



# 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<sup>1</sup>
- CMT affects roughly 1 in 2,500 individuals in the general population and about 125,000 individuals in the US<sup>2</sup>
- Limited studies detail patient-reported impact of muscle weakness on functional activities

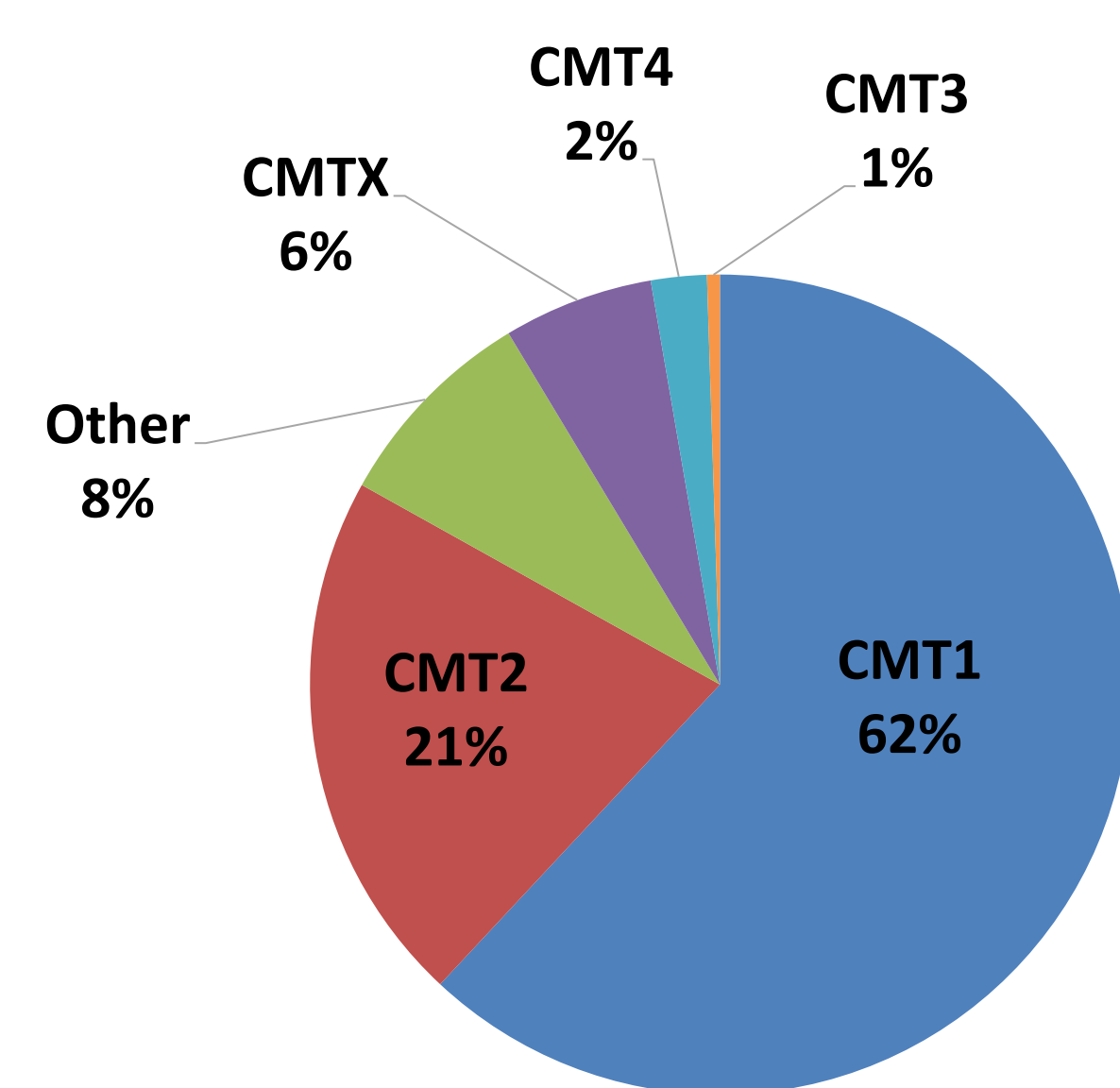
## Methods

- 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 the HNF patient contact database of 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

- A majority of patients were female (66%) 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

Figure 1: Patient-Reported CMT Type (n=591)



\* "Other" did not fit any of the included categories.

