Euro-Ataxia is an association whose members work together to give people with hereditary ataxia as normal a life as possible. It does this by building a strong organisation which represents people with hereditary ataxia’s throughout Europe, by encouraging scientific research into causes, treatments and by campaigning for treatments to be made available.

**Friedreich’s Ataxia receives EU funding**

It was the best Christmas present that FA sufferers could receive when they heard in December 20, 2009 that the Friedreich’s ataxia proposal, presented for funding to the EU - VII Framework Programme for Research and Technological Development (FP7) has succeeded in obtaining financing of around €6,000 over a four-year period.

This proposed project will involve the participation of 13 centres from the United Kingdom, Belgium, France, Germany, Austria, Spain, and Italy and a meeting took place on 3rd May 2010 in Brussels to discuss the project which has become known as European Friedreich’s Ataxia Consortium for Translational Studies or EFACTS. Several European scientists, researchers and clinicians came to the meeting from all over Europe and Prof Gottesfeld from USA, The meeting covered areas ranging from the structure and management of the project to specific details on Friedreich ataxia including

- FA database and registry (– you can read more about that on page 17);
- development of biomarkers
- functions of frataxin
- new disease models of Friedreich ataxia
- frataxin silencing mechanism

This project had its beginnings four years ago in Spain, under the impetus of the Spanish patient organisation, FEDAES who worked actively during 2007/08 to modify the original EU call and with the help of other patient organisations eventually obtained the inclusion of rare neurological diseases, among them the ataxias.

Researchers and neurologists at the European Friedreich’s ataxia clinical network June 2009 Turin.
The importance of spreading awareness of ATAXIA

All over the world there are problems with the diagnosis of ataxia. Dr Susan Perlman has published a booklet (see euro-ATAXIA newsletter 33 September 2008) to guide physicians in this regard. However, it is not possible to cover every eventuality as the two cases below demonstrate.

In February 2010 “Neurogenetics” medical magazine Dr Diehl, National Hospital for Neurology and Neurosurgery, London published a report of a 41 year old man who developed

- profound loss of vision
- episodic complete blindness
- spastic paraparesis i.e. the body develops spasticity and weakness
- sensory neuropathy i.e. reduced or absent touch sensation, reduced or absent pain sensation and inability to stand when eyes are closed

Such symptoms would generally signal a diagnosis of multiple sclerosis. Dr Diehl went on to say the case emphasises the need to consider Friedreich Ataxia for individuals with significant vision deficit.

In March 2010 Dr NiCuerica, Hospital for Sick Children, Toronto reported in Science Direct about a 4 year old boy who suffered a sudden cardiac death. At post mortem he was found to have a dilated cardiomyopathy (see article in this newsletter on cardiomyopathy explained) and left ventricular hypertrophy. Molecular genetic testing subsequently confirmed the diagnosis of Friedreich Ataxia.

I am sure the diagnosis of Friedreich Ataxia came as a terrific shock to the boy’s family, I am sure that they will realise in the future how fortunate they have been that a definitive diagnosis was made. It is now possible for the parents and other family members to receive genetic counselling.

Could we in euro-ATAXIA do more to highlight such incidences. Would an ataxia stand at a general medical conference help?

Recent publication on SCA

“Self-rated health status in spinocerebellar ataxia - Results from a European multicenter study”

The Movement Disorder magazine published a review on SCA from 18 different centres involving 24 different neurologists and funded by more than 40 different sources. Dr Tanja Schmitz-Hubsch, Bonn was the lead author of the article.

The study was trying to determine the variability and predictors of subjective health ratings in a possible target group of patients with SCA. Subjective health rating is how the patient feels about their own health.

ED-5D is a standardised instrument for use as a measure of health outcome. It is applicable to a wide range of health conditions and treatment. It consists of a 2 page questionnaire so it is considered short. It comprises of 5 different sections: mobility, self-care, usual activities, pain/discomfort and anxiety/depression. In addition a neurological examination was performed and a patient health questionnaire.

Results: Patients reported that their health was affected in all types of SCA. Specifically problems were reported in the dimensions of mobility (89%)

Usual activities (61%); pain/discomfort (50%); depression/anxiety 46% and self care (38%). I am sure this project would not have been possible without the initially setting up of the EUROSCA project.

EU looking for experts on rare diseases

In the Autumn of 2009 the European Commission announced that it would be appointing Members to a new EU Committee of Experts on Rare Diseases as per Commission Decision (30/11/09). It put out a call for expression of interest to be a member of this Committee - from members of Industry, patient organisations etc. It also outlined the expected role of the Committee, composition of the Committee (from the different stakeholders) and Term of Office.
Editorial

Welcome to our latest edition of euro-ATAXIA newsletter. It is wonderful to hear of the clinical trials taking place all over the world in Friedreich’s Ataxia and to read about the ever increasing knowledge of SCAs’. However it is disappointing that MICONOS trial using idebenone has failed to meet its primary endpoint (see page 9).

Our thought over the last few months have been with those who have lost a loved one through ataxia. In particular we think of Oscar Stenholm, Sweden who lost his older brother Gustav (FA) within a few days of having a scoliosis operation. Oscar is a Michael Jackson fan and he also loves to play video games. Oscar has had to deal with the reality of Friedreich ataxia and the loss of his older brother from it while he himself is still only a child of 13 years. We remember other euro-ATAXIA members including Peter Reussner, Germany and Claudia Baylier, France who have buried a loved one recently.

We also think of the Bartek Family, USA who lost their son Keith. He was in his 20’s. Ron continues his work for FARA- the American patient organisation for Friedreich Ataxia.

Many thanks to all who contributed to this newsletter and if you have information you would like to share with euro-ATAXIA from the science, politics or legislation please feel free to inform me at newsletter@euro-ataxia.eu.

