# DISEASE:
# Autosomal dominant Charcot-Marie-Tooth disease type 2C

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| **NAME:** | Autosomal dominant Charcot-Marie-Tooth disease type 2C |
| **DESCRIPTION:** | A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by the association of vocal cord anomalies, impairment of respiratory muscles and sensorineural hearing loss with the distal hands and feet weakness. Onset is between infancy and the 6th decade. |
| **ORPHACODE:** | 99937 |
| **SYNONYMS:** | CMT2C |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | TRPV4 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Neuropathy (gene panel)

## Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

## Related Analytes

- transient receptor potential cation channel subfamily V member 4

## Related Gene Panels

- Neuropathy (148 genes) - IPG