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DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy – RETIRED CHAPTER, FOR HISTORICAL REFERENCE ONLY

Synonym: Dominant Intermediate Charcot-Marie-Tooth Neuropathy Type B (DI-CMTB)

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Summary

NOTE: THIS PUBLICATION HAS BEEN RETIRED. THIS ARCHIVAL VERSION IS FOR HISTORICAL REFERENCE ONLY, AND THE INFORMATION MAY BE OUT OF DATE.

Clinical characteristics

DNM2-related intermediate Charcot-Marie-Tooth neuropathy (DI-CMTB) has a classic, mild to moderately severe Charcot-Marie-Tooth hereditary neuropathy phenotype that often includes *pes cavus* foot deformity, depressed tendon reflexes, distal muscle weakness and atrophy, and sensory loss. Age of onset varies greatly among affected individuals and ranges from age two to 50 years. It is unusual for individuals with DI-CMTB to become wheelchair bound. Other findings include asymptomatic neutropenia and early-onset cataracts (often noted in childhood before age 15 years).

Diagnosis/testing

The diagnosis is suspected in individuals with typical findings of CMT hereditary neuropathy and intermediate or axonal motor median nerve conduction velocities (NCV) ranging from 26 m/s to normal. Diagnosis requires identification of a heterozygous pathogenic variant in *DNM2*, the only gene known to be associated with DI-CMTB.

Management

Treatment of manifestations: Treatment of DI-CMTB is symptomatic and involves evaluation and management by a multidisciplinary team that includes neurologists, orthopedic surgeons, and physical and occupational

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therapists. Treatment may include ankle/foot orthoses, orthopedic surgery, forearm crutches or canes, wheelchairs, acetaminophen or nonsteroidal anti-inflammatory agents (NSAIDs) for musculoskeletal pain, and career and employment counseling.

Prevention of secondary complications: Physical therapy to prevent foot contractures, acquired foot deformities, and difficulty walking.

Surveillance: Regular evaluation by the multidisciplinary team to determine neurologic status and functional disability.

Pregnancy management: In general there appears to be an increased occurrence of abnormal fetal presentation and maternal postpartum bleeding in women with Charcot-Marie-Tooth disease.

Agents/circumstances to avoid: All drugs or agents known to be hazardous for peripheral neuropathies.

Genetic counseling

DI-CMTB is inherited in an autosomal dominant manner. Most individuals diagnosed with DI-CMTB have an affected parent. The proportion of cases caused by a heterozygous *de novo* pathogenic variant is unknown. Each child of an individual with DI-CMTB has a 50% chance of inheriting the pathogenic variant. Prenatal testing for pregnancies at increased risk is possible if the pathogenic variant has been identified in an affected family member. Requests for prenatal testing for conditions which (like DI-CMTB) do not affect intellect and have some treatment available are not common.

Diagnosis

Suggestive Findings

DNM2-related intermediate Charcot-Marie-Tooth neuropathy (DI-CMTB) **should be suspected** in individuals with the following clinical findings, nerve conduction velocities, and neuropathology:

Clinical manifestations

- Sensory and motor deficiencies involving the lower legs
  - Sensory loss
  - Depressed tendon reflexes
  - Distal muscle weakness and atrophy
  - *Pes cavus* foot deformity
- Asymptomatic neutropenia
- Early-onset cataracts (noted before age 15 years)

Nerve conduction velocities (NCVs) are "intermediate" (i.e., 25-45 m/s) between a demyelinating and axonal neuropathy using strict electrophysiologic criteria [Davis et al 1978, Nicholson & Myers 2006].

Neuropathology. Sural nerve biopsy has shown diffuse loss of large myelinated fibers, clusters of regenerating myelinated axons, and fibers with focal myelin thickenings [Kennerson et al 2001, Claeys et al 2009].

Establishing the Diagnosis

The diagnosis of DI-CMTB **is established** in a proband by the identification of a heterozygous pathogenic variant in *DNM2*.

Molecular testing approaches can include **serial single-gene testing**, use of a **multigene panel**, and **comprehensive genomic testing**.

