## Open Peer Review on Qeios

## Charcot-Marie-Tooth Disease Type 2E

National Cancer Institute

## Source

*National Cancer Institute. <u>Charcot-Marie-Tooth Disease Type 2E</u>. NCI Thesaurus. Code C134953.* 

A form of Charcot-Marie-Tooth disease which is inherited in an autosomal dominant manner. It is caused by mutation(s) in the NEFL gene, encoding neurofilament light polypeptide. It results in peripheral axonal neuropathy.