

# Newsletter

## Issue 6



# ATAXIA

## Ireland

Chy No. 7076 CLG 566322

## E-health Era Emerging

On November 16<sup>th</sup>, the Health Informatics Society of Ireland (HISI) held their annual conference in the Aviva stadium. The theme of the two day event was the Digital Patient-Empowerment through Digital Technology. Chief Information Officer of the HSE, Richard Corbridge spoke about the challenges and progress of implementing an electronic patient record (EPR) in the Irish Healthcare Industry.

Dame Fiona Caldicott who is the National Data Guardian of the NHS highlighted the importance of maintaining patient confidentiality and trust at all times.

Ingrid Brindle, who is a patient in a UK health practice which uses an electronic database that gives patients full access to their medical history; spoke of the personal benefits of accessing this system.

Electronic records will allow patient registries to be generated more easily which will make clinical trials and research more efficient and streamlined

### Key Features

- Online Database (Healthlink) linking Healthcare providers to patient medical history
- App technology providing access for patients to medical records, appointments etc
- 'Lighthouse projects' in 3 areas of health (Epilepsy, Hemophilia and Bipolar Disorder) to trial the system and target pitfalls

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## Apps for Ataxia

If you have a smartphone, there are some really useful apps for managing and monitoring your ataxia.

### Dexterity on Android/iPhone

This app was designed for stroke victims to aid with recovery, however its designed to improve fine motor skills and improve muscle tone in the lower arms and hands. Therefore people with ataxia could find it very beneficial. It allows you to track your progress in terms of accuracy and speed.



### Medhelper On IOS and Android

This app was designed by patients for patients. It allows you to record all your medications, their dose and when they are due. It also allows you to keep track of your health appointments (GP, physio etc).

## In Focus: MSA-c

Multiple System Atrophy (MSA) is a progressive neurological disorder that affects both the autonomic nervous system (parts of the nervous system that control involuntary functions such as blood pressure and digestion) and movement.

[www.orpha.net/MSA-c](http://www.orpha.net/MSA-c)

Multiple System Atrophy , cerebellar type is a form of MSA with predominant cerebellar features such as gait and limb ataxia, visual disturbance and speech problems (dysarthria) .

It's highest occurring prevalence is in Asia. Disease onset is typically in the fifth decade. The cause is unknown but people with MSA-c have a build up of synuclein complexes in many of their cells. These complexes are proteins found in high concentrations in the human brain. Mutations have also been reported in the COQ2 gene, which is needed for coenzyme Q10 production.

## Research

*Vitamin B1 (Thiamine) has potential as treatment for FA, a recent Italian study shows*

Constantini et al (2016) Long-term treatment with thiamine as possible medical therapy for Friedreich ataxia

Journal of Neurology Vol. 263 (11): 2170-2178

34 patients who were recruited in the study, received biweekly injections of 100mg of thiamine (intramuscularly). At the start of the trial, these participants were assessed using the rating scale for ataxia, SARA. This assessment was repeated at certain intervals during the trial (1,3,6,9,12, and 24 months). The treatment significantly improved parameters such as movement, speech and fatigue and it also lowered the rate of progression. There were also no side effects reported from the treatment. Further studies are required to test efficacy.



## Our Contribution to Research

### Conferences

- UCD, 1984 (hosted in Dublin)
- Co-hosted International Conferences in 2008, 2012 and 2015
- Contributed to International Conferences since 2008
- Contributed to National Ataxia Foundations annual investigator meetings in 2012, 2014 and 2016

### Irish Projects

- FA Prevalence 1985
- Heart Disease & Ataxia 1992
- Gene Prevalence 1997
- Proteomics of Ataxia 2007
- Episodic Ataxia 2011
- We have part funded the salary of the Ataxia Registrar at the Ataxia Clinic in Tallaght since 2012 (€20,000 annually)

### International Projects

- Scripps Institute, US. HDAC Inhibitors 2010
- Hospital Robert Debre, Paris. Target genes in FA 2010
- University of Oxford, SCA RNAi therapy 2011
- Hertie Institute, Germany. Videogame therapy for Ataxia 2012, 2014 – second installment (£2,500)
- University of Rome, Frataxin Ubiquitination 2012
- Imperial College London, HDAC Inhibitors in FA 2011-2013
- University of Melbourne, Listening devices for people with ataxia 2013

### Since 2015

- EFACTS FA Database (€20,000) and ongoing
- €4,000 - Pfizer FA drug development study (grant to Dr Giunti, UCL)
- €4,000 – FA gene therapy project (grant to Drs Pook and Themis, Brunel University, UK).
- £3,000 to Ataxia UK supported trial on Interferon Gamma for FA treatment

## News & Events

### Unite for Rights Conference

November 4<sup>th</sup>, Carrickdale Hotel, Dundalk

Sinn Fein Hosted a conference on issues relating to inequality of services and facilities in Ireland for disabled people. EU representatives discussed the emerging policies which will put pressure on member states to implement fairer and more accessible services. Ireland is the only EU state to fail to ratify the UN Convention on the rights of those with disabilities.

November 24<sup>th</sup>, Leinster House

Politicians met with patient representatives from organizations such as Ataxia Ireland to discuss the following issues

- Disability services & facilities
- Genetic services in Ireland
- Ratification on UN mandate for disabled rights
- Rare disease research



### Christmas Socials

December 3<sup>rd</sup>, Talbot Hotel Stillorgan, Dublin

- A Great day was had with music, raffles and even a visit from Santa.
- We launched our new awareness video at the Dublin Social
- Some attendees comments...

“Thanks for your excellent Social. It was the best Social for a long time.”

“One of the best and really liked the music”

December 9<sup>th</sup>, Oriel Hotel Ballincollig, Cork

### Active with Ataxia: Video Launch

Wednesday December 14<sup>th</sup> at 2pm on our facebook page

Week 1- Alan will introduce some basic exercises and explain how they benefit the core. Member Redmond O’ Hanlon will participate in the session.

### Interview with Paddy Doyle

December 20<sup>th</sup> at 10am on our facebook page

The writer will discuss how creative writing is a great outlet and therapy for people with physical impairments. He has a rare neurological condition called Dystonia and is an active campaigner for disabled rights.