THE PATH TO DIAGNOSING CHARCOT MARIE TOOTH DISEASE: THE PATIENT EXPERIENCE

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Abstract

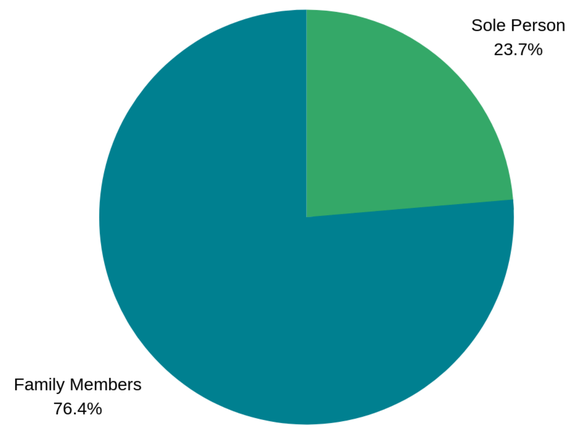
OBJECTIVES: Charcot-Marie-Tooth disease (CMT) is one of the most common inherited neurological disorders, affecting approximately 1 in 2,500 people worldwide. CMT comprises a group of disorders that affect peripheral nerves.
Charcot-Marie-Tooth (CMT) patients cite a long path to obtaining an accurate diagnosis of their disease, even with a family history of CMT. This study assessed the path to diagnosis experienced by CMT patients, considering family history of disease, initial presentation of symptoms, and length of time to obtain a diagnosis.

METHODS: Hereditary Neuropathy Foundation, in association with Hannah's Hope Fund, created the Global Registry for Inherited Neuropathy (GRIN), to capture detailed Inherited Neuropathy (IN) patient history via an online, IRB approved patient survey from 2013Q1-2019Q1. IN patients (N=2,195) engaged in an eight question survey regarding family history of CMT and diagnosis.

RESULTS: 76% of CMT patients are aware of having a family history. Once medical attention is sought, 59% of patients take over one year to get an accurate diagnosis of CMT, with 23% of patients taking five years or more to get diagnosed. 42% become aware of symptoms of age 15 years or younger, with 26% of patients being 30 years or older. 30% of patients were the first to notice their CMT symptoms, while 27% of patients were first identified by a healthcare practitioner (HCP). Neurologists were overwhelming identified as the HCP who first diagnosed CMT at 55%. Genetic testing was the leading method for obtaining an accurate diagnosis at 42%, with electrodiagnostic studies (EDX) next at 23%

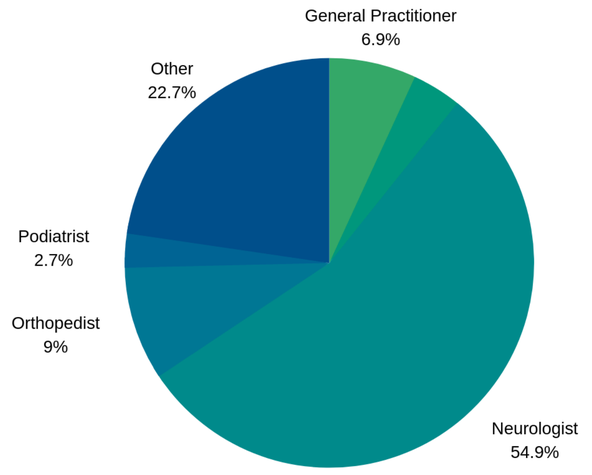
CONCLUSIONS: CMT patients can present symptoms early in life, yet it can still take several years to obtain a definitive diagnosis, even with a family history of the disease. While HCP's early identification of patient symptoms is sizably represented, given the large cohort of youthful manifestation of the disease coupled with the length of time it takes to obtain a definitive diagnosis, increase symptom awareness across the spectrum of HCP's is indicated.

Prevalence of CMT Within Families



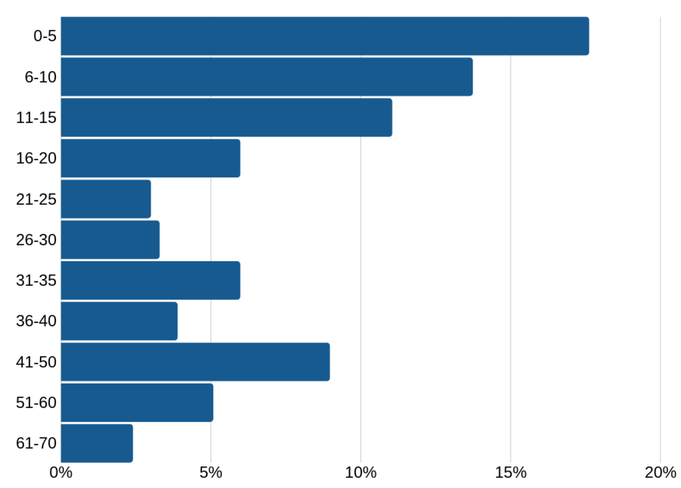
Over 76% of CMT patients have a history of the disease within their family. Individual diagnosis can occur due to spontaneous mutation. It can result in other previously undiagnosed family members also having the disease.

