

*From the European federation of hereditary ataxias (Euro-Ataxia)*

### **Promoting awareness of hereditary ataxia in medicine, science and society.**

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

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#### **Euro-Ataxia at European congress of neurologists**

### **11th Congress of the European Federation of Neurological Societies**

**Brussels, Saturday 25 – Tuesday 28 August 2007**

Some years ago, we joined the European Federation of Neurological Associations (EFNA, erfna.net). This umbrella organisation of neurological patient advocacy groups aims at improving the quality of life of people living with neurological conditions, their families and carers. It maintains a close relationship with the European Federation of Neurological Societies (EFNS, efns.org). One of EFNS's aims is to support research and the dissemination of research results throughout Europe. Currently, EFNS represents 40 European national neurological societies and more than 12,000 European neurologists.

To help us raise awareness, EFNA kindly offered us a booth in the exhibition hall, which we accepted. Based on a document agreed upon by our Medical and

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Scientific committee at their last meeting, Mary had made several folders and brought a lot of these with her to Brussels. There, we filled our booth with our folders and newsletters, and prominently displayed folders of our member organisations. Thus it happened that Mary, Helen and myself found ourselves talking to neurologist from everywhere but the north and south poles, handing out information and referring some of them to our member organisations.

Our folders were in popular demand and we ran out of them on the second day already. Fortunately, Mary and Helen managed to have more copies made.

One neurologist surprised us by saying that he thought there were only 5,000 people with Friedreich's Ataxia in the world – whereas it is estimated that there are at least 11,250 in Europe alone... not to mention other ataxias!

Looking back at the event, we conclude that we made a lot of neurologists aware of our existence.

*Helen Rikken-van Wijk, president*  
*Dr. Mary Kearney, secretary-general*  
*Marco Meinders, treasurer*

### **Friedreich's Ataxia patient associations adopt strategy**

#### **Result of the Friedreich's Ataxia (FA) Associations' Workshop 30-31st March 2007**

As it is important that

1. we have valid successful clinical trials,
2. that we enrol sufficient patient participants for clinical trial
3. we clearly identify to the patient population, parameters of each clinical trial
  - a. time lines \ schedule
  - b. inclusion \ exclusion criteria
  - c. explanation of therapeutic intervention
  - d. primary and secondary end-points
  - e. consent \ assent (for children)
  - f. full and prompt report to patient participants of results and to the public regardless of success or failure
4. We insist on prompt application to the appropriate regulatory agencies for approval of successful interventions (medication or devices)
5. Friedreich's Ataxia (FA) patients have no treatment, so we must get the fastest possible effective treatment.

Therefore our group undertakes the following:

1. to strongly encourage all our investigators around the world, to work closely together, to advance these clinical trials in the shortest time.
2. to ensure that our patient organisations around the world work together to enrol the maximum number of patients interested in participating in these

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trials. We commit:

- a) to get all patients on 1 register, we believe that FARA registry facilitates this currently
  - b) to encourage patient organisations to place a link on each of their web sites to the FARA registry with an encouragement and explanation of the importance of registration
  - c) to provide information from the FARA registry to all clinical trials around the world.
  - d) to seek additional detailed information on each patient during the initial portion of each clinical trial led by the clinical trial physicians
  - e) to monitor the progress of each clinical trial and report as fully as possible to all of our patients by providing information to the patient associations
3. to encourage the development of pilot trials, in particular, for drugs which have been already approved for other diseases, for their potential efficacy in FA
  4. to keep additional potential drugs in the "pipeline", at the same time as these current drugs are in clinical trials, for potential trials in the future, until we have a fully effective treatment for FA.
  5. to insist that researchers and pharmaceutical companies, before starting a trial must involve all the appropriate patient representatives (at least 2) with a goal of fully involving them in the early planning stages as well as implementation. "Involvement" is intended to include genuine participation in decisions regarding clinical trial planning, design and implementation that impacts on FA patient participation, safety and benefits

We look forward to continue building and nurturing global public-private partnerships that involve patients, patient advocates, governments, pharmaceutical companies and investigators, that will provide full support and motivation for accelerated progress in discovering, developing and testing potential therapeutic treatments.

We recognise Bernardo Ruggeri as European representative, Ron Bartek as American and Australasia representative or other representatives as they themselves may appoint.

