Charcot Marie Tooth Disease

- Most common inherited neuromuscular disorder (15 per 100,000)
- Group of disorders referred to as hereditary motor and sensory neuropathy

- Type 1 = autosomal dominant, with nerve biopsy revealing “onion” hypertrophy
- Type 2 = autosomal recessive, presenting during adolescence
- Type X = X-linked and presents during childhood

- Slowly progressive but patients have a normal life expectancy, but 95% have characteristic foot involvement: weakness of tibialis anterior and peroneus brevis and a cavovarus foot.
  - Unopposed action of peroneus longus (by weak tibialis anterior) results in flexion of 1st ray → high arch / cavus
  - Unopposed tibialis posterior (by weak peroneus brevis) results in hindfoot cavus.
  - Weakness of tibialis anterior predisposes to a foot drop, with compensatory high stepping gait.

- Nerve conduction tests can confirm peripheral neuropathy. Check for family history – but nerve biopsy may clinch diagnosis.

- Treatment:
  - Stretching and strengthening with physiotherapy
  - AFO brace – reduces sprains and energy requirements of gait
  - Tall toe-box shoes
  - Surgery reserved for those with refractory disease progression, or inability to get foot to plantargrade position
    - TA lengthening
    - Peroneus longus to peroneus brevis transfer
    - Dorsiflexion osteotomy of 1st MT, or dorsiflexed fusion of 1st MTC joint
    - Lateral calcaneal slide
    - Ankle arthrodesis