



Friedreich's
Ataxia
Research
Alliance
IRELAND

Friedreich's Ataxia - The need for research and funding

Friedreich's ataxia (FA) is a debilitating, life shortening, degenerative, rare, neuro-muscular, genetic, disorder. Onset of symptoms can vary from childhood to adulthood. They initially include clumsiness of movement, weakness and muscle loss which causes an unsteadiness in standing and walking which can be mistaken for drunkenness.

As the disorder progresses, it causes vision impairment, hearing loss, slurred speech, aggressive scoliosis (curvature of the spine). Diabetes mellitus and a serious heart condition often co-exist. Sufferers eventually are wheelchair dependent in their late teens and sometimes earlier. As time progresses, they become totally physical dependent.

Treatment

There is currently no treatment or cure for FA which is one of over 50 different types of ataxia. It is one of the more aggressive forms of hereditary ataxia. One year long clinical trials have been carried out using idebenone, Vitamin E and EPO but no positive results have been proven. Shorted clinical trials and stem cell therapies have also been unsuccessful to date.

Research in Friedreich's Ataxia (FA)

Several different approaches to finding a treatment are in place internationally at present. These include:

- 1) Developing collaboration between pharmaceutical companies interested in working in FA, academic researchers and clinicians,
- 2) Grant funding potential drugs, cell and animal models which are potential candidates for FA treatment. **This is where most of the money raised in Ireland goes.**
- 3) Providing a patient registry for people with FA which can be used to recruit individuals for clinical trials
- 4) FARA advocates for policy initiatives that provide greater awareness, financial resources and other support for research in rare diseases like FA.

How can you help?

- Befriend an ataxia
- Spread awareness of ataxia
- Encourage the ataxian to register on an international register for Friedreich's Ataxia (FA)
- Fundraise for research. A member of FARA Ireland can help you - faraireland@yahoo.ie or marykearney@gmail.com if you have any difficulty with international register for FA.

Research FARA Ireland supported in 2019

\$7,000 was given to Dr. Helene Puccio, Strasbourg in collaboration with Prof. M. Pandolfo, Brussels who are carrying out unbiased proteomic analysis and focused analysis of metabolic and inflammation markers in plasma and cerebro-spinal fluid of those with FA. They hope to identify and validate related biomarkers to be used as candidate surrogate outcomes in clinical trials. They plan to use human induced-pluripotent stem cell derived neurons from FA patients and affected neural tissues from two mouse models,

FARA Ireland are a silver sponsor for the International Ataxia research conference due to be held in November 2019 in Washington DC, USA. It is a good forum for researchers to meet and debate. Dr Mary Kearney, secretary FARA Ireland attended this conference and presented 2 posters:

- Results of 1 year long randomised clinical trials in FA
- European Reference Network – What is in it for ataxia?

\$7,000 was given to Dr Vania Broccoli and Prof Franca Codazzi, San Raffaele University, Milan, Italy for work in advancing stem cell therapy. They are working on the neurophysiology of a miniature and simplified version (organoid) of FA. All these projects have other co-funders.

In total FARA Ireland gave €14,529 in 2019 to research.

The Bandon ataxia shop and our Christmas card fundraiser are essential to help the research into Friedreich's Ataxia continue. We are very grateful to all our donors and thank everyone for their continued support.



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Friedreich's Ataxia Research Alliance Ireland (FARA Ireland)

FARA Ireland is a non-profit, non-governmental, Irish organisation representing people with Friedreich's Ataxia (www.faraireland.ie). It was initiated by parents whose children have Friedreich's Ataxia. They were spurred on by the success and determination on FARA, based in the USA (www.curefa.org) and FARA Australasia (www.fara.org.au) to contribute to funding research into Friedreich's Ataxia from Ireland.

Aims of FARA Ireland:

- 1) to develop a strong connection with international research on Friedreich's Ataxia.
- 2) to raise funds for research and
- 3) to increase public awareness of the plight and condition of people with Friedreich's Ataxia.

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February 2020