



# What harms the peripheral nervous system?

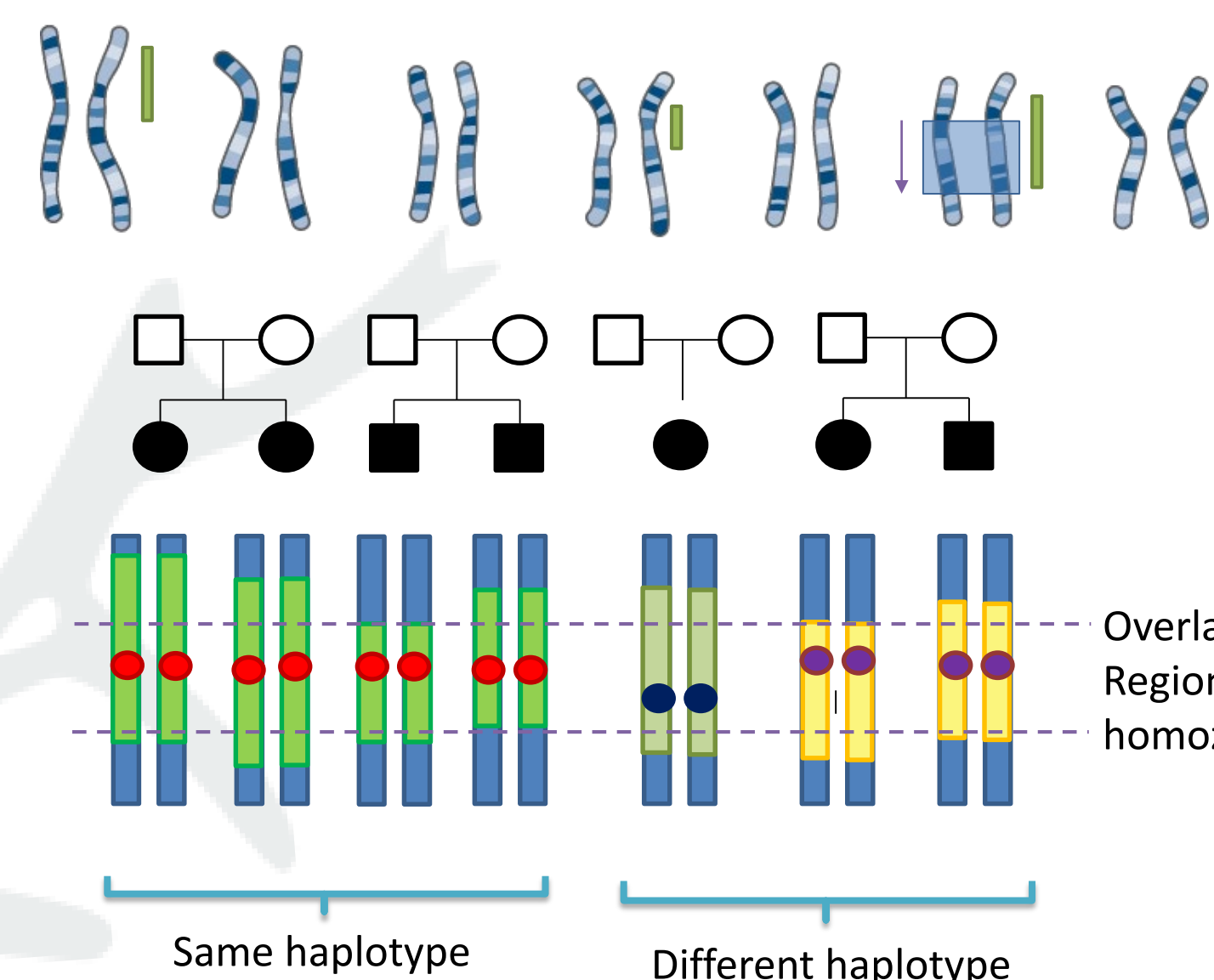
Molecular Neurogenetics Lab, Ghent University, Prof. Dr. Hildebert

