

Full name:	Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
Version number:	WESAT_v3
Laboratory:	<u>Centrum Medische Genetica - UZ Antwerpen</u>
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Related Diseases

- Ataxia with vitamin E deficiency
- Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome
- Autosomal dominant spastic paraplegia type 10
- Autosomal dominant spastic paraplegia type 12
- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 3
- Autosomal dominant spastic paraplegia type 31
- Autosomal dominant spastic paraplegia type 4
- Autosomal dominant spastic paraplegia type 73
- Autosomal dominant spastic paraplegia type 8
- Autosomal dominant spastic paraplegia type 9A
- Autosomal dominant spastic paraplegia type 9B
- Autosomal recessive ataxia, Beauce type
- Autosomal recessive cerebellar ataxia due to STUB1 deficiency
- Autosomal recessive cerebellar ataxia with late-onset spasticity
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay
- Autosomal recessive spastic paraplegia type 11
- Autosomal recessive spastic paraplegia type 15
- Autosomal recessive spastic paraplegia type 21
- Autosomal recessive spastic paraplegia type 26
- Autosomal recessive spastic paraplegia type 28
- Autosomal recessive spastic paraplegia type 35
- Autosomal recessive spastic paraplegia type 39

- [Autosomal recessive spastic paraplegia type 46](#)
- [Autosomal recessive spastic paraplegia type 48](#)
- [Autosomal recessive spastic paraplegia type 5A](#)
- [Autosomal recessive spastic paraplegia type 62](#)
- [Autosomal recessive spastic paraplegia type 76](#)
- [Autosomal recessive spastic paraplegia type 77](#)
- [Autosomal recessive spastic paraplegia type 78](#)
- [Autosomal recessive spastic paraplegia type 9B](#)
- [Autosomal spastic paraplegia type 18](#)
- [Autosomal spastic paraplegia type 30](#)
- [Autosomal spastic paraplegia type 58](#)
- [Autosomal spastic paraplegia type 72](#)
- [Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome](#)
- [Severe intellectual disability and progressive spastic paraplegia](#)
- [Spastic paraplegia type 2](#)
- [Spastic paraplegia-severe developmental delay-epilepsy syndrome](#)
- [Spectrin-associated autosomal recessive cerebellar ataxia](#)
- [Spinocerebellar ataxia with axonal neuropathy type 2](#)
- [Wilson disease](#)
- [X-linked complicated spastic paraplegia type 1](#)

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
AAAS	99.98	1	
ABCB7	98.62	1	
ABCD1	99.78	1	
ABHD12	99.11	1	
ABHD16A	100.00	1	
