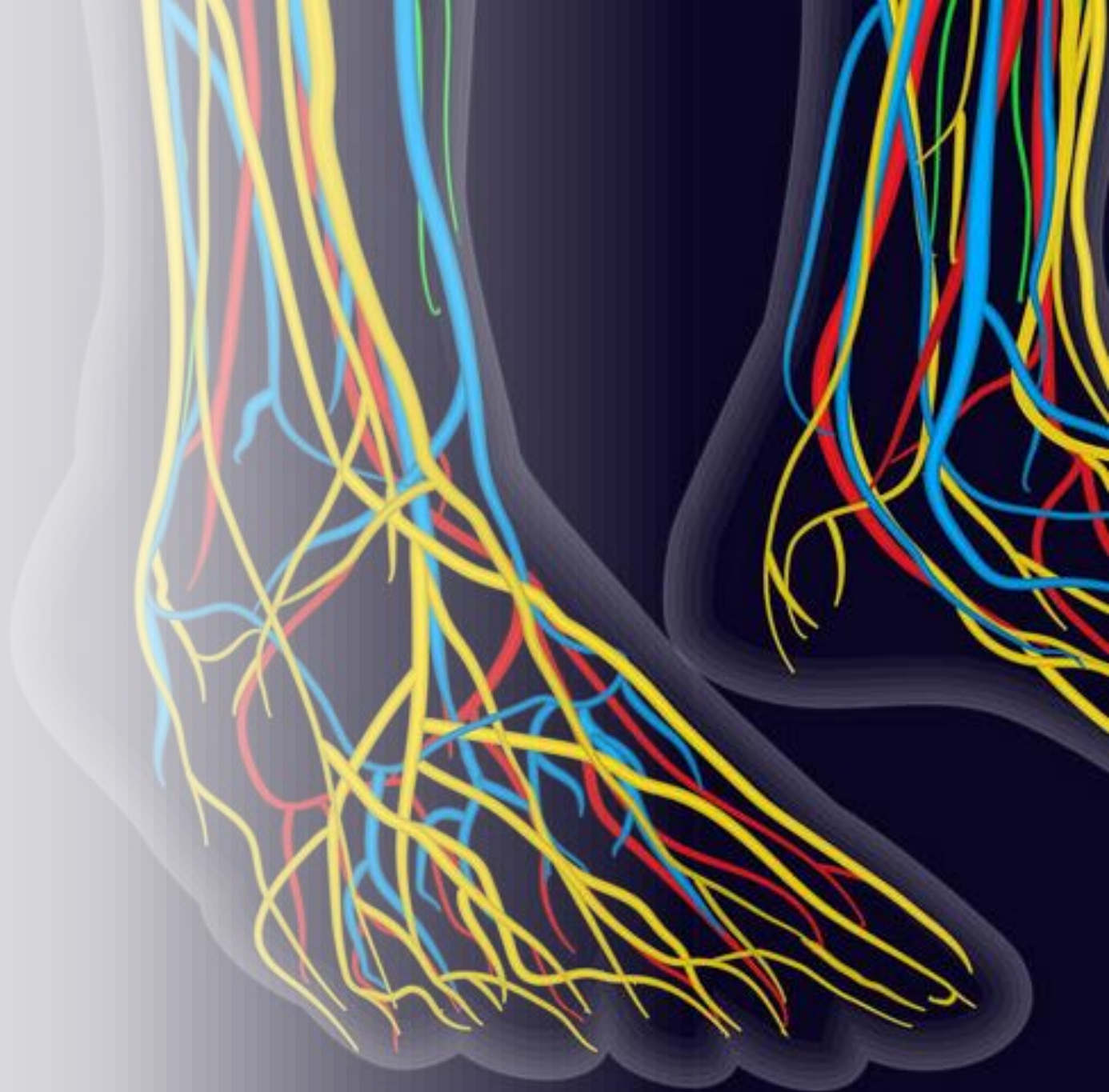




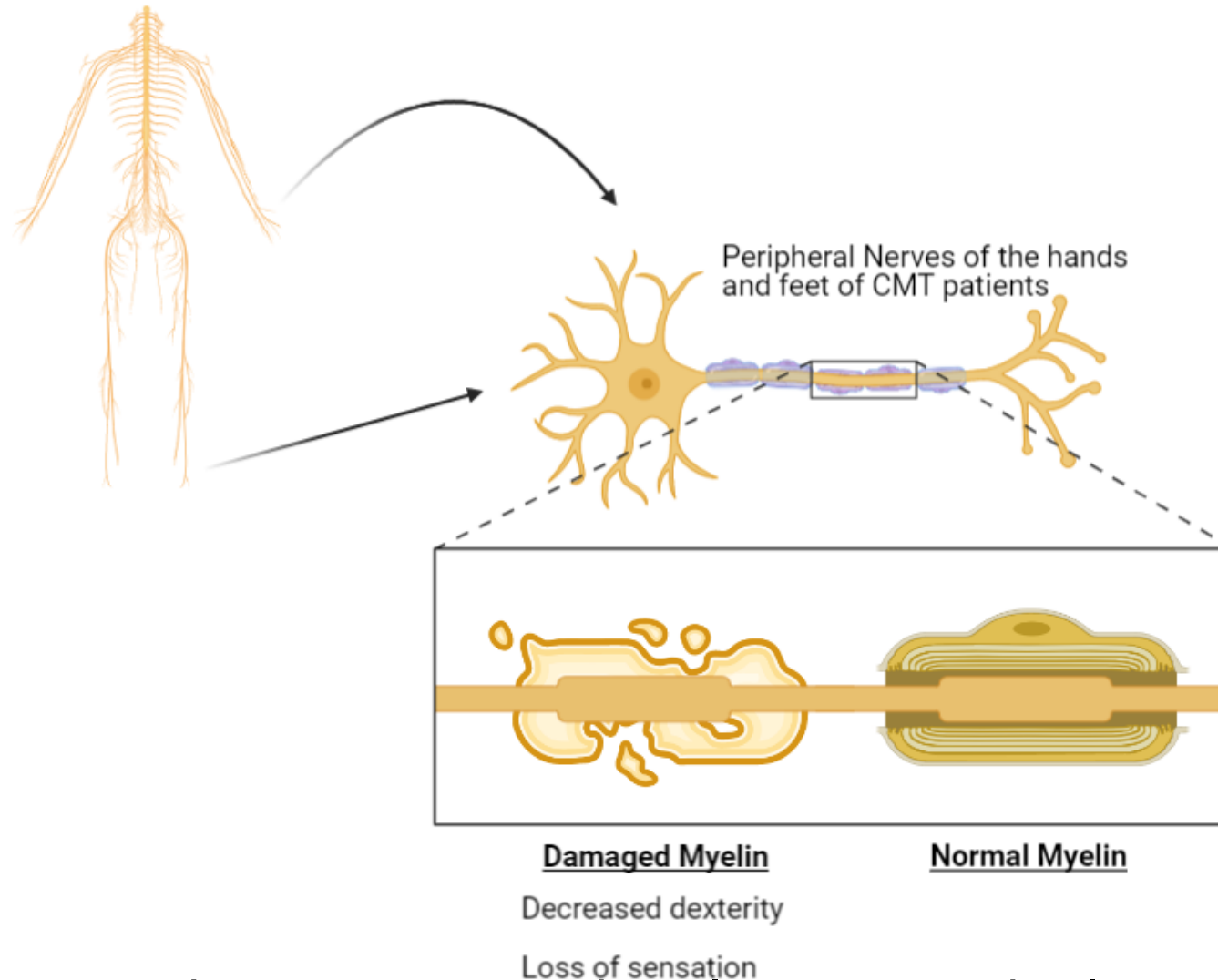
# Charcot-Marie-Tooth Disease

---

Collin Nguyen



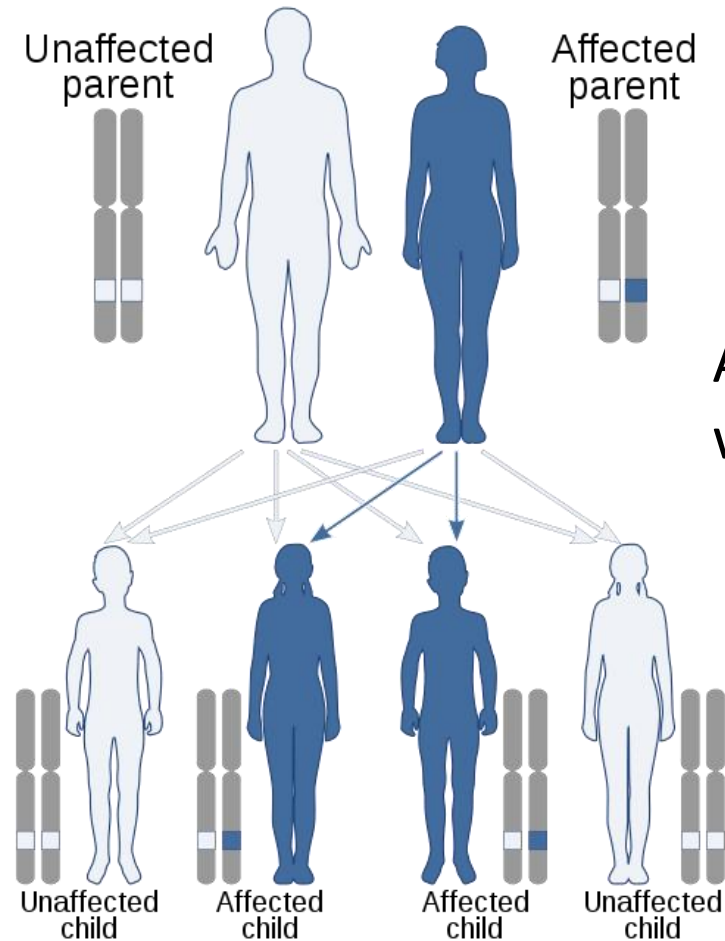
# What is Charcot-Marie-Tooth (CMT) Disease?



Results in nerve damage to peripheral nerves in the legs and arms resulting in an inability to properly use the muscles in these areas