Mary Kearney

EXPLANATION OF ABBREVIATIONS

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tr>
<td>ADCA</td>
<td>Autosomal Dominant Cerebellar Ataxia (Dutch Patient Organisation)</td>
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<td>AGM</td>
<td>Annual General Meeting</td>
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<td>EU</td>
<td>European Union</td>
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<td>EURODIS</td>
<td>European Organisation for Rare Disease</td>
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<td>FA</td>
<td>Friedreich’s Ataxia</td>
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<td>FARA</td>
<td>Friedreich Ataxia research Alliance (USA patient organization)</td>
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<td>iPSC</td>
<td>Induced Pluripotent Stem Cells</td>
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<td>NAF</td>
<td>National Ataxia Foundation (USA patient organisation)</td>
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Editor and lay-out by
Mary Kearney, Boherboy, Dunlavin, Co. Wicklow, Ireland
E-mail: newsletter@euro-ataxia.eu
EURORDIS & NORD join forces

The European Organisation for Rare Diseases (EURORDIS) and the National Organisation for Rare Disorders (NORD) in the USA have signed a Memorandum of Understanding to join forces on several key strategic initiatives relating to rare diseases.

"The EURORDIS/NORD partnership aims to strengthen the common international voice of people living with rare diseases, promote rare disease as an international public health and research priority, promote the development of new treatments, enlarge outreach to more patient groups and spread out networking and knowledge sharing beyond European borders.

As part of their Strategic Partnership EURORDIS and NORD will:

- Co-organise an annual global Rare Disease Day
- Establish common positions on key advocacy priorities
- Play a pivotal role in the International Conferences for Rare Diseases and Orphan Drugs (ICORD) to help expand the movement of rare diseases
- Collaborate in the development of international web media and social networking information and communication services
- Implement an international mentoring project for rare disease patient organisations
- Coordinate more closely their activities to enhance relations with the pharmaceutical and biotechnology industry

EURORDIS and NORD will kick off their collaboration with a joint public policy advocacy blog and shared online communities”.

Additional information on the new initiative can be obtained by contacting: paloma.tejada@eurordis.org

The value of diagnosis in rare diseases

Health professionals have long been so helpless in did not even dare to make a diagnosis when presented with a patient with a rare disease. Not any more. Diagnosis is now available for about 1,900 diseases, though not one single European country can provide all existing tests. Use of existing treatment and drugs has improved. Increased numbers of orphan drugs are now on the market. Rapid progress is made in advanced therapies. Today no one should have the right to say to a family: “nothing can be done”.

Eurordis Care study showed that diagnosis alone considerably improves quality of life and life expectancy by reducing unnecessary medical interventions, inappropriate treatments (including psychiatric), the birth of other affected children, maladapted family behaviour and the loss of confidence in medicine.

No diagnosis: no treatment. For example: gene therapy, recently successful for children born with adrenoleukodystrophy, must be implemented as early as possible in life to prevent irreversible brain damage.

National plans for rare disease

How to give isolated patients and families equal access to early diagnosis and care? Is it by bringing together all available expertise and resources. In 1993, Denmark organised two centres of expertise for rare diseases, then Sweden and Norway. In 2001, Italy designated regional centres. France implemented the first national plan for rare diseases from 2005 to 2008, soon to be followed by Bulgaria, Romania, Spain and Portugal. Following the Recommendations of the European Council of Ministers, plans are currently in preparation in Germany, Hungary, Ireland, Denmark, Sweden...Everywhere, patients and their associations are playing a key role throughout the process.
What do we want in a national plan or strategy?

Integration of all initiatives and resources: improved diagnosis linked to better awareness, information, training of professionals, clinical laboratories and centres of expertise; centres of expertise instrumental for research, registries and clinical trials, good practice guidelines, improved social care. Last but not least, long term sustainability of appropriate health and social policies, vital necessity for 30 millions patients affected in Europe by one of the 6,000 to 8,000 rare diseases.

**Update from France on its rare disease plan**

France's first national plan - which ran from 2005-2008 and was the first of its kind in the world. The 10 major priorities in it were outlined in the euro-ATAXIA newsletter July 2009. These have now been updated and the Second French National Plan for Rare Diseases (a four year plan to run from 2010-2014) has been streamlined to comprise 7 defined priority areas for action.

The priorities for the second plan are as follows:

"**Development Indicators**" will work for the development of diverse complex tools providing the necessary knowledge to better monitor the impact of the various actions of the plan.

"**Health Care Coverage**" will seek to better coordinate medicinal and related services usage and ensure equity.

"**Information and Training**" will reinforce support to Orphanet, improve information on disability by coordinating with appropriate national counterparts, and ameliorate rare disease training in France.

"**Organisation of Care and Diagnostics**" will pursue support for diagnostics and will tweak the actions taken vis-a-vis the centres of reference in order to meet any needs not currently covered in this area. Resources will be structured to implicate extra-hospital services as well as the country's rare disability schema under preparation.

"**Targeted Medicines**" will address the development of specific products for rare conditions and will also focus on issues concerning price and commercialisation.

"**Research**" will seek an evolution of the Institute for Rare Diseases, France's main clearinghouse for calls for proposals in the field of rare disease research. Research will continue on the epidemiology and natural history of rare diseases, genetics, and physiopathological mechanisms.

"**European and International Cooperation**" will seek enhanced collaboration with industrialised and developing countries in the areas of information, public/private partnerships, research, epidemiology, networks of expertise, etc.

The call for expression of interest outlined the required qualifications and conditions to become a member of the Committee - deadline for receipt of expression of interest 21 December 2009. We await to hear who was appointed.

**EURORDIS conference in Krakow**

Eurordis have its annual meeting in Krakow. Several members of euro-ATAXIA hope to attend. On day 3 of the conference, euro-ATAXIA vice president, Dr Paco Palau will present during the session on “Determinants for research on rare diseases”.

This session will be co-chaired Patrick Kolar from EU commission and Dr Birgit Werreraue, from Ministry of Education and Research in Germany. It is hoped that the session looks at developments in research of 7000 rare diseases, Research is one of the two main pillars in EU and national strategies. I am sure Dr Paco will be well able to justify money the EU spent on SCA research.

**Editor:** I understand that several members from euro-ATAXIA will attend this conference. I look forward to their report.
Cardiomyopathy explained

By kind permission Mary Lisa Orth in conjunction with
Dr Mark Payne & Dr David Lynch
Pennsylvania USA

Cardiomyopathy is a disease of the heart muscle and it can be classified into different types. Cardiomyopathy can be life threatening for FA sufferers. As FA is rare and cardiomyopathy in FA is rarer it is difficult for all cardiologists to be au-fait with every aspect of cardiomyopathy. Mary Lisa parent of twin boys, one of whom died from cardiomyopathy has provided the following details.