AFG3L2	99.82	1	
AIFM1	99.43	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>AIMP1</u>	99.90	1	
<u>ALDH18A1</u>	99.96	1	
<u>ALS2</u>	99.91	1	
<u>AMPD2</u>	99.99	1	
<u>ANO10</u>	99.92	1	
<u>AP1S2</u>	97.43	1	
<u>AP4B1</u>	99.97	1	
<u>AP4E1</u>	99.89	1	
<u>AP4M1</u>	100.00	1	
<u>AP4S1</u>	87.70	1	
<u>AP5Z1</u>	99.94	1	
<u>APTX</u>	99.92	1	
<u>ARG1</u>	99.98	1	
<u>ARL6IP1</u>	99.84	1	
<u>ARSA</u>	100.00	1	
<u>ATCAY</u>	99.99	1	
<u>ATG7</u>	99.98	1	
<u>ATL1</u>	99.95	1	
<u>ATM</u>	99.83	1	
<u>ATP13A2</u>	99.98	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ATP1A3</u>	99.99	1	
<u>ATP2B3</u>	99.82	1	
<u>ATP5MC3</u>	100.00	1	
<u>ATP7B</u>	99.99	1	
<u>ATP8A2</u>	99.96	1	
<u>AUH</u>	99.95	1	
<u>B4GALNT1</u>	99.95	1	
<u>BSCL2</u>	100.00	1	
<u>BTD</u>	99.99	1	
<u>C19ORF12</u>	99.36	1	
<u>CA8</u>	99.97	1	
<u>CACNA1A</u>	99.66	1	
<u>CACNA1G</u>	99.89	1	
<u>CACNA2D2</u>	99.32	1	
<u>CAMTA1</u>	99.97	1	
<u>CAPN1</u>	100.00	1	
<u>CASK</u>	99.40	1	
<u>CCT5</u>	99.97	1	
<u>CHMP1A</u>	100.00	1	
<u>CLCN2</u>	99.97	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CLN6</u>	98.04	1	
<u>CLP1</u>	100.00	1	
<u>COA7</u>	100.00	1	
<u>COASY</u>	99.98	1	
<u>COG5</u>	99.92	1	
<u>COQ8A</u>	100.00	1	
<u>COX20</u>	99.40	1	
<u>CPT1C</u>	99.99	1	
<u>CTNNB1</u>	99.98	1	
<u>CWF19L1</u>	99.89	1	
<u>CYP27A1</u>	99.96	1	
<u>CYP2U1</u>	97.44	1	
<u>CYP7B1</u>	99.87	1	
<u>DARS1</u>	99.87	1	
<u>DARS2</u>	99.86	1	
<u>DDHD1</u>	99.98	1	
<u>DDHD2</u>	99.74	1	
<u>DNAJC5</u>	99.99	1	
<u>DNMT1</u>	99.88	1	
<u>EEF2</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>EIF2B1</u>	99.96	1	
<u>EIF2B2</u>	99.84	1	
<u>EIF2B3</u>	99.95	1	
<u>EIF2B4</u>	99.94	1	
<u>EIF2B5</u>	99.95	1	
<u>ELOVL4</u>	99.92	1	
<u>ELOVL5</u>	99.95	1	
<u>ENTPD1</u>	99.90	1	
<u>ERLIN1</u>	99.96	1	
<u>ERLIN2</u>	99.94	1	
<u>EXOSC3</u>	99.98	1	
<u>EXOSC8</u>	99.69	1	
<u>EXOSC9</u>	99.86	1	
<u>FA2H</u>	99.93	1	
<u>FAR1</u>	99.83	1	
<u>FARS2</u>	99.98	1	
<u>FAT2</u>	99.94	1	
<u>FGF14</u>	99.79	1	
<u>FLVCR1</u>	99.93	1	
<u>GALC</u>	99.88	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>GBA2</u>	99.97	1	
<u>GCH1</u>	99.74	1	
<u>GDAP2</u>	99.96	1	
<u>GFAP</u>	99.94	1	
<u>GJC2</u>	96.12	1	
<u>GLRX5</u>	98.29	1	
<u>GOSR2</u>	99.75	1	
<u>GPAA1</u>	99.88	1	
<u>GRID2</u>	99.98	1	
<u>GRM1</u>	99.87	1	
<u>HACE1</u>	99.91	1	
<u>HEXA</u>	99.98	1	
<u>HEXB</u>	99.91	1	
<u>HIKESHI</u>	99.89	1	
<u>HPDL</u>	100.00	1	
<u>HSPD1</u>	99.95	1	
<u>IBA57</u>	96.45	1	
<u>IFRD1</u>	99.96	1	
<u>INPP5E</u>	99.75	1	
<u>IRF2BPL</u>	98.65	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ITPR1</u>	99.94	1	
