Newsletter

Issue 4 Chy No: 7076 CLG No: 566322

ATAXIA Ireland

International Ataxia Awareness Day

September 25th provided a useful platform for us to spread awareness of ataxia to the general public. Neurologist Dr Richard Walsh and our Information officer Clodagh appeared on TV3's Sunday AM to discuss the condition. They demonstrated how coordination and balance are distorted with simulation goggles, recently acquired from the US. We also had a feature in the Herald about the patient perspective, whereby Clodagh spoke about living with ataxia.

Several of our members also held coffee mornings to raise proceeds for the charity and we'd like to extend a warm thanks to those who did. Thank you to the all the volunteer bakers also; you would rival the great British bake off any day!

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Living with Ataxia in Ireland

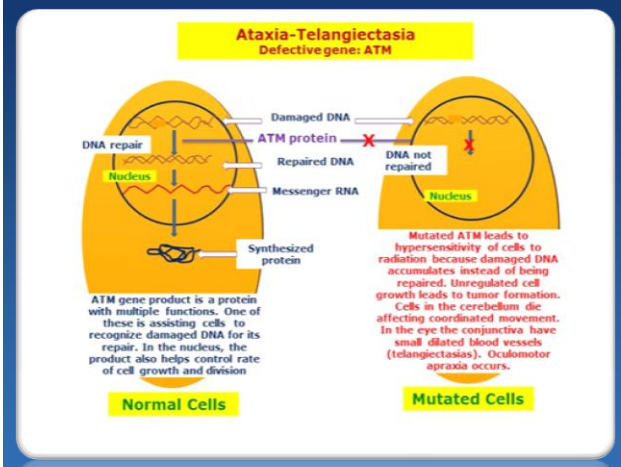
We would like to remind members to find the time to complete the survey if you haven't done so already (all data is anonymous). We are gathering information about quality of life and the health and social care costs associated with ataxia in Ireland. Similar studies have been done in multiple sclerosis, motor neuron disease and Parkinson's disease in Ireland and the results have been used successfully to improve the care of people living with those conditions. We have had a great response already, with 128 surveys completed to date, but the more people who take part, the more useful information we can gather. This kind of information is vital in order to improve access to care, make better use of limited resources, improve public awareness of ataxia and, of course, lobby for more resources. Once the study is complete, we plan to disseminate the results as widely as possible by presenting the data at national and international conferences and publishing in peer reviewed journals

Dr Petya Bogdanova-Mihaylova, Dr Richard Walsh and Dr Sinéad Murphy

National Ataxia Clinic, Tallaght Hospital

The survey can be completed online at https://www.surveymonkey.com/r/FGP9D89 or for a paper copy please contact the Ataxia Ireland office

In Focus: Ataxia Telangiectasia



This condition is a combination of progressive cerebellar ataxia and immunodeficiency. The severity of the neurological and immune system manifestations is variable from patient to patient. Prevalence is estimated to be 1/100,000 children.

A-T is an autosomal recessive disease caused by mutations of the ATM gene. This gene is important for coding a protein involved in double-strand-break DNA repair, particularly in neurons in the Cerebellum. Onset for the condition is usually early childhood but again this is variable. Management involves physiotherapy, speech therapy and sometimes Immunotherapy is useful for keeping infections to a minimum. People with A-T should minimize exposure of cells to X-Rays and UV light as this increases their vulnerability to damage.

FA Study on UCM's (Rufini et al Neurobiology of Disease, Vol. 75: 91-99)

A study which was based in **Tor Vergata**, at the **University of Rome** looked at one strategy of increasing frataxin levels in the cell by preventing degradation of existing frataxin.

In people with FA the protein is 'tagged' by an ubiquitin molecule which essentially labels it for degradation by cellular cutting enzymes. The scientists here identified small molecules which can dock on the same site as ubiquitin molecules, thereby preventing their attachment and thus the subsequent degradation of the frataxin protein. They called these **ubiquitin competing compounds (UCM)**. They hope these UCM's could offer a novel therapeutic strategy for FA. Professor Roberto Testi was one of the researchers on the study and we spoke with him about ataxia research.

Interview with…. Roberto Testi, University of Rome

1) **Is it better to focus research on one particular therapy or explore several different therapeutic options for Friedreich's ataxia and why?**
   It is a good idea to explore several different therapeutic options for a number of reasons. In fact, there is no guarantee that any single therapeutic program, no matter how smart or expensive, will eventually succeed in generating an effective and approved drug or treatment. Second, a given approved treatment might be not well tolerated by all patients, or even be effective in all patients, so we need a portfolio of possible effective treatments available. Third, it is quite likely that a successful therapeutic approach will eventually require more than one drug simultaneously, with therapeutic combinations that might vary from patient to patient.