**This document has been read and approved in principal by the people below and they now seek full agreement of their organisations as soon as possible no later than April 18th 2007 in preparation for the organisational meeting for the clinical research network in Brussels on April 20-21 2007.**

- GoFAR Mina D'Agostino
- FARA Australasia Varlli Beetham
- FARA- USA Ron Bartek
- AFAF Robert Breniaux
- FASI Tom Kelleher
- *euro-ATAXIA* Mary Kearney

## Rare Diseases in Europe: EURORDIS

### Report from EURORDIS Annual General meeting 4 & 5 May 2007 Paris

At the EURORDIS AGM in Paris, organisations of people with ataxia were represented by (amongst others) Dr Mary Kearney (Ireland), Dr Juliette Dieusaert (Hirson, France) and Christian Gabrillague (Paris, France).

#### What is EURORDIS ?

EURORDIS is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe.

It was founded in 1997 by four patient groups from different therapeutic fields including Muscular Dystrophy and Cystic Fibroses, AIDS and a Cancer patient Organisation called, Ligue Nationale Contre le Cancer. Today, it is supported by its members and by the French Muscular Dystrophy Association (AFM), the European Commission, corporate foundations, and the health industry. Currently EURORDIS represents a vast number of people and organisations. At the recent conference there were patients representatives from Argentina, Africa, Europe and America.

#### Ten years Eurordis

The Eurordis 10 year anniversary Membership Meeting included a European Workshop on "Gaining access to rare disease research resources" on 4 and 5 May 2007 at the **Pasteur Institute** in Paris saw 270 participants from 27 countries getting together and discussing issues at the heart of patients. The audience was made of 75% rare disease patient representatives and 25% researchers, healthcare professionals, industry representatives and public policy makers. The European workshop was part of the *Capacity Building for Patient Organisations in Research Activities* project (name code: CAPOIRA). 'It was a milestone in the empowerment of patient groups on the way to playing a greater role in the European research agenda,' says Fabrizia Bignami, Eurordis, who was in charge of the workshop. The workshop, which lasted a day and a half, saw presentations on rare disease research resources and integration of patient groups in research. Two sub-groups were asked to meet and debate on the needs and wishes of patient groups on two different topics: "European tools for rare disease research", and "getting involved in research".

The following main points and needs came out of the discussions:

Basic requirements:

- Push for national plans for rare diseases (including research policy) in Member States
- Raise awareness on rare disease research amongst EU decision makers
- Provide more opportunities for patients and EU decision makers to work

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together

- Establish EU-wide Committees for rare diseases rather than national ones
- Keep providing information in national languages.

European project applications:

- Simplify the procedure for getting support during the preparatory phase;
- Implement a two step approach: expression of interest first, and full application when selected;
- Create a permanent advisory/support desk at EU level within Eurordis, dedicated to rare disease project development;
- Publish a who's who of the European Commission, with phone numbers and email addresses
- Organise training sessions on constraints of research, drug development process, clinical trials and related issues for patient representatives;
- Disseminate information on research resources possibilities in the EU;
- Allow for core funding of patient groups;
- Create longer-term support for research projects with long term aims (eg projects on natural history of the disease)
- Develop more research projects on quality of life of patients
- Calls for proposals: provide help at EU level; develop tools to help patient groups organise their own international calls for proposal

Research tools:

- Rare disease biobanks: use EuroBioBank as a starting point for the creation of new high-quality biobanks; make researchers using biobanks understand that they need to share results with patients
- Patient databases and registries: organise training sessions for patient representatives; write and disseminate EU guidelines on the creation of databases; develop databases linking genotypes and phenotypes that can be operated or supervised by patient groups with the support of specialists
- Clinical trials: create an EU centralised registry of clinical trials and publish anonymised results; organise training sessions on protocols, methodology and analysis for patient representatives; ensure results of clinical trials are fully used by the research community

The results of this Eurordis workshop were presented at another European policy development workshop on rare disease research organised by the European Commission in Brussels on 14 June 2007. 'It was an opportunity to further stress the role of patients in EU research,' says Fabrizia Bignami, who participated in this meeting. The outcomes of the 14 June discussions will in turn be presented at a conference entitled 'Rare Diseases Research: Building on Success', which will take place in Brussels on 13 September 2007, with members of the EU Parliament, representatives of rare disease research funding institutions, national and European health and regulatory authorities, researchers, and patient organisations. Another opportunity for the rare disease community to express its needs in term of research, and another step towards full participation of rare disease patients in the European research agenda and decision making process - something Eurordis has

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long fought for, and always will.

