Charcot Marie Tooth Syndrome

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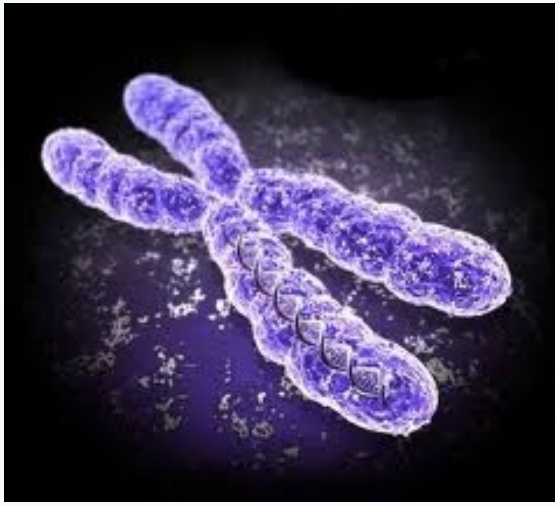
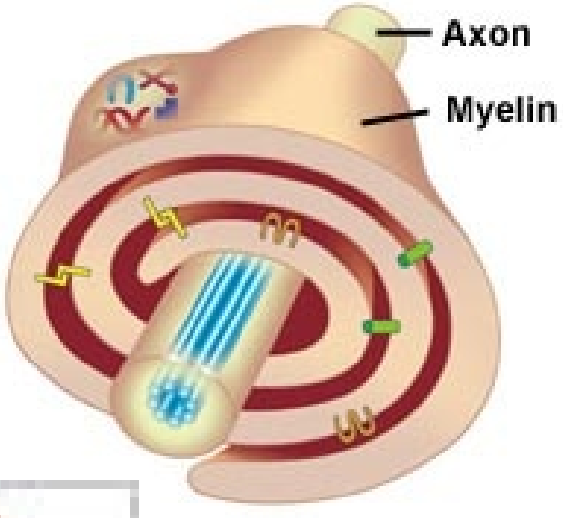
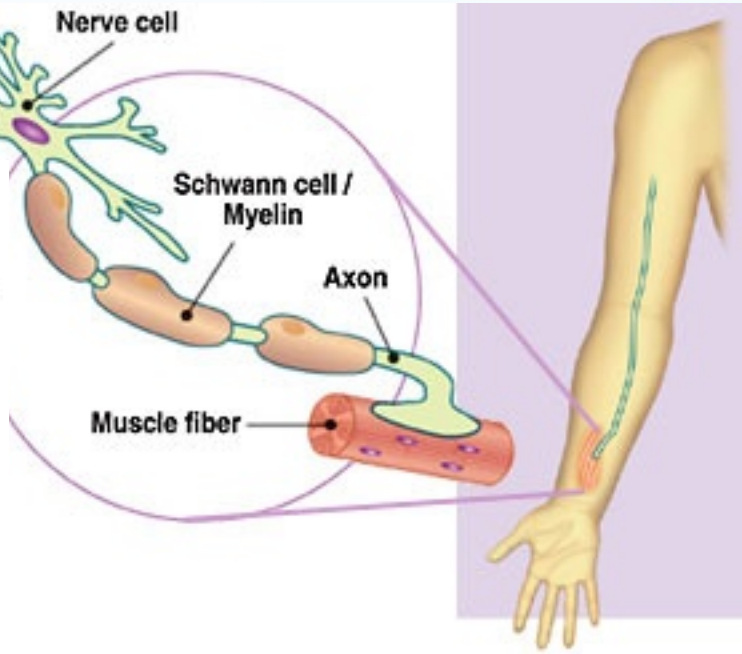
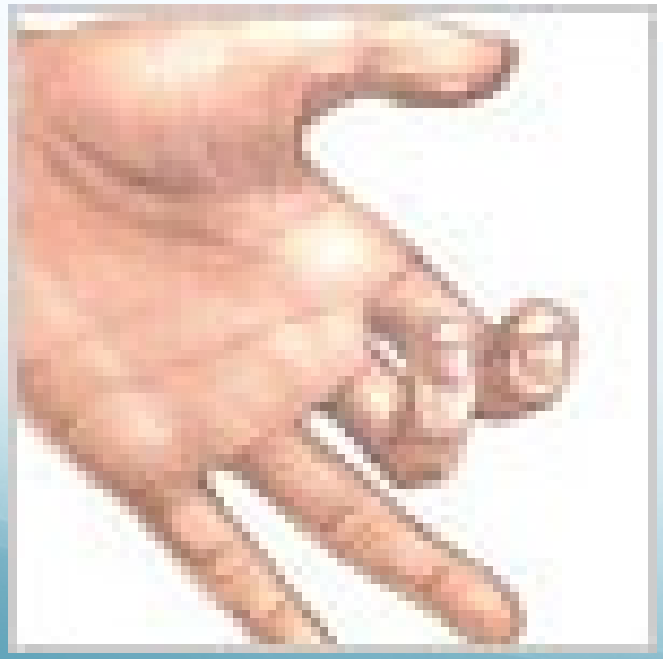
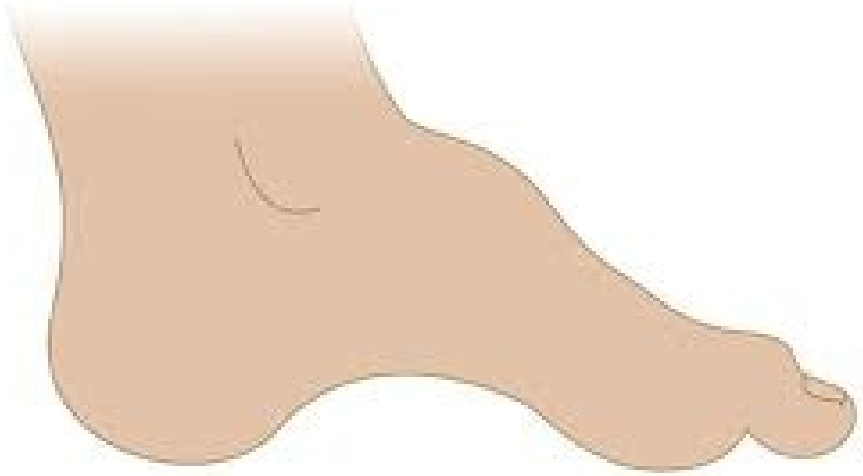
BIOCHEM 118




