CHARCOT-MARIE-TOOTH 1A (CMT1A) AND IMPAIRED PATIENT MOBILITY-EXPRESSIONS, REMEDIES AND IMPACT ON QUALITY-OF-LIFE



RN Moore¹; AT Moore²; FP Thomas, MD, PhD³; JM Aldrich²

¹True Reply LLC, New York NY, USA, ²Hereditary Neuropathy Foundation, New York NY, USA, ³Hackensack University Medical Center and Hackensack Meridian School of Medicine, Hackensack NJ, USA



Abstract

OBJECTIVES: Charcot-Marie-Tooth disease (CMT) is one of the most common inherited neurological disorders, affecting approximately 1 in 2,500 people in the United States. CMT comprises a group of disorders that affect peripheral nerves. The peripheral nerves lie outside the brain and spinal cord and supply the muscles and sensory organs in the limbs.

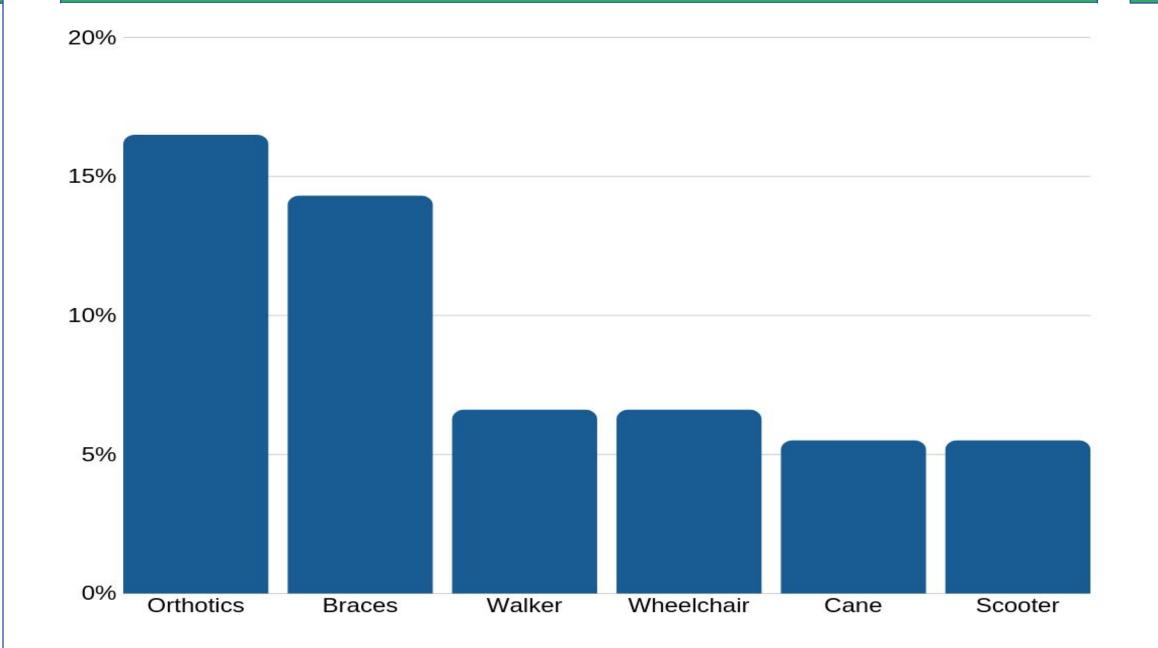
Inherited neuropathies (IN) compromises muscle strength, activities of daily living (ADL) and quality of life (QOL). One subtype, CMT1A, accounts for half of all patients. This study assessed the frequency of specific symptoms and signs in CMT1A and their impact on ADL and QOL. In addition, it identified the remedies patients use to cope with their mobility impairment.

METHODS: The Hereditary Neuropathy Foundation, in association with Hannah's Hope Fund, created the Global Registry for Inherited Neuropathies (GRIN), to capture detailed IN patient history via an online, IRB approved patient survey from 2013-2019. 2,142 IN patients were surveyed, yielding a 34% CMT1A (N=770) cohort. This cohort was queried against a subset of registry questions to test for relationships between disease state, level of impairment and QOL.