There was a number of satellite meetings organised on Thursday 3 May 2007. Dr Mary Kearney took the opportunity to attend Kick-off meeting of the Drug information, Transparency and access Task Force (DIT-TF). There were 12 participants at this meeting with a representative from Sweden, Finland, Germany, UK, Spain, Romania and 4 representatives from France. This task force was set up to

- Train its members on the respective roles of national and European regulatory agencies and proposes actions to train other patient organisations
- Learn the marketing authorisation procedures, their timelines (for first indication and variations), and review the implementation guidance documents
- Make proposals to improve the provision of medicinal product related information adapted to patients and consumers needs, for example, package leaflets, labelling, summary of product characteristics
- Make the Patients Consumer Working Party and European Medicines agency work plan clear
- Access what patient organisations can achieve

A document on guidelines on clinical trials in small population was also made available to the group. A work plan was drafted and reading was recommended. The next meeting takes place in conjunction with EURORDIS

Part of the text of this summary has been given to by EURORDIS, many thanks to them for their help in this regard.

*Mary Kearney*

### **EURO-ATAXIA at annual meetings of French and Spanish member organisations.**

#### **The French association for Friedreich's ataxia**

AFAF annual meeting took place on the 12-13th May 2007 in Nounant. This is a building about 50 years old. It is wheelchair accessible and is situated in a forested area. It is 150kms far from Paris and it is not served by any public transport. It is a very peaceful place and has been chosen by AFAF as it is almost in the centre of France.

This year AFAF were pleased to welcome Mary Kearney (EA general secretary) and people from Luxemburg, Belgium, Switzerland, and Germany. Isabel Campos send her apologies. 180 people from A.F.A.F (ataxians and members of their families) were present.

Saturday afternoon was dedicated to associative or practical problems, Sunday to scientific and medical presentations. These included presentation from Prof Michel

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Koenig & Helene Puccio laboratory, Prof Pandolfo, Dr Isabel Husson and members of Prof Testi laboratory in Rome but to mention a few. There were representatives from Hopital Necker, La Pitie Salpetriere, Inserm and of course Dr Pierre Rustin. Takeda was well represented and many of the helpers were workers from Takeda. I had the pleasure of private transport back to Paris and then a quick personal tour of Paris.

### **The Spanish ataxia federation**

Friday, 15th June, started the FEDAES annual general meeting in the evening. After the food there was an exhibition of mobility aids. Throughout afternoon they are arriving more people coming from different parts from the country. After the supper, there was a small celebration and it gave the opportunity for all to chat, renew acquaintances and make new friendships. Saturday, 16 thJune, was devoted to the Scientific presentations. Topic varied from varied from the latest information from the laboratory, gene therapy, the progress of investigation and the possible clinical trials in dominant ataxias, to he latest discoveries at Dr Javier Arpa clinic in Madrid using idebenone. Dr Mary Kearney from euro-ATAXIA was there to tell us what was happening worldwide. A intense discussion followed and it was agreed that it would be helpful if a Spanish registry for ataxia was instigated.

I look forward to attending more of the National meetings and I am impressed with the dedication and determination of all groups in their fight against this disease.

*Mary Kearney, Euro-Ataxia's Secretary General*

### **EU consortium fighting Spino-Cerebellar Ataxias (EUROSCA)**

#### **RARE DISEASES - Concentrating European research for the few**

##### **What is a rare disease?**

So called rare diseases individually affect small groups of people within the general population. In European Union, a rare disease is considered rare when it affects less than one person in 2,000. Theses rare diseases are often of genetic origin, usually life-threatening or chronic debilitating. The number of rare disease is estimated at some 6000 to 7000. Because of the low prevalence of each rare individual rare disease, patients affected with a particular rare disease in any one country are scarce as are specialized clinicians. The causes of the disease are often poorly understood, diagnosis is frequently delayed and for the majority of rare diseases therapeutic (treatment) options are minimal or non-existent.

The impact of a rare disease on the quality of life of affected patients and their family members is significant. Elucidating the pathology of a rare disease can provide insights into more common diseases. In contrast with more common disease, which are generally multi-factorial in their causes, rare disease often result from a particular pathway (for example a defective gene or protein). An

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understanding of the impact of a single defect may hence yield insights into more complex pathways involved in common diseases.

