

Full name:	Neuropathy (genepanel) - UZA
Description:	https://labogidsmedgen.uza.be/analyses/perifere-neuropathie-pn
Version number:	PN_versie 3
Laboratory:	Centrum Medische Genetica - UZ Antwerpen
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Related Diseases

- [Autosomal dominant Charcot-Marie-Tooth disease type 2D](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2DD](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2F](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2K](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2L](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2N](#)
- [Autosomal dominant Charcot-Marie-Tooth disease type 2W](#)
- [Autosomal dominant intermediate Charcot-Marie-Tooth disease type F](#)
- [Autosomal recessive Charcot-Marie-Tooth disease with hoarseness](#)
- [Autosomal recessive axonal neuropathy with neuromyotonia](#)
- [Autosomal recessive intermediate Charcot-Marie-Tooth disease type A](#)
- [Autosomal recessive intermediate Charcot-Marie-Tooth disease type D](#)
- [Charcot-Marie-Tooth disease type 1D](#)
- [Charcot-Marie-Tooth disease type 2H](#)
- [Charcot-Marie-Tooth disease type 4A](#)
- [Charcot-Marie-Tooth disease type 4E](#)
- [Charcot-Marie-Tooth disease type 4G](#)
- [Charcot-Marie-Tooth disease type 4H](#)
- [Charcot-Marie-Tooth disease type 4J](#)
- [Dejerine-Sottas syndrome](#)
- [Distal hereditary motor neuropathy type 2](#)

- [Distal hereditary motor neuropathy type 5](#)
- [Muscular dystrophy, Selcen type](#)
- [Neuropathy with hearing impairment](#)
- [Severe neurodegenerative syndrome with lipodystrophy](#)
- [X-linked Charcot-Marie-Tooth disease type 1](#)
- [X-linked Charcot-Marie-Tooth disease type 4](#)