Serial single-gene testing can be considered based on the order in which pathogenic variants most commonly occur in individuals with the above suggestive findings:

- In a person with a CMT phenotype and very slow NCV (<30 m/s), perform molecular genetic testing of *PMP22* first to determine if a *PMP22* duplication, the most common cause of this demyelinating phenotype, is present.
- In a person with a CMT phenotype and intermediate to normal NCV, perform molecular genetic testing of the *MPZ*, *GJB1* (encoding the protein connexin 32), and *MFN2* genes first because mutation of one of these genes is a common cause of this phenotype.
- In a person with a CMT phenotype and NCV between 30 and 45 m/s in whom testing for the above genes has not identified a pathogenic variant, molecular genetic testing of *DNM2* is appropriate.

A multigene panel that includes *DNM2* and other genes of interest (see Differential Diagnosis) may also be considered. Note: (1) The genes included in the panel and the diagnostic sensitivity of the testing used for each gene vary by laboratory and are likely to change over time. (2) Some multigene panels may include genes not associated with the condition discussed in this *GeneReview*; thus, clinicians need to determine which multigene panel is most likely to identify the genetic cause of the condition at the most reasonable cost while limiting identification of variants of uncertain significance and pathogenic variants in genes that do not explain the underlying phenotype. (3) In some laboratories, panel options may include a custom laboratory-designed panel and/or custom phenotype-focused exome analysis that includes genes specified by the clinician. (4) Methods used in a panel may include sequence analysis, deletion/duplication analysis, and/or other non-sequencing-based tests.

For an introduction to multigene panels click here. More detailed information for clinicians ordering genetic tests can be found here.

Comprehensive genomic testing may be considered if serial single-gene testing (and/or use of a multigene panel) has not confirmed a diagnosis in an individual with features of DI-CMTB. Such testing may include exome sequencing, genome sequencing, and mitochondrial sequencing.

For an introduction to comprehensive genomic testing click here. More detailed information for clinicians ordering genomic testing can be found here.

Table 1. Molecular Genetic Testing Used in *DNM2*-Related Intermediate Charcot-Marie-Tooth Neuropathy

Gene [1]	Method	Proportion of Probands with a Pathogenic Variant [2] Detectable by Method
DNM2	Sequence analysis [3]	Nearly 100%
	Gene-targeted deletion/duplication analysis [4]	None reported [5]

1. See Table A. Genes and Databases for chromosome locus and protein.
2. See Molecular Genetics for information on allelic variants.
3. Sequence analysis detects variants that are benign, likely benign, of uncertain significance, likely pathogenic, or pathogenic. Pathogenic variants may include small intragenic deletions/insertions and missense, nonsense, and splice site variants; typically, exon or whole-gene deletions/duplications are not detected. For issues to consider in interpretation of sequence analysis results, click here.
4. Gene-targeted deletion/duplication analysis detects intragenic deletions or duplications. Methods used may include quantitative PCR, long-range PCR, multiplex ligation-dependent probe amplification (MLPA), and gene-targeted microarray designed to detect single-exon deletions or duplications.
5. No deletions or duplications involving *DNM2* as causative of DI-CMTB have been reported.

Clinical Characteristics

Clinical Description

DNM2-related intermediate Charcot-Marie-Tooth neuropathy (DI-CMTB) is a so-called "dominant intermediate form" of CMT neuropathy because it is inherited in an autosomal dominant manner and it is "intermediate" between a demyelinating and axonal neuropathy using strict electrophysiologic criteria for nerve conduction velocities (NCVs). The condition is characterized by a classic, mild to moderately severe Charcot-Marie-Tooth hereditary neuropathy phenotype that often includes *pes cavus* foot deformity, depressed tendon reflexes, distal muscle weakness and atrophy, and sensory loss.

Age of onset varies greatly among affected individuals and ranges from age two to 50 years. Some persons require AFO braces or other walking aids. Three percent of affected individuals become wheelchair bound; one person in the Claeys et al [2009] study required a wheelchair at age 61 years.

Other findings include asymptomatic neutropenia and early-onset cataracts (often noted in childhood before age 15 years).

Electrophysiologic studies indicate intermediate or axonal motor median nerve conduction velocities (NCV) ranging from 26 m/s to normal values.