### **EU policy on rare disease research**

As a consequence of the very nature of rare diseases- their low prevalence- substantial benefits towards the development of new diagnostics and treatments could be achieved by increasing the number of patients available for each study and bringing together the scattered specialists with complementary expertise to allow for multidisciplinary approaches and the exchange of good practices.

Recognising the importance to stimulate rare disease research activities at the European level, the EU provides direct financial support for selected research projects through its Sixth Framework Programme for research, Technological Development and Demonstration Activities (FP6). Support for rare diseases from 2002-2006 was mainly from Thematic Priority 1- Life Sciences, Genomics and Biotechnology for Health. EUROSCA was funded as one of the projects in this area. EUROSCA's aim was to translate basic research into clinical applications.

### **EU consortium fighting Spino-Cerebellar Ataxias (EUROSCA)**

*European integrated project on spino-cerebellar ataxias - Pathogenesis, genetics, animals models and therapy.*

Twenty-two European groups from nine countries with an excellent reputation in clinical, clinical-genetic and basic research on Spino-Cerebellar Ataxias (SCA) jointly form an integrated Project to develop a treatment for patients suffering from these rare, late manifesting, and autosomal dominantly inherited group of neuro-degenerative diseases. To date (June 2007) there are now 29 different types of SCA's. Seven new genes have been identified since 2001. Initially it was decided that an international standard on the clinical evaluation, in the form of a Core Assessment Programme for the Interventional Therapies of SCA (CAPITSCA), be developed based on the clinical rating scales, structural imaging and electrophysiology.

Next, the generation of the world's largest collection of information on SCA, the European SCA Registry recruitment of SCA Registry (EUROSCA-R) was developed to ensure standardized data acquisition. This powerful tool facilitates continuous recruitment of SCA patients throughout Europe for linkage analysis, identification of novel ataxia genes and natural history studies. The potential to include larger European families into linkage Genotype-phenotype correlations has followed. This has facilitated a systematic large-scale search for a genetic modifier factors in SCA, allowing for a better comprehension of factors accounting for wide clinical variability, with application for prognosis and to identify new potential targets (modifier genes). EUROSCA has also implemented strong research projects to generate and characterize cells and transgenic mice models. This has enabled a more defined study of the pathogenesis and has served as a tool for first therapeutic studies. Nine European research groups have been supported by five core facilities undertaking transgenic Drosophila work, Expression-Chip-Technology,

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Proteomics yeast two-hybrid technology and monoclonal antibodies. Training programmes have complemented research efforts and clinical work.

Written by Euro-SCA.

## Going Santhera

### **SANTHERA phase III clinical trial with Idebenone in Friedreich's Ataxia**

Authors from the company:

- M.Sc. Clara Fehrmann, Clinical Research Associate;
- Dr. med. Geoffrey Holder, Senior Clinical Research Physician;
- Dr. Thomas Meier, Chief Scientific Officer;
- Thomas Staffelbach, Head Investor Relations & Public Relations
- lic. phil. Daniela Iser, editor Euroataxia Newsletter once after and before Michael Morgan

**Idebenone** and **Santhera** are two expressions to be uttered in one breath. Santhera is the name of the pharmaceutical company conducting the phase III clinical trial with idebenone in Friedreich's Ataxia (FA) in several countries in Europe. The company collaborated already with the National Institutes of Health (NIH) in Washington, USA in the phase II trial conducted by Dr. N. Di Prospero.

I had the pleasure to be invited to Liestal near Basel, the pharma epicentre in Switzerland, the seat of Santhera Pharmaceuticals (Switzerland) Ltd. It is now four years ago, in 2003, that the Euroataxia AGM took place in Switzerland. Back then, I contacted Dr. Th. Meier of a small company named MyoContract that was rather engaged in the field of neuromuscular diseases and asked him to give a presentation in Euroataxia's field of interest. At the time MyoContract worked together with the team of Prof. M. Koenig and was already interested in evaluating a treatment for Friedreich's Ataxia. These were the early days. In 2004 Dr. Meier has merged his company with the German company Graffinity to form Santhera Pharmaceuticals. Today as the Chief Scientific Officer of a public company with 70 employees, he took the opportunity to pay me back - in the best of positive senses! Friendliness, professional dynamics and enthusiasm in the course of planning, organising and conducting a clinical study as well as motivation in its best expression and dedication of the heart are to be summarised in what could well be termed as "santheraism" – I was absolutely amazed as to what extent this team of medical professionals, researchers, chemists and biologists have the substantial and unpretentious benefit of FA patients and other persons affected by neuromuscular pathologies at the back of every single one of their thoughts and actions.

