Title: Charcot-Marie-Tooth Neuropathy Type 2 *GeneReview* Molecular Genetics: Less Commonly Involved Genes

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KIF1B

Gene structure. *KIF1B* comprises 47 exons and 167.13 kb of DNA.

Pathogenic allelic variants. See Table A, **Locus Specific** and **HGMD**

Normal gene product. Kinesin-like protein KIF1B is involved in axonal transport of synaptic vesicle precursors [Zhao et al 2001]. The kinesin superfamily of proteins is essential for intracellular transport along microtubules.

Abnormal gene product. There may be a defect in the transport of synaptic vesicles.

RAB7A

Gene structure. *RAB7A* has six exons and 87.9 kb of DNA.

Pathogenic allelic variants. See Table A.

Normal gene product. Ras-related protein Rab-7a belongs to the RAB family of Ras-related GTPases essential for the regulation of intracellular membrane trafficking. Rab-7a is involved in transport between late endosomes and lysosomes. RAB-interacting lysosomal protein (RILP) induces the recruitment of dynein-dynactin motors and regulates transport toward the minus-end of microtubules [Verhoeven et al 2003].

Abnormal gene product. Abnormal Rab-7a may cause malfunction of lysosomes and inhibit neurite outgrowth [Spinosa et al 2008, Bucci & Deluca 2012].

LMNA

Gene structure. *LMNA* has 12 exons spread over 24 kb of genomic DNA.

Pathogenic allelic variants. The most common pathogenic variant found in individuals with CMT2B1 is p.Arg298Cys, a founder mutation in North Africa [Bouhouche et al 2007, De Sandre-Giovannoli et al 2002].

See also Table A.

Table 5. Selected *LMNA* Variants

Class of Variant Allele	DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
Benign	c.1908C>T	p.= [1]	NM_170707.2 NP_733821.1
Pathogenic	c.398G>T	p.Arg133Leu	
	c.892C>T	p.Arg298Cys	
	c.1411C>T	p.Arg471Cys	
	c.1579C>T	p.Arg527Cys	

Note on variant classification: Variants listed in the table have been provided by the author. *GeneReviews* staff have not independently verified the classification of variants.

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1. p.= signifies that protein has been analyzed but no amino acid change is expected.

Normal gene product. Lamins are the principal component of the nuclear lamina, a major portion of the nuclear envelope. Two A-type lamins exist: A and C. Lamins play a role in DNA replication, chromatin organization, spatial arrangement of nuclear pore complexes, nuclear growth, mechanical stabilization of the nucleus, and anchorage of the nuclear envelope protein.

Abnormal gene product. Position 29 is located in the lamin-A/C rod domain. The manner in which disruption of this domain adversely affects peripheral nerve function is unknown. Other *LMNA* pathogenic variants are associated with a wide variety of disorders (see Genetically Related Disorders).

MED25

Gene structure. *MED25* has 18 exons.

Pathogenic allelic variants. In the one family reported, affected individuals were homozygous for NM_030973.2:c.1004C>T (p.Ala335Val) [Leal et al 2009].

Normal gene product. *MED25* encodes a 747-amino acid protein designated the mediator complex subunit 25 protein. This protein is a subunit of the human activator-recruited cofactor (ARC), a family of large transcriptional coactivator complexes. Its precise function in transcriptional regulation is unknown.

Abnormal gene product. The p.Ala335Val substitution is located in a proline-rich region with high affinity for SH3 domains of the Abelson type. The pathogenic variant causes a decrease in binding specificity leading to the recognition of a broader range of SH3 domain proteins.

TRPV4

Gene structure. *TRPV4* has 16 exons; exon 1 of NM_021625.3 is non-coding.

Pathogenic allelic variants. The pathogenic variants in Table 6 have been associated with CMT2C.

Table 6. Selected *TRPV4* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.805C>T	p.Arg269Cys	NM_021625.3 NP_067638.3
c.806G>A	p.Arg269His	
c.943C>T	p.Arg315Trp	
c.946C>T	p.Arg316Cys	

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Normal gene product. TRPV is a vanilloid receptor-related transient receptor potential channel which plays an important role in neural signal. The protein is composed of a cytosolic N-terminal region and six transmembrane domains, including the pore region and an intracellular C-terminal tail. The N-terminal region contains the ankyrin repeat domain (ARD).

Abnormal gene product. Landouré et al [2010] demonstrated cellular toxicity and increased constitutive and activated channel currents in TRPV4-transected cells. Deng et al [2010] showed increased calcium channel activity resulting from the two pathogenic variants found in two families with CMT2C. The effect of pathogenic variants on molecular functions like oligomerization, surface expression and ubiquitination are reviewed by Verma et al [2010].

GARS

Gene structure. *GARS* is a 40-kb gene with 17 exons.