RESULTS: CMT1A patients identify mobility (63%) as the ADL impacted most by their disease. Multiple features of their disease impact their mobility, weak ankles being most often cited (79%). 58% of patients can neither walk on their heels or toes. Even though 55% use some type of leg brace or mobility device (cane, walker, wheelchair, scooter), 60% suffer from frequent falls which can result in fractures. 54% have had orthopedic surgery to improve mobility or foot deformities. Imbalance and walking fatigue were cited as *most* challenging by 28%. Fatigue, a important corollary of impaired strength and mobility was reported by 92%, pain by 78%.

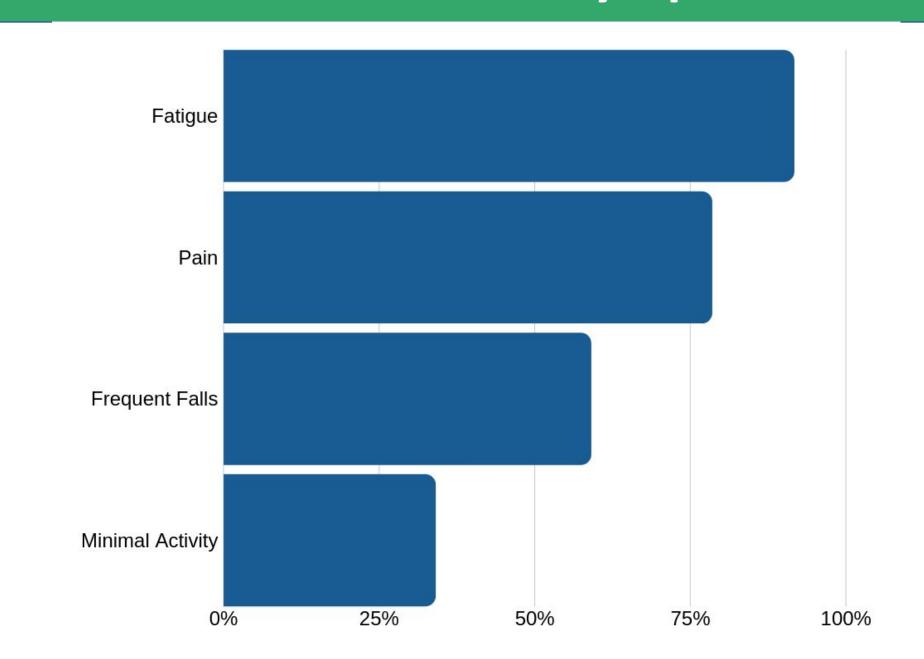
CONCLUSIONS: Mobility impairments were represented in a large cohort of CMT1A patients, affecting their physical and psychological (pain, fatigue) well-being. While use of orthotics, mobility devices, and surgery can have a positive impact on mobility, CMT1A patients need a more cohesive and comprehensive approach to addressing mobility impairments.