Our group focuses on the identification and characterization of genes and pathways implicated in the molecular etiology of inherited motor and sensory peripheral neuropathies (known as Charcot-Marie-Tooth disease). Our research strategy is characterized by a multidisciplinary experimental approach:

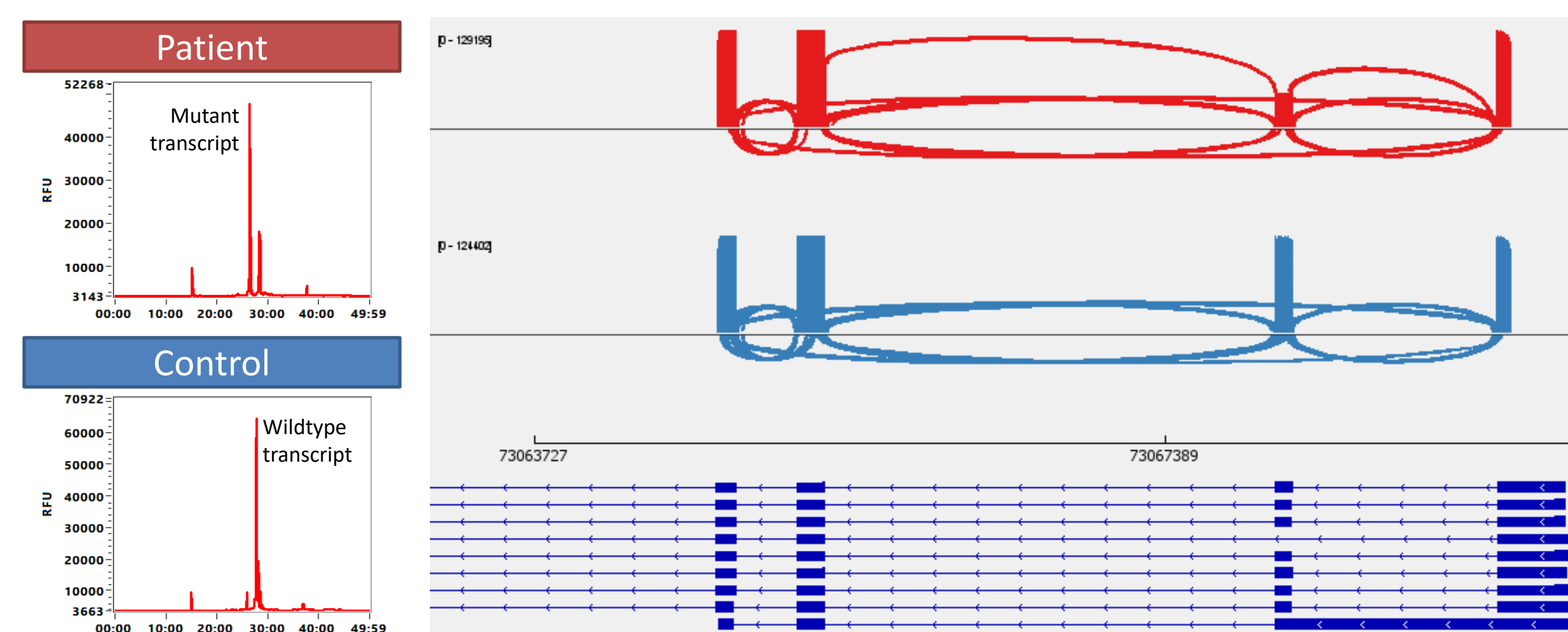


## Disease gene discovery using next-generation sequencing technologies

Whole exome sequencing coupled with linkage analysis or homozygosity mapping (HOMWES)

