Concord Hospital researchers lead world-first discovery of cause behind mysterious motor neurone disorder

In a world-first discovery, researchers at Concord Hospital’s ANZAC Research Institute and the University of Sydney have identified the cause of a disorder that degenerates motor nerves in men, raising the potential of new treatments.

Led by Principal Scientist Dr Marina Kennerson and in association with Professor Garth Nicholson, the research team has been investigating the X-linked distal hereditary motor neuropathy—a non-fatal disorder on chromosome X where the motor neurons of the peripheral nervous system progressively disintegrate.

Affected men cannot run properly and lose strength in their hands and feet, becoming increasingly disabled through later life.

About one in 2500 people suffer from the inherited peripheral nerve disease, with X-linked distal hereditary motor neuropathy representing one form of this disorder.

Dr Kennerson said the research team was studying the DNA in families with hereditary motor neuropathy to identify the gene causing the disorder, when they came across an unexpected discovery.

“We discovered mutations in the ATP7A gene, which is responsible for controlling copper levels in our bodies,” Dr Kennerson said.

“The discovery of this gene produced a big surprise because it turned out to be the same gene responsible for a completely different disease—a severe infantile multisystem disorder of copper metabolism called Menkes Disease.

We now believe that this gene abnormality causes the distal hereditary motor neuropathy by impairing the body’s ability to control copper levels, resulting in damage to the ends of extremely long motor neurone cells”.

Dr Kennerson said the discovery highlighted the importance of copper metabolism in the maintenance and function of motor neurons and would raise the possibility of new treatments for this disorder.

The discovery was published in the February edition of the American Journal of Human Genetics. The research work also involved collaborations with research groups in Australia, Brazil, the USA, Switzerland and Belgium.

CERA Recognised as One of World’s Best

The Centre for Education and Research on Ageing (CERA) based at Concord Hospital recently received notification of its selection as an international collaborating centre of the International Association of Gerontology and Geriatrics (IAGG).

The IAGG program aims to identify centres involved in research, teaching and the provision of services in Gerontology and Geriatrics, to establish a world community of those who are devoted to improve living in old age. The IAGG Collaborating Centre program seeks to be a forum to exchange information and expertise and Centres were selected on the basis of their international reputation and contribution to ageing research and education.

Concord Hospital is honoured that CERA was one of only eight centres in Asia, Australia and Oceania selected to be an IAGG Collaborating Centre.