Preliminary Results for Charcot-Marie-Tooth Patient-Reported Survey

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Background

- Charcot-Marie-Tooth (CMT) disease is described as an inherited neuropathy affecting the peripheral nerves and resulting in distal muscle weakness.
- CMT affects roughly 1 in 2,500 individuals in the general population and about 125,000 individuals in the US.
- Limited studies detail patient-reported impact of muscle weakness on daily activities.

Methods

- This anonymous survey was developed with input from clinical experts, the Hereditary Neuropathy Foundation (HNF), and patient interviews.
- The 44 questions were designed to assess patient characteristics (23), disease impact (12), and clinical trial participation (9).
- The online survey was offered to all HNF CMT community support contact databases totaling over 4,000 individuals.
- As of 22 May 2017, there were 1,137 surveys completed by 1,068 patients and 69 caregivers/friends.
- Patient-only responses (n=1,068) collected from 17 February 2017 to 22 May 2017 are presented here.

Patient Characteristics and Population

- The majority of patients were female (60%) and from the US (79%).
- 75% of patients knew their CMT subtype.
- 59% of patients had been genetically tested for CMT.
- Of patients with known disease type, the most common diagnoses were CMT1 (62%) and CMT2 (21%). (Fig 1)
- Mean age of symptom onset was 21 yrs, compared with mean age of diagnosis of 35 yrs. (Table 1, by CMT type)
- 57% of patients had bilateral disease presentation.

Disease Challenges and Quality of Life

- "Pain" (19%) and "weakness" (8%) were the single words that most often "came to mind when patients think of CMT." (Fig 3)
- Physical factors that affected patients’ quality of life "very much" were problems with balance (67%), foot drop (64%), and fatigue (52%). (Table 2)
- Psychosocial factors that affected patients’ quality of life "very much" were "People’s lack of understanding" (35%), and "Change in appearance/body image" (27%). (Table 2)

Healthcare Utilization

- 658 patients (61%) reported using at least one of the following supplemental therapies: vitamin supplements (62%), pain medication (36%), anti-inflammatory medication (39%), and herbal supplements (44%).
- 22% of patients took depression medication and 16% took anxiety medication.
- Most patients (79%) used an assistive device for ambulation, including ankle-foot orthotics (40%), cane/walking sticks (33%), and custom foot orthotics (24%).

Clinical Trial Participation

- Top motivators for patients to participate in clinical trials included "potential that the treatment and study will help others some day" and "idea of advancing science." (Table 3)

Conclusions

- An observational study was the most common type of clinical trial that patients had participated in previously (71%). (Fig 6)
- Most patients were interested in participating in future clinical trials (96%), with participation in an exercise/physical therapy trial being the most preferred (68%). (Fig 6)

Disease Signs and Symptoms

- The most frequently reported physical/clinical manifestations due to CMT were problems with balance (89%), foot drop (84%), loss of or abnormal sensation in the lower leg/foot (80%), and hand muscle weakness (80%). (Fig 2)

Ankle Weakness / Foot Drop

- The 3 most common challenges associated with foot drop were maintaining balance (80%), walking long distances (77%), and climbing up and down stairs (73%). (Fig 4)
- Walking long distances (31%) was reported as the primary challenge of foot drop. (Fig 4)

References