Genotype-Phenotype Correlations

Strong genotype-phenotype correlations have not been reported.

The majority of *DNM2* pathogenic variants appear to be in the domain encoding homology to pleckstrin [Züchner et al 2005, Fabrizi et al 2007]; however, exceptions have been identified [Claeys et al 2009, Susman et al 2010].

Penetrance

DNM2 pathogenic variants are penetrant over a wide range of ages (age 2-50 years) [Claeys et al 2009, Haberlová et al 2011].

Anticipation

Anticipation is not observed.

Prevalence

DI-CMTB is a rare cause of CMT. Up to 3.4% of CMT (in which CMT1A, 1B, and 1X have already been excluded) is caused by a *DNM2* pathogenic variant [Claeys et al 2009].

Genetically Related (Allelic) Disorders

Charcot-Marie-Tooth Type 2M (CMT2M) is a purely axonal CMT caused by heterozygous pathogenic variants in *DNM2*; it is distinguished from DI-CMT by the lack of demyelinating changes. CMT2M is inherited in an autosomal dominant manner [Fabrizi et al 2007].

Autosomal dominant centronuclear myopathy (*DNM2*-related CNM) is also associated with pathogenic variants in *DNM2* [Bitoun et al 2005]. *DNM2*-related CNM usually presents with neonatal hypotonia, weak suck, poor feeding, and progressive muscle weakness with respiratory problems [Jungbluth et al 2010, Melberg et al 2010].

Some individuals with CNM have clinical findings that overlap with DI-CMTB [Fischer et al 2006, Bitoun et al 2008, Susman et al 2010]. The findings shared between DI-CMTB and CNM are muscle weakness and sometimes cataract. CNM does not have peripheral nerve involvement and often has proximal muscle weakness that is not seen in DI-CMTB.

Differential Diagnosis

Other forms of intermediate CMT:

- DI-CMTA, linked to the 10q24-q25.1 region
- DI-CMTC, caused by heterozygous pathogenic variants in *YARS1* (formerly *TyrRS*)
- DI-CMTD, caused by heterozygous pathogenic variants in *MPZ*
- *GNB4*-related CMT, also inherited in an autosomal dominant manner

It is usually not possible to differentiate between DI-CMTB, other intermediate forms of CMT, and most CMT2 types based on clinical findings [Nicholson & Myers 2006], unless cataract and/or neutropenia (occasional findings in DI-CMTB) are present.

See CMT Overview, particularly to exclude potentially treatable causes of acquired neuropathy.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease and needs in an individual diagnosed with *DNM2*-related intermediate Charcot-Marie-Tooth neuropathy (DI-CMTB), the following evaluations are recommended:

- Neurologic examination
- Electrophysiologic studies to establish a baseline for further monitoring of disease progression
- Complete blood count (CBC) with absolute neutrophil count (ANC) to evaluate for neutropenia
- Ophthalmologic examination for cataract
- Consultation with a clinical geneticist and/or genetic counselor

Treatment of Manifestations

Treatment of DI-CMTB is symptomatic and involves evaluation and management by a multidisciplinary team that includes neurologists, orthopedic surgeons, and physical and occupational therapists. Due to the great phenotypic variability, disease treatment should be tailored to the individual's needs.

Treatment may include:

- Ankle/foot orthoses
- Orthopedic surgery
- Forearm crutches or canes; rarely, wheelchairs
- Treatment of musculoskeletal pain with acetaminophen or nonsteroidal anti-inflammatory agents (NSAIDs)
- Career and employment counseling

Prevention of Secondary Complications

The most common secondary complications include foot contractures and acquired foot deformities, difficulty walking, and, in severe cases, inability to ambulate. Physical therapies such as stretching and exercise are recommended to prevent these secondary complications.

Surveillance

Surveillance includes regular evaluation by the multidisciplinary team to determine neurologic status and functional disability.

Agents/Circumstances to Avoid

Medications that are toxic or potentially toxic to persons with CMT comprise a spectrum of risk ranging from definite high risk to negligible risk. See the Charcot-Marie-Tooth Association website (pdf) for an up-to-date list.