# How is CMT inherited?

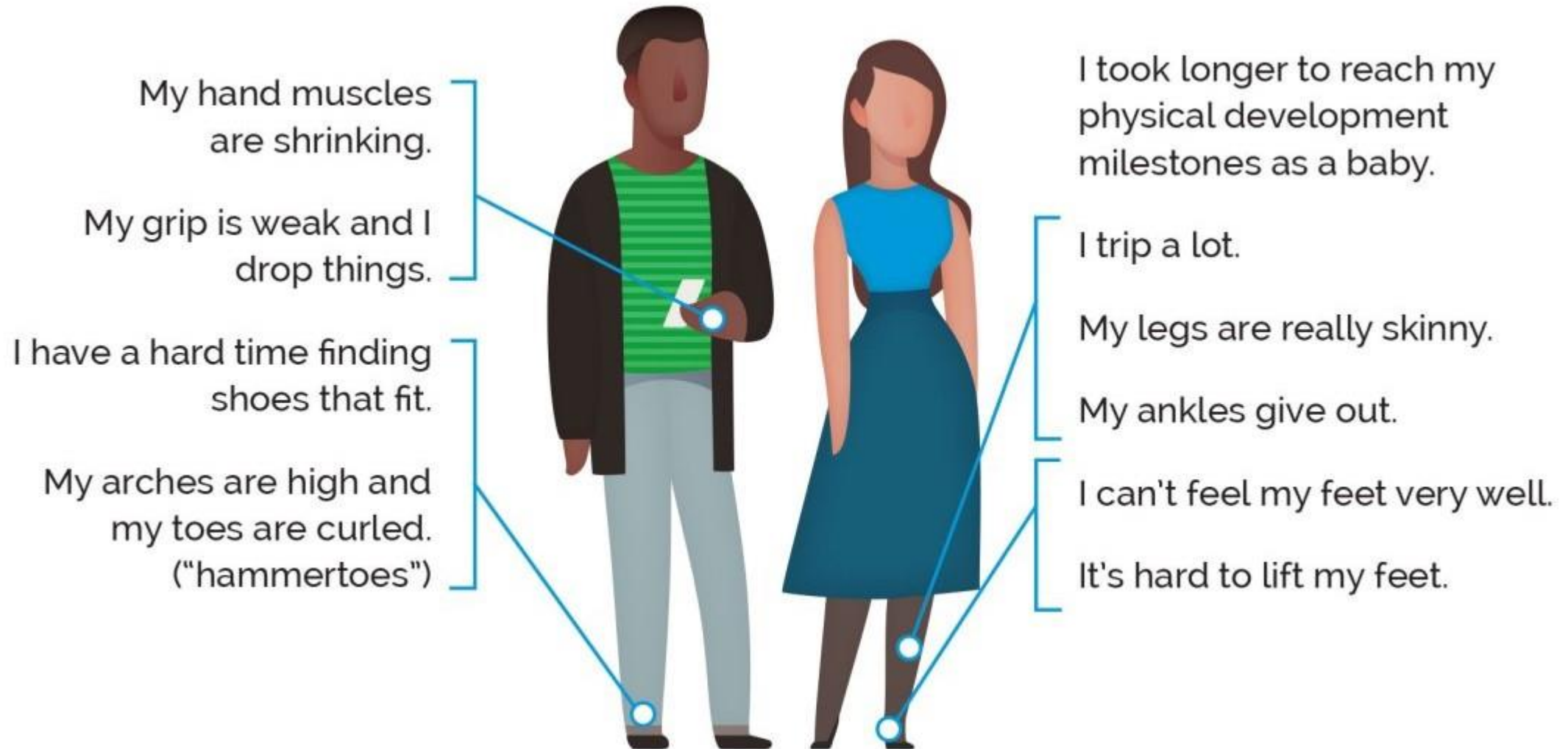
## Autosomal dominant



