# A Prospective Natural History and Outcome Measure Discovery Study of Charcot-Marie-Tooth Disease, Type 4J

NCT06151600

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| **Status** | RECRUITING |
| **Sponsor** | Elpida Therapeutics SPC |
| **Enrollment** | 20 participants |

## Key Eligibility Criteria

### Inclusion (5)

- Male or female, all ages
- A molecularly-confirmed diagnosis of CMT4J (confirmed by a CLIA certified, CE-marked, or equivalent lab): Genomic DNA mutation analysis demonstrating 1) bi-allelic pathogenic and/or likely pathogenic variants (by ACMG criteria) in the FIG4 gene, or 2) bi-allelic variants with one pathogenic and/or likely pathogenic variant in trans with a variant of uncertain significance if laboratory evidence and expert consensus exits in support of loss of FIG4 function exists.
- Informed consent from patients 18 years or older who are able to provide consent and from caregivers; parent(s)/guardian(s) providing consent for subjects younger than 18 years at Screening and patients older than 18 years unable to provide informed consent
- Informed assent of patients younger than 18 years at Screening who are able to provide assent
- Able and willing to comply with the study protocol, including travel to Study Center, procedures, measurements and visits

### Exclusion (6)

- Any known genetic abnormality, including chromosomal aberrations that confound the clinical phenotype
- Current participation in an interventional or therapeutic study
- Receiving an investigational drug within 90 days of the Baseline Visit
- Prior or current treatment with gene or stem cell therapy
- Any other diseases which may significantly interfere with the assessment of CMT4J

... and 1 more (see full listing online)

## Locations (3 total)

Stanford University, San Francisco, California, United States
University of Iowa, Iowa City, Iowa, United States
University of Texas Southwestern, Dallas, Texas, United States