

# Skin provides Australia's first adult stem cells for rare genetic disease

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(PhysOrg.com) -- Scientists have developed Australia's first adult induced pluripotent stem cell lines using skin biopsies from patients with the rare genetic disease Friedreich Ataxia (FA).

The study was conducted by the University of Melbourne and Monash Institute of Medical Research and is published in the current online edition of the international journal *Stem Cell Reviews and Reports*. It is the first time adult pluripotent [stem cells](#), known as iPS cells have been developed for a specific disease in [Australia](#), allowing for the development of new treatments for FA and related conditions such as diabetes and heart disease.

Induced pluripotent stem (iPS) cells result from the reprogramming of adult cells, such as skin cells, and are similar to embryonic stem cells in that they have the potential to generate any cell type of the body.

Dr Alice Pébay and Dr Mirella Dottori, co leaders of the study from University of Melbourne, characterized and directed the Friedreich Ataxia iPS cells to become specific cell types, including heart cells and nerves, which are normally not functioning well in the disease.

“By focusing on the heart and nerve cell types, we hope to be able to develop treatments to improve heart function and the loss of movement experienced by patients with FA,” Dr Pébay said.

Friedreich Ataxia affects one in 30,000 people globally, and Dr Paul Verma of the Monash Institute of Medical Research said this research

could be applied to other diseases.

“Due to the number of symptoms experienced by people with FA, including diabetes and heart disease, this resource could be applied to developing treatment for those conditions and helping even more people,” he said.

Dr Dottori said the research could not have been achieved without a significant network of experts and support from the Friedreich Ataxia Research Association (Australasia) (FARA-A) and the Friedreich Ataxia Research Alliance (FARA) in the United States.

“It is the collective effort of clinicians, scientists, patients and FARA that has made this discovery possible,” she said.

Ms Varlli Beetham, Executive Director of FARA said the finding provided real hope for people suffering the debilitating condition. “We are proud to have supported this research effort and look forward to the next stage of research, the development of new trial treatments,” she said.

Provided by University of Melbourne

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