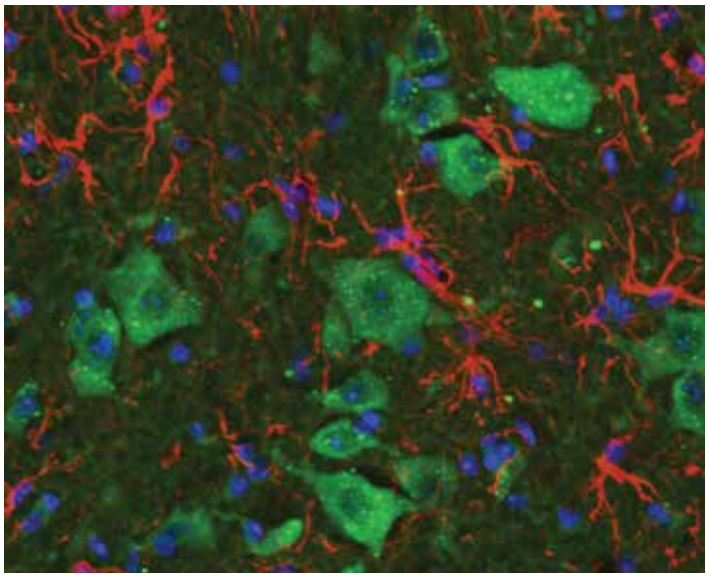
7. DISEASE MODELS

PROJECT:
Developing a validated C9orf72 mouse model of ALS/FTD using genome editing MND

PROJECT LEAD:
Associate Professor Bradley Turner
The University of Melbourne, VIC

Associate Professor Bradley Turner



Spinal cord of mice carrying the familial MND C9ORF72 mutation

Although defects in the C9orf72 gene are the most common cause of hereditary MND, there is still no effective model available to study this type of MND. Investigators in this project will create a new model that develops symptoms and motor neuron loss mimicking C9orf72-related MND. They will do this using powerful gene modifying technology that causes defects in the C9orf72 gene. The new model of MND will allow investigators to research the cause of C9orf72-related MND and test the effectiveness of drugs with promise for treating this most common genetic form of MND.

KEY HIGHLIGHTS:
This project will address a key gap in the MND research field by developing a new model of MND that mimics the most common genetic cause of MND.

AMOUNT INVESTED BY FIGHTMND IN THIS PROJECT:
$248,787

Q&A:
What problem are you trying to solve with this project?
Despite the discovery of C9ORF72 mutations in MND over a decade ago, it remains unclear exactly how abnormal C9ORF72 triggers MND which has hampered development of treatments. A major gap in our knowledge has been the lack of a robust and reproducible animal model of C9ORF72. This project will overcome this problem by developing a new and rigorously validated C9ORF72 animal model using powerful genetic engineering.

"This model will provide an invaluable resource to the global research community for testing disease hypotheses, pathology and therapeutic agents in the most common genetic form of disease broadly applicable to the MND population."
– Associate Professor Bradley Turner