Pathogenic allelic variants. See Table A.

Normal gene product. Glycyl-tRNA synthetase ligates amino acids to their cognate tRNA.

Abnormal gene product. The missense variants in this gene may produce a loss of function that allows the incorporation of the wrong amino acid in the place of glycine [Motley et al 2010]. These variants alter the binding of GlyRS (CMT2D) that antagonizes the VEGF-Nrp1 interaction [He et al 2015].

NEFL

Gene structure. *NEFL* contains four coding exons; the 5' UTR is highly conserved.

Pathogenic allelic variants. One family has a deletion/insertion variant in exon 1 (c.22_23delCCinsAG) [De Jonghe et al 2001]. See also Table A.

Table 7. Selected *NEFL* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.22_23delCCinsAG	p.Pro8Arg	NM_006158.1 NP_006149.2

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Normal gene product. Neurofilament light polypeptide, the protein encoded by *NEFL*, contains 543 amino acids with a head, rod, and tail domain. Neurofilaments form the cytoskeletal component of myelinated axons.

Abnormal gene product. Knockout mice lacking neurofilaments have diminished axon caliber and delayed regeneration of myelinated axons following crush injury. A mouse mutation in *Nefl* has massive degeneration of spinal motor neurons and abnormal neurofilament accumulation with severe neurogenic skeletal muscle atrophy. Defects in transport and assembly of neurofilaments have been reported [Perez-Olle et al 2004].

GDAP1

Gene structure. *GDAP1* has six exons, 13.9 kb of DNA, and a 1007-nucleotide open reading frame.

Pathogenic allelic variants. See also Table A.

Table 8. Selected *GDAP1* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.358C>T	p.Arg120Trp	NM_018972.2 NP_061845.2

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Normal gene product. Ganglioside-induced differentiation-associated protein-1 [Baxter et al 2002]

Abnormal gene product. It is speculated that pathogenic variants may prevent the correct catalyzing S conjugation of reduced GCH, resulting in progressive attrition of both axons and Schwann cells.

HSPB8 (HSP22)

Gene structure. *HSPB8* has three exons and spans about 16 kb.

Pathogenic allelic variants. Three pathogenic variants have been reported. See Table 9 [Irobi et al 2004b, Tang et al 2005].

Table 9. Selected *HSPB8* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.423G>T	p.Lys141Asn	NM_014365.2 NP_055180.1
c.423G>C	p.Lys141Asn	
c.421A>G	p.Lys141Glu	

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Normal gene product. HSPB8 (also called HSP22) is a phosphor protein that interacts with HSPB1.

Abnormal gene product. Mutant HSPB8 proteins interact with HSPB1 and form aggregates that may lead to dysfunctional axonal transport and dysregulation of the cytoskeleton [Irobi et al 2004b].

AARS

Gene structure. *AARS* has 21 exons and is located on chromosome 16.

Pathogenic allelic variants. Two pathogenic variants have been associated with CMT (p.Arg329His and p.Glu778Ala) [Latour et al 2010, McLaughlin et al 2012]. See Table 10.

The variant p.Asn71Tyr (c.211A>T) may also be pathogenic.

Table 10. Selected *AARS* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.986G>A	p.Arg329His	NM_001605.2 NP_001596.2
c.2333A>C	p.Glu778Ala	

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Normal gene product. Alanyl-tRNA synthetase attaches alanine to tRNA molecules in cytoplasm and mitochondria completing the first step in protein translation.

Abnormal gene product. Functional studies suggest these are loss-of-function mutations [McLaughlin et al 2012].

DYNC1H1

Gene structure. *DYNC1H1 has* 78 exons.

Pathogenic allelic variants. One pathogenic variant has been described (p.His306Arg) by Weedon et al [2011]. See Table 11.Ten novel missense variants have been reported by Scoto et al [2015].

Table 11. Selected *DYNC1H1* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.917A>G	p.His306Arg	NM_001376.4 NP_001367.2

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Normal gene product. 4,646-amino acid protein. DYNC1H1 is a subunit of cytoplasmic dynein, the primary motor protein producing retrograde axonal transport in neurons.

Abnormal gene product. Presumably the abnormal protein produces defective retrograde axonal transport in peripheral nerves.

LRSAM1

Gene structure. *LRSAM1* has 25 exons.

Pathogenic allelic variants. A single autosomal dominant and a single autosomal recessive pathogenic variant have been described. See Table 12.

Table 12. Selected *LRSAM1* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.2121_2122 dupGC	p.Leu708ArgfsTer28	NM_138361.5 NP_612370.3
c.1913-1G>A	--	

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Normal gene product. A ubiquitin ligase (E3) involved with sorting ubiquitinylated cytoplasmic cargo (TSG101); 723-amino acid protein.

Abnormal gene product. Disturbs sorting of ubiquitinylated cargo in neuronal cytoplasm.