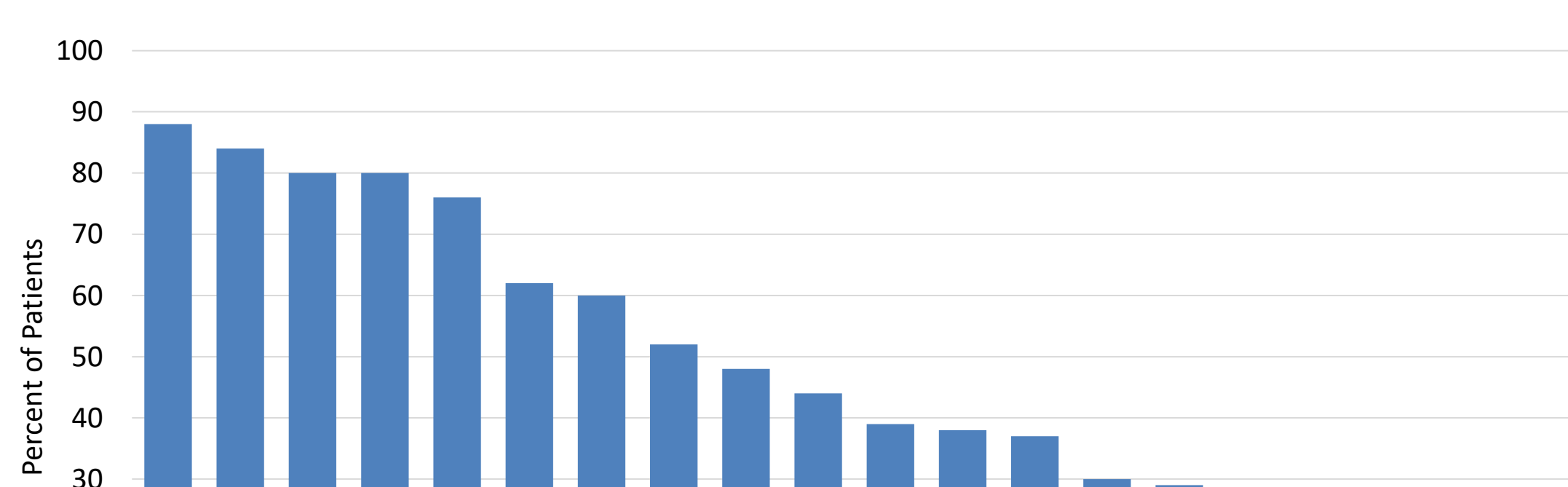
Table 1: Age of Onset and Diagnosis

Mean (SD) Age, yr	CMT1 (n=361)	CMT2 (n=125)	CMT3 (n=3)	CMT4 (n=13)	CMTX (n=33)
Symptom onset	16 (16)	24 (20)	19 (29)	23 (19)	18 (11)
Diagnosis	31 (17)	37 (19)	24 (27)	32 (20)	28 (13)
Current	52 (13)	54 (14)	49 (16)	51 (14)	48 (12)

## Disease Signs and Symptoms

- The most frequently reported physical/clinical manifestations due to CMT were problems with balance (88%), foot drop (84%), loss of or abnormal sensation in the lower leg/foot (80%), and hand muscle weakness (80%) (Fig 2)