Mobility Devices Used by CMT1A Patients



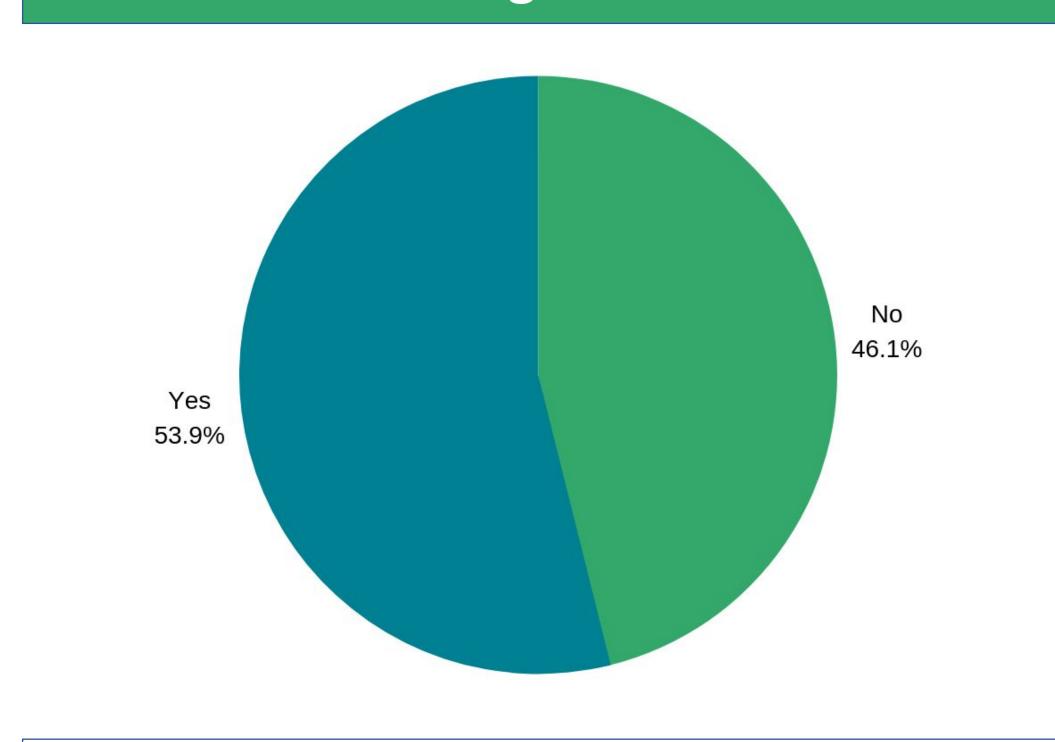
Over half (55%) of CMT1A patients use some sort of device to mitigate the impact of their disease on mobility, with orthotics and bracing cited by over 30% of patients.

Additional Patient Symptoms



CMT1A patients that suffer from mobility impairment over index for other ancillary disease symptoms such as fatigue (92%), pain (78%) and frequent falls (59%). Physical activity can be greatly curtailed.

Surgeries



Almost 54% of CMT1A patients have had some type of orthopedic surgery to maintain or regain function lost because of the expression of their disease.

Although surgery can have significant benefits for CMT1A patients, its impact on QoL can be significant in the short to medium term due to risk of anesthesia and complications, long recovery times, physical therapy, loss of productivity, and the cost burden borne by patients and their families.

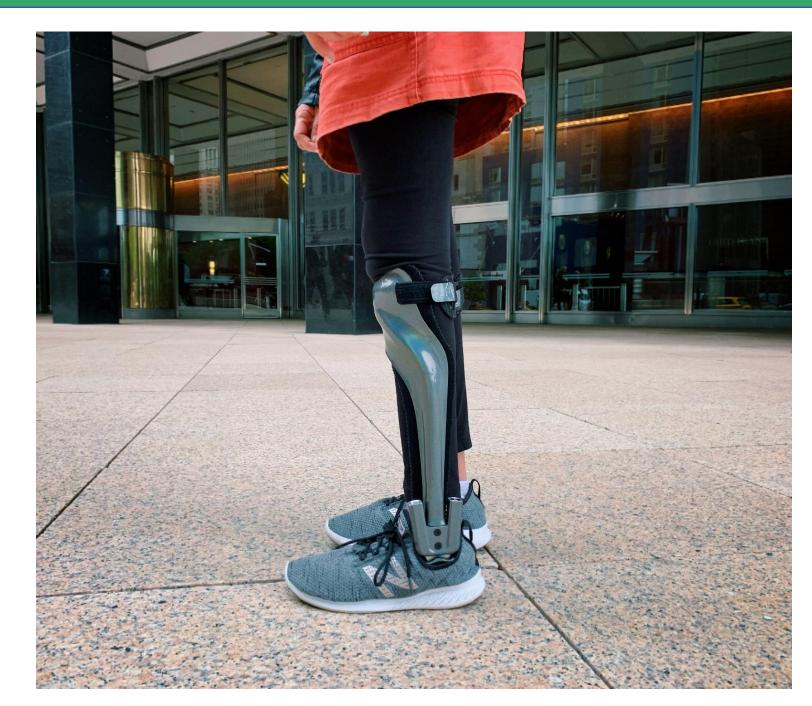
Voice Survey - Impact of Disease



Using Voice Activation Technology (VAT) powered by True Reply, CMT1A patients were asked to name the one symptom of their disease that has the biggest impact on QoL.