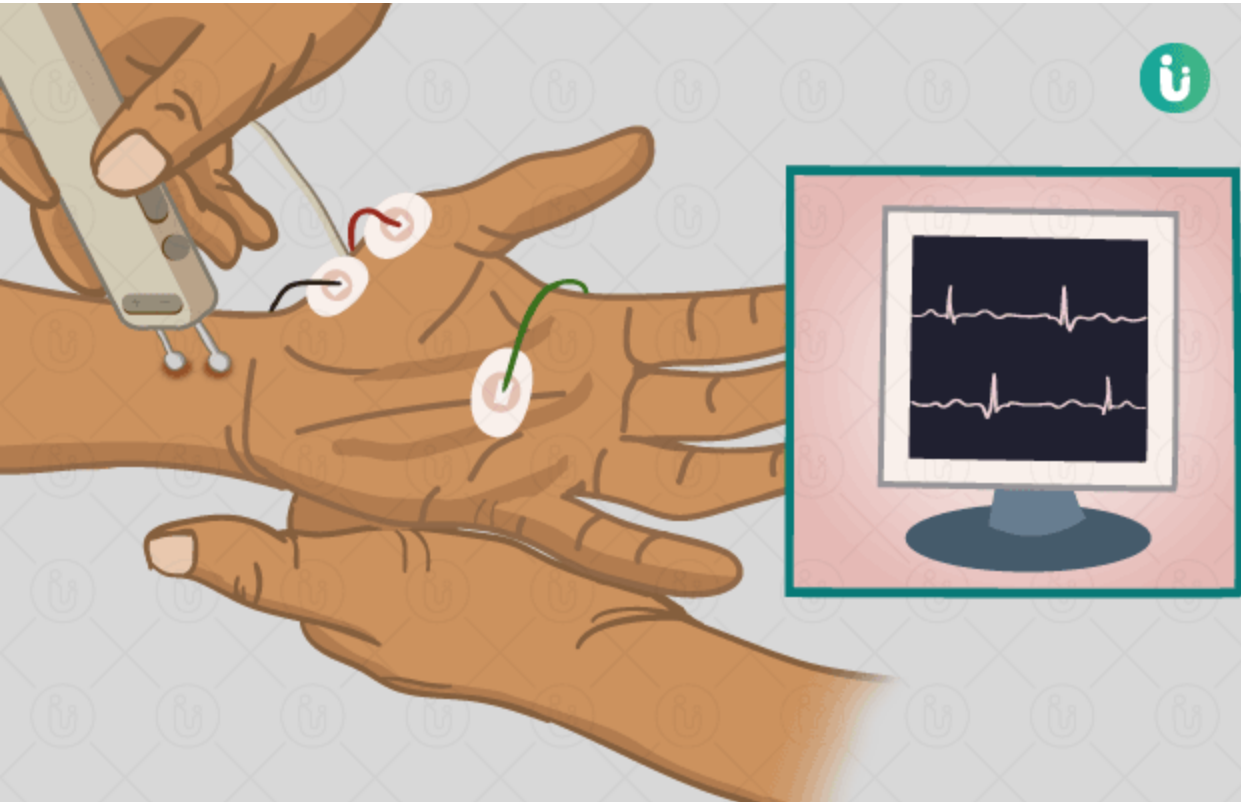
Autosomal dominant inheritance as well as de novo mutations