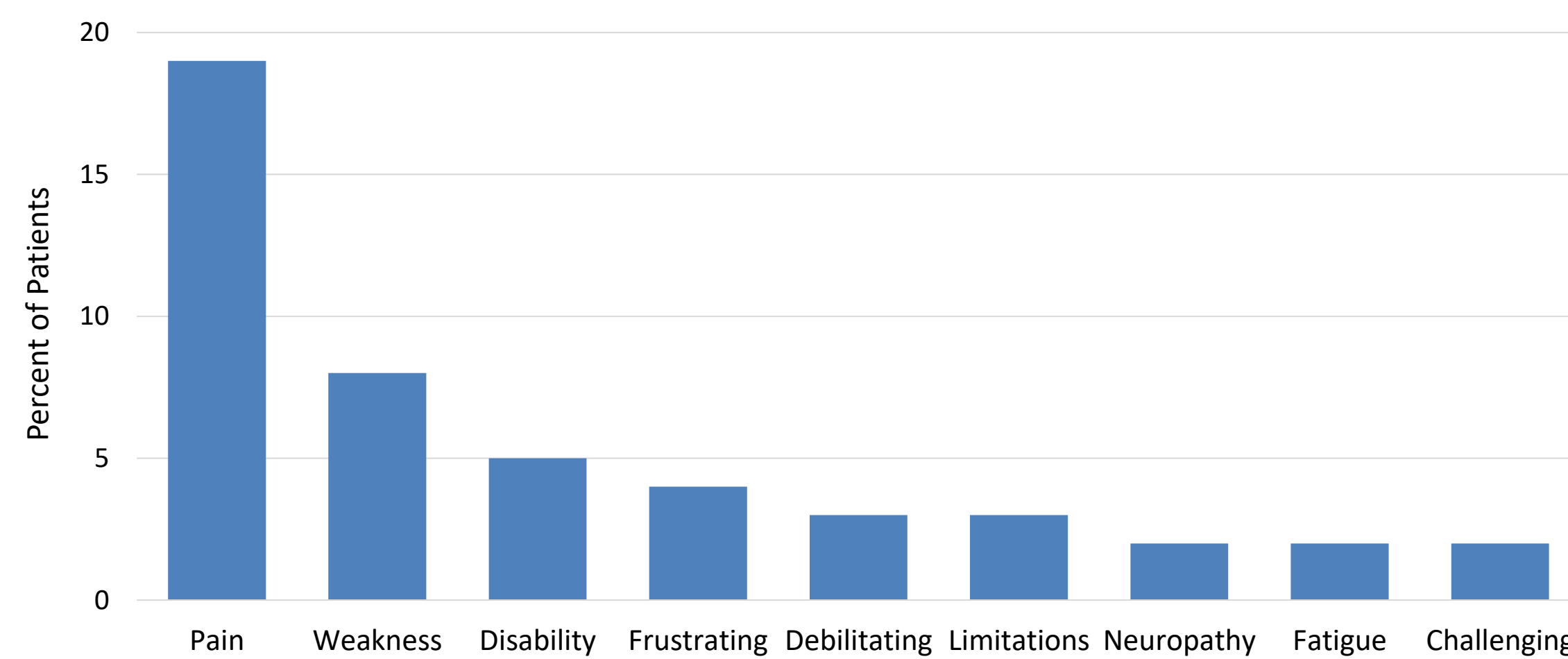
Figure 2: CMT Physical/Clinical Manifestations (n=1068)



## Disease Challenges and Quality of Life

- "Pain" (19%) and "weakness" (8%) were the single words that most often "come to mind when patients think of CMT" (Fig 3)

Figure 3: CMT Word Association (n=1018)



- 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" (31%) (Table 2)

Table 2: Factors Affecting Patient Quality of Life (%) (n=1068)