Along with me came my personal assistant Thom, a student of architecture soon with a diploma, who makes just the gentlest of PAs (a topic for itself... See my other essay...). We were picked up at the train station by two smiling and just perfect lady samples of "santheraism" and welcomed warmly by Dr. Th. Meier. He introduced us to members of the Santhera team who see to it that innovation has

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its most effective results for people who are affected by FA as well as other neuromuscular diseases. The first thing that drew Thom's attention were the plans on the wall showing the site of a still larger firm, the near future home of the growing company.

Both phase I and II clinical studies with idebenone were accomplished between 2005 and 2006. Santhera conducted four Phase I trials in healthy volunteers to characterize in great detail the pharmacokinetics as well as safety and tolerability of idebenone including at high doses (i.e. at doses of up to 45 mg of idebenone per kg body weight and day; 45 mg/kg/d). These studies were complemented by a Phase I program that was conducted independently by the NIH in children, adolescents and adult FA patients in which a dose of up to 60 mg/kg/d was tested for 1 month. Taken together, all Phase I studies demonstrated good safety and tolerability of idebenone; the data of the NIH Phase I program are now published (di Prospero et al., 2007).

Based on their Phase I program, Santhera concluded that the pharmacokinetic profile (i.e. the way the drug is metabolized in the body) of idebenone indicated that higher doses could be more efficacious and that at a dose of 5mg/kg/d of idebenone, which is what most patients who are not participating in a clinical trial are taking, is probably much too low to produce a recognisable effect in FA patients, at least for the neurological part of symptoms. As idebenone was well tolerated at doses 9 times higher (i.e. 45 mg/kg/d) the door was open to investigate the efficacy of higher doses in clinical trials with FA.

The completion of Phase II was achieved by transatlantic and combined efforts of the NIH on the one hand and Santhera on the other. In general, the aim of a Phase II clinical trial is to evaluate if a drug actually works in a double-blind, placebo-controlled configuration and if it has potentials – and to what extent – for positive effects, but also for side effects. In the case of idebenone, 48 patients were divided into four groups and given either one of three different doses – or a placebo. The results were rather encouraging. Those patients who had the middle (~ 15 mg/kg/day) or the highest (~ 45 mg/kg/day) dose showed an improvement of daily living activities and improved scores with ICARS (International Cooperative Ataxia Rating Scale) after six months of treatment. The effect on the neurological parameters was particularly striking for a subgroup of patients who still were ambulatory at the beginning of the trial.

These results support data obtained in previous studies that have shown already that idebenone may improve the degree of cardiomyopathy in FA even in lower doses. Cardiac hypertrophy is an often life threatening manifestation in FA. In the literature it has been shown that Idebenone has a beneficial impact on the cardiac part of the disease.

In Europe Santhera is currently conducting a Phase III trial targeting ~200 FA patients in a double-blind, placebo-controlled study (neither participants nor professionals know if the drug or a placebo is given). Participating FA patients will receive either a low (~5mg/kg/d), a middle (~15 mg/kg/d) or a high dose (~45 mg/kg/d) of idebenone, or, if luck so has it, a "flour pill" (placebo). This is when the

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price to pay goes cynical: scientific evidence and humanity do not precisely go together. But such a setting is required by regulatory authorities in order to evaluate a compound's efficacy before the product receives marketing approval. Not all patients with FA will immediately profit for themselves if they join in the study. Still, it is to be hoped that the "unlucky quarter" won't get very much worse – and receive the high dose in an extension study being offered to all patients who have participated and completed the "core study".

Patient recruitment is currently ongoing in Germany (Deutsche Heredo-Ataxie Gesellschaft), the UK (Ataxia Group of Great Britain and Northern Ireland, Ataxia UK), France (Association Française de l'Ataxie de Friedreich) as well as Belgium and The Netherlands, with Austria to also join in soon. Treatment duration will be 12 months and results are expected later in 2008.