□ Unaffected  
■ Affected

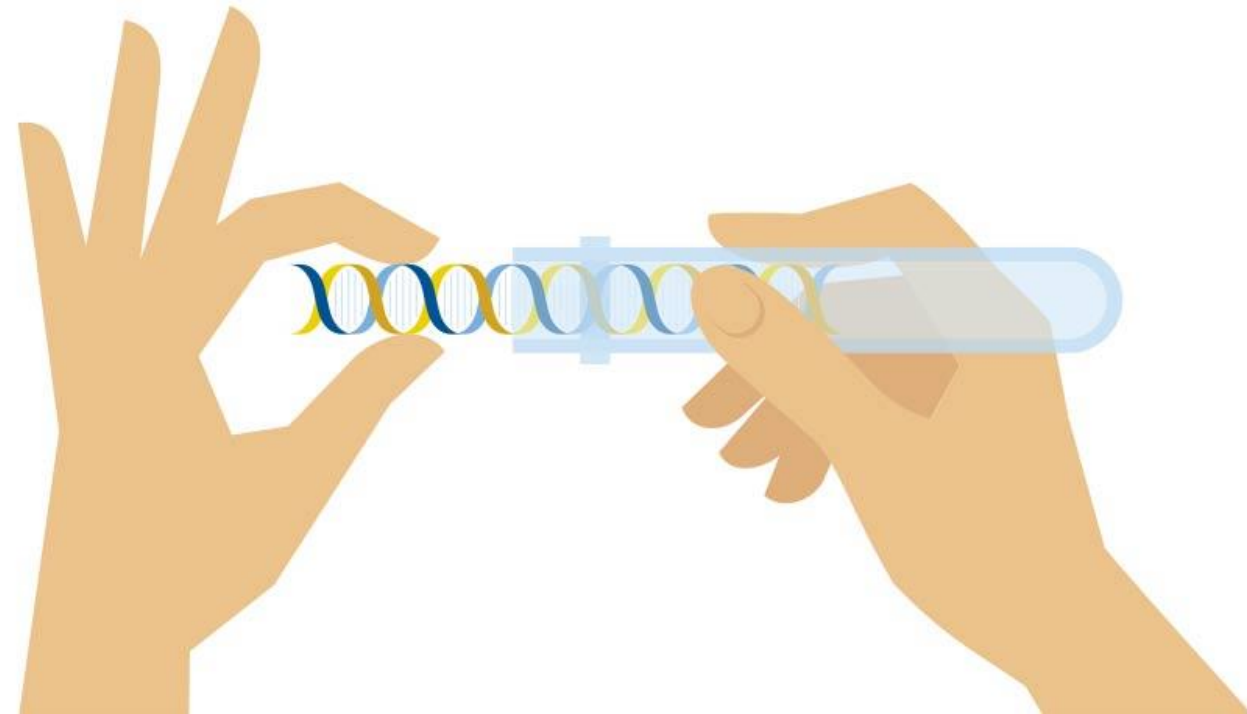
# What are the symptoms of CMT?



