www.ataxia.ie July 2016

Newsletter

Issue 2

ATAXIA Ireland

Cuisle 2016

This year's holiday took place from July 2nd to 8th in our usual venue at Cuisle Holiday Centre, Co. Roscommon. Over 30 of our members came along and they were joined by helpers and volunteers who really helped to make it a memorable experience.

There were various activities organized throughout the week such as bowling, yoga and cookery competitions.

We also had a visit from Wild Encounters who brought along some exotic creatures for us to handle and interact with.

There were daytrips to nearby venues such as the racecourse and Knock.

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Living with Ataxia Survey

We had a great response from our members to the survey we recently distributed. We'd like to thank all those that responded so diligently to completing and returning them to our office. We'd also like to encourage anyone that has yet to fill out their survey to please do so as soon as possible.

The questionnaires are anonymous so you do not need to provide name or contact details. The data will be collected by Dr Petya Mihaylova in Tallaght and used to enhance our knowledge of services and care provided to our members by the state.

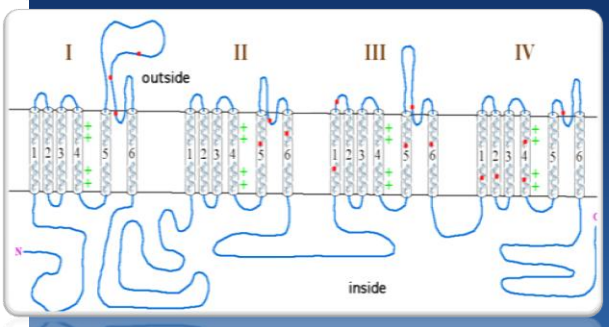
International Ataxia Awareness Day Sep 25th

If you'd like to raise the profile of Ataxia and increase awareness; why not host a coffee morning in your home or at your place of work. Contact the office for more information.

Redmond O'Hanlon is getting ready for his 2nd jump this year. watch video

Skydive 2016

It's not too late to sign up or encourage friends to do so for this year's skydive. Abbeyschrule in Longford will once again be our base on September 3rd.
Contact geoff@ataxia.ie for more information.



Episodic Ataxia Type II is caused by mutations (red dots) in calcium channel genes. This means Calcium is not transported into and out of the cell membranes adequately and this affects nerve cell conduction.

In Focus: Episodic Ataxia

There are two main types; both of which are autosomal dominant in terms of inheritance pattern. This means an affected individual has a 50% chance of passing the gene on to their children. Type I and II episodic ataxia, are as their name suggests, sporadic in nature.

Individuals can have episodes of ataxia ranging from hours to days. Symptoms include nausea, vertigo, migraine and fatigue. Acetozolamide can help those with type II as it is thought to stem from problems in the calcium channels in the brain.

Type I is even rarer and is characterised by Myokymia (muscle twitching). This is believed to arise due to problems with the potassium channels involved in nerve conduction.

Cuisle 2016

Angela making Cocktails

Brid's right hand man- meerkat

Catherine catching dinner

Ready Steady Cook

Self Service

Ken's eclectic tastes

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We also ran a murder mystery, which had a cocktail theme as Cuisle became Hotel Cabana for the week. All the members and helpers were given characters and many were involved in illicit side stories so there was more than one suspect in the frame for the killing.

On the last night our two comedians improvised as local gardai and arrested the offender, one Thornton the 3rd, a millionaire guest who let power go to his head. Congratulations to Katherine Herbrich who figured out the perpetrator

SPATAX, Paris June 23rd-25th 2016

Paris played host to the EURO's but more importantly it was also where scientists converged for the conference on rare ataxias.

There has been a lot of progress in identifying more advanced and selective methods of screening genes, such as in next generation sequencing (NGS).

Although many ataxias have common genes involved there are other mutations which this sequencing allows to be identified.

It is only by finding these that scientists can go on to test potential treatments.

SCA3, which was highlighted in last months newsletter was one particular ataxia whereby sequencing techniques facilitated improved understanding.

For example Riluzole was ringfenced as a potential treatment but using NGS, researchers in Tubengern University, Germany have shown that KPNA3 is a more useful strategy for keeping ataxin-3 away from the nucleus.

Research

Iron Toxicity induces Neurodegeneration in FA Model

Researchers at Bayer College of Medicine in Houston, Texas showed that loss of the frataxin gene in FA causes iron toxicity which is responsible for the neurodegeneration that people with this condition experience.

This is important as there is some dispute over whether or not it is Iron build up or free radical accumulation that contributes to neuronal death.

Although researchers used a fruit fly as a model for their study, there is a huge similarity in terms of molecular pathways between both species.

When they restricted some of the pathways that Iron build-up affected they saw neuronal death was reduced and neurodegeneration slowed.

This will hopefully have implications for future therapeutic strategies.

Chen et al/elife 2016

HDAC Drug restores frataxin functionality

Researchers at the University of Oklahoma College of medicine have shown in a phase 1b trial that a drug named 109 can help to restore normal levels of the frataxin protein.

The mutation of the gene in individuals with FA prevents it from being made into the protein. However using cells from 12 people with FA who had mutations ranging from 200 to 1122 triplet repeats, the researchers were able to reactivate the gene and get messenger RNA, the intermediate product between gene and protein.

The mechanism by which it is initiated is not understood but similar drug compounds could not generate the same results so it is definitely a promising candidate.

Chutake et al /Nucleic Acid Resolutions 2016

Gene identified in Cerebellar Ataxia

Researchers at Western University of Health Sciences have discovered an important gene mutation which is implicated in ataxia in humans and dogs.

The CAPN1gene mutation impairs the function of its enzyme calpain-1 which subsequently causes abnormal brain development.

The fact that this gene is conserved in both species shows its role in ataxia is very important and this makes it an important marker for future studies.

This study was the first to look at the affect of calpain-1 inhibitors exclusively. Previous research involved inhibiting both calpain-1 and calpain-2 mutually and so any compounds used to treat these mutations didn't discriminate between the two molecules.

This means more specific drugs can now be developed with the required neuroprotective properties that prevent ataxias.

Wang et al. /Cell Reports 2016

Charities under fire

It has been a time of upheaval and uncertainty for the charity sector with the negative press coverage of late. Accountability and regulation are the key concerns.

Fundraising is no easy feat and any money needs to be channeled in an appropriate manner for people to have assurance it is been properly used.

Ataxia Ireland has recently become a registered limited company. As part of that transition it will also adopt SORP, which is the highest level of reporting and regulation a charity can adhere to. The directors of the board are John Bradshaw, Jennifer Peacocke, Clare Shine and Cormac Murphy.

Although it's primary role remains the same; that of supporting its members and spreading awareness; the transition will help further to give the charity full transparency, which will be a key factor in restoring public trust in the charity sector.

Future issues will give more information on SORP

Get in touch:

T: 353 1 299 9033

E: info@ataxia.ie

Ataxia Ireland

4 Leopardstown Business centre

Ballyogan Road

Dublin 18