This information is designed to give the parents a better basic understanding of the heart so that appointments with the cardiologist can be better understood and hopefully this information will help you ask the right questions during the appointment.

The knowledge about cardiomyopathy in FA is incomplete. Dr David Lynch, Children’s Hospital of Pennsylvania (CHOP) is hoping to have a greater understanding of cardiomyopathy in FA after he completes the study of the echocardiograms of more than 300 FA patients. He also feels that more systematic studies of the heart in FA are needed. This would require multiple centres to collect the data due to the small numbers at each centre.

Dr David Lynch has also said most of our current understanding is anecdotal, meaning, whatever we saw happen the last time we saw a case like this and not an organized or systematic review of the heart disease. This is partly due to

1) FA being a relatively rare disease, and

2) the fact that most patients don't care about the heart until it is way late in the game.
For FA patients the neurologic impact is far more concerning at first, even though the heart has the final say.
The following information was complied using information from the National Institute of Health (NIH) website, the American Heart Association website and the Cleveland Clinic website, and is only as accurate as these sources.

http://www.nlm.nih.gov/medlineplus/
http://www.americanheart.org/
http://my.clevelandclinic.org/

**Cardiac Terminology**

- The term “cardio” means heart.
- The term “myopathy” means muscle related disease.
- The term “cardiomyopathy” simply means disease of the muscle of the heart.
- The term “hypertrophic” simply means enlargement.
- The term “hypotrophic” means progressive degeneration of an organ or tissue caused by loss of cells.
- The term “idiopathic” means that the cause is unknown.
- The term “arrhythmia” means a disorder of the heart rate (pulse) or heart rhythm, such as beating too fast (tachycardia), too slow (bradycardia), or irregularly.
- The term “supraventricular tachycardia” is a fast heart rate that originates in the upper chambers right or left atrium. A supraventricular tachycardia can also include atrial fibrillation.
- The term “atrial fibrillation/flutter” is a heart rhythm disorder (arrhythmia). It usually involves a rapid heart rate, in which the upper heart chambers (atrium) are stimulated to contract in a very disorganized and abnormal manner. This is caused by a disorder in the heart’s electrical system.
- The term “myocardial infarction” is what your doctor calls a heart attack.

**Hypertrophic cardiomyopathy**

Hypertrophic Cardiomyopathy (HCM) is a pathological description of the condition of the heart when there is a thickening of the heart muscle. The thickening makes it harder for blood to leave the heart, forcing the heart to work harder to pump.

In FA, this is usually seen in the left ventricular wall and the left ventricular septum where measurements are taken. HCM is a manifestation of FA, not a separate condition.

Symptoms of Hypertrophic Cardiomyopathy can include:

- Chest pain
- Dizziness
- Fainting
- Heart Failure
- High blood pressure
- Palpitations
- Shortness of breath
- Fatigue
- OR no symptoms at all.

**Dilated cardiomyopathy**

Dilated cardiomyopathy is a condition in which the heart becomes weakened and enlarged, and it cannot pump blood efficiently. The decreased heart function can affect the lungs, liver, and other body systems. The heart’s ability to pump blood is decreased because the heart's main pumping chamber, the left ventricle, is enlarged, dilated and weak.
At first, the chambers of the heart respond by stretching to hold more blood to pump through the body. This helps to strengthen the heart’s contraction and keep the blood moving for a short while. With time, the heart muscle walls weaken and are not able to pump as strongly. The kidneys often respond by retaining fluid (water) and sodium. If fluid builds up in the legs, ankles, feet, lungs or other organs, the body becomes congested, and congestive heart failure is the term used to describe this condition.

Symptoms can include:

- Shortness of breath
- Swelling of feet and ankles (in adults)
- Irregular or rapid pulse
- Fatigue, weakness, faintness
- Swelling of the abdomen (in adults)
- Loss of appetite
- Cough
- Chest pain
- Decreased alertness
- Failure to thrive (in children)
- Low urine production

Atrial Fibrillation
Atrial fibrillation (AF or a-fib) is the most common type of arrhythmia. The cause is a disorder in the heart’s electrical system. Often, people who have AF may not even feel symptoms or can have:

- palpitations -- an abnormal rapid heartbeat
- shortness of breath
- weakness or difficulty exercising
- chest pain
- dizziness or fainting
- fatigue
- confusion

AF can lead to an increased risk of stroke. In many patients, it can also cause chest pain, heart attack, or heart failure.

Congestive Heart Failure
Congestive heart failure (CHF), or heart failure, is a condition in which the heart can't pump enough blood to the body's other organs. The "failing" heart keeps working but not as efficiently as it should. People with heart failure can't exert themselves because they become short of breath and tired.

As blood flow out of the heart slows, blood returning to the heart through the veins backs up, causing congestion in the tissues. Often swelling (odema) results. Most often there's swelling in the legs and ankles, but it can happen in other parts of the body, too. Sometimes fluid collects in the lungs and interferes with breathing, causing shortness of breath, especially when a person is lying down.

Heart failure also affects the kidneys' ability to dispose of sodium and water. The retained water increases the oedema.

Your doctor is the best person to make the diagnosis. The most common signs of congestive heart failure are swollen legs or ankles or difficulty breathing. Another symptom is weight gain when fluid builds up.

Comment from Mary Lisa Orth: I hope this helps to clear up some of the questions about the heart, but when in doubt, consult your doctor. I have simply summarized and plagiarized from the sources listed above, in the interest of helping us to understand the cardiac issues and to facilitate your formulations of questions to ask your cardiologist. (And technically, since I am not passing this off as my own work, it is not plagiarism).
IDEBENONE CLINICAL TRIAL RESULTS –
Disappointment for Friedreich Ataxia sufferers

*MICONOS Clinical trial fails to meet its primary endpoint*

The results of the Phase III, MICONOS idebenone trial have been eagerly awaited by anyone associated with Friedreich’s ataxia especially in those countries where the government would not fund idebenone for FA sufferers. You will have read in the *euro-ATAXIA* newsletter 33 (September 2008) how Santhera had filed for approval to the European Medicines Agency (EMEA) for a licence to use idebenone for Friedreich ataxia. You will also remember that Canada did licence idebenone for use at that time. As a result idebenone became extremely expensive to purchase in Canada.