# How is CMT diagnosed and treated?



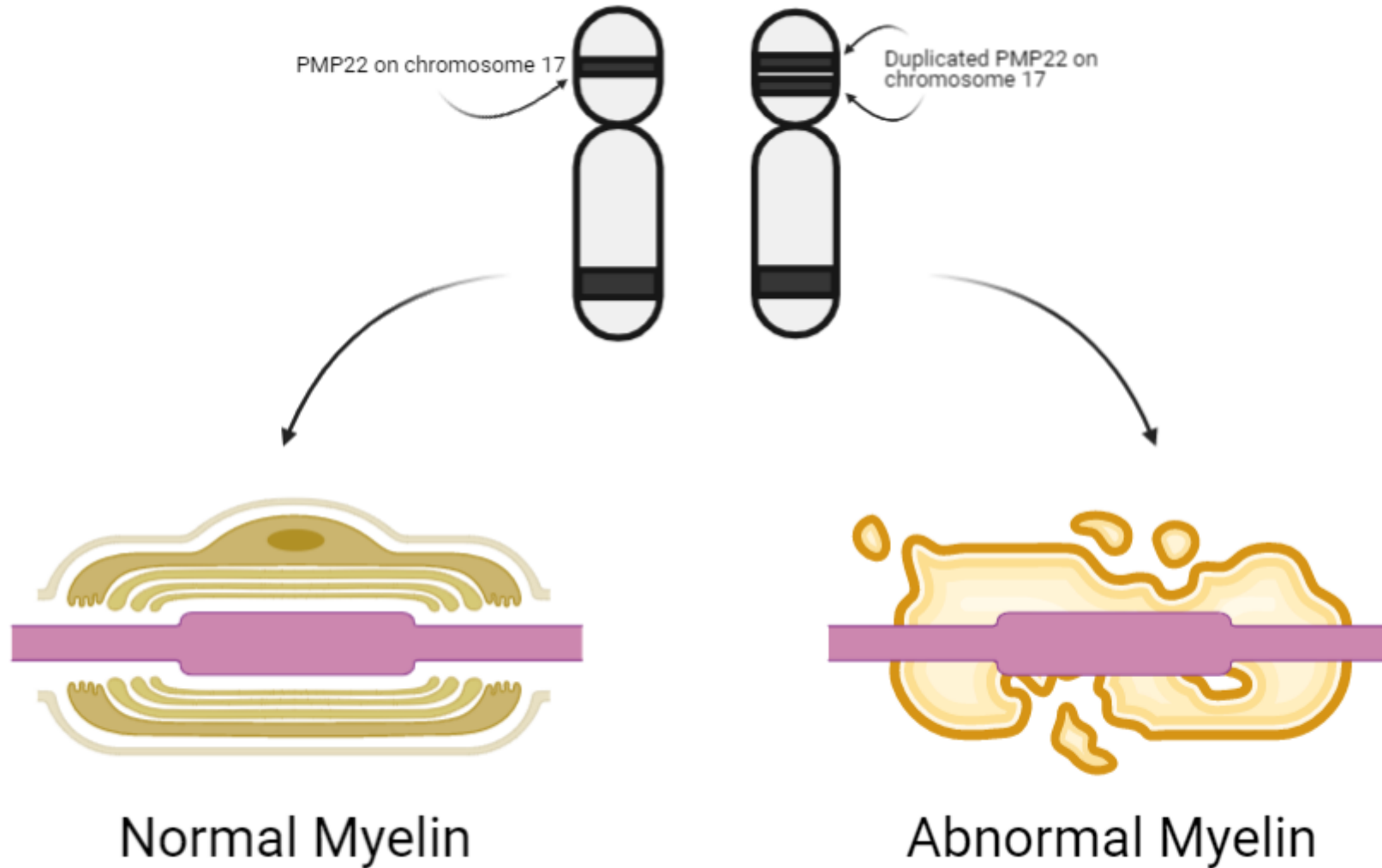
**Nerve Conduction Studies**



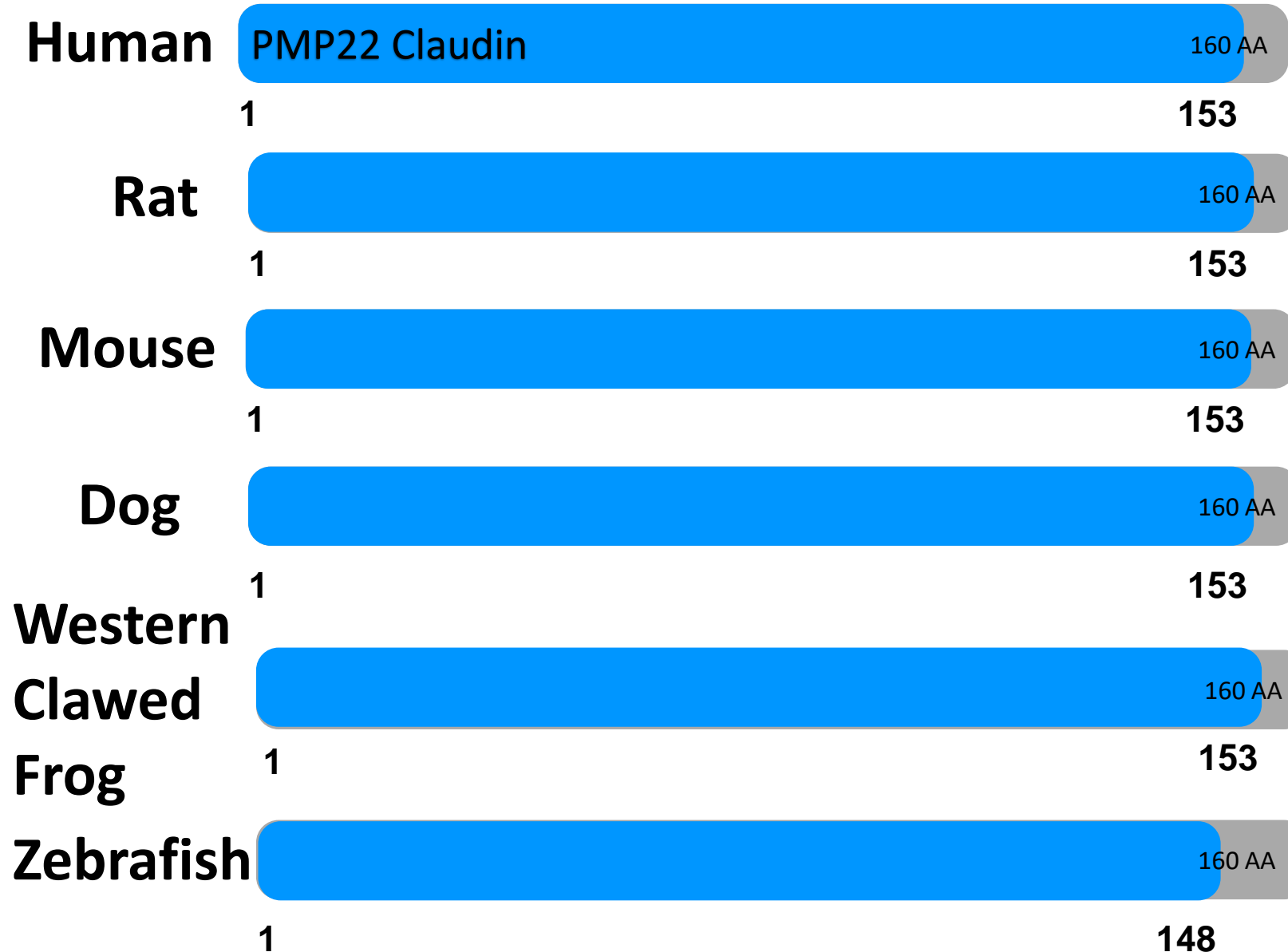
**Genetic Testing**



# What causes CMT1A?



# What domains are conserved in PMP22?



# CMT1A and PMP22

Human