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
AARS1	99.94	1	
ABCA1	99.92	1	
ABHD12	99.11	1	
ADPRS	100.00	1	
AGTPBP1	99.40	1	
AIFM1	99.43	1	
ARHGEF10	100.00	1	
ARSA	100.00	1	
ATL1	99.95	1	
ATL3	99.86	1	
ATM	99.83	1	
ATP1A1	99.94	1	
ATP7A	99.35	1	
B4GALNT1	99.95	1	
BAG3	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>BICD2</u>	100.00	1	
<u>BSCL2</u>	100.00	1	
<u>CADM3</u>	99.94	1	
<u>CD59</u>	99.97	1	
<u>CFAP276</u>	99.73	1	
<u>CHCHD10</u>	97.06	1	
<u>CLTCL1</u>	99.98	1	
<u>CNTNAP1</u>	99.80	1	
<u>COA7</u>	100.00	1	
<u>COX20</u>	99.40	1	
<u>COX6A1</u>	99.99	1	
<u>CTDP1</u>	98.02	1	
<u>CTNNB1</u>	99.98	1	
<u>CYP27A1</u>	99.96	1	
<u>DARS2</u>	99.86	1	
<u>DCAF8</u>	99.98	1	
<u>DCTN1</u>	99.95	1	
<u>DGAT2</u>	99.99	1	
<u>DGUOK</u>	99.76	1	
<u>DHTKD1</u>	99.88	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DNAJB2</u>	100.00	1	
<u>DNAJC3</u>	99.58	1	
<u>DNM2</u>	99.96	1	
<u>DNMT1</u>	99.88	1	
<u>DRP2</u>	99.45	1	
<u>DST</u>	99.92	1	
<u>DYNC1H1</u>	99.97	1	
<u>EGR2</u>	100.00	1	
<u>ELP1</u>	99.96	1	
<u>ETFDH</u>	99.76	1	
<u>FBLN5</u>	92.79	1	
<u>FBXO38</u>	99.93	1	
<u>FGD4</u>	99.89	1	
<u>FIG4</u>	99.94	1	
<u>FLVCR1</u>	99.93	1	
<u>FXN</u>	97.92	1	
<u>GALC</u>	99.88	1	
<u>GAN</u>	99.84	1	
<u>GARS1</u>	99.86	1	
<u>GBA2</u>	99.97	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>GBE1</u>	99.84	1	
<u>GBF1</u>	99.95	1	
<u>GDAP1</u>	99.93	1	
<u>GJB1</u>	99.99	1	
<u>GJB3</u>	100.00	1	
<u>GJC2</u>	96.12	1	
<u>GLA</u>	99.64	1	
<u>GNB4</u>	99.95	1	
<u>GSN</u>	97.83	1	
<u>HADHA</u>	99.96	1	
<u>HADHB</u>	99.97	1	
<u>HARS1</u>	99.99	1	
<u>HINT1</u>	99.96	1	
<u>HK1</u>	96.86	1	
<u>HNRNPA1</u>	100.00	1	
<u>HOXD10</u>	100.00	1	
<u>HSPB1</u>	99.83	1	
<u>HSPB3</u>	100.00	1	
<u>HSPB8</u>	99.90	1	
<u>IARS2</u>	99.96	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IFRD1</u>	99.96	1	
<u>IGHMBP2</u>	99.99	1	
<u>INF2</u>	97.15	1	
<u>ITPR3</u>	99.88	1	
<u>JAG1</u>	99.92	1	
<u>KIF1A</u>	99.96	1	
<u>KIF5A</u>	99.93	1	
<u>LITAF</u>	100.00	1	
<u>LMNA</u>	99.98	1	
<u>LRSAM1</u>	99.97	1	
<u>LYST</u>	99.92	1	
<u>MCM3AP</u>	99.96	1	
<u>MFN2</u>	99.96	1	
<u>MMACHC</u>	99.99	1	
<u>MME</u>	97.69	1	
<u>MORC2</u>	99.98	1	
<u>MPV17</u>	100.00	1	
<u>MPZ</u>	99.97	1	
<u>MTMR2</u>	99.98	1	
<u>MTRFR</u>	99.85	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MYH14</u>	99.79	1	
<u>NAGA</u>	99.99	1	
<u>NAGLU</u>	98.00	1	
<u>NARS1</u>	99.74	1	
<u>NDRG1</u>	100.00	1	
<u>NEFH</u>	98.77	1	
<u>NEFL</u>	99.99	1	
<u>NGF</u>	99.99	1	
<u>NTRK1</u>	99.67	1	
<u>PCYT2</u>	97.60	1	
<u>PDK3</u>	98.86	1	
<u>PEX7</u>	99.87	1	
<u>PHYH</u>	99.65	1	
<u>PIEZO2</u>	99.88	1	
<u>PLD3</u>	99.99	1	
<u>PLEKHG5</u>	99.66	1	
<u>PLP1</u>	99.93	1	
<u>PMM2</u>	99.94	1	
<u>PMP2</u>	99.95	1	
<u>PMP22</u>	100.00	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PNKP</u>	100.00	1	
<u>PNPLA6</u>	99.88	1	
<u>POLG</u>	99.98	1	
<u>POLR3B</u>	99.88	1	
<u>PPOX</u>	99.94	1	
<u>PRDM12</u>	99.60	1	
<u>PRKCG</u>	99.90	1	
<u>PRNP</u>	100.00	1	
<u>PRPS1</u>	99.79	1	
<u>PRX</u>	99.94	1	
<u>PTEN</u>	99.15	1	
<u>PTPN11</u>	99.91	1	
<u>PTRH2</u>	99.97	1	
<u>RAB7A</u>	99.94	1	
<u>REEP1</u>	99.99	1	
<u>RETREG1</u>	99.92	1	
<u>RFC1</u>	99.44	1	
<u>SACS</u>	99.98	1	
<u>SBF1</u>	99.97	1	
<u>SBF2</u>	99.92	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SCARB2</u>	99.94	1	
<u>SCN10A</u>	99.97	1	
<u>SCN11A</u>	99.77	1	
<u>SCN9A</u>	99.96	1	
<u>SCO2</u>	100.00	1	
<u>SCYL1</u>	99.64	1	
<u>SEPTIN9</u>	99.94	1	
<u>SETX</u>	99.96	1	
<u>SH3TC2</u>	99.98	1	
<u>SIGMAR1</u>	99.99	1	
<u>SLC12A6</u>	99.88	1	
<u>SLC25A19</u>	99.99	1	
<u>SLC25A46</u>	99.93	1	
<u>SLC52A2</u>	100.00	1	
<u>SLC52A3</u>	100.00	1	
<u>SLC5A7</u>	99.97	1	
<u>SORD</u>	92.86	1	
<u>SOX10</u>	100.00	1	
<u>SPAST</u>	99.83	1	
<u>SPG11</u>	99.94	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SPG7</u>	99.99	1	
<u>SPTAN1</u>	99.97	1	
<u>SPTBN4</u>	99.19	1	
<u>SPTLC1</u>	99.98	1	
<u>SPTLC2</u>	99.94	1	
<u>SPTLC3</u>	99.94	1	
<u>SURF1</u>	97.64	1	
<u>SYT2</u>	99.89	1	
<u>TBCE</u>	99.69	1	
<u>TBCK</u>	99.76	1	
<u>TECPR2</u>	99.90	1	
<u>TFG</u>	99.34	1	
<u>TRIM2</u>	99.99	1	
<u>TRPV4</u>	99.96	1	
<u>TTPA</u>	99.97	1	
<u>TTR</u>	100.00	1	
<u>TUBB3</u>	100.00	1	
<u>TWINK</u>	100.00	1	
<u>UCHL1</u>	99.99	1	
<u>VAPB</u>	99.94	1	

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>VCP</u>	99.92	1	
<u>VPS13A</u>	99.60	1	
<u>VRK1</u>	99.94	1	
<u>VWA1</u>	91.35	1	
<u>WARS1</u>	99.79	1	
<u>WNK1</u>	99.94	1	
<u>XRCC1</u>	99.94	1	
<u>YARS1</u>	99.96	1	
<u>ZFHX2</u>	99.98	1	
<u>ZFYVE26</u>	99.96	1	