Research into axon degeneration hits a nerve

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Dr Massimo Hilliard of the Queensland Brain Institute.

(Medical Xpress)—University of Queensland (UQ) researchers have made a significant discovery that could one day halt a number of neurodegenerative diseases.

Scientists at the Queensland Brain Institute (QBI) have identified a gene

that protects against spontaneous, adult-onset progressive nerve degeneration.

Dr Massimo Hilliard said that the discovery of gene mec-17 causing axon (nerve fibre) degeneration could open the door to better understand the mechanisms of neuronal injury and neurodegenerative diseases characterised by axonal pathology, such as motor neuron disease, Parkinson's, Alzheimer's and Huntington's diseases.

"This is an important step to fully understand how axonal degeneration occurs, and thus facilitates development of therapies to prevent or halt this damaging biological event," Dr Hilliard said.

Dr Hilliard runs a laboratory at QBI specialising in neuronal development, and focuses on how nerves both degenerate and regenerate.

The team found that mec-17 protects the neuron by stabilising its cytoskeletal structure, allowing proper transport of essential molecules and organelles, including mitochondria, throughout the axon.

This discovery has also the potential to accelerate the identification of human neurodegenerative conditions caused by mutations in genes similar to mec-17.

"It's our hope that this could one day lead to more effective treatments for patients suffering from conditions causing neuronal degeneration," Dr Hilliard said.

This discovery highlights the axon as a major focal point for the health of the neuron.

Findings of the research have been published in journal *Cell Reports*, and

lead author Dr Brent Neumann anticipates that the research into the gene will soon lead to further discoveries.

"This study demonstrates that mec-17 normally functions to protect the nervous system from damage," Dr Neumann said.

"This knowledge can now be used to understand precisely how the gene achieves this and to discover other molecules that are used by the nervous system for similar protective functions," he said.

"We can now start to look into means of bypassing the function of mec-17, such as activating other genes or alternative mechanisms that can protect the nervous system from damage."

Previous research has shown that mec-17 is conserved across species, including humans, suggesting a possible shared function of protection.

"We identified mec-17 from a genetic screening method aimed at identifying molecules that cause axonal degeneration when they become inactive through genetic mutations," Dr Neumann said.

This project was conducted in the tiny nematode worm C. elegans, which is a very powerful genetic model system commonly used for addressing and understanding neurobiology questions at a basic biological level.

The project was funded the National Health and Medical Research Council and an Australian Research Council Future Fellowship.

More information: Findings of the research appear in the journal *Cell Reports* published on 26 December 2013.

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