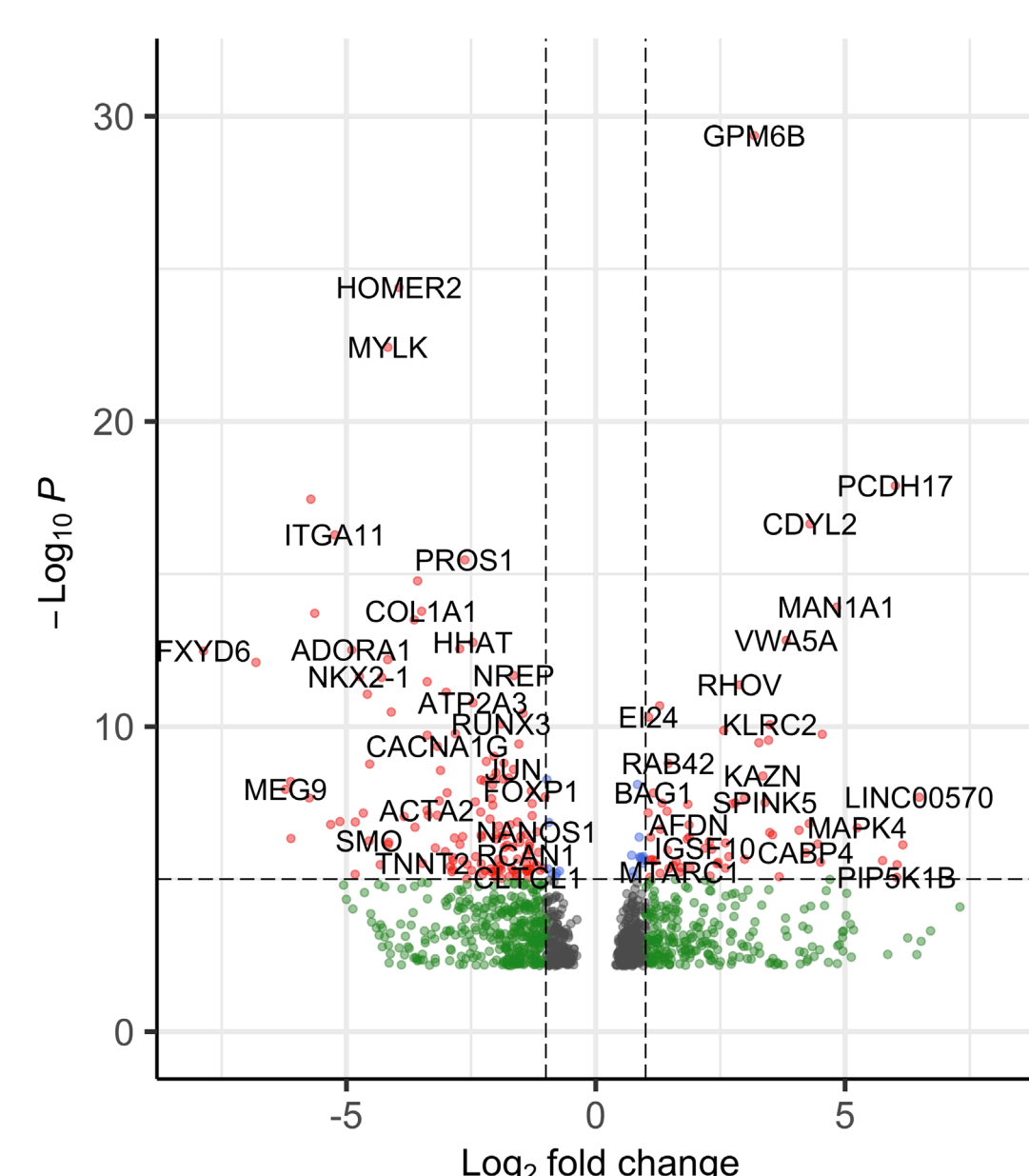


Follow-up techniques: Sanger sequencing, qPCR, long read sequencing, cDNA analysis.