Evaluation of Relatives at Risk

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Pregnancy Management

Although no systemic studies have been done concerning pregnancy in women with DI-CMTB, there are reports of an increased occurrence of abnormal fetal presentation and maternal postpartum bleeding in women with CMT in general [Hoff et al 2005]. The early miscarriage rate is **not** increased in women with CMT [Argov & de Visser 2009].

Therapies Under Investigation

Search ClinicalTrials.gov in the US and EU Clinical Trials Register in Europe for access to information on clinical studies for a wide range of diseases and conditions. Note: There may not be clinical trials for this disorder.

Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, mode(s) of inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members; it is not meant to address all personal, cultural, or ethical issues that may arise or to substitute for consultation with a genetics professional. —ED.

Mode of Inheritance

DNM2-related intermediate Charcot-Marie-Tooth neuropathy (DI-CMTB) is inherited in an autosomal dominant manner.

Risk to Family Members

Parents of a proband

- Most individuals diagnosed with DI-CMTB have an affected parent.
- A proband with DI-CMTB may have the disorder as the result of *de novo DNM2* mutation. The proportion of cases caused by a *de novo* pathogenic variant is unknown.
- If the pathogenic variant found in the proband cannot be detected in the leukocyte DNA of either parent, two possible explanations are germline mosaicism in a parent or *de novo* mutation in the proband. Although no instances of germline mosaicism have been reported, it remains a possibility.
- Recommendations for the evaluation of parents of a proband with an apparent *de novo* pathogenic variant include *DNM2* molecular genetic testing for the variant identified in the proband. Evaluation of parents

may determine that one is affected but has escaped previous diagnosis because of failure by health care professionals to recognize the syndrome and/or a milder phenotypic presentation. Therefore, an apparently negative family history cannot be confirmed until appropriate evaluations have been performed.

Note: (1) Although most individuals diagnosed with DI-CMTB have an affected parent, the family history may appear to be negative because of failure to recognize the disorder in family members, early death of the parent before the onset of symptoms, or late onset of the disease in the affected parent. (2) If the parent is the individual in whom the pathogenic variant first occurred s/he may have somatic mosaicism for the variant and may be mildly/minimally affected.

Sibs of a proband

- The risk to the sibs of the proband depends on the genetic status of the proband's parents.
- If a parent of the proband is affected and/or has a pathogenic variant, the risk to the sibs of inheriting the variant is 50%.
- When the parents are clinically unaffected, the risk to the sibs of a proband appears to be low.
- The sibs of a proband with clinically unaffected parents are still at increased risk for DI-CMTB because of the possibility of reduced penetrance in a parent.
- If the pathogenic variant found in the proband cannot be detected in the leukocyte DNA of either parent, the empiric recurrence risk to sibs is approximately 1% because of the theoretic possibility of parental germline mosaicism.

Offspring of a proband. Each child of an individual with DI-CMTB has a 50% chance of inheriting the *DNM2* pathogenic variant.

Other family members

- The risk to other family members depends on the status of the proband's parents.
- If a parent is affected and/or has a pathogenic variant, his or her family members may be at risk.

Related Genetic Counseling Issues

Considerations in families with an apparent *de novo* pathogenic variant. When neither parent of a proband with an autosomal dominant condition has clinical evidence of the disorder or the pathogenic variant, it is likely that the proband has a *de novo* pathogenic variant. However, possible non-medical explanations including alternate paternity or maternity (e.g., with assisted reproduction) or undisclosed adoption could also be explored.

Family planning

- The optimal time for determination of genetic risk and discussion of the availability of prenatal testing is before pregnancy.
- It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected or at risk of being affected.

DNA banking is the storage of DNA (typically extracted from white blood cells) for possible future use. Because it is likely that testing methodology and our understanding of genes, allelic variants, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals.

Prenatal Testing and Preimplantation Genetic Testing

Once the *DNM2* pathogenic variant has been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic testing for *DNM2*-related intermediate Charcot-Marie-Tooth neuropathy are possible.

Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing, particularly if the testing is being considered for the purpose of pregnancy termination rather than early diagnosis. While most centers would consider decisions regarding prenatal testing to be the choice of the parents, discussion of these issues is appropriate.