<u>KCNA1</u>	99.96	1	
<u>KCNA2</u>	100.00	1	
<u>KCNC3</u>	94.29	1	
<u>KCND3</u>	100.00	1	
<u>KIDINS220</u>	99.97	1	
<u>KIF1A</u>	99.96	1	
<u>KIF1C</u>	99.99	1	
<u>KIF5A</u>	99.93	1	
<u>KPNA3</u>	99.96	1	
<u>L1CAM</u>	99.90	1	
<u>MAG</u>	99.81	1	
<u>MARS2</u>	100.00	1	
<u>MMACHC</u>	99.99	1	
<u>MRE11</u>	99.90	1	
<u>MSTO1</u>	98.87	1	
<u>MTRFR</u>	99.85	1	
<u>MTTP</u>	99.96	1	
<u>NIPA1</u>	96.26	1	
<u>NKX6-2</u>	96.38	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>NPC1</u>	99.84	1	
<u>NPC2</u>	99.99	1	
<u>NPTX1</u>	99.35	1	
<u>NT5C2</u>	99.89	1	
<u>OPA1</u>	99.87	1	
<u>OPA3</u>	100.00	1	
<u>PCDH12</u>	100.00	1	
<u>PCYT2</u>	97.60	1	
<u>PDYN</u>	99.98	1	
<u>PEX7</u>	99.87	1	
<u>PGAP1</u>	99.64	1	
<u>PHYH</u>	99.65	1	
<u>PLA2G6</u>	99.98	1	
<u>PLP1</u>	99.93	1	
<u>PMPCA</u>	99.99	1	
<u>PMPCB</u>	99.98	1	
<u>PNKP</u>	100.00	1	
<u>PNPLA6</u>	99.88	1	
<u>POLG</u>	99.98	1	
<u>POLR3A</u>	99.95	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PRICKLE1</u>	99.97	1	
<u>PRKCG</u>	99.90	1	
<u>PUM1</u>	99.95	1	
<u>RARS2</u>	97.34	1	
<u>REEP1</u>	99.99	1	
<u>REEP2</u>	100.00	1	
<u>RFC1</u>	99.44	1	
<u>RNF170</u>	99.94	1	
<u>RNF216</u>	99.99	1	
<u>RNF220</u>	99.95	1	
<u>RTN2</u>	99.99	1	
<u>RUBCN</u>	99.94	1	
<u>SACS</u>	99.98	1	
<u>SAMD9L</u>	99.98	1	
<u>SCN8A</u>	99.90	1	
<u>SCYL1</u>	99.64	1	
<u>SELENOI</u>	99.88	1	
<u>SEPSECS</u>	99.84	1	
<u>SETX</u>	99.96	1	
<u>SIL1</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SLC1A3</u>	99.90	1	
<u>SLC1A4</u>	99.95	1	
<u>SLC25A46</u>	99.93	1	
<u>SLC33A1</u>	99.82	1	
<u>SLC9A1</u>	99.86	1	
<u>SNX14</u>	99.73	1	
<u>SPART</u>	99.99	1	
<u>SPAST</u>	99.83	1	
<u>SPG11</u>	99.94	1	
<u>SPG21</u>	99.99	1	
<u>SPG7</u>	99.99	1	
<u>SPTAN1</u>	99.97	1	
<u>SPTBN2</u>	99.99	1	
<u>SQSTM1</u>	100.00	1	
<u>STUB1</u>	99.73	1	
<u>SYNE1</u>	99.96	1	
<u>TDP2</u>	99.85	1	
<u>TFG</u>	99.34	1	
<u>TGM6</u>	100.00	1	
<u>THG1L</u>	99.95	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TMEM240</u>	99.83	1	
<u>TMEM63C</u>	99.99	1	
<u>TOE1</u>	99.99	1	
<u>TPP1</u>	99.88	1	
<u>TSEN2</u>	86.92	1	
<u>TSEN54</u>	98.24	1	
<u>TTBK2</u>	99.88	1	
<u>TTPA</u>	99.97	1	
<u>TUBB4A</u>	98.81	1	
<u>UBAP1</u>	99.96	1	
<u>UCHL1</u>	99.99	1	
<u>VAMP1</u>	99.70	1	
<u>VLDLR</u>	99.93	1	
<u>VPS13D</u>	99.85	1	
<u>VPS41</u>	99.95	1	
<u>VPS53</u>	99.98	1	
<u>VRK1</u>	99.94	1	
<u>WASHC5</u>	99.90	1	
<u>WDR45B</u>	99.96	1	
<u>WDR73</u>	99.99	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>WDR81</u>	100.00	1	
<u>ZFYVE26</u>	99.96	1	