

Newsletter

Issue 1

Chy No:7076



ATAXIA

Ireland

New Logo

Ataxia Ireland has recently changed its logo. The new look, comes as the charity makes the transition to a limited company. The organization will still have the same role, that of supporting its members and raising the profile and awareness of ataxia.

The logo is a depiction of a DNA strand, this helps to illustrate the basis of ataxia but it also shows people with the condition are more than their disability. Our DNA codes our personality traits, our appearance and our ability to cope with life's challenges. Ataxia is only part of the picture!

Dates

June 11th	Summer Social Stillorgan Hotel
July 2 nd - 8th	Annual Respite Week Cuisle, Roscommon
Sep 3rd	Skydive Abbeyschrule, Longford
Sep 23rd	Gala Fundraising Ball Stillorgan Hotel
Sep 25th	International Ataxia Awareness Day



**“Before you there was no possibility, ahead of you there is every possibility” Alastair Kent, Genetic Alliance, UK
EURORDIS 2016**

Researchers Award

Dr Petya Bogdanova, an ataxia registrar at Tallaght Ataxia Clinic, recently presented on her work with families with rare undiagnosed ataxias at a researchers conference in Limerick.

She received the **Mark Gibson Prize for Best Movement Disorder related presentation** at the 52nd Irish Neurological Association Meeting 2016, May 5-6th, Limerick.

The work that Petya and her colleagues carried out described clinical characteristics for three rare autosomal recessive cerebellar ataxias.

This allowed them to make diagnosis using a selective gene sequencing method. It also showed that these particular ataxias were more common than previously thought, with two of the mutations in the cohort accounting for the second and third most common ataxias after Friedreich's ataxia. See our website for more information.

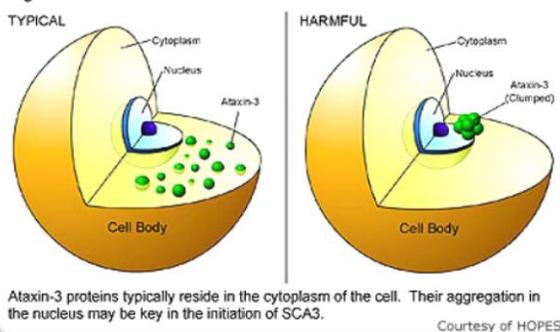


National Ataxia Clinic Tallaght Hospital

This service is held on the first Friday of every month. It is run by Neurologists Dr Richard Walsh, Dr Sinead Murphy and Dr Petya Bogdanova, who are all expert in the assessment and care of people with ataxia.

It is a multi-disciplinary approach with input from other specialists such as cardiology and physiotherapy. People who wish to be seen here should request a referral from their GP.

Figure F-12: Location of Ataxin-3



It is thought that clumping of this protein in the nucleus is the cause of the condition.

In Focus: SCA3

Spinocerebellar Ataxia type 3 is an autosomal dominant cerebellar ataxia. It's also known as Machado-Joseph disease. It can present in later adulthood depending on which form an individual has. It is associated with a CAG repeat expansion mutation in the ATXN3 gene. Genetic testing is available for it.

It's believed 1-2 in 100,000 people have the condition and prevalence is most common in Portugal, Germany, The Netherlands and parts of Asia. Riluzole is currently being trialled in Italy as a possible therapy for this and other cerebellar ataxias.

News

Welcome

New staff member, Geoffrey Kane, is now working with Ataxia Ireland. He is a great addition to the team and we look forward to working with him.



EURORDIS

The 8th conference on Rare Diseases & Orphan Products took place in Edinburgh, May 25th-28th. It was hosted by EURORDIS, Rare Disease Europe, which is the platform for bringing together patient organizations, people from academia and industry.

The conference was a great success, where delegates voted on new board members and new legislation. There were long discussions on how to develop trials for small patient groups and how to bring rare disease to the forefront of public policy. The event really highlighted how rare disease organizations collectively hold more authority and potential for progress.



HORIZON PHARMA

The company, whose headquarters are in Dublin, announced in May that they have bought the right for Interferon Gamma in Europe. If their current US based trials are successful in FA, this will allow them to conduct future trials here in Europe.



Nationwide Survey

The National Ataxia Clinic in Tallaght Hospital is launching a new survey, with the support of Ataxia Ireland, for people *living with Ataxia in Ireland*.

The focus is to examine the impact of the condition on costs and quality of life. The survey will collect the information anonymously, but the data generated will be crucial for future research. We encourage all our members to take part in the survey and we are happy to assist in completing them.

EFACTS

Ataxia Ireland are delighted to announce that the National Ataxia Clinic is now a designated centre for the European Friedreich's Ataxia Natural History Database and we will be updating members on future study dates.



RETROTOPE

This US based pharmaceutical company has completed phase I (first dose) of RT001. This is a fatty acid, which stops lipid peroxidation (free radical damage). Preliminary results are promising, suggesting it is well tolerated with minimal side effects.



Gala Fundraising Ball September 23rd



Stillorgan Hotel

This will be a corporate event aimed at attracting business and commercial enterprise. Businesses can purchase tickets individually or a table for a discounted price.

All proceeds raised will go to Ataxia Ireland.

Details to follow in the near future.

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Skydive September 3rd

There is still time to register for this year's skydive. Contact Geoff at geoff@ataxia.ie for more details.

The event takes place in Abbeyschrulle, Co. Longford.

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