Resources

GeneReviews staff has selected the following disease-specific and/or umbrella support organizations and/or registries for the benefit of individuals with this disorder and their families. GeneReviews is not responsible for the information provided by other organizations. For information on selection criteria, click here.

- **Association CMT France**
  France
  Phone: 820 077 540; 2 47 27 96 41
  www.cmt-france.org
- **Charcot-Marie-Tooth Association (CMTA)**
  PO Box 105
  Glenolden PA 19036
  Phone: 800-606-2682 (toll-free); 610-499-9264
  Fax: 610-499-9267
  Email: info@cmtausa.org
  www.cmtausa.org
- **European Charcot-Marie-Tooth Consortium**
  Department of Molecular Genetics
  University of Antwerp
  Antwerp Antwerpen B-2610
  Belgium
  Fax: 03 2651002
  Email: gisele.smeyers@ua.ac.be
- **Hereditary Neuropathy Foundation, Inc.**
  432 Park Avenue South
  4th Floor
  New York NY 10016
  Phone: 855-435-7268 (toll-free); 212-722-8396
  Fax: 917-591-2758
  Email: info@hnf-cure.org

www.hnf-cure.org

- **My46 Trait Profile**
  Charcot Marie Tooth disease
- **National Library of Medicine Genetics Home Reference**
  Charcot-Marie-Tooth disease
- **NCBI Genes and Disease**
  Charcot-Marie-Tooth syndrome
- **TREAT-NMD**
  Institute of Genetic Medicine
  University of Newcastle upon Tyne
  International Centre for Life
  Newcastle upon Tyne NE1 3BZ
  United Kingdom
  Phone: 44 (0)191 241 8617
  Fax: 44 (0)191 241 8770
  Email: info@treat-nmd.eu
  Charcot-Marie-Tooth Disease
- **Association Francaise contre les Myopathies (AFM)**
  1 Rue de l'International
  BP59
  Evry cedex 91002
  France
  Phone: +33 01 69 47 28 28
  Email: dmc@afm.genethon.fr
  www.afm-telethon.fr
- **European Neuromuscular Centre (ENMC)**
  Lt Gen van Heutszlaan 6
  3743 JN Baarn
  Netherlands
  Phone: 31 35 5480481
  Fax: 31 35 5480499
  Email: enmc@enmc.org
  www.enmc.org
- **Muscular Dystrophy Association - USA (MDA)**
  222 South Riverside Plaza
  Suite 1500

Chicago IL 60606
Phone: 800-572-1717
Email: mda@mdausa.org
www.mda.org

- **Muscular Dystrophy UK**
  61A Great Suffolk Street
  London SE1 0BU
  United Kingdom
  Phone: 0800 652 6352 (toll-free); 020 7803 4800
  Email: info@musculardystrophyuk.org
  www.musculardystrophyuk.org
- **RDCRN Patient Contact Registry: Inherited Neuropathies Consortium**
  Patient Contact Registry

Molecular Genetics

Information in the Molecular Genetics and OMIM tables may differ from that elsewhere in the GeneReview: tables may contain more recent information. —ED.

Table A. DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy: Genes and Databases

Locus Name	Gene	Chromosome Locus	Protein	Locus-Specific Databases	HGMD	ClinVar
DI-CMTB	*DNM2*	19p13.2	Dynamin-2	IPN Mutations, DNM2 DNM2 homepage - Leiden Muscular Dystrophy pages	DNM2	DNM2

Data are compiled from the following standard references: gene from HGNC; chromosome locus from OMIM; protein from UniProt. For a description of databases (Locus Specific, HGMD, ClinVar) to which links are provided, click here.

Table B. OMIM Entries for DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy (View All in OMIM)

602378	DYNAMIN 2; DNM2
606482	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE B; CMTDIB

Gene structure. *DNM2* has several isoforms; the longest transcript is isoform 1 (NM_001005360.1), which has 22 exons. See Entrez Gene for a description of isoforms.

Pathogenic variants. Pathogenic missense variants and small deletions in the coding region have been described.

Normal gene product. Isoform 1 encodes the dynamin-2 protein of 870 amino acid residues (NP_001005360.1).

Abnormal gene product. The identification of small deletions suggests that haploinsufficiency is the cause of the disorder.

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Chapter Notes

Revision History

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