Santhera issued a press release on the 20th May 2010 see www.santhera.com. Look under media events. In this press release the results of European Phase III clinical trial of Idebenone in Friedreich Ataxia showed

1) failure to meet its primary endpoint as viewed by change in International Cooperative Ataxia Rating Scales (ICARS).

2) Data from the Friedreich's Ataxia Rating Scales (FARS) also failed to show any significant difference between the drug and placebo arms.

3) a detailed analysis of the cardiac endpoint data is still under way, no significant difference between placebo and drug arm was detected in the key secondary endpoint involving cardiac anatomy and function.

**Positive trends:** Santhera officials emphasized that some positive trends (as you would expect any pharmaceutical company that had spent over €1,000,000 on a clinical trial.

- MICONOS also confirmed that idebenone is safe and well tolerated at doses of up to 2250 mg/day.

- "Trends towards improvement in the key neurological endpoint were identified by a sophisticated statistical analysis called, meta-analysis …"

and Santhera's Thomas Meier added that “Considering the data from all three clinical studies, Phase I, Phase II, and Phase III (MICONOS) we are convinced that individual patients benefit from idebenone, although we were not able to demonstrate this conclusively in this clinical trial of still limited size and duration.”

I wonder what country will licence idebenone for treatment of Friedreich’s Ataxia in the light of this slim evidence. How would the experts from EURORDIS who have been involved in the Drug Information conference view the information from Santhera. EURORDIS have been advising patients who suffer from rare diseases that evidence of improvement must be supported by a double blind clinical trial.

While, like most in the Friedreich’s Ataxia family, I am disappointed with the results of the MICONOS double blind clinical trial, I am not discouraged. There are a number of positives to be drawn from it:

- (a) The FA family was able to recruit all the patients needed to conduct all these trials in record time.

We knew from the beginning that most drug trials fail, that many of them fail because of insufficient patient participation and that we could not afford to let that happen to us. Because so many FA patients stepped up to this challenge so promptly, we were able to give Idebenone a fair trial that was not slowed by "recruiting problems."
- (b) We have established what was necessary to get into place if the data analysis was sufficiently positive to take to the regulatory agencies to seek approval.

The Friedreich’s Ataxia family is a strong and effective family which performed extremely well in these Idebenone trials and will prevail to help other double blind clinical trials.

So, the good news is that we are still barking up one of the right trees in this mitochondrial space and we have other very promising branches on that tree.

A0001 is now in an exciting phase II trial in FA that is, once again, moving quickly because FA patients are eager to participate. In May 2010, Repligen announced that they are close to giving a date for a clinical trial.

*Editor’s note* I, personally, will be keeping a close “eye” on the exact results of the clinical trial when they are published in the medical press. I will be happy to update you at any time and I can be contacted by email at newsletter@euro-ataxia.eu

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**Changes in Dutch Dominant Ataxia patient organisation -ADCA**

*Thanks to the dutch government, the association can now really focus on the future & offer better services to its members and the public.*

*By Marco Meinders*

Founded in 1994, the Autosomal Dominant Cerebellar ataxia (ADCA) association has 500 ataxia members who have ataxia or have a family member with ataxia and 100 non ataxia members who give money to the organisation. Initially, funding came from contributions from members, non-ataxic members, and subsidies from the Ministry of Health and private organisations.

Now, the Ministry of Health provides structural funding for the organisation itself and for informational services, meetings, lobbying and representation. It also gives a subsidy for structural organisation. This has some profound effects on the association.

The association now has an office, a staff member, an organisation development advisor, and a lot of volunteers. This enables the board to focus on steering the association into the future. Before, the board members were too busy with day-to-day activities. It also means that a board member can now leave the board, knowing that another volunteer will step in. My parents, two of the founding members, are the first to use this opportunity. Originally, they thought that they would be on the board for a maximum period of five years. That was seventeen years ago… Of the original group of eight founding members, only two are still on the board.

You may have noticed that I have not mentioned funding research. We have been involved in the margins of research. Since 2004, we give money to cover some costs related to obtaining and preparing ataxic brains from post mortem pathology. (Brunt, De Vries, Rüb). Sometimes, we pay a portion of the printing costs of a thesis or dissertation. We have supported research funding applications such as ‘Protein clotting and the formation of ataxia’ (Kampinga et al, 2005) and research into fatigue, mood and sleeping (Brusse, 2010). The money comes from gifts from individuals, labelled exclusively for research.

*Further details can be obtained from ADCA Association of The Netherlands*

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Dr Ewout Brunt retires

by Balthasar Schaap

On 29 October 2009, Dr. Ewout Brunt retired from his position at the University Medical Centre in Groningen, The Netherlands. It was an impressive farewell with many patients, patient organisation representatives, colleagues and employees.

Over the years Dr Brunt had given several lectures on varied topics from Dystonia, Parkinson, Immunology and of course Ataxia. The many speakers at his farewell praised Dr. Brunt for his dedication, perseverance and communication skills with patients and employees.

The ADCA Association, of which Dr. Brunt is an honorary member, gave him a framed certificate to express their appreciation for all his help. Dr Brunt was always on hand to help euro-ATAXIA as you can see from the picture above

On the night many stories about Dr Brunt were told but the ones that fascinated me most I will share with you. Dr. Brunt did the bulk of his medical training in Groningen and graduated in Rotterdam. After several years in Malawi, he specialized as a neurologist in Rotterdam under Professor Steel. After several years he settled as a neurologist in Winterswijk. One of the first patients he had was a woman with ataxia. SCA 3, as it turned out. This condition fascinated him so that he soon started a family tree and did research on this disease.

