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Full name:	Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
Abbreviation:	IPN
Version number:	v5-2020
Laboratory:	Centrum Menselijke Erfelijkheid - KUL
Created:	28 Jun 2019 - 09:53
Changed:	26 Jul 2023 - 10:05

Related Diseases

- ATTRV122I amyloidosis
- ATTRV30M amyloidosis
- Amyotrophic lateral sclerosis
- Autosomal dominant Charcot-Marie-Tooth disease type 2 due to DGAT2 mutation
- Autosomal dominant Charcot-Marie-Tooth disease type 2DD
- Autosomal dominant Charcot-Marie-Tooth disease type 2E
- Autosomal dominant Charcot-Marie-Tooth disease type 2F
- Autosomal dominant Charcot-Marie-Tooth disease type 2I
- Autosomal dominant Charcot-Marie-Tooth disease type 2J
- Autosomal dominant Charcot-Marie-Tooth disease type 2K
- Autosomal dominant Charcot-Marie-Tooth disease type 2M
- Autosomal dominant Charcot-Marie-Tooth disease type 2N
- Autosomal dominant Charcot-Marie-Tooth disease type 2O
- Autosomal dominant Charcot-Marie-Tooth disease type 2V
- Autosomal dominant Charcot-Marie-Tooth disease type 2W
- Autosomal dominant Charcot-Marie-Tooth disease type 2Y
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type B
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type C
- Autosomal dominant intermediate Charcot-Marie-Tooth disease type D
- Autosomal dominant progressive external ophthalmoplegia
- Autosomal dominant slowed nerve conduction velocity

- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 3
- 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 B
- Autosomal recessive progressive external ophthalmoplegia
- Autosomal recessive spastic paraplegia type 55
- Autosomal spastic paraplegia type 30
- CADDS
- Charcot-Marie-Tooth disease type 1A
- Charcot-Marie-Tooth disease type 1B
- Charcot-Marie-Tooth disease type 1D
- Charcot-Marie-Tooth disease type 1E
- Charcot-Marie-Tooth disease type 1F
- Charcot-Marie-Tooth disease type 2B2
- Charcot-Marie-Tooth disease type 2B5
- Charcot-Marie-Tooth disease type 2H
- Charcot-Marie-Tooth disease type 4A
- Charcot-Marie-Tooth disease type 4B3
- Charcot-Marie-Tooth disease type 4E
- Charcot-Marie-Tooth disease type 4H
- DYNC1H1-related autosomal dominant childhood-onset proximal spinal muscular atrophy
- Dejerine-Sottas syndrome
- Distal hereditary motor neuropathy type 2
- Distal hereditary motor neuropathy type 5
- Fabry disease
- Giant axonal neuropathy
- Hereditary neuropathy with liability to pressure palsies
- Hereditary sensorimotor neuropathy with hyperelastic skin
- Hereditary sensory and autonomic neuropathy type 1
- Hereditary sensory and autonomic neuropathy type 2
- Hereditary sensory and autonomic neuropathy type 6
- Hereditary sensory and autonomic neuropathy type 8
- Hypomyelination neuropathy-arthrogryposis syndrome
- Muscular dystrophy, Selcen type

- Mutilating hereditary sensory neuropathy with spastic paraplegia
- Neuralgic amyotrophy
- Neuropathy with hearing impairment
- Palmoplantar keratoderma-hereditary motor and sensory neuropathy syndrome
- Perrault syndrome
- Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome
- Pontocerebellar hypoplasia type 1
- SURF1-related Charcot-Marie-Tooth disease type 4
- X-linked Charcot-Marie-Tooth disease type 1
- X-linked Charcot-Marie-Tooth disease type 6

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
AARS1	100.00	0	NM_001605.2
ABCD1	100.00	0	NM_000033.3
ABHD12	98.00	0	NM_001042472.2
AGTPBP1	99.90	0	NM_001286715.1
AIFM1	100.00	0	NM_004208.3
APTX	99.00	0	NM_175073.2
ARHGEF10	100.00	0	NM_014629.3
ATL1	100.00	0	NM_015915.4
ATL3	100.00	0	NM_015459.4
ATP1A1	100.00	0	NM_000701.7
ATP7A	100.00	0	NM_000052.6
BAG3	100.00	0	NM_004281.3
BICD2	100.00	0	NM_001003800.1

