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NeuropathySelect Genes and Associated Diseases | April 2019

NeuropathySelect is a 81 gene panel designed for patients experiencing symptoms associated with polyneuropathy but lacking a clear diagnosis. Given the overlap in genetic causes and variability in clinical symptoms and presentation, NeuropathySelect represents an effective way of identifying at-risk individuals, or confirming a diagnosis.

Gene Name	Associated Disease
AARS	CMT
AIFM1	Cowchock syndrome
APOA1	Amyloidotic neuropathy
ATL1	Sensory neuropathy/SPG
ATL3	Sensory neuropathy
ATP7A	ATP7A-related distal motor neuropathy/SMA
BICD2	SMA
BSCL2	HMN/SMA/SPG
CHCHD10	ALS
DCTN1	HMN
DNAJB2	SMA
DNM2	CMT
DNMT1	Sensory neuropathy
DST	Sensory and autonomic neuropathy
DYNC1H1	CMT/SMA
EGR2	CMT/DSS
FAM134B (RETREG1)	Sensory and autonomic neuropathy
FBXO38	HMN
FGD4	CMT
FIG4	CMT
FUS	ALS
GAN	Giant axonal neuropathy
GARS	CMT/HMN
GDAP1	CMT
GJB1	CMT
GNB4	CMT
GSN	Amyloidosis, Finnish type
HARS	CMT
HINT1	Neuromyotonia and axonal neuropathy
HSPB1	CMT/HMN
HSPB8	CMT/HMN
IGHMBP2	CMT/HMN
IKBKAP (ELP1)	Familial dysautonomia
INF2	CMT
KIF1A	Sensory neuropathy/SPG
LITAF	CMT
LMNA	CMT
LRSAM1	CMT
MARS	CMT
MFN2	CMT/HMN
MORC2	CMT

Gene Name	Associated Disease
MPZ	CMT/DSS
MTMR2	CMT
NDRG1	CMT
NEFH	CMT
NEFL	CMT
NGF	Sensory and autonomic neuropathy
NTRK1	CIP/Sensory and autonomic neuropathy
OPTN	ALS
PDK3	CMT
PLEKHG5	CMT
PMP22	CMT/DSS/HNPP
PRDM12	Sensory and autonomic neuropathy
PRPS1	CMT
PRX	CMT/DSS
RAB7A	CMT
REEP1	CMT/HMN/SPG
SBF2	CMT
SCN10A	Episodic pain syndrome
SCN11A	CIP/Episodic pain syndrome/Sensory and autonomic neuropathy
SCN9A	CIP/Erythromelalgia/Sensory and autonomic neuropathy
SETX	ALS/Spinocerebellar ataxia
SH3TC2	CMT
SIGMAR1	ALS/SMA
SLC25A46	Motor and sensory neuropathy
SLC52A2	BVVL/RTD
SLC52A3	BVVL/RTD
SLC5A7	Congenital myasthenic syndrome/HMN
SPG11	ALS/CMT/SPG
SPTLC1	Sensory and autonomic neuropathy
SPTLC2	Sensory and autonomic neuropathy
TARDBP	ALS
TFG	Sensory and motor neuropathy/SPG
TRPV4	CMT/Motor and sensory neuropathy/SMA
TTR	hATTR amyloidosis
UBA1	SMA
VAPB	ALS
VCP	ALS/CMT
VRK1	Motor and sensory neuropathy
WNK1 (HSN2 isoform)	Sensory and autonomic neuropathy
YARS	CMT

References available upon request.
ALS = Amyotrophic lateral sclerosis, BVVL = Brown Vialetto Van Laere syndrome, CIP = Congenital insensitivity to pain, CMT = Charcot-Marie-Tooth, DSS = Dejerine-Scottas syndrome, HMN = Hereditary Motor Neuropathy, HNRP = Hereditary neuropathy with liability to pressure palsies, RTD = Riboflavin transporter deficiency, SMA = Spinal muscular atrophy, SPG = Spastic paraplegia

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