After Dr Brunt was appointed to the University Medical Centre in Groningen, he could immerse himself in research and formed a research group with a pathologist from Winterswijk (NL), California (USA) and Frankfurt (D). For research, he needed brains of deceased persons with Ataxia. Dr Brunt had the courage, motivation, communication skills and network to convince people in life to allow after death that their brains could be used for his research. Few could ignore this request.

Through this research, we know much more about the various SCAs, the size and amount of damage they cause. There is not only damage in the small brain or cerebellum, but also in the medulla oblongata and the nerves that come from the spinal cord and brains. Also, a method has been developed to detect the damage in the brain and this has helped identify the natural course of the disease. This will help clinical trails for dominant ataxias when they do eventually come.

I very much appreciated my colleague Ewout for his perseverance, passion and research into a chronic degenerative rare disease like Ataxia.

Ewout, “I wish you relaxing retirement years with lots of time to walk. Thanks for your efforts as a neurologist. Hopefully, you do not stop with promoting the study of Ataxia and your volunteer job as a medical advisor to the ADCA Association”.

Editor’s note: I am sure I speak for all euro-ATAXIA members when I wish you well in your retirement.
Friedreich Ataxia Research Alliance (FARA) (www.curefa.org)

FARA presented a therapeutics symposium at The Children’s Hospital of Philadelphia, USA on 17-18 July 2009. Dr Alison Stevenson, research officer, Ataxia UK did an extensive summary which is available on the website www.ataxia.org.uk. The main news for Friedreich ataxia sufferers was that the idebenone trial had been extended on an open basis for a further 12 months. Progress was reported on the Histone deacetylase inhibitors but no date has been set for a clinical trial.

FA - Symposium Nov 2009 – Philadelphia, USA

A parent’s view
The second annual Friedreich’s Ataxia symposium was held on Saturday November 14th 2009 in Philadelphia. Organised by the Children’s Hospital of Philadelphia the symposium began with a ‘meet and greet’ session on Friday evening. Families young and old were in attendance. Dr David Lynch welcomed everyone and introduced Dinah, who suffers from Friedreich’s Ataxia (FA). Dinah gave a superb demonstration of wheelchair ballroom dancing along with her able bodied dance partner Rob. Light refreshments were served and people had an opportunity to catch up with old friends and meet new families.

‘Positive Exposure’
Saturday morning commenced at 7.30am with registration and breakfast. Following this Rick Guidotti, an award winning former fashion photographer addressed the meeting. Rick is the founder and director of ‘Positive Exposure’ an innovative arts, education and advocacy organisation working with individuals living with genetic difference. Rick’s photographs challenge us to see the individual living with genetic difference as human beings first and foremost, with their own special needs rather than as their disease. He took photos of adults and children with Friedreich’s Ataxia and intends to place them on his website, www.positiveexposure.org

Following a short break Richard Currier, Keith Dalton, Jennifer D’Antoni and Allison Rice addressed the audience. All four suffer from FA. They talked about the impact that FA has on their lives and careers. Issues such as the importance of honesty, dealing with fatigue and coping with difficulties were discussed.

Biochemical mechanisms in FA
Dr Robert Wilson then spoke about biochemical mechanisms in FA. He outlined the structure of DNA and the various biochemical issues that arise when a person has FA. He explained that FA results in an underlying vicious cycle and that there are a number of therapeutic approaches under investigation. These include Parabenzoquinones (CoQ10, Idebenone, A0001), Iron Chelators, Histone deacetylase (HDAC) inhibitors, Erythropoietin (EPO) and Tat-frataxin. He noted that as there are pros and cons to every therapeutic approach the Friedreich’s Ataxia Research Alliance (FARA) has cast a wide net in searching for a cure.

Scoliosis in FA.
Orthopaedic surgeon Dr John Flynn talked about scoliosis. He noted that there is limited information on scoliosis in Friedreich’s Ataxia patients and that research needs to be conducted. He addressed the issue of bracing and was of the opinion that to date it did not seem to be of benefit when treating scoliosis in FA patients. He also talked briefly about surgery and the fact that post surgery recovery is significantly longer for the FA patient. The danger of fluid overload during scoliosis surgery was emphasised. Mary-Lisa Orth, whose son died following scoliosis surgery, has written an article on this matter.

Neurological perspective of FA
Dr David Lynch guided us through neurological perspectives on FA after lunch. He explained the classic description of neuro-degeneration in FA. Of significance is the loss of balance due to a lack of input on where the limbs are located in space. In addition there are speech articulation difficulties.
and subtle eye movement abnormalities. He noted
the there is normal cognition. Dr Lynch believes
that there are clear differences between early and
late onset FA and those with point mutations. These
differences lead neurologists to re-evaluate and ask
new questions. It is hoped that this will prompt new
approaches to possible treatments. Finally the issue
of regaining function was addressed. Recent
research has shown that physical therapy has a
beneficial impact on motor performance.

**Eye symptoms in FA**
Dr Laura Balcer dealt with the neuro
ophthalmological issues in FA. She advised that FA
does have visual manifestations and that patients
should have an annual eye test. She noted the
necessity of getting the best corrective glasses or
lenses and the importance of using proper lighting.
The issue of diabetic control is also of significance.

**Heart problems in FA**
After a short break Dr Robert Shaddy discussed
cardiac issues in FA. Left ventricular hypertrophy is
the most common clinical manifestation in FA.
Patients should undergo annual ECG and
echocardiogram and where necessary, cardiac MRI.
An indebt analysis of cardiac treatment in FA can
be assessed on a recent review, October 2009 at
www2.cochrane.org/reviews/en/ab007791.html.

**Research pipeline in FA**
Jennifer Farmer took us through the research
pipeline and pathways to clinical trials. Of
significant importance is that there are 7 clinical
trials and 8 approaches underway at present. She
reminded the audience that they should include their
details on the FARA patients register. It is clear that
there is great cooperation between the different
researchers around the world and that this
willingness to share information will help to speed
up the search for a cure.

**Kyle Bryant fundraising ride**
The inspirational Kyle Bryant concluded the
conference. Suffering from FA, Kyle founded Ride
Ataxia three years ago and has raised over $800,000
for research. Photos from the Philadelphia Ride
Ataxia event highlighted the success of this
endeavour. Kyle’s ride usually takes place in
conjunction with the NAF conference held annually
in March. This year, 2010, it was decided in view of
the variability of weather in Chicago in March to
postpone the ride until the summer.