Because of the suggestively positive effects of idebenone on the neurological aspects of FA when taken at higher doses, it was recently decided by Santhera to declare the neurological outcomes more important in the success of the trial than the cardiological outcomes. This means that cardiomyopathy is now no longer an inclusion criterion for participation of patients in the EU trial. Also, patients who already take the drug need to stop taking it only one month prior to enrolment into the trial.

In January 2007 Santhera announced that, after discussions with The European Agency for the Evaluation of Medicinal Products (EMA - who is responsible for reviewing designation applications from companies who intend to develop medicines) it plans to file for Marketing Approval, ahead of schedule. This means potentially idebenone could be on the market earlier than expected, potentially already next year and as a fully reimbursed drug! In Switzerland, a parallel process is going on (the relevant administration is Swissmedic).

If you are interested in participating in the EU Phase III trial, please contact the Ataxia association nearest to you.

Reference:

Di Prospero N., et al. (2007) Safety, tolerability, and pharmacokinetics of high-dose idebenone in patients with Friedreich Ataxia. Arch Neurology 64:803-808.

## **Euro-Ataxia sponsored GeNeMove Symposium**

**May 10-12 2007, Bonn**

In our last newsletter Peter Reussner had advertised the GeNeMove symposium which took place in Bonn on 10-12 May 2007. He was requesting sponsorship for the event and euro-ATAXIA was delighted to be in a position to be one of the sponsors. I was fortunate to be able to attend.

The day was started with Dr Harry Orr from Minneapolis, USA telling us of his work on SCA1. In this SCA the function of the nerve cells, the purkinje cell is disturbed. Currently there is a phase 2 trial in progress in USA for SCA1. Dr Hank Paulson

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from Iowa then took the floor and he spoke about the pathogenesis SCA3 and told us a lot of work has been done using RNA therapy in the laboratory for SCA3.

This was followed by Dr Steve Pulst, who received his medical degrees at Hanover University, Germany and currently working on Ataxias in Los Angeles. He got a special welcome to his home country. He spoke about his work in SCA2, which is the most common SCA in Cuba. It is an unstable repeat disease, so parents and their affected children have different gene abnormalities. He told us that SCA 13 has been seen in Philippines in children. These children have also been noted to have mental retardation. In Europe SCA13 has shown to have an early onset and a late onset. Such variation in the age of onset is really a great discovery for scientists to make.

The morning session continued with Dr Laura Ranum from Minneapolis, who was just recovering from jet lag, giving us a good overview on SCA 8 She told us it is a multi-system disease. The same is said for Friedreich's ataxia. It has also been found to have reduced penetrance in upcoming generations when a lot of members are affected. This is similar to what we heard earlier about SCA 2.

The session finished, by departing slightly from the scheduled programme, to hear from Dr Chris Gomez. He has recently moved from Minnesota to Chicago. He spoke of how ion channels are involved in SCA6, episodic ataxia, migraine, and epilepsy. He has evidence to prove that episodic ataxia is worsened by fatigue, exercise and caffeine. Dr Thomas Klockgether chaired the lively ensuing discussion. I felt that it was a very fruitful session for the scientists.

The next session concentrated on Friedreich's Ataxia (FA). Prof Pandolfo, Brussels gave a good overview of FA, including the way the abnormal DNA folds and causes problems at a cellular level. Dr H  l  ne Puccio, Illkirch, France spoke at length about iron metabolism and her work in mice. She told us that Dr Mark Pook mice were more humanised. Dr Michel Ristow, Berlin continued this session by telling us that frataxin, the protein reduced in FA, is directly responsible for causing the diabetes. He said that reduced frataxin results in people not being able to manage a high glucose meal and reduces the body sensitivity to insulin. For me this raised a further query in my mind. Would carriers of FA be more likely to develop diabetes. Prof Michel Koenig spoke of his work in other recessive ataxias and of his latest gene discovery Juvenile ataxia with sensitive neuropathy.

This finished the morning session. Everyone welcomed our lunch box and break. There were over 50 posters on display to browse over during lunch. I attended a satellite meeting with Prof Schult, Dr Brunt and Peter on the progress on the FA register. There was a lively exchange of views and Prof Schult seemed to continue his work in this area to reach a solution which would serve all FA sufferers best.