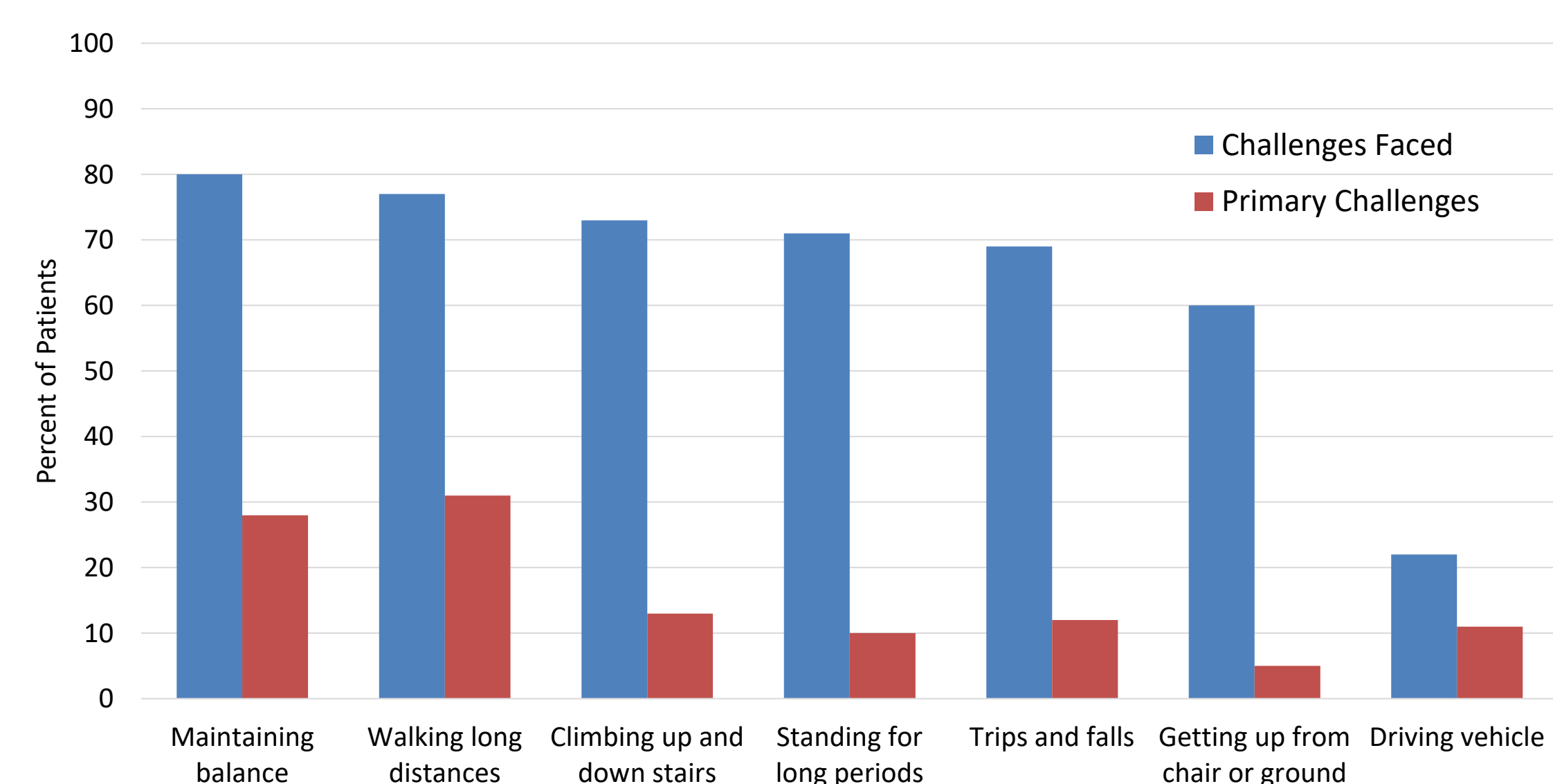
Factors	Very Much	Moderate	Slightly	Not at all
Problems with balance	67	20	11	2
Ankle weakness (foot drop)	64	24	8	4
Fatigue	52	28	13	6
Abnormal sensation in lower leg/foot	44	30	18	8
Foot contractures/deformities	37	26	20	18
Pain in lower leg/foot	35	30	17	18
People's lack of understanding	35	28	22	15
Hand muscle weakness	31	33	27	9
Change in appearance/body image	27	27	29	16
Awkward social interactions	26	26	28	20
Muscle cramps or spasms in lower leg/foot	24	28	13	6
Anxiety/fear	24	27	30	20
Pain in back	24	27	27	22
Depression	20	24	30	26
Sleeping issues	19	23	24	34
Abnormal sensation in hand/forearm	16	23	31	31
Hand contractures/deformities	14	18	24	44
Forearm muscle weakness	13	25	38	25
Pain in hand/forearm	13	22	29	36
Keeping my CMT a secret from others	11	14	16	58
Muscle cramps or spasms in hand/forearm	9	17	36	37
Tremor	8	17	28	48
Carpal tunnel syndrome	8	15	22	55
Respiratory/breathing issues	7	12	25	56
Curvature of spine (scoliosis)	6	12	20	63
Hip dysplasia	4	7	18	70

Note: Not all subjects responded for each quality of life factor

## 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)

Figure 4: Challenges due to Ankle Weakness/Foot Drop (n=1068)



- Patients reported frequent near falls (mean 19.8/month) and falls to ground (mean 2.5/month)
- The primary physical factor contributing to patients' near falls and falls was ankle weakness/foot drop (Fig 5)

Figure 5: Primary Physical Factors that Contribute to Patients' Near Falls and Falls (n=1068)



## Healthcare Utilization

- 658 patients (61%) reported at least one of the following supplemental therapies, of which included vitamin supplements (61%), pain medication (55%), anti-inflammatory medication (39%), and herbal (14%)
- 22% of patients took depression medication and 16% took anxiety medication
- Most patients (75%) use an assistive device for ambulation including ankle-foot orthotics (40%), canes/walking sticks (33%), and custom foot orthotics (24%)

