# Charcot-Marie-Tooth disease

## Introduction

- Charcot-Marie-Tooth (CMT) affects the peripheral nerves.
- These are the nerves outside the main central nervous system (CNS). They control the muscles and relay data from the arms and legs to the brain, allowing a person to sense touch.
- Symptoms of Charcot-Marie-Tooth disease typically appear in adolescence or early adulthood, but may also develop in midlife

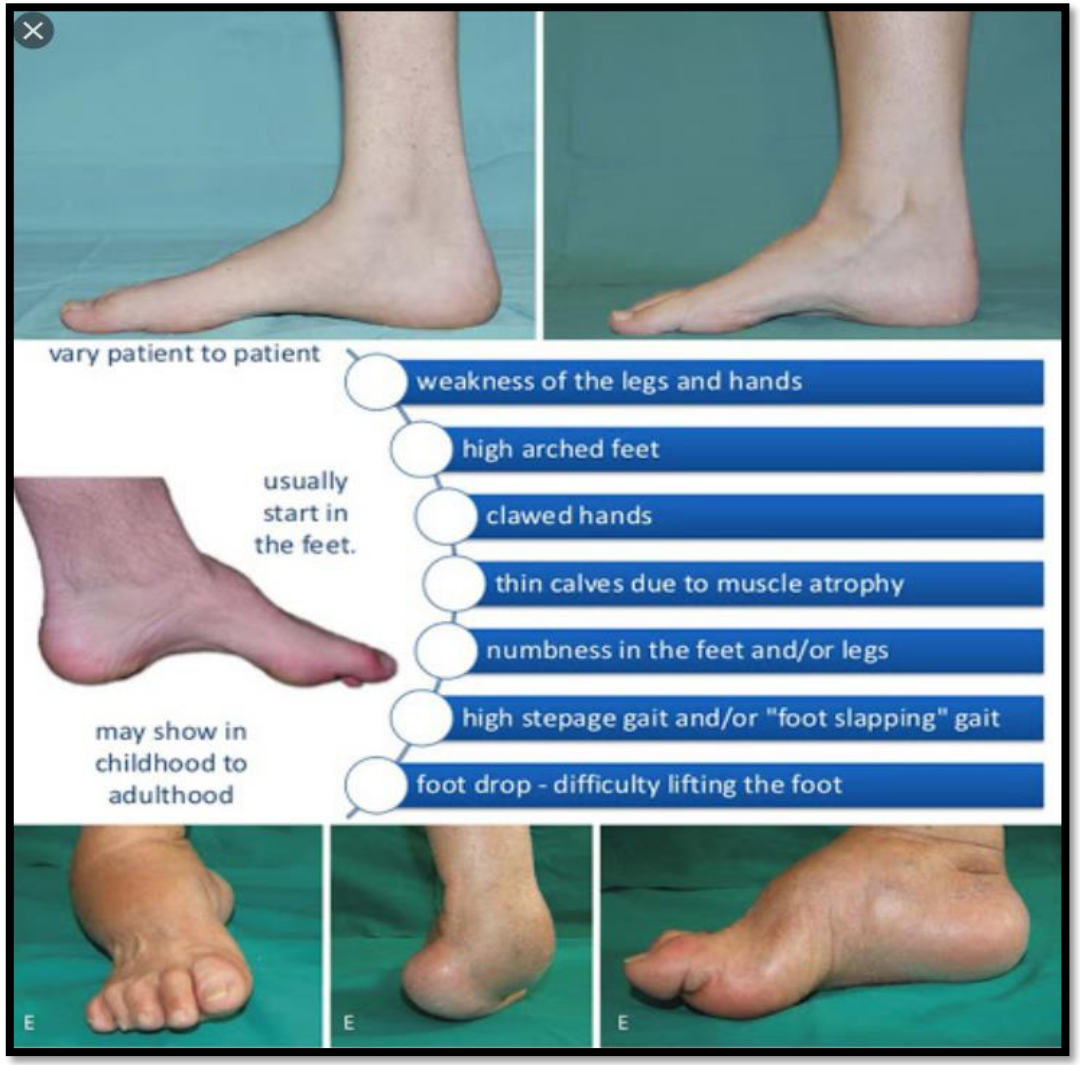
# Charcot-Marie-Tooth disease

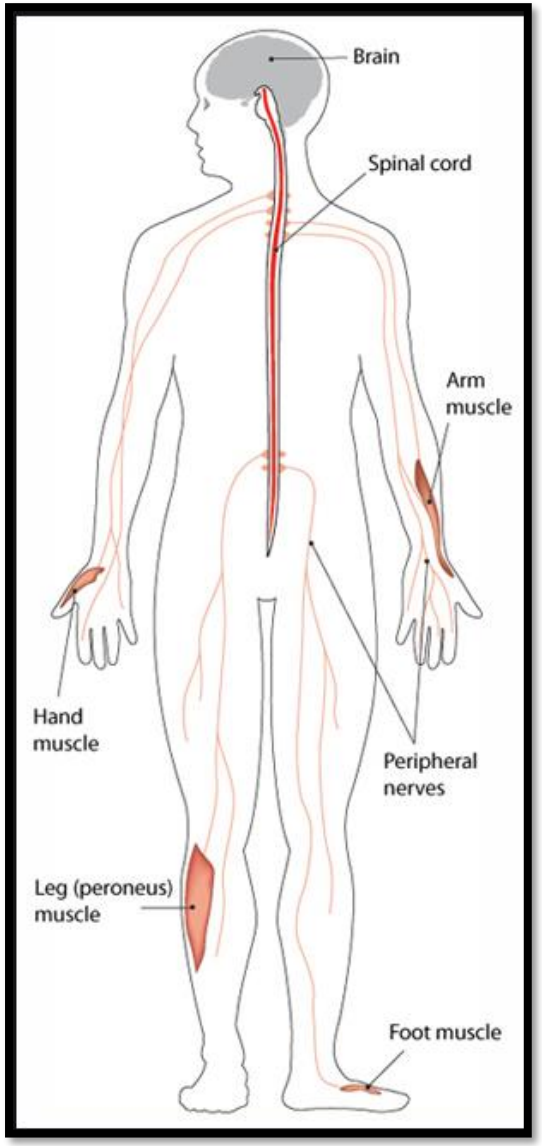
## Clinical manifestation /Symptoms

- Weakness in your legs, ankles and feet
- Loss of muscle bulk in your legs and feet
- High foot arches
- Curled toes (hammertoes)
- Decreased ability to run
- Difficulty lifting your foot at the ankle (footdrop)
- Awkward or higher than normal step (gait)
- Frequent tripping or falling
- Decreased sensation or a loss of feeling in your legs and feet

# Charcot-Marie-Tooth disease

## Clinical manifestation /Symptoms



# Charcot-Marie-Tooth disease

## Causes

❖Charcot-Marie-Tooth disease can be caused by mutations in many different genes.

❖These genes provide instructions for making proteins that are involved in the function of peripheral nerves in the feet, legs, and hands.