In the afternoon we heard from Dr Rubinsztein, Cambridge, UK. This lecture was very difficult for a non scientist to follow. He has been fortunate to get some phenotypical mice from Dr Olaf Reiss. The scientists present very impressed his findings and there was a lively exchange of views. Dr Gottesfeld followed and described his work on histone deacetylase inhibitors (HBDAC) as treatment for FA.

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He told us that these compounds are still in the laboratory but hold out considerable promise for the future.

Dr Kurt Fischbeck, Bethesda, USA was involved in the phase 2 idebenone trial in USA. He presented his findings prior to publishing them in a very easy to follow manner. The slide below interested me most. It shows that if you treat a FA sufferer with a placebo and intermediate dose idebenone you get the same improvement in their neurological condition.

It is hoped that the phase 3 trial currently in progress in Europe will explain this difference.

Dr Thomas Klockgether, Bonn presented some of the findings from the EUROSCA project and his hopes for the future. Dr Mascalchi, Florence, Italy guided us through the latest advances in radiology. We finished the programme at 7pm and made our way to the Rhine for our well earned evening meal. We were all delighted with the German hospitality and enjoyed the entertainment.

The conference continued on Saturday with talks on Huntingtons, Disease, Hereditary spastic paraparesis, hereditary dystonia, and Wilson's disease.

### **Last farewell to Michael Morgan, a friend to remember**

Sad news has arrived. On Friday 27 April 2007 Michael Morgan suddenly passed away at the age of 51. As with so many Fa'ers it was his heart that finally lost the battle.

Michael has always been one of the thriving forces of Euro-Ataxia. For many years he edited the newsletter, until a few years ago he decided to step down as editor. But he was always there to advise and help out. Last year he took the newsletter over again when no-one was willing to do the job. He was busy working on a new issue when his heart gave away.

Apart from being a great inspirer for Euro-Ataxia, Michael was a very dear personal friend. I remember with fondness our get-togethers on Euro-Ataxia meetings and especially the holidays we spend in Donegal a few years ago, when Michael had invited us over.

It sure feels like the end of an era now that he too is no longer with us.

Sláinte,

*Hans Doré*  
1 May 2007

## What makes a personal assistant a good personal assistant?

*lic. phil. Daniela Iser, Zurich*

A lady from a public health care service once told me that she thinks she is doing her job well if I forget her as soon as she walks out of my apartment. Apart from being amazed at hearing such wisdom from someone who works in a public organization I had to agree – to a certain extent, though.

A personal assistant (PA) is not a carer. The main difference is probably to be found in the question of responsibility: It is you who knows what is good for yourself. It is you who knows how and when and how things are to be done, since you live in your own apartment which is your private area. It is you who makes decisions, which is already where things start getting complicated, because there are right decisions and wrong decisions, too, and the responsibility is yours.... Another major difference is the kind of relationship between the person receiving services on the one side and the help providing person on the other: It is you as employer who pays an employee a salary for services defined in a contract. You are not a client of a health service and the carer is not its employee, but he or she is your employee. Even though it is that care giving person who provides help and even though it is you who receive help, you are the boss - his, or hers, and especially *your own*. The third big point is that it is you yourself to choose. Whom do you want to be your helper? To be in your apartment, to be with you in the bathroom, to wash your underwear, to get you into bed, to feed your cat? It will not be a person picked and sent by the health care organisation. It will be someone answering your ad, someone who, rather likely, is not a professional. Someone looking for a job who will try to do what you judge to be a good job. Someone you'd like to have at your side.

In very few countries in Europe, this is a familiar scenario. Sweden and Austria have known in for some time, Germany just put something of this kind into legislation and Switzerland is testing in a pilot project if the "assistance system" actually works (as though it didn't. It has been working in the USA, for example, for decades already...). – But this is politics. I'd like to talk about what a good assistant could be like.

The area is huge. Let me meditate on people affected by ataxia, like myself, and the concept of independent living with the assistance of one or several employees. You, an ataxian, are a person – a character, with moods, with qualities, needs, weaknesses, and with a hell of a disease that nobody knows as well as yourself. The care giving person has a mind with certain ideas, has a lot of characteristics, tendencies, affections and probably no idea whatsoever what ataxia is, let alone what it means.

You are going to be a teacher.