DHTKD1

Gene structure. The *DHTKD* transcript NM_018706.6 has 17 exons.

Pathogenic allelic variants. A large Chinese family segregated the nonsense pathogenic variant NM_018706.6:c.1455T>G (p.Tyr485Ter) in exon 8 [Xu et al 2012]

Normal gene product. *DHTKD* encodes the DNA-binding protein SMUBP-2 (DHTKD1), which has 919 amino acids. DHTKD1 contributes to mitochondrial biogenesis and function maintenance and plays a critical role in energy production [Xu et al 2012].

Abnormal gene product. Decreased functional DHTKD1 leads to retarded cell growth and increased cell apoptosis, a result of impaired mitochondrial biogenesis and increased reactive oxygen species.

IGHMBP2

Gene structure. *IGHMBP2* has 15 exons.

Pathogenic allelic variants. See Table 13.

Table 13. Selected *IGHMBP2* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.138T>A	p.Cys46Ter	NM_002180.2 NP_002171.2
c.604T>G	p.Phe202Val	
c.2911_2912delAG	p.Arg97GlufsTer4	
c.1591C>A	p.Pro531Thr	
c.1738G>A	p.Val580Ile	
c.449+1G>T		
c.2784+1G>T		

Note on variant classification: Variants listed in the table have been provided by the author. *GeneReviews* staff have not independently verified the classification of variants.

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Normal gene product. The 993-amino acid protein has seven putative helicase motifs and a DEAD box-like motif typical for RNA helicases.

Abnormal gene product. Mutation may lead to dysfunction of helicase activity.

DNAJB2

Gene structure. Transcript variant 1 (NM_001039550.1) has ten exons and is the predominant transcript.

Pathogenic allelic variants. See Table 14.

Table 14. Selected *DNAJB2* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.229+1G>A		NM_001039550.1 NP_001034639.1
c.14A>G	p.Tyr5Cys	

Note on variant classification: Variants listed in the table have been provided by the author. *GeneReviews* staff have not independently verified the classification of variants.

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Normal gene product. Transcript variant 1 encodes a protein of 277 amino acids. It belongs to the HSP40 chaperone protein family involved with protein folding. It may clear toxic proteins involved in disease aggregations such as those associated with ALS (SOD1) and Huntington.

Abnormal gene product. Mutation results in an inability to clear toxic protein aggregates.

MARS

Gene structure. The gene has 21 exons.

Pathogenic allelic variants. Two missense pathogenic variants have been reported. See Table 15.

Table 15. Selected *MARS* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.1852C>T	p.Arg618Cys	NM_004990.3 NP_004981.2
c.2398C>A	p.Pro800Thr	

Note on variant classification: Variants listed in the table have been provided by the author. *GeneReviews* staff have not independently verified the classification of variants.

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Normal gene product. Methionine--tRNA ligase, cytoplasmic has 900 amino acids.

Abnormal gene product. The two reported pathogenic variants likely cause loss of function [Gonzalez et al 2013].

NAGLU

Gene structure. The gene has 6 exons (NM_000263.3).

Pathogenic allelic variants. In affected members of a large French Canadian kindred, Tetreault et al [2015] identified a heterozygous c.1208T-C transition (c.1208T-C,

NM_000263.3) in exon 6, resulting in an ile403-to-thr (I403T) substitution at a highly conserved residue in the Tim-barrel domain. The pathogenic variant segregated with the disorder in the family and was not found in the dbSNP (build 137), Exome Variant server databases, or in over 50 French Canadian controls.

Table 16. *NAGLU* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
c.1208T>C	p.Ile403Thr	NM_000263.3

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Normal gene product. *NAGLU* encodes the 743-amino acid enzyme alpha-N-acetylglucosaminidase that degrades heparan sulfate by hydrolysis of terminal N-acetyl-D-glucosamine residues in N-acetyl-alpha-D-glucosaminides.

Abnormal gene product. Leukocytes from affected individuals showed significantly decreased NAGLU enzyme activity (36-54% of controls) [Tetreault et al 2015].

Haploinsufficiency of alpha-N-acetylglucosaminidase is thought to cause CMT2 phenotype.

HARS

Gene structure. The longest *HARS* transcript variant NM_002109.5 has 13 exons.

Pathogenic allelic variants. Heterozygous *HARS* pathogenic variants have been reported in five unrelated families with CMT2 [Vester et al 2013, Safka Brozkova et al 2015].

Normal gene product. The longest isoform NP_002100.2 has 509 amino acids. HARS encodes histidine--tRNA ligase, cytoplasmic, the enzyme responsible for the synthesis of histidyl-transfer RNA, which is essential for the incorporation of histidine into proteins.

Abnormal gene product. Haploinsufficiency is the presumed cause of the CMT2 phenotype.

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