

Media Release

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**BREAKTHROUGH IN UNDERSTANDING INHERITED DESTRUCTIVE
DISEASE IN TEENAGERS**

On every morning peak hour train in Australia there are around a dozen carriers of Friedreich's ataxia. This rare inherited disease affects the heart and nervous system with symptoms often showing in the teenage years.

Parents must both be carriers to pass on the condition and the teenager can present to their doctor with clumsy, awkward movements and unsteadiness. The terrible consequence is that these children grow up knowing the joys of self-sufficiency, being able to walk and function normally before they are suddenly struck down. The disease leads to the patient becoming bound to a wheelchair and suffering an early death due to heart problems. Currently, there is no treatment for this condition.

Bosch Institute scientist, Professor Des Richardson, has been conducting research to better understand the disease and his internationally ground breaking findings have been published in the latest issue of the prestigious *Proceedings of the National Academy of Sciences of the United States of America*¹.

"Every cell in our body is divided into little compartments, each performing a unique function. The mitochondrion, is the powerhouse that is responsible for generating the energy for the cell," said Professor Richardson.

"Our studies have shown that the mitochondrion in Friedreich's ataxia takes up too much iron, releases less, as well as using less iron than normal. This is toxic to the cell.

"Previously, we knew that the powerhouse in the cells of Friedreich's patients became iron loaded, but we didn't know how.

"The identification of the reason for the iron-loading will lead to further research into how we can help to treat this disease," he concluded.

This disease, although rare, strikes without warning and is devastating for families. This research is a huge step in understanding the disease and what makes it destroy lives.

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¹ <http://www.pnas.org/content/early/recent>

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