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
BSCL2	100.00	0	NM_032667.6
CCT5	100.00	0	NM_012073.4
CD59	100.00	0	NM_203330.2
CNTNAP1	100.00	0	NM_003632.2
COA7	100.00	0	NM_023077.2
COX6A1	100.00	0	NM_004373.3
CTDP1	98.90	0	NM_004715.4
DCAF8	100.00	0	NM_015726.3
DCTN1	100.00	0	NM_004082.4
DGAT2	100.00	0	NM_032564.4
DHTKD1	99.90	0	NM_018706.6
DNAJB2	100.00	0	NM_001039550.1
DNM2	100.00	0	NM_001005360.2
DNMT1	99.50	0	NM_001130823.2
DRP2	100.00	0	NM_001939.2
DST	100.00	0	NM_001723.5
DYNC1H1	100.00	0	NM_001376.4
EGR2	100.00	0	NM_000399.4
ELP1	100.00	0	NM_003640.4
FBLN5	100.00	0	NM_006329.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
FBXO38	100.00	0	NM_030793.4
FGD4	100.00	0	NM_139241.3
FIG4	100.00	0	NM_014845.5
FLVCR1	100.00	0	NM_014053.3
GAN	100.00	0	NM_022041.3
GARS1	100.00	0	NM_002047.3
GDAP1	100.00	0	NM_018972.3
GJB1	100.00	0	NM_000166.5
GJB3	100.00	0	NM_024009.2
GLA	100.00	0	NM_000169.2
GNB4	100.00	0	NM_021629.3
HADHA	100.00	0	NM_000182.4
HADHB	100.00	0	NM_000183.2
HARS1	100.00	0	NM_002109.5
HINT1	100.00	0	NM_005340.6
HK1	100.00	0	NM_000188.2
HOXD10	100.00	0	NM_002148.3
HSPB1	99.00	0	NM_001540.4
HSPB3	100.00	0	NM_006308.2
HSPB8	100.00	0	NM_014365.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
IARS2	100.00	0	NM_018060.3
IFRD1	99.90	0	NM_001550.3
IGHMBP2	100.00	0	NM_002180.2
INF2	98.80	0	NM_022489.3
ITPR3	99.50	0	NM_002224.3
KARS1	100.00	0	NM_001130089.1
KIF1A	99.90	0	NM_004321.7
KIF1B	100.00	0	NM_015074.3
KIF5A	100.00	0	NM_004984.3
KLHL13	100.00	0	NM_033495.3
LITAF	100.00	0	NM_004862.3
LMNA	99.90	0	NM_170707.3
LRSAM1	100.00	0	NM_138361.5
MARS1	100.00	0	NM_004990.3
MCM3AP	100.00	0	NM_003906.4
MED12	100.00	0	NM_015560.2
MED25	100.00	0	NM_030973.3
MFN2	100.00	0	NM_014874.3
MME	100.00	0	NM_007289.3
MORC2	100.00	0	NM_001303256.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
MPV17	100.00	0	NM_002437.4
MPZ	100.00	0	NM_000530.7
MTMR2	100.00	0	NM_016156.5
MTRFR	100.00	0	NM_152269.4
MYH7B	99.80	0	NM_024729.3
NAGLU	99.70	0	NM_000263.3
NDRG1	100.00	0	NM_006096.3
NEFH	99.50	0	NM_021076.3
NEFL	99.60	0	NM_006158.4
NGF	100.00	0	NM_002506.2
NTRK1	100.00	0	NM_001012331.1
OPA3	100.00	0	NM_025136.3
PDK3	100.00	0	NM_001142386.2
PDXK	100.00	0	NM_003681.4
PEX1	100.00	0	NM_000466.2
PEX7	100.00	0	NM_000288.3
PHYH	100.00	0	NM_006214.3
PLEKHG5	99.80	0	NM_020631.4
PMP2	100.00	0	NM_002677.4
PMP22	100.00	0	NM_000304.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
PNKP	99.80	0	NM_007254.3
POLG	100.00	0	NM_002693.2
PRDM12	90.30	0	NM_021619.2
PRNP	100.00	0	NM_000311.4
PRPS1	100.00	0	NM_002764.3
PRX	100.00	0	NM_181882.2
RAB7A	100.00	0	NM_004637.5
REEP1	100.00	0	NM_022912.2
RETREG1	99.40	0	NM_001034850.2
SBF1	99.70	0	NM_002972.3
SBF2	100.00	0	NM_030962.3
SCN10A	100.00	0	NM_006514.3
SCN11A	99.80	0	NM_014139.2
SCN9A	100.00	0	NM_002977.3
SCO2	100.00	0	NM_005138.2
SCP2	100.00	0	NM_002979.4
SEPTIN9	99.20	0	NM_006640.4
SETX	100.00	0	NM_015046.6
SGPL1	100.00	0	NM_003901.3
SH3BP4	100.00	0	NM_014521.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
SH3TC2	100.00	0	NM_024577.3
SIGMAR1	100.00	0	NM_005866.3
SLC12A6	100.00	0	NM_133647.1
SLC25A46	100.00	0	NM_138773.3
SLC5A7	100.00	0	NM_021815.4
SORD	91.20	0	NM_003104.5
SOX10	100.00	0	NM_006941.3
SPG11	100.00	0	NM_025137.3
SPTLC1	100.00	0	NM_006415.3
SPTLC2	100.00	0	NM_004863.3
SURF1	99.00	0	NM_003172.3
TDP1	100.00	0	NM_018319.3
TECPR2	100.00	0	NM_014844.4
TFG	100.00	0	NM_006070.5
TRIM2	100.00	0	NM_001130067.1
TRPA1	100.00	0	NM_007332.2
TRPV4	99.90	0	NM_021625.4
TTR	100.00	0	NM_000371.3
TUBB3	100.00	0	NM_006086.3
TWNK	100.00	0	NM_021830.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
TYMP	100.00	0	NM_001953.4
VCP	99.90	0	NM_007126.4
VRK1	100.00	0	NM_003384.2
WARS1	99.80	0	NM_004184.3
WNK1	99.80	0	NM_018979.3
YARS1	100.00	0	NM_003680.3