**In conclusion the FARA symposium…**
was excellently arranged and presented. There is no
doubt that some of the detailed scientific
information is challenging and, at times, difficult to
understand. That said it behoves parents to learn all
that they can about this rare condition. Even taking
into account the distances to be travelled and the
expenses involved this symposium is great benefit
to anyone who has or who cares for someone with
FA.

The fact that the number of people
attending doubled since the first symposium in
December ’08 highlights the need for such a
conference. Plans are already underway for next
year’s event!

**Association Française de l’Ataxie de
Friedreich (AFAF)**
AFAF held their annual conference in Nouant on
10-11 April 2010. It was well attended and
participants were able to hear of the latest
developments in France on fundamental research
form Drs’ Pierre Rustin and Helens Puccio.

Prof Munnich was there to talk about the clinical
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**Federación de Ataxias de España
(FEEA)** will hold their annual meeting on 25-27
June 2010 in Villagarcia de Camopos, Valladolid.
There is a session on basic research and clinical
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National Ataxia Foundation (NAF) Conference

NAF held its annual meeting in March 2010 in Chicago with the title “Winds of Progress”. This year it was preceded by the Ataxia investigators meeting (AIM). At the AIM meeting Episodic ataxias were discussed at one of the morning sessions. SCA 2,3,5,6,7,8,10 &17 were dealt with in detail. The keynote address was given by Dr Ole Isacson, Harvard University, USA on “Stem cells and Induced Pluripotent stem cells (iPSC): Paradigm shifts in cell therapeutics and discovery for neurodegenerative diseases.

What are iPSC?

The ground breaking discovery that mousse (Takahashi and Yamanaka 2006) and more recently human (Takahashi et al 2007; Yu et al 2007; Park et all 2008) skin fibroblastic cells have the capacity to be reverted to an embryonic stem cell fate has significantly advanced the field of stem cell research as well as the study of neurodegenerative diseases.

By employing gene altering technologies specific “stem cell” genes were forced into DNA of the fibroblasts, resulting in reprogramming and an adoption of human embryonic stem cell (hESC) characteristic at the expense of fibroblast cell properties. Since it is now possible to have induced pluripotent stem cells (iPSC) from adult skin, four very important implications arise.

1) a fertilized embryo is no needed for production of iPSC – thus reducing ethical concerns about stem cell treatment

2) iPSC could theoretically be generated from any patient population to study ataxia.

3) As the skin cells carry the code of the patient, there is no more need to artificially create any genetic abnormalities

4) The potential of using patient specific iPSC for small molecule screening and therapeutic testing could vastly improve the developments of treatments

Dr Allison Ebert, University Wisconsin, Madison, USA has also studied this area intensively.

At the AIM four groups meet to collaborate on processing and making available for the first time Friedreich ataxia heart cells and nerve cells. How skin fibroblasts will be harvested from FA sufferers and then be teased to become pluripotent skin stem cells. These will then be directed to form heart and nerve cells, which will be used for research. In the words of Paul Konanz, Californian father of FA and whom spoke at euro-ATAXIA’s Paris conference ”This is a big deal”.

The annual membership meeting took place on Friday, Saturday and Sunday with researchers and neurologists giving freely of their time and energy. The varied and interesting presentations for this meeting are available on the web at www.ataxia.org/events/Presentations-Chicago2010.aspx. Many thanks to all at NAF for making them available to us all.

FARA Australasia www.fara.org.au

The big news from “down under” is the ability of scientists at the Monash Institute of medical research, Melbourne making iPSC (induced pluripotent stem cells, from the skin of FA sufferers. This was also discussed at length at the National Ataxia Foundation conference held in March 2010 in Chicago.

The Australians also investigated the upper limb function in Friedreich ataxia suffers. A report from this research is discussed on page 18 of this newsletter.
Cerebellar -C7 Network –Meeting in London

By Julie Greenfield
Ataxia UK

C7 is Cerebellar cortical control: cells, circuits, computation and clinic

This project is funded by European Commission (FP7) University College London - Marie-Curie Initial training network (FP7). A group of European researchers have been awarded a large grant to study the cerebellum. They held their first meeting 11-12 March 2010 at the Institute of Cognitive Neuroscience, University College London. euro-ATAXIA patient representatives Julie Greenfield Ataxia UK and Peter Reussner (Deutsche Heredo-Ataxie Gesellschaft – DHAG) were invited and their patient organisation are named partners in this project.

Introduction
This project involves nine groups from Institutions around Europe and it was successful in getting funding from a Training funding stream of the European Commission called Marie Curie, who was a physicist and chemist of polish upbringing. Marie Curie She was a pioneer in the field of radioactivity and was the first person to be honoured with two Nobel prizes in chemistry and physics. Thus, a major focus is on training of PhD students in different techniques and integrating research in academic setting with industry.

The overall aim of this project is increasing our understanding of the cerebellum. The programme runs from November 2009- October 2013 and the budget is €3,600,000. The grant provides funding for 17 posts. Although it involves some clinical work the emphasis is on basic research.

Scientific Presentations
At the meeting the majority of the talks focused on understanding how the cerebellum works and which parts of the cerebellum are involved in different functions. Some researchers will be working on understanding what happens at the cellular level and others will focus on how the neural networks work and involve computer modelling work. There were projects dissecting the coordination of eye movements, arm movements and individual finger movements and how these are disrupted in people with cerebellar ataxia.

One clinician in Germany will be doing a physiotherapy intervention trial in people with cerebellar ataxia. He will try and relate the effect of exercises on changes seen within the cerebellum. In another project, collaboration with researchers in the UK will study people who have had cerebellar lesions after a stroke and see how their recovery in time can be related to changes within the cerebellum and to identify which parts of the cerebellum are showing changes as a result of recovery. This may have implications for patients with degenerative cerebellar ataxias too.

For more information on the project visit the website: http://www.cerebellumc7.eu/

Outreach discussion
Outreach work is this context means reaching out and involving groups directly affected by ataxia in this project. This is done so that researchers will know what affects ataxia patients and a useful collaboration can be established.

