	0	interpretable range CS1>95%
<u>MTPAP</u>	95.00	0	interpretable range CS1>95%
<u>MTRFR</u>	95.00	0	interpretable range CS1>95%
<u>NIPA1</u>	95.00	0	interpretable range CS1>95%
<u>NKX6-2</u>	95.00	0	interpretable range CS1>95%
<u>NRAS</u>	95.00	0	interpretable range CS1>95%
<u>NT5C2</u>	95.00	0	interpretable range CS1>95%
<u>OPA3</u>	95.00	0	interpretable range CS1>95%
<u>PCYT2</u>	95.00	0	interpretable range CS1>95%
<u>PGAP1</u>	95.00	0	interpretable range CS1>95%
<u>PLP1</u>	95.00	0	interpretable range CS1>95%
<u>PNPLA6</u>	95.00	0	interpretable range CS1>95%
<u>POLR3A</u>	95.00	0	interpretable range CS1>95%
<u>PTPN11</u>	95.00	0	interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>RAB3GAP2</u>	95.00	0	interpretable range CS1>95%
<u>RAF1</u>	95.00	0	interpretable range CS1>95%
<u>REEP1</u>	95.00	0	interpretable range CS1>95%
<u>REEP2</u>	95.00	0	interpretable range CS1>95%
<u>RIT1</u>	95.00	0	interpretable range CS1>95%
<u>RNASEH2B</u>	95.00	0	interpretable range CS1>95%
<u>RTN2</u>	95.00	0	interpretable range CS1>95%
<u>SACS</u>	95.00	0	interpretable range CS1>95%
<u>SERAC1</u>	95.00	0	interpretable range CS1>95%
<u>SHOC2</u>	95.00	0	interpretable range CS1>95%
<u>SLC16A2</u>	95.00	0	interpretable range CS1>95%
<u>SLC1A4</u>	95.00	0	interpretable range CS1>95%
<u>SLC2A1</u>	95.00	0	interpretable range CS1>95%
<u>SLC33A1</u>	95.00	0	interpretable range CS1>95%
<u>SOS1</u>	95.00	0	interpretable range CS1>95%
<u>SPART</u>	95.00	0	interpretable range CS1>95%
<u>SPAST</u>	95.00	0	interpretable range CS1>95%
<u>SPG11</u>	95.00	0	interpretable range CS1>95%
<u>SPG21</u>	95.00	0	interpretable range CS1>95%
<u>SPG7</u>	95.00	0	interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TECPR2</u>	95.00	0	interpretable range CS1>95%
<u>TFG</u>	95.00	0	interpretable range CS1>95%
<u>TUBB4A</u>	95.00	0	interpretable range CS1>95%
<u>UBAP1</u>	95.00	0	interpretable range CS1>95%
<u>UCHL1</u>	95.00	0	interpretable range CS1>95%
<u>USP8</u>	95.00	0	interpretable range CS1>95%
<u>VAMP1</u>	95.00	0	interpretable range CS1>95%
<u>VPS37A</u>	95.00	0	interpretable range CS1>95%
<u>WASHC5</u>	95.00	0	interpretable range CS1>95%
<u>WDR48</u>	95.00	0	interpretable range CS1>95%
<u>ZFR</u>	95.00	0	interpretable range CS1>95%
<u>ZFYVE26</u>	95.00	0	interpretable range CS1>95%
<u>ZFYVE27</u>	95.00	0	interpretable range CS1>95%