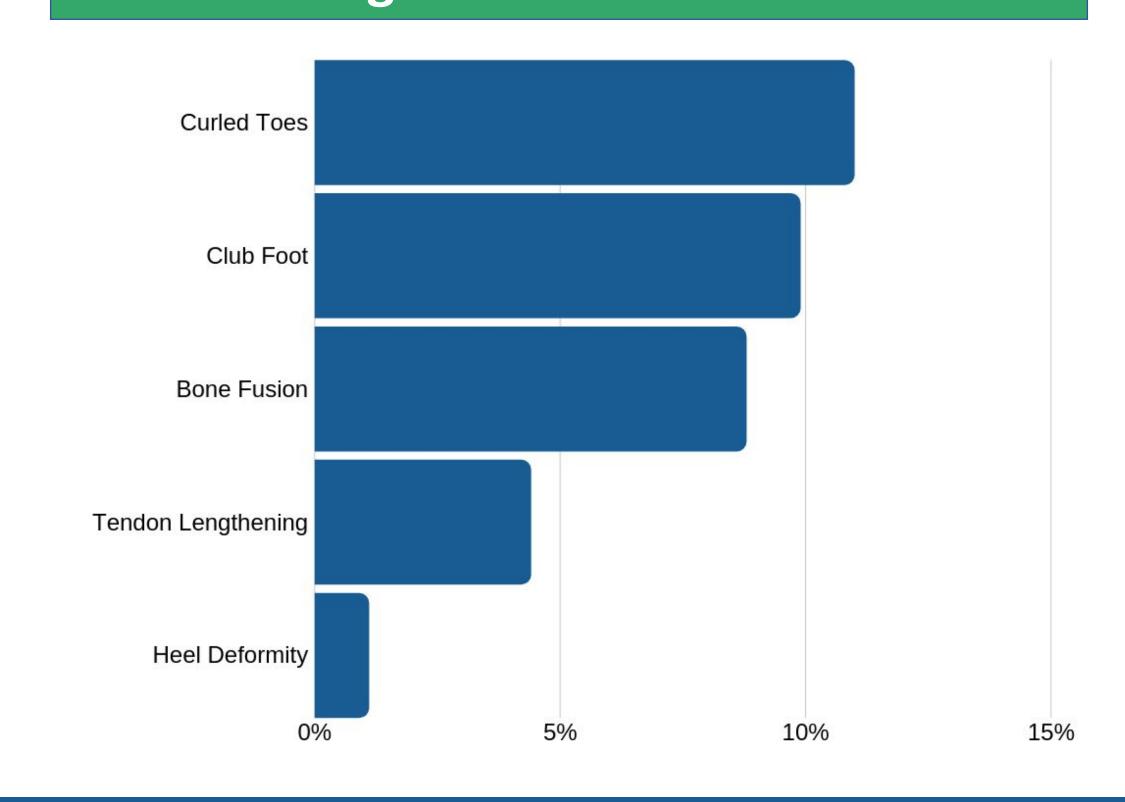
Keyword density was noted around words like walk, balance, feet, pain and fatigue, supporting natural history collected by GRIN regarding patient mobility issues.

Neuroswing Leg Braces



31% of CMT1A patients use leg braces or orthotics to assist with mobility.

Foot Surgeries in CMT1A Patients



Conclusions

Patient mobility was the #1 issue for patients with CMT1A; it has a significant impact on multiple aspects of a patient's QoL.

Patient phenotype, like curled toes, pes cavus feet and muscle atrophy, contributes directly to impaired mobility. Patients that do not opt for corrective surgery use multiple modalities to assist with mobility, with bracing and orthotics used by a large cohort of patients.

Pain and fatigue are also a serious by-product of patient mobility issues, along with frequent falls.

Contact:
Allison Moore
Founder & CEO
Hereditary Neuropathy Foundation
Allison@HNF-Cure.org

www.hnf-cure.org

Contact:
Robert Moore
Founder & CMO
True Reply, LLC
Robert@TrueReply.com