Charcot-marie-tooth disease: Exploring the genetics, symptoms, and treatment options.

Antoni Suntar*

Department of Molecular Pathobiology of Brain Disease, Kyoto Prefectural University of Medicine, Japan

Introduction

Charcot-Marie-Tooth Disease (CMT) is a hereditary neuropathy named after the three physicians who first described it in 1886: Jean-Martin Charcot, Pierre Marie, and Howard Henry Tooth. CMT is one of the most common inherited neurological disorders, affecting about 1 in 2,500 people. This progressive condition affects the peripheral nerves, leading to muscle weakness, atrophy, and sensory loss in the limbs. Understanding the pathophysiology, symptoms, diagnosis, causes, treatment, and living with CMT provides a comprehensive view of this complex disorder [1].

CMT primarily affects the peripheral nerves, which are responsible for transmitting signals between the central nervous system (brain and spinal cord) and the muscles and sensory organs. The disease is caused by mutations in various genes that affect the structure and function of peripheral nerves. These mutations lead to the degeneration of the myelin sheath (the protective covering of nerves) or the axons (the core part of the nerve fiber that transmits signals) [2].

The symptoms of CMT can vary widely in severity and age of onset, even within the same family. Common symptoms include: Muscle Weakness and Atrophy: Typically starts in the feet and lower legs and progresses to the hands and forearms. This can lead to difficulty walking, foot drop, and a high-stepping gait. Sensory Loss: Numbness, tingling, or loss of sensation in the feet, legs, hands, and arms [3].

Foot Deformities: High arches (pes cavus) and hammer toes due to muscle imbalance. Difficulty with Fine Motor Skills: Problems with tasks requiring hand coordination, such as buttoning clothes or writing. Balance Problems: Due to muscle weakness and sensory loss, individuals may have difficulty maintaining balance. Pain: Muscle cramps, spasms, and nerve pain can occur, although this varies among individuals [4].

CMT is a genetic disorder with various inheritance patterns, including autosomal dominant, autosomal recessive, and X-linked. The specific cause depends on the type and subtype of CMT: Autosomal Dominant: The most common inheritance pattern, where only one mutated copy of the gene is needed to cause the disease. Examples include CMT1A (PMP22 duplication) and CMT2A (MFN2 mutations) [5].

Autosomal Recessive: Both copies of the gene must be mutated for the disease to manifest. Examples include some subtypes of CMT4. X-Linked: The mutated gene is located on the X chromosome. Males are typically more severely affected, while females may have milder symptoms or be carriers. An example is CMTX1 (GJB1 mutations) [6].

There is currently no cure for CMT, and treatment focuses on managing symptoms, maintaining mobility, and improving quality of life. A multidisciplinary approach involving neurologists, physical therapists, occupational therapists, and other specialists is crucial. Key treatment strategies include: Physical Therapy: To maintain muscle strength, flexibility, and joint mobility. Tailored exercise programs can help slow the progression of muscle weakness [7].

Occupational Therapy: To assist with daily activities and recommend adaptive devices to improve independence and safety. Orthopedic Interventions: Use of braces, orthotic devices, and custom footwear to correct foot deformities, improve walking, and prevent falls [8].

There is currently no cure for CMT, and treatment focuses on managing symptoms, maintaining mobility, and improving quality of life. A multidisciplinary approach involving neurologists, physical therapists, occupational therapists, and other specialists is crucial. Key treatment strategies include: Physical Therapy: To maintain muscle strength, flexibility, and joint mobility. Tailored exercise programs can help slow the progression of muscle weakness [9].

Occupational Therapy: To assist with daily activities and recommend adaptive devices to improve independence and safety. Orthopedic Interventions: Use of braces, orthotic devices, and custom footwear to correct foot deformities, improve walking, and prevent falls. Pain Management: Medications such as analgesics, anti-inflammatory drugs, and neuropathic pain medications to manage pain [10].

Conclusion

Charcot-Marie-Tooth Disease is a complex genetic disorder that affects the peripheral nerves, leading to progressive muscle weakness, atrophy, and sensory loss. While there is no cure, advances in medical care and supportive therapies have improved the management and prognosis of the disease. A comprehensive approach to treatment, including physical therapy, occupational therapy, orthopedic interventions, and pain management, can help individuals with CMT maintain mobility and quality of life. Ongoing research holds promise

***Correspondence to:** Antoni Suntar, Department of Molecular Pathobiology of Brain Disease, Kyoto Prefectural University of Medicine, Japan. E-mail: Antonio@kpu.jp.com

Received*:** 27-Dec-2023, Manuscript No. JNNR-24-137399; ***Editor assigned: 29-Dec-2023, Pre QC No. JNNR-24-137399(PQ); ***Reviewed*:** 12-Jan-2024, QC No. JNNR-24-137399; ***Revised:*** 17-Jan-2024, Manuscript No. JNNR-24-137399(R); ***Published:*** 24-Jan-2024, DOI: 10.35841/aajnnr-9.1.186

Citation: *Suntar A. Charcot-marie-tooth disease: Exploring the genetics, symptoms, and treatment options. J Neurol Neurorehab Res.2024;9(1):186*

for more effective treatments and a deeper understanding of this challenging condition. Through continued efforts in scientific research, patient care, and advocacy, the outlook for those living with CMT continues to improve.

References

1. Yiu EM, Bray P, Baets J, et al. Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. J Neurol Neurosurg Psychiatry. 2022;93(5):530-8.
2. Okamoto Y, Takashima H. The current state of Charcot–Marie–Tooth disease treatment. Genes (Basel). 2023;14(7):1391.
3. McCray BA, Scherer SS. Axonal Charcot-Marie-Tooth disease: from common pathogenic mechanisms to emerging treatment opportunities. Neurotherapeutics. 2021;18(4):2269-85.
4. Pfeffer GB, Michalski MP. Charcot-Marie-Tooth Disease: A Surgical Algorithm. Foot Ankle Clin. 2023;28(4):857-71.
5. Itani R, Elmallahi N, Al Ibrahim A. Charcot–Marie–Tooth (CMT) disease and pregnancy: a case report and literature review. J Obstet Gynaecol. 2020;40(2):275-6.
6. Higuchi Y, Takashima H. Clinical genetics of Charcot–Marie–Tooth disease. J Hum Genet. 2023;68(3):199-214.
7. Laurá M, Pipis M, Rossor AM, et al. Charcot–Marie–Tooth disease and related disorders: an evolving landscape. Curr Opin Neurol. 2019;32(5):641-50.
8. Kotaich F, Caillol D, Bomont P. Neurofilaments in health and Charcot-Marie-Tooth disease. Front Cell Dev Biol. 2023;11.
9. Singh D, Gray J, Laura M, et al. Charcot neuroarthropathy in patients with Charcot Marie tooth disease. Foot Ankle Surg. 2021;27(8):865-8.
10. Kitani-Morii F, Noto YI. Recent advances in Drosophila models of Charcot-Marie-Tooth disease. Int J Mol Sci. 2020;21(19):7419.

Citation: *Suntar A. Charcot-marie-tooth disease: Exploring the genetics, symptoms, and treatment options. J Neurol Neurorehab Res.2024;9(1):186*