Some funding is available for outreach activities and a very useful discussion was held on the types of outreach activities that could be done. This session was attended by the two patient representatives and most of the researchers present. It is now considered that outreach activities are an integral training aim for the young researchers. The countries involved in this project are the following: Belgium, Germany, Netherlands, Israel and UK.
Outreach activities for C7

Ataxia awareness day – it was agreed this would be a good place to do outreach activities. Some funds could be used to facilitate press work (Ataxia UK had had some success in this during last awareness day). It was agreed that an event around awareness day should be held in 2011. Each University from the countries taking part in this project would organise an event in September 2011, such as an Open Day. This would involve both people with cerebellar ataxia and the general public.

Press work – In order to get information out to the general public press releases on each project should be ready in advance that could be used by the press when the timing was right.

Attendance at euro-ATAXIA meetings – It was felt that this would be a good forum for dissemination of this research to people with ataxia around Europe. The researchers have been put in touch with AISA, the organisers of the 2010 euro-ATAXIA conference to discuss the possibility of attending and presenting the work.

Presentations at national ataxia meetings – This would be a useful forum for research dissemination to people with ataxia and each researchers could contact charities in its country to do this. Representation at future meetings of this project – Peter Reussner suggested inviting ataxia charities from the country in which the meeting was taking place to future meetings.

Dissemination via website – It was agreed that for each article written there would be a lay summary and information on why the research is important to patients. Ataxia UK could provide support with this. Translation of material should be considered and funding could be used for this.

Dissemination via other organised events – It would be useful to have presentations at other events in each country that are already taking place, eg: British Science Week.

Conclusions
This was a very informative initial meeting with plenty of time for discussion. The project should yield much useful information on how the cerebellum works and what goes wrong in people with cerebellar ataxia. Patient representation and input is very welcome. For more information on how to get involved contact: Joern Diedrichsen (j.diedrichsen@ucl.ac.uk).

Speech problems -dysarthria in Ataxia

It has been noted for a long time that ataxia sufferers have problems with their speech. Symptoms have been described in detail in the dutch produced “Revalidatie geneeskundige richtlijn” Medical guidelines for Friedreich Ataxia. This has recently been translated to English and is available from newsletter@euro-ataxia.eu

As we all know scientists and clinicians are trying to get reliable methods of measuring the progress of Friedreich Ataxia over 12-24 months. In this article we look at the different problems associated with speech in ataxia and the latest research in this area of documenting speech problems. To date they have not been able to assess speech disorders in this regard.

Speech problems vary from
1) loudness
2) melody of speech
3) poor accuracy of sounds
4) difficulty in timing speech.

The coordination of various systems which work together to produce speech is affected.

Prosody

Prosody is the rhythm, stress and intonation of speech. Prosody reflects various features of the speaker or the words that are being communicated; the emotional state of the speaker, whether the communication is a statement, a question or a command, whether the speaker is sarcastic or ironic. Therefore a problem with prosody can lead to misunderstandings in everyday life for ataxia sufferers.
UK - Ataxia and speech research

Dr Anja Lowit, Bangor, North Wales published results of a 3 year study on speech disturbances in ten ataxia sufferers (did not specify which type) and 10 healthy volunteers.

Results:
The main findings were

a) ataxia sufferers spoke slow than the healthy people
b) the more severe the speech problem the more likely the ataxia sufferer had rhythmic disturbances
c) ataxians found it more difficult to put the correct emphasis on words in sentences i.e. object and object; and The lawyer came to London versus The lawyer came to London.

The data highlighted that these problems were caused by a combination on controlling speech timing (i.e. the duration of syllables and words) as well as loudness and pitch variation.

Australia – Friedreich Ataxia and speech

Meanwhile in Australia at the University of Queensland Two papers on speech were published by the same group. In February 2010 Dr Rosen reported that he recorded the speech of 13 patients and 18 healthy participants. They measured speech in 38 FA patients, and 20 healthy controls

They used 30 different dimensions of speech and the AIDS speech test.

Results:
An analysis of results showed 3 sub-groups
1) mild dysarthric symptoms
2) increased velopharyngeal involvement
3) increased laryngeal dysfunction
They did not say that their findings confirmed that speech could be used a method of assessment disease severity in clinical trial.

Conclusion:
They found that dysarthria severity showed a significant correlation to disease duration but to no other clinical outcome. This conclusion is similar the conclusion which is presented in the Dutch document “Revalidatie geneeskundige richtlijn”

USA - Friedreich ataxia and speech

In Jan 2010 Dr Singh in conjunction with Dr David Lynch, Jennifer Farmer and others at University Pennsylvania, USA published clinical measures of dysarthria in FA. The study had 22 FA sufferers and 16 control participants. Five 5 different ways to assess speech in an effort to see if they were reliable and would all give a consistent result.

1) PATA
2) PATAKA subjects were asked to repeat these words as many times as possible in a defined amount of time
3) Oral motor examination involves repeated articulation of specific words
4) Assessment of intelligibility of dysarthria speech(AIDS) is carried out by asking participants to say 50 specific words and assess them for intelligibility. The words were recorded and assessed by two blinded observers.
5) Cookie theft picture description task - In this test the subject is asked to describe the scene involved in a picture of a child taking a cookie

Results:
The first 4 methods demonstrated significantly lower scores for patients with FA compared with controls so any of them could be used in patients with FA particularly those who are unable to be assessed using measures of arm or leg function. However other problems did emerge.

PATA & PATAKA speech test had a limited range of score. The score for this test does not change significantly until late in the disease. The oral examination is easily graded and relatively short but also has a limited range of score. The AIDS speech test is highly specific and reproducible but it is time consuming. Long examinations are not ideal for FA patients

Conclusions:
The phonetic profile of dysarthria in FA is currently being better elucidated by speech pathologists and hopefully they may be able to come up with an easily administered measure which can assess dysarthria in FA.
The latest on Ataxia Rating scales

The ataxia clinicians are constantly looking for better ways to assess ataxia. As anyone who has attended ataxia conferences will tell you, there is always an interactive discussion when the topic is mentioned. Recently, Europe (Germany and Austria), Australia and USA have published information about ataxia rating scales.