PMP22 Claudin

160 AA

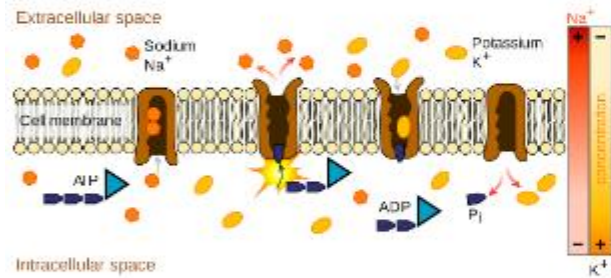
1

153

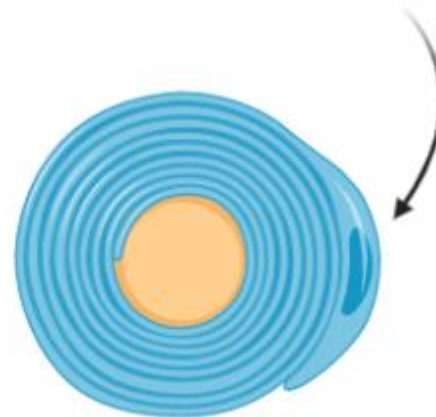
Molecular Function

Cellular Component

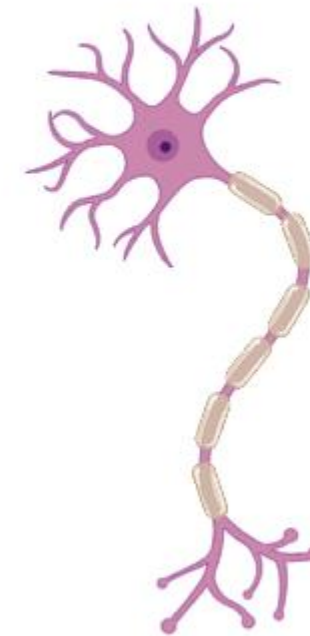
Biological Process