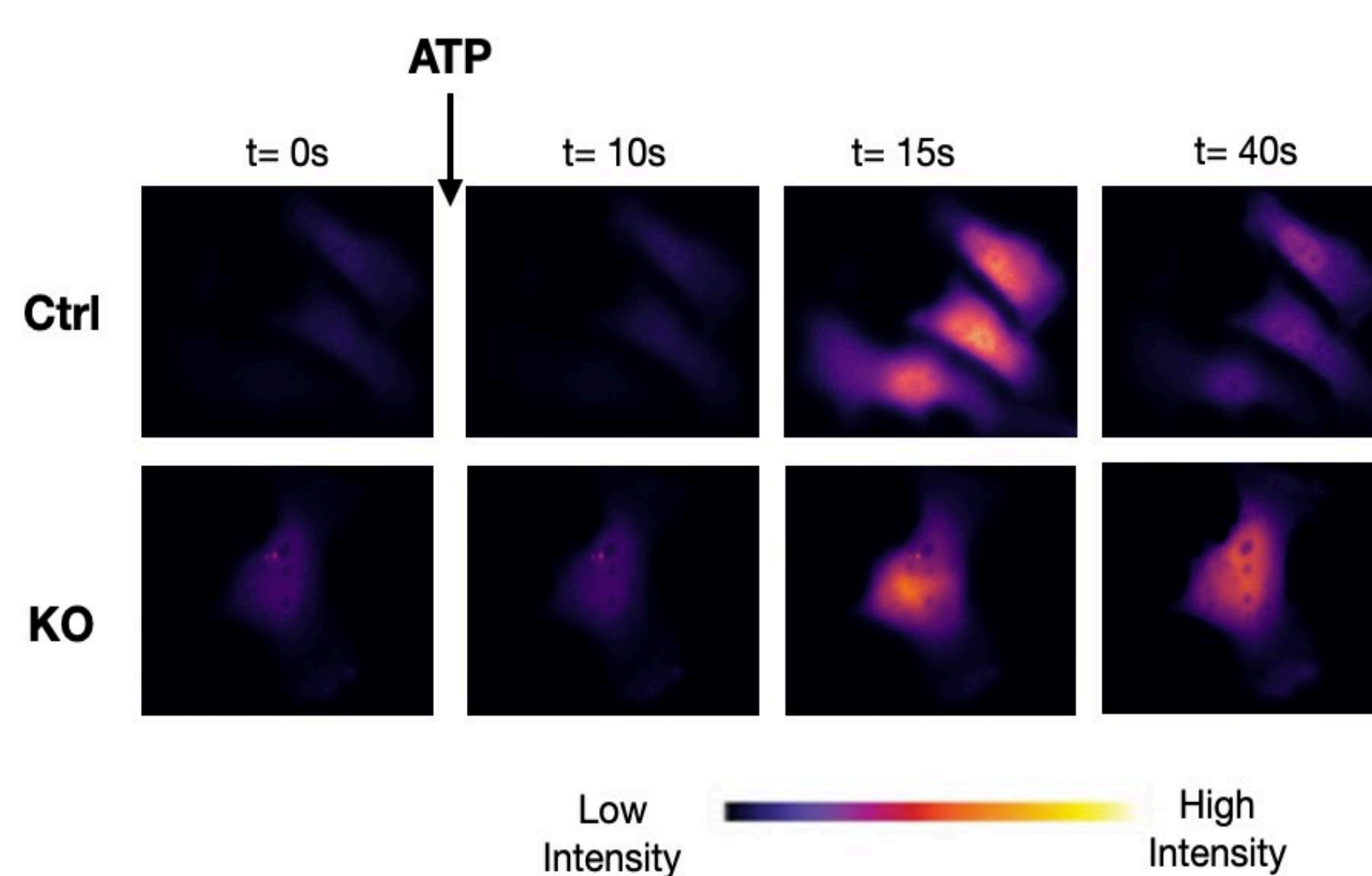


## Identification and validation of molecular pathways *in cellulo*: Crispr/Cas9 and patient-derived iPSCs

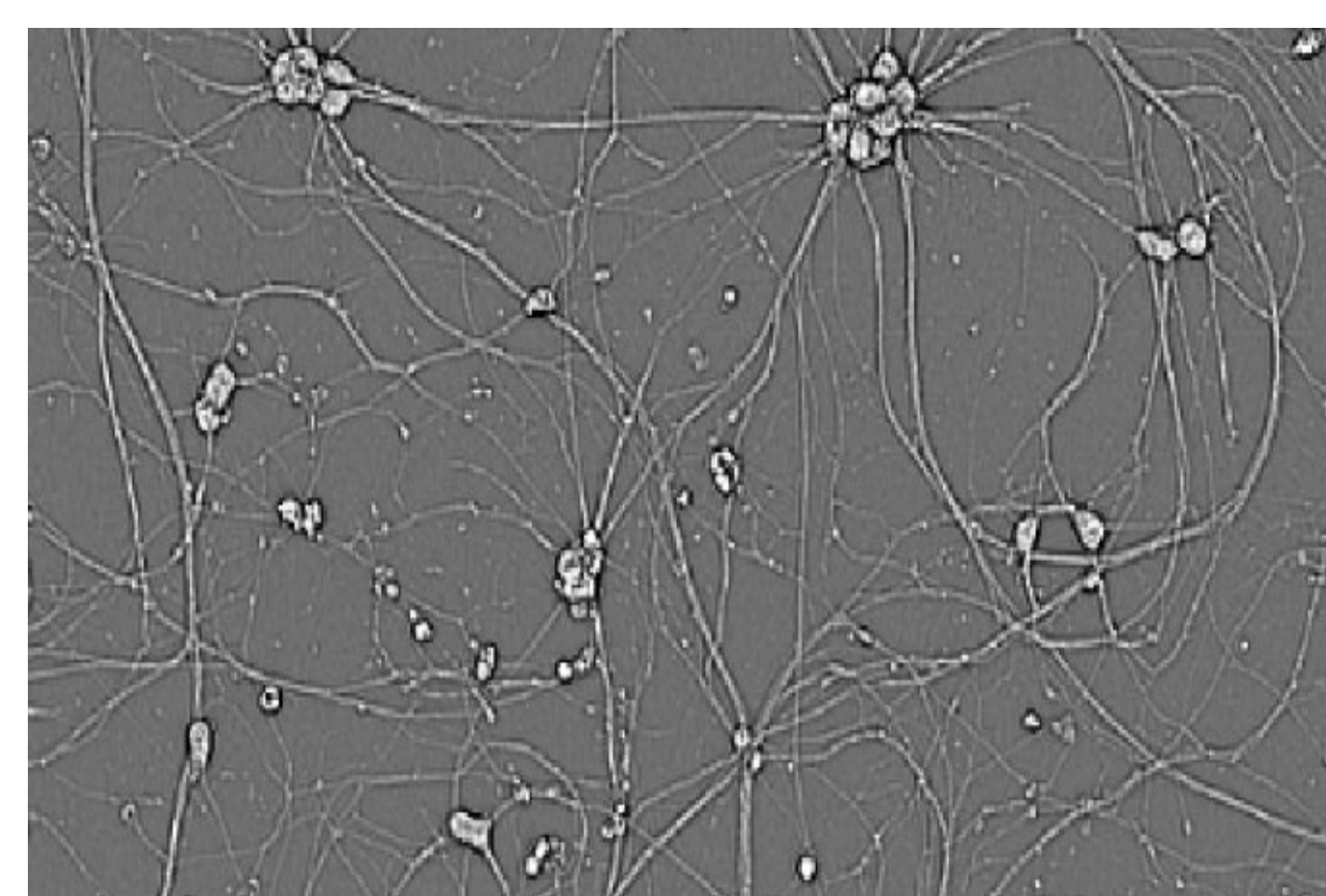
Transcriptome analysis for the identification of affected pathways in CMT



Functional characterization of identified misregulated pathways, e.g., Calcium signaling