Axon
Myelin


Nerve cell
Schwann cell / Myelin
Axon
Muscle fiber



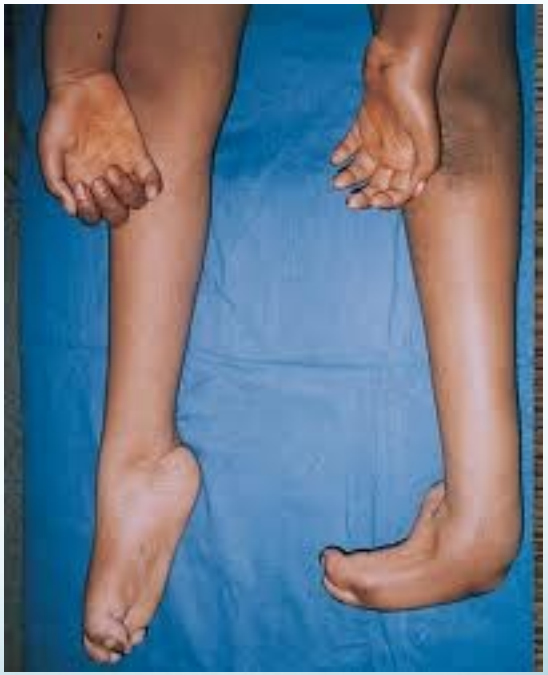


Charcot Marie Tooth Syndrome

- Discovered in 1886
- Several Types of CMT
- 70-80% CMT patients have Type 1
- Does not shorten lifespan
- Symptoms appear from age 5 to 25
- Causes muscle atrophy in hands and feet

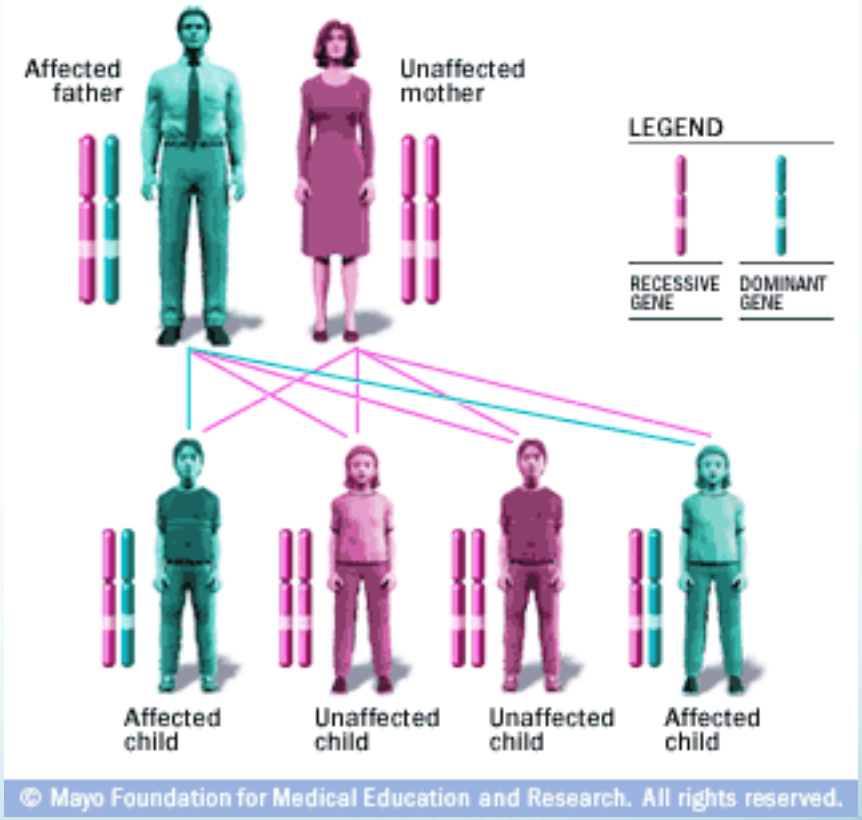
Diagnosis

- Progressive peripheral and motor neuropathy
- Slow nerve conduction
- Enlarged nerves, especially ulnar nerve
- Family history



Genetics

- Autosomal dominant
- Chromosome 17 mutation
- Genetic testing can identify syndrome
- Genetic counseling is advised



Treatment

- No cure yet
- Team of neurologists, orthopedic surgeons, psychical therapists, physiatrists.
- Special shoes and splints to aid walking
- Surgery as needed

Novel Testing

- New PCR Testing
  - duplication or deletion of gene
  - Breakpoint of misalignment in myelin
  - Amplifies breakpoint region of duplication or deletion
  - Quicker diagnosis

Novel Treatments

- Experimental Studies
  - Identify more causal genes for syndrome
  - Deliver genes to Schwann Cells
  - Vitamin C

http://www.ncbi.nlm.nih.gov/books/NBK1358/

http://omim.org/entry/118220

http://www.ncbi.nlm.nih.gov/books/NBK22241/

http://www.ncbi.nlm.nih.gov/pubmed/1044268