2. RESCUING THE BLOCKAGE OF CRITICAL FUNCTIONS IN MOTOR NEURONS CAUSED BY GENE DEFECTS

PROJECT:
Trouble at the ribosome
in C9ORF-72-driven MND

PROJECT LEAD:
Dr Danny Hatters
The University of Melbourne, VIC

Defects in the C9orf72 gene are the most common cause of hereditary MND. In MND arising because of this defect, harmful proteins, called dipeptide repeats, form in motor neurons and block critical machinery needed for them to function appropriately. The research team will use motor neurons made from stem cells obtained from patients with C9orf72-MND to investigate why these blocks occur, why they are harmful to motor neurons and how they contribute to the onset of MND. To do this, the team will use the latest microscopic technology allowing them to view the atomic structure of the dipeptide repeat proteins and determine how they interact with pathways critical to the function of motor neurons.

KEY HIGHLIGHTS:
Dr Hatters is a first-time recipient of research support from FightMND. Successful outcomes will identify new therapeutic targets that prevent the formation of proteins that block the function of machinery critical to the viability of motor neurons.

AMOUNT INVESTED BY FIGHTMND IN THIS PROJECT:
$852,446

Q&A:
What are you hoping to uncover from this project?
We hope to identify the molecular events involved in the blockages that harm motor neurons. By clarifying these details, we hope to unearth mechanisms that could be targeted therapeutically. This could, for example, be through boosting mechanisms involved in their clearance mechanisms.

***"More insight is needed at the fundamental level to understand what drives the earliest steps of pathogenesis to enable new therapeutic strategies to be developed. Our research is directed at this goal."* – Dr Danny Hatters**

Right: Dr Danny Hatters
Left: Dr Danny Hatters and Postdoctoral Fellows Dr Christian Makhoul and Dr Chloe Gerak examining data from an electron microscope