2) **How can patient representative organisations help to improve research activity in their own countries?**
   Patient organizations can play a number of key roles in supporting the research effort. They may directly fund discovery programs and clinical trials run by small companies, academic groups and research centres. They may also act as opportunity platforms where investors, biotech/pharmaceutical companies and research groups interact and plan drug development strategies. Patient organizations also can help in the dissemination of research information and in promoting the interaction among research stakeholders through the organization of scientific meetings and conventions. Moreover, they can be instrumental for patient recruitment and support in clinical trials.

Patient organizations might exercise influence on national and international research agencies to help allocate their budget on programs focused on the disease of interest. Finally they might use their influence over local regulatory agencies to implement procedures and policies that reduce the time and costs necessary to bring new drugs to the patients.

3) **What can patients themselves do, to increase the amount of research in their condition?**
Probably the best way patients and their families can increase the research effort is by collaborating with their local patients organizations. Most if not all patient organizations have been started by the impulse and commitment of patients and their families.

4) **Can therapies in ataxia be applied to other conditions and therefore make them more attractive for funding?**
In general, the more specific a therapeutic approach is and tailored on a given disease, the less it can be applied to other diseases. Attractiveness of a potential treatment for funding by investors and biotech/pharmaceutical companies, however, may depend on a number of additional factors besides the number of potentially reachable patients. These factors include the possible final cost of treatment to the users, the distance to market along the development process, ongoing competition, market penetration costs and expectations, IPR and regulatory issues, specific features of the potential treatment, etc…

5) **What time frame do you think it will take to find a successful treatment for FA?**
As outlined in the Research Pipeline made available by FARA (http://www.curefa.org/pipeline), the potential treatment that is closer to the finish line is interferon gamma. This drug is currently in final clinical testing. We might learn about the efficacy of interferon gamma in FA patients in 2017. If effective, therefore, the drug might be made available to FA patients rather soon. Other potential treatments are also in clinical testing, while others are way behind in the development pipeline. It is reasonable to assume that within the next 10 years we will have several therapeutic options available to FA patients, perhaps to be used in different combinations, depending on each individual patient needs.

6) **Will the therapies for other ataxias be related to FA or will they require novel approaches?**
In general, therapies that address common symptoms or pathologic features that are shared among different ataxias might be effective across different groups of patients. Therapeutic approaches that address molecular aspects that are specific for individual types of ataxia, are likely to remain restricted to patients affected by that particular disease.

PRESENTING...

WINE RECEPTION FOLLOWED BY 5 COURSE MEAL AND BAND ''THE PRICKLY PEARS'

BLACK TIE EVENT

AUCTION INCLUDING RUGBY TICKETS, F1 TICKETS AND SIGNED FOOTBALL JERSEYS

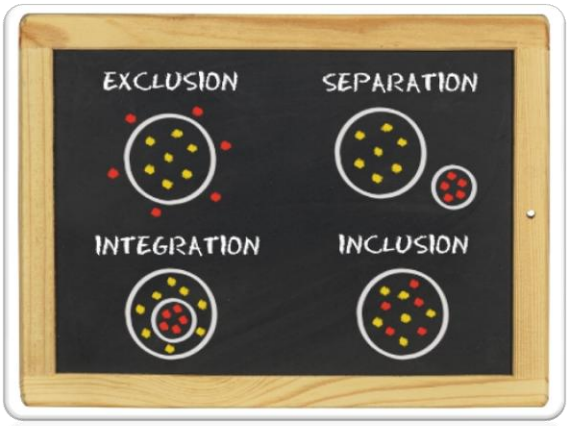
GUEST SPEAKER AND SHOWING NEW 'AWARENESS VIDEO' ON THE NIGHT

Disability Matters

At the recent seminar **"Enabling Access to Education & Employment -Digital strategies for sustainable community building"**, John Dolan, CEO of the Disability Federation of Ireland showcased DigiPlace4all
This is an online peer support community for people with disabilities developing digital skills for accessing education and employment. The seminar highlighted the discrimination against disabled people in terms of accessing employment.
Mainstream activation and employment measures such as JobBridge, Momentum, Gateway, and Community Employment for those on the Live Register exclude people on Disability Allowance. We are the only EU country to systematically exclude young disabled people from the youth guarantee.

Disability in Ireland: Some key facts

- The National Disability Survey (2006) recorded a disability rate of 18% of the Irish population
- 1 in 10 adults of working age have a disability
- 13% of disabled people experience high levels of poverty compared to 2% of those at work
- 24.5% of people with a disability have completed third level education compared to 38.7% of the general population
- Two thirds of people with a disability aged 18-34 would like to be able to work but they don't feel they'd receive the adequate support in employment.



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