Nucleoside Triphosphatase Activity



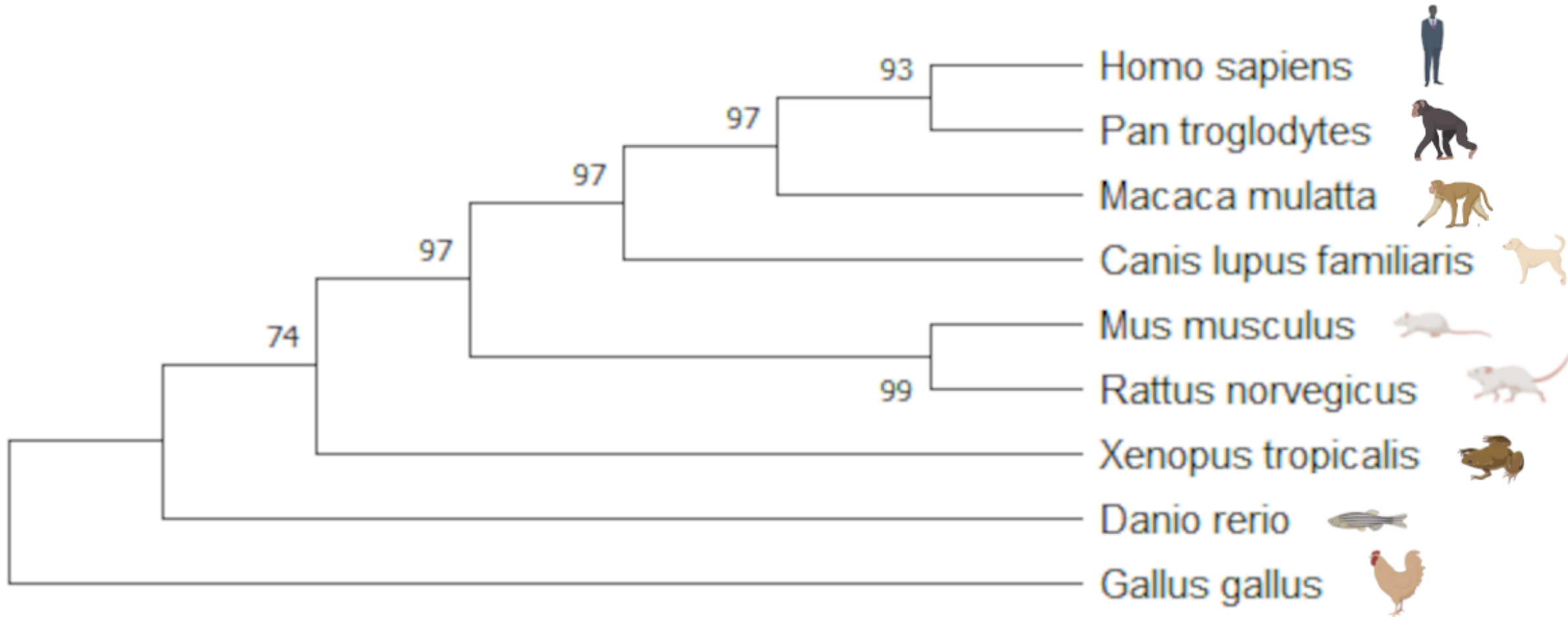
Scwann Cells



Myelination



# What are the PMP22 homologs?



# What is the gap in knowledge?

How exactly PMP22 causes abnormal myelin function resulting in CMT1A