❖The gene mutations that cause Charcot-Marie-Tooth disease affect the function of the proteins in ways that are not fully understood; however, they likely impair axons, which transmit nerve impulses, or affect the specialized cells that produce myelin.

❖various types of Charcot-Marie-Tooth disease, subtypes (such as CMT1A, CMT1B, CMT2A, CMT4A, and CMTX1) indicate different genetic causes.

# Charcot-Marie-Tooth disease

## Inheritance Pattern

❖CMT1, most cases of CMT2, and most intermediate forms are inherited in an autosomal dominant pattern.

❖CMT4, a few CMT2 subtypes, and some intermediate forms are inherited in an autosomal recessive pattern.

❖CMTX is inherited in an X-linked dominant pattern.

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## Gene Panel

AARS, AIFM1, ATL1, ATP7A, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, ELP1, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SH3TC2, SLC12A6, SPTLC1, SPTLC2, TRIM2, TRPV4, TTR, WNK1, YARS

No of Genes : 59
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

# Diagnostic Tests

anderson
Clinical Genetics

# Management

# Duchenne muscular dystrophy

## Reference


https://rarediseases.info.nih.gov/diseases/6291/duchenne-muscular-dystrophy

https://ghr.nlm.nih.gov/gene/DMD

https://www.healthline.com/health/duchenne-muscular-dystrophy#treatment