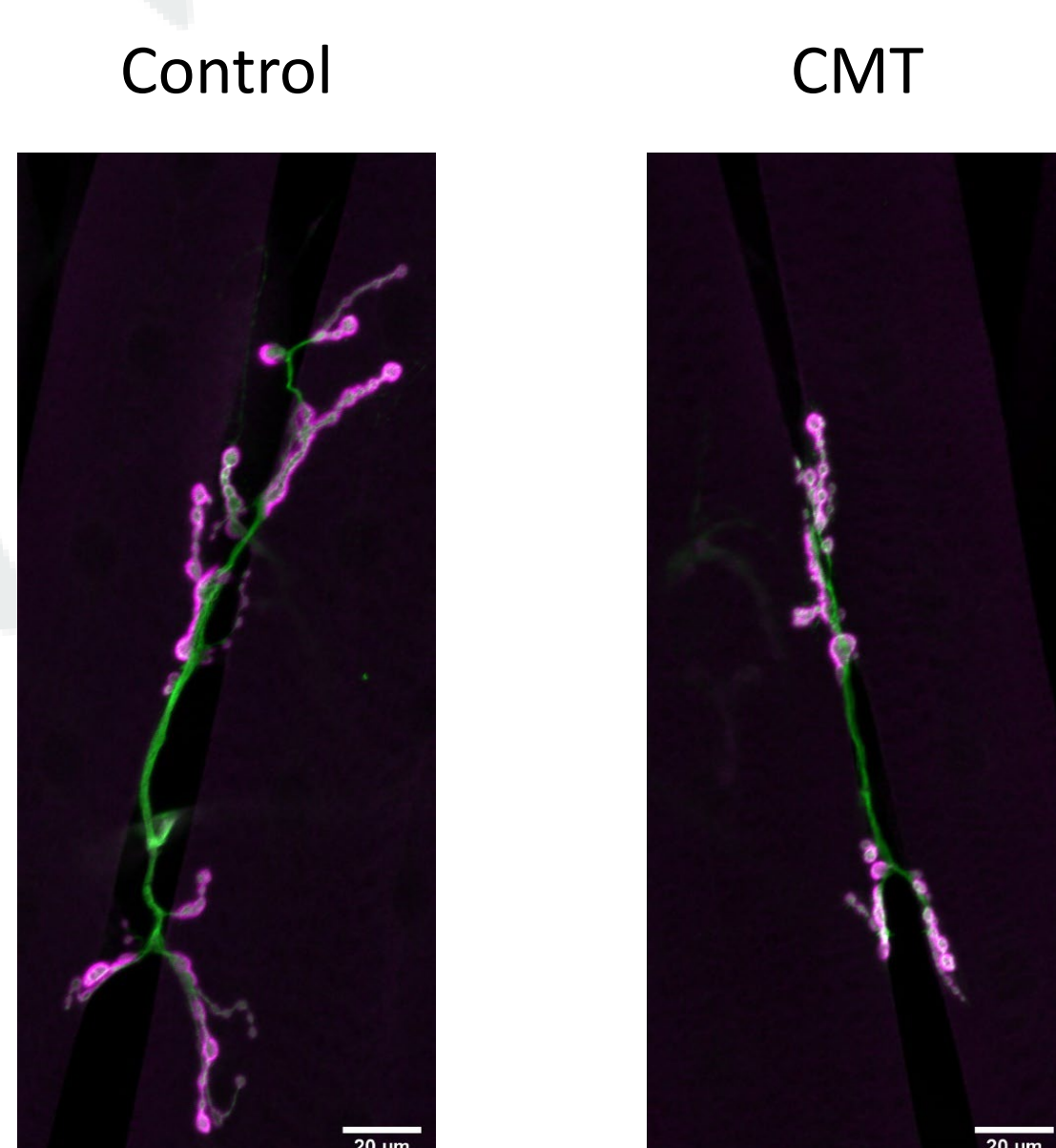


Differentiation and characterization of iPSC-derived motor neurons

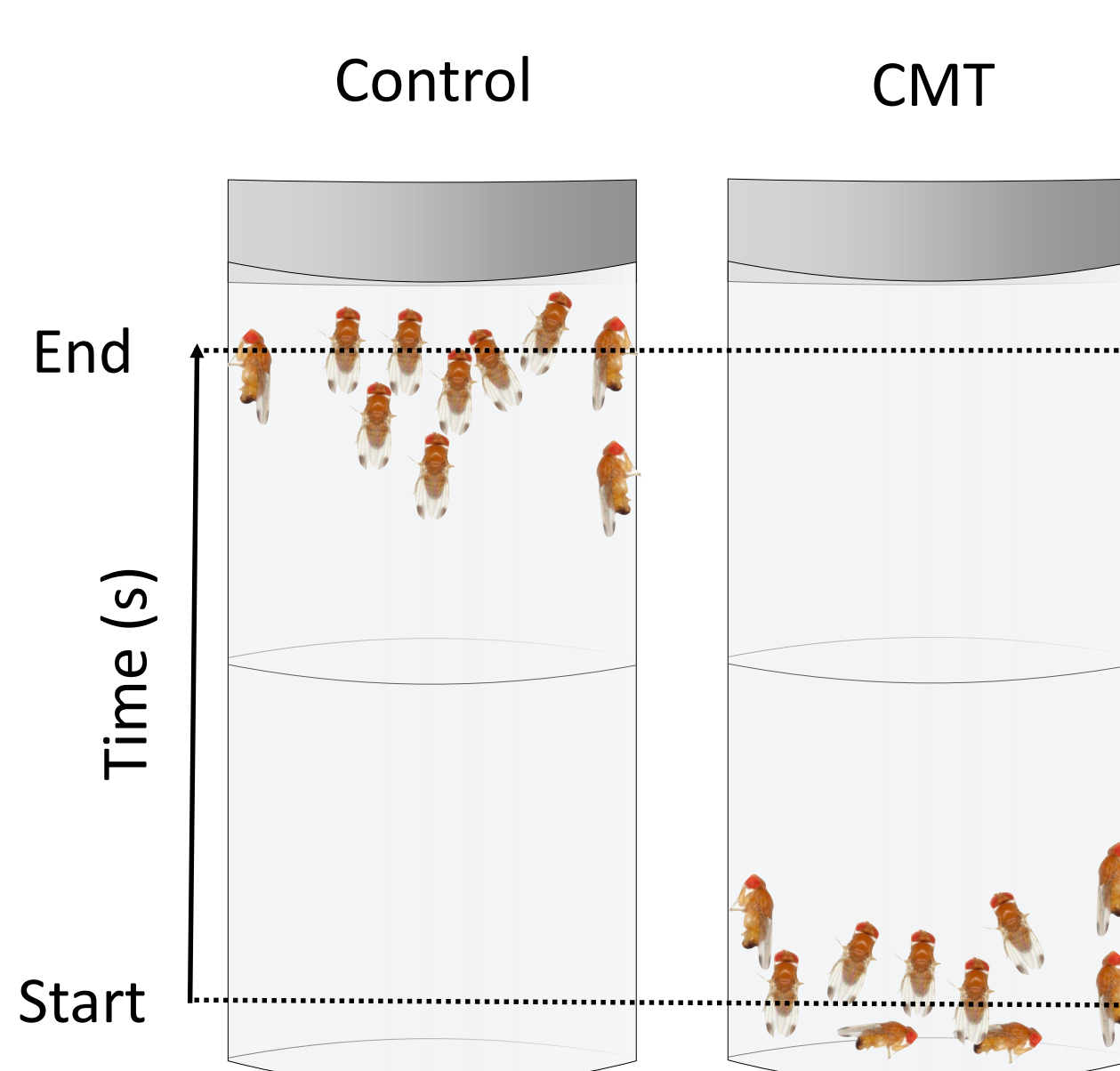


## Modeling Charcot-Marie-Tooth mutations in *Drosophila melanogaster*

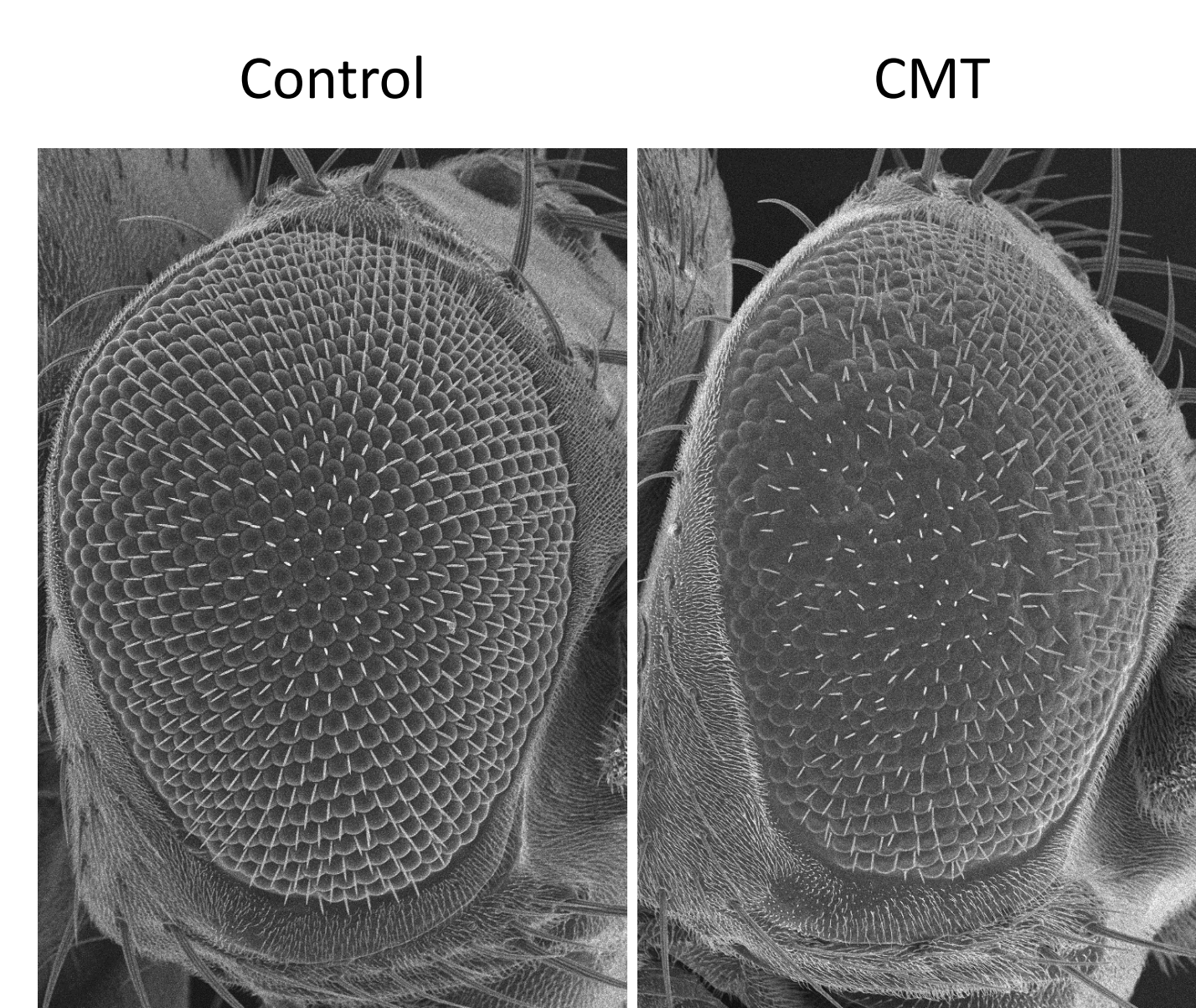
Neuromuscular junction analysis in larvae using immunocytochemistry



Locomotor performance of ageing flies using automated negative geotaxis assay



The fly eye as a system for high-throughput genetic screening using scanning electron microscopy



➤ Phenotypes in CMT fly models recapitulate the main hallmarks of the human disease