So, first of all, a good assistant is a good *student* (which is why professionals hardly ever are good PAs – they already "know" things). He or she will have to *listen*. To your words: your needs, your ways (which is why parents rarely are good

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PAs...). He or she will have to *observe*. Observe you. To know how your mind works, how your body works (this concerns carers who think they know how everything is with you if they know how things are with some other client – but two individuals are never alike and ataxia in two persons does not mean more than a bunch of similar problems and will never refer directly to your personality). No, medical details about ataxia are not necessarily part of your student's knowledge (of course this can help). The main "object of study" is your person. Your ataxia may be a predominant feature in your being, but of course it is not the only one and maybe not even the most important one.

Your assistant is not simply a worker (even though some people think that this is precisely what he or she ought to be, no more, no less). Along with the working part (the ability to be practical and useful) comes the personality part. The former is easy to judge: It won't take too long before you see if he or she cleans up properly, is careful with your dishes and your clothes, delicate with your body, a good cook. The latter, however, presents an area full of snares and ditches and questions. *How* will your newly recruited PA listen to you? This is basic for relationships with ataxians. *How* will he or she observe you? Patiently? Friendly? Attentively? Superficially? Kindly? Not really...? That one is so complex in ataxians and asks for the ability to differentiate and identify properly all kinds of appearances. The person in question will show a polite and friendly face at the beginning, will demonstrate eagerness at the start – only to reveal herself as an alcoholic who won't mind waking you up in the middle of the night or not showing up at your house if you need her to accompany you to a therapy session (because she has a hangover). Or to make you realize that she doesn't think your needs are all that important – a luxury variation of disrespect. Such black holes in a character cannot be foreseen. There are things you cannot anticipate when you give a person a job that is so extremely close to your own personality. Other employees hardly ever work so closely together with their boss... It is not your mistake if, at the very beginning of your time with a PA, you do not recognize all tiny signs of possible weaknesses. There is no way to avoid collisions. Maybe it is a matter of experience: Having had several PAs already might give you a feeling of how to interpret early signs of what will develop as unacceptable feature. On the other hand, someone can reveal herself or himself as a truthful and thoroughly reliable person even though you had doubts about these and other positive qualities at the beginning of your "relationship". Such positive surprises can happen just as well as negative ones. As an employer of PAs, you are not only your own specialist – you are your own administration office, your own advisor, your own salaries' office and most importantly, your own personnel department. It means quite a lot of work, but it's worth it!

I have been following the independent living path for several years now, have seen quite a few assistants come and go, have cried after one or two, fired three. At the moment, there are ten women and men supporting me in my daily routines, enabling me to continue a self-conscious life.

Let me return to the qualities that students need to have: The very first requirement is the talent to listen to observe well. Combined with a natural eagerness and easiness to learn about the mechanisms and individual ways of other people

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(which is a curious variation of curiosity!), these prerequisites may produce what can be called an *unpretentious kind of help*. What qualifies a PA candidate is certainly not his or her wish to help (this is, anyway, a rather suspicious claim for most people to make for themselves). Any agreeable, smooth, even delightful sort of help will come forth from a simple and light attitude in your assistant. Moreover, a multitude of “adverbs” may describe more closely the best required actions: assistants listening patiently, waiting for you to finish your sentence, never trying to take the words out of your mouth; assistants observing quietly your way to move your body, to treat your clothes, to touch your pet – observe precisely – *you*. Waiting can be done with tenderness (unlike taxi-drivers or mechanics or service men who can drive you mad with their impertinence). Asking questions can be done with respect and a fine sense for delicacy (or with a sneer, like some people on the other end of the line shouting “whooo is there?!?” as soon as you produce the first slurred words). Processing the learned things can be done roughly, superficially, not really bothering – or light-heartedly and with the best intention to please you. And then, what is suggested to result in proper actions, in *help*, may be the very kind of help which help really should be: the help *you* define as help.

You will spend many hours of your days with your assistant. If luck will have it, a couple of hours are added... Because you share personal interests, because you start having a tight relationship that is no longer defined strictly by correctness, but by an atmosphere that is simply comforting and marked by confidence. And by countless fits of laughter! ;)

The woman from the public health service was right in the “work half” of an assistant’s personal abilities, but she seemed to forget (what most people tend to ignore): We are not objects to be helped, but individual persons.

*Many thoughts in this essay are inspired by my young assistant Thom, who taught me that sweetness and respectfulness in a student can well go together, that my idea of what help really is can be realized in a way more tender and truthful than I could have imagined and that grace and dignity can be oscillating echoes in two independent minds.*

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