## Clinical Trial Participation

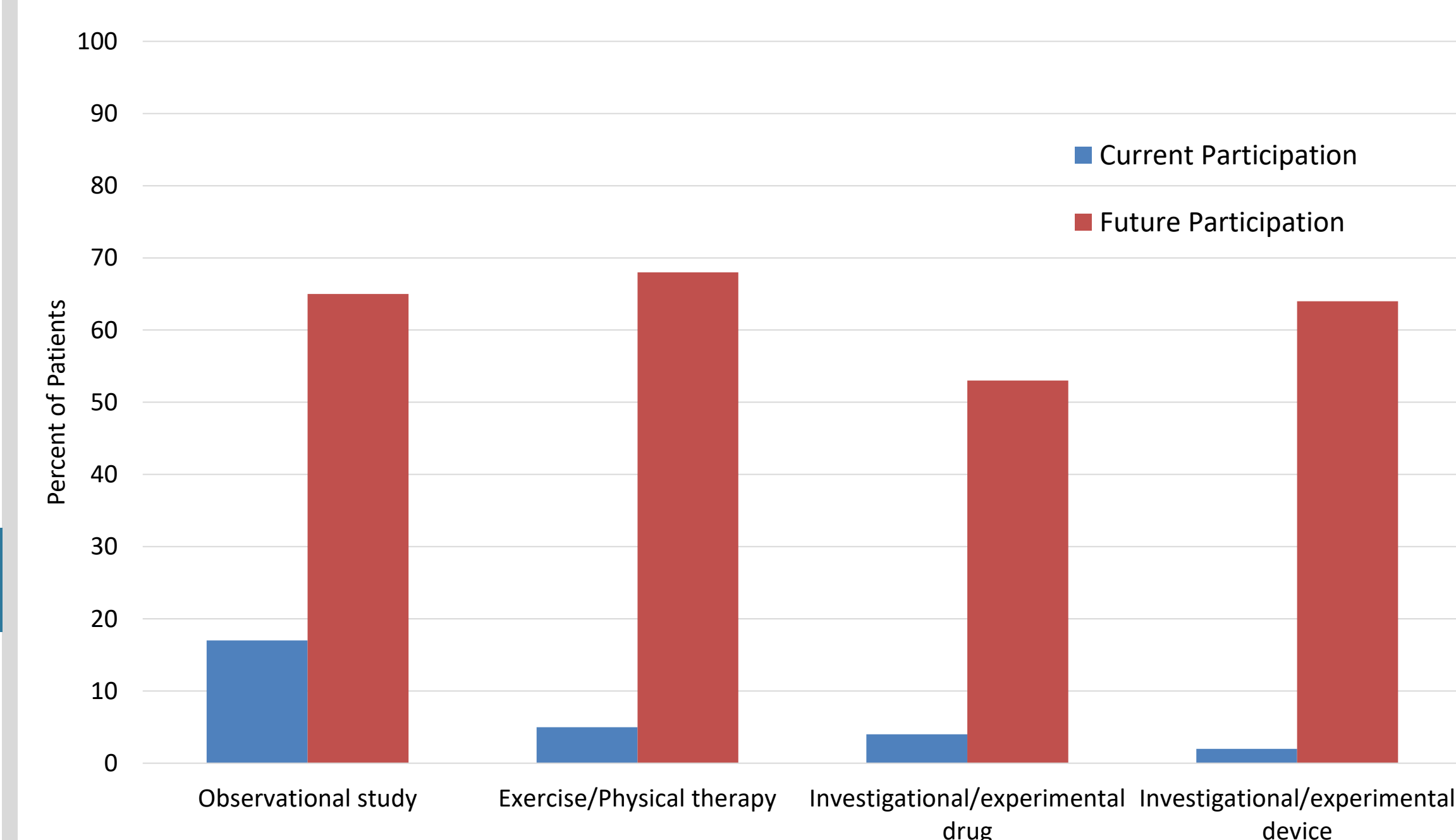
- Top motivators for patients to participate in clinical trials included "potential that the treatment and study will help others some day" and "idea of advancing science" (Table 3)

Table 3: Motivators for Clinical Trial Participation (n=1068)

The potential that the treatment and study will help others some day	80%
The idea of advancing science	64%
The feeling that I'm doing everything I can do to help	62%
Receiving my personal data results from the study/studies that I participate in	53%
The chance to be the first in line to try a treatment	44%
I am not interested in participating in a clinical study	6%
Other	4%

- An observational study was the most common type of clinical trial that patients had participated in previously (17%) (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)

Figure 6: Current and Potential Future Clinical Trial Participation (n= 1068)



## Conclusions

- The distribution of patient-reported CMT types was representative of the CMT population
- Mean age of respondents was 53 years
- Problems with balance and foot drop were the most common physical/clinical manifestations and most affected quality of life in patients with CMT
- Ankle weakness/foot drop was considered a primary factor contributing to near falls and falls
- "Pain" was a top-of-mind concern for many patients
- Most patients had not previously participated in a clinical trial, but demonstrated interest in participating in various clinical trials in the future
- These results support development of therapies to treat foot drop in patients with CMT

## References

- Carter GT, Jensen MP, Galer BS, Kraft GH, Crabtree LD, Beardsley RM, Abresch RT, Bird TD. Neuropathic pain in Charcot-Marie-Tooth disease. Arch Phys Med