Diagnosis by HCP Practice



Neurologists diagnose CMT by a significant majority at 54.93%; orthopedists were the next largest cohort at almost 9%. Given the large number of patients diagnosed at a young age, pediatricians were under represented.

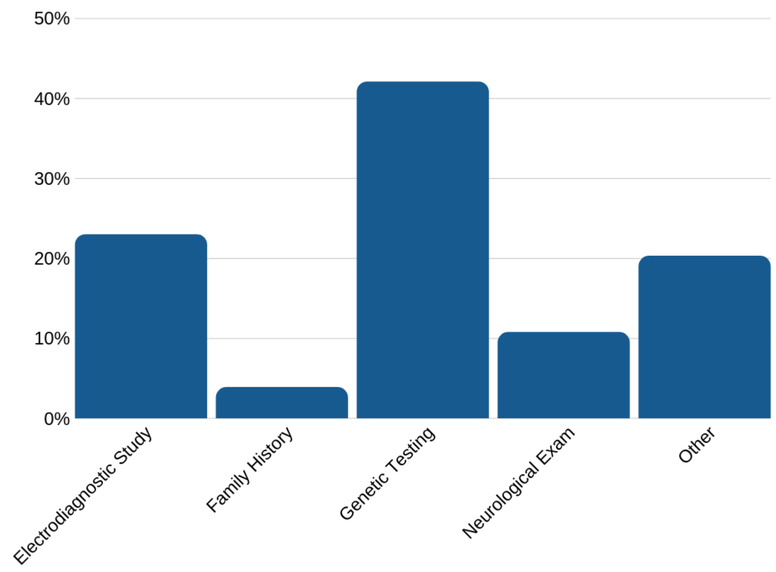
Age When CMT Symptoms Presented



CMT symptoms can present quite early in life, with the largest cohort found in the 0-5 age group at over 17%. By age 20, almost half (48.36%) of patients have started to manifest symptoms.

Another statistically significant cohort was found in patients ages 41-50 at almost 9%. Within this group, 43% took 3 or more years to obtain a confirmed diagnosis, vs. 30% of the patient populations as a whole, indicating that patients that manifest symptoms later in life experience a longer journey to obtain a confirmed diagnosis.

Testing Used to Confirm Diagnosis



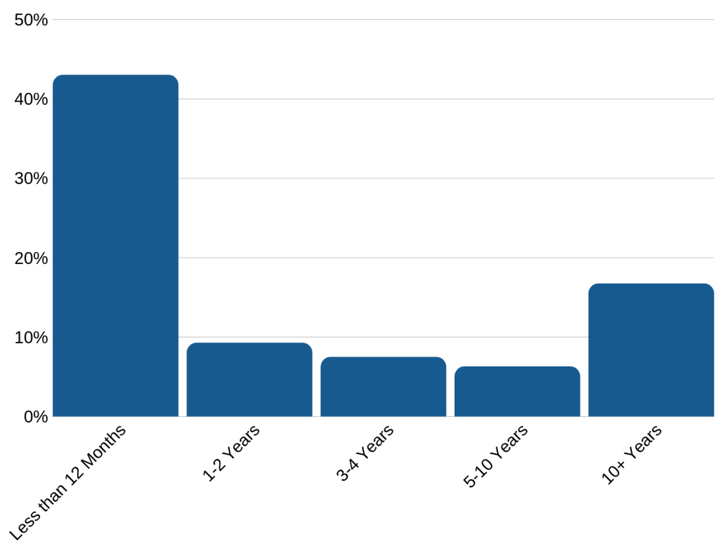
The majority of CMT patients received their confirmed diagnosis via genetic testing at just over 42%. Electrodiagnostic studies accounted for almost 23% of diagnosis.

Other methods reported by patients included biopsies, physical exams, and clinical observation. Cost of genetic testing was cited by patients as barrier to participation.

CMT Can Take Years to be Diagnosed

23% of CMT patients take over 5 years to get an accurate diagnosis.

Time Taken to Get Confirmed Diagnosis



Conclusions

Although many CMT patients obtain a confirmed diagnosis of their disease within 12 months of manifesting symptoms, a significant cohort of patients can take several years to obtain an accurate diagnosis. This is particularly prevalent in older patients that manifest symptoms later in life.

Given the significant patient population under 20 years of age that are diagnosed, pediatricians were underrepresented in our patient survey as the HCP that first recognized patient symptoms.

Although the majority of patients received their diagnosis via genetic testing, cost of testing was noted as a barrier.

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