**Europe compares 3 ataxia rating scales**

This time report come from Dr Jorg Schutz, a well known researcher and neurologist in Göttingen, Germany had an interest is developing a database for ataxia patients. Dr Schutz recently published a paper on rating scales in Friedreich ataxia. The publication is a result of the research activities of the German Network of Hereditary Movement (GeNe Move) Disorders that was funded from 2003 to 2009 by the German Ministry of Research and Education. With the euro-ATAXIA money, some additional money from the German national patient organisation (DHAG) and the pharmaceutical company Takeda Dr Schutz build a European database of Friedreich’s ataxians.

The initial results from the database were presented at the EFACTS meeting (see page 1) in May 2010 in Brussels.

This study compared three different rating scales

1) Friedreich ataxia rating scale (FARS) - This is an elaborate disease specific rating scale that was developed specifically for the sensory abnormalities associated with FA

2) International co-operative ataxia rating scale (ICARS) - This was the first scale convened for ataxic symptoms in general by a group of international neurologists. To date it has been used in most clinical trials for FA and has good inter-rater reliability. Concerns have been raised over its practicability in daily routine and its subscale structure with special emphasis on limb ataxia

3) Scale for assessment and rating of ataxia (SARA) - This was generated for use in dominantly inherited ataxias by the EUROSCA group. It is shorter and simpler than ICARS

The quality of a rating scale is not only defined by its inter-rater reliability and validity but also by its user friendliness. SARA takes only 15 minutes per patient, so this may be a consideration in the long term. SARA examination included a section on eye examination which may not be relevant in FA where eye problems are commonly moderate even in severe ataxia.

**Results:**

The recommendation from this study is that SARA is well suited and shows good reliability for FA and could be used in clinical trails for FA.

Ireland was not included in the EU application of EFACTS but it was decided at the EFACTS meeting to open that database for Ireland as well. What other are our Swedish colleagues included? Are other countries excluded?

**USA measures rate of progression of Friedreich’s Ataxia (FA)**

Meanwhile the Americans were measuring the rate of progression in FA with a view to assessing the implications for clinical trial design. This study had patients from 12 different centres in USA over a 2 year period. Initially 236 patients were examined but only 168 patients presented for follow-up 1 year later. This study used

1) FARS

2) Multiple Sclerosis Functional composite - 9 hole peg test for arm function; a time 25 foot walk for ambulation; speech test using phrase "PATA", and a low contrast letter vision test(LCLA)
Study on Progression of FA continued:

Results:
The PATA speech test was not a good way of measuring the progress of FA over a 2 year period. Neither did the vision test, LCLA, The 9 hole peg test and the 25 foot walk results reflected disease progression and revealed more linear progress over the two years than the FARS.

Conclusion
This research suggests that studies designed to assess FA progression in unselected population (as was the case in this study) will almost certainly require a two year duration. These results help to establish norms for progression in FA that can be useful in measuring the long-term success of therapeutic agents and defining sample size calculations for double blind clinical trials.

Australia measures upper limb function in Friedreich ataxia

This study was carried out in Brisbane in conjunction with Prof Delatycki who you will know from his visit to the euro-ATAXIA conference in Dublin in September 2008. Dr Corben led the group which examined upper limb function in 38 genetically confirmed FA patients. (On note there were 38 patients in the speech study done in Brisbane and most of the authors for both studies are similar).

The recommendation from upper limb study was that the 9 hole peg board test was a reliable measure of hand function particularly when performed by the non dominant hand.

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A.I.S.A. – Associatiazione Italiana per la lotta alle Sindromi Atassiche

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**euro-ATAXIA board member** – Fedricia Crimaldi has advised us that the Italian patient organization plan to host the 2010 euro-ATAXIA meeting in Cervia. Cervia is 27 km from Rimini and I understand that Ryan air fly from Bristol, Frankfort- Hahn, Stockholm Skavsta to Rimini on a regular basis. I am sure other airlines also fly into Rimini and Forli. We look forward to further details.
You will have read in an earlier euro-ATAXIA newsletter of how an Israeli boy who had ataxia travelled to China for what his family were told “life saving stem cell injections”. These stem cell injections caused the boy to develop several brain tumours. Cells of this brain tumour were of female origin proving that they were caused by the stem cell injections rather than by the ataxia itself.

In response to such situations The International Society for Stem Cell Research (ISSCR) has convened a new committee tasked with weeding out companies that offer unapproved stem cell ‘therapies’, the ISSCR’s new president Irving Weissman announced at the World Stem Cell Summit in Baltimore, Maryland.

In December 2009, Weissman, who also directs the Stanford Institute for Stem Cell Biology and Regenerative Medicine in Palo Alto, California, wrote an opinion article in *Cell Stem Cell* calling for stem cell centres to be judged on three criteria.

- **First**, the company should be able to cite peer-reviewed papers from third party investigators showing that the therapy is possible.
- **Second**, there should be institutional review board oversight of the treatment.
- **Third**, the US Food and Drug Administration or an equivalent agency should give the final green light. "That's the minimum beginning," he said at the meeting.

Weissman revealed that he had convened an 18-member panel of lawyers, FDA regulators, medical ethicists, and stem cell scientists last week to look into the feasibility of establishing an online registry of wayward companies. His idea is for the ISSCR supervisory body to request documentation of the three requirements from all stem cell providers. Companies that don't comply would get blacklisted.

In December 2009, The ISSCR committee saw the National Institute for Health announce the first hESC line eligible for funding.

The ISSCR did issue “Guidelines for the clinical translation of Stem cells” in December 2008 see www.isscr.org. The guidelines off recommendation founded on general principles for scientific, clinical, and ethical conduct that should be followed by all translation stem cell researchers, clinician-scientists and regulators in the international community. The were developed by a multi-disciplinary group of stem cell researchers, clinicians, ethicists and regulatory officials from 23 countries.

**About ISSCR** It is s an independent, non-profit membership organisation established to:
1) promote and foster the exchanges and dissemination of information and ideas relating to stem cells
2) encourage the field of research involving stem cells
3) promote professional and public education in all areas of stem cells research and application.