Friedreich’s Ataxia Research Alliance (FARA) Ireland is a non-profit, non-governmental, Irish organisation representing people with Friedreich’s Ataxia. It aims to develop a strong connection with international research on Friedreich’s Ataxia, raise funds for research and to increase public awareness of the plight and condition of people with Friedreich’s Ataxia.

BRANCHING OUT INTO EUROPE

European Patient Advocate Group (ePAG)

Feedback needed from other patient groups

It is with great pleasure that I welcome you to read our first newsletter. As the political landscape changes all over Europe, we as a patient organisation must be ready to change with it. In 2017, European Reference Networks (ERNs’ see page 9) will be created as legal entities by the European Union. Information on patients will be exchanged in a secure internet system. Patients will not have to travel. As patients and patient organizations are experts in their own rare disease, they would have a special place in these new European networks.

I have been privileged to be accepted as European patient advocate (see more page 10) since May 2016. I have attended several webinars and seminars to upgrade me on the role as ePAG. However I need your feedback so my contribution to this network is maximised.

In this newsletter, I would like to share with you what I have learnt from EURORDIS trying to serve all those without a diagnosis for their rare disease to helping those for whom a treatment is in sight. In recent weeks we have seen a treatment come for some of those with spinal muscular atrophy, a life-limiting and progressive condition (see page 14).

In May 2016 I was privileged to be able to attend the European ataxia annual meeting and the Friedreich’s Ataxia symposium in Philadelphia. The challenges in both continents are quite similar.

CONTENTS

The latest in research
Study on fatigue, physiotherapy, psychotherapy in Friedreich’s Ataxia …2
E-RARE - EU research programme……5
What is happening in Ireland……………16

From Europe
Report from EURORDIS conference ..3
ERNs’ – what are they all about? ……9
European Patient Advocate (ePAG) …10
Data exchange in ERNs’………………12

Caring for patients
Kids suffer when patients clash with medical staff ………………………7
Tips for FAers’ ……………………15
Are there risks to in clinical trials?…14

What’s up and coming?
New treatment for Spinal Muscular atrophy ……………………14
Levels of Engagement ……………..13
Official launch of ERN ……………10

Abbreviations ……………………..8

Editor and lay-out by
Mary Kearney,
Dunlavin, Co. Wicklow, Ireland
E-mail: marykearney@gmail.com
Study on fatigue, physiotherapy and psychotherapy in Friedreich’s Ataxia

Dr Husson from Paris gave an overview of her work with children and young people with Friedreich’s Ataxia (FA). She had been doing a clinical trial using pioglitazone between December 2009 – 2012. It was a 2 year study in Paris. She has followed patients and has been doing a natural history study on 60 people with FA ranging from 6 years of age to 25. At the European and American ataxia conferences, she highlighted the importance of muscle strengthening exercises and stretching as part of a physiotherapy programme which can lead to improvements physically and psychological for people with an otherwise progressive condition. Practical benefits because increased muscle strength in legs improves gait, posture, and autonomy. For example it can improve the quality and security of transfers, the ability to get dressed alone (especially shoes, socks, pants). These rehabilitation choices may decrease fatigue, and improve body image with the corresponding positive psychological effects.

Muscle strengthening exercises, stretching, and aerobic exercises, may be possible for patients, even if a wheelchair or rollator is used. Rehabilitation can be adapted for each person, taking into account their capabilities. Dr Husson thinks that verticalisation (standing up straight with support, see equipment page 15) is of the utmost importance for strengthening and stretching leg muscles. She insists on individual rehabilitation, adapted sports for aerobic and strength improvement, which has a psychological and social effect.

Aerobic and endurance exercises are important. This importance was illustrated with a video of a young girl. Her gait was much better before muscle fatigue in the first meters of walking compared with fatigue after due to 200m. With better endurance, the gait quality was better on distance.

Another important area is the use of foot orthoses, orthopaedic shoes, or ankle braces, to prevent falls, ankle sprains and foot pain. In association with stretching and strengthening, they can improve gait, posture, stability and autonomy.

Dr Husson focused on the importance of rollators which help prevent falls, improve gait, give posture stability and increase independence and so help prevent social isolation. In her experience, it is important to choose an anterior rollator with forearm supports (better position, high comfort and stability, low fatigue due to the high stability). The use of a rollator can postpone significantly the loss of walking ability. Combined use of a wheelchair or rollator depending on circumstances is possible, and it helps in terms of autonomy and quality of life.

• Using videos was a good way of showing the improvement to patients who may be reluctant to wear such foot orthoses with or without a rollator. Again, a video case study was presented showing clear improvements in walking in a girl after wearing orthoses and using a rollator with forearm supports.

• The importance of a psychologist for acceptance of assistive devices is totally clear. The goal is to avoid a progressive decrease of activities and socialisation.

• A large percentage (69%) of her young patients with FA have urinary symptoms such as urgency and urge urinary incontinence, which can have huge psychological impact, especially in teenagers. She has been testing transcutaneous tibial nerve stimulation for overactive bladder, and positive results were seen in 3 patients, hence a larger study will now be done.

• Scoliosis was found in 86% of her young patients. Braces had been used as a treatment in nearly a third of these, which can be helpful in reducing back pain, improving posture, gait and fatigue. A study is beginning on this topic.

Dr Husson finally reflected on how a psychologist should have a major role in supporting the young patient and their family in accepting the diagnosis, adapting to adjustments needed to daily perspectives and to restoring trust in their own capabilities. A study on fatigue run by the psychologist is ongoing.
EURORDIS Conference Edinburgh
European Conference on Rare Disease & Orphan products (ECRD)

Delivering 21st century healthcare to rare disease patients:
Together we can change the future  May 2016

Following on from seven previous European Conference on Rare Diseases and Orphan Products (ECRD), 2016 was a wonderful event which started from small beginnings. This conference, which is held every 2 years, is a unique opportunity to bring together all rare disease stakeholders from all European nations:

1) patients and patient representatives,
2) healthcare professionals and researchers,
3) industry, pharmaceutical companies,
4) regulators and policy makers alike - in the fight against rare diseases. The ECRD brought together over 80 speakers and more than 800 participants, covering six themes:

- Diagnosis,
- Research,
- Drug development,
- authorisation and access,
- Care provision,
- Social policy and Global society

New meeting formats to enhance the on-site learning experience were showcased for the first time in Edinburgh and included a speed networking session to connect patients, researchers and industry, an open-house “soap box” lunch session, interactive roundtables, audience polling, networking lunches / coffee breaks and dedicated poster sessions

**Game Changers in diagnosis:**

Accurate diagnosis is essential as without a diagnosis, families and the clinicians who support the family are “groping in the dark”, possibly missing out on interventions that might improve the situation for the affected patients. It can take 5-8 years for the correct diagnosis (EURORDIS survey). If the incorrect diagnosis is made, it can lead to incorrect treatment.

An area where social media has helped in diagnosis is the a global “Matchmaker exchange”. The phenotype of a disease is given 5 different characteristics for example in ataxia unsteady gait, loss of sensation, scoliosis, diabetes and heart irregularities.

The genotype is also described. It is not uncommon for some people to have abnormalities in their genotype which are not the cause of the rare disease.

The match-maker programme, pictured above tries to match the abnormalities of the genotype with and the phenotype. To date quite a few countries and pharmaceutical companies have joined this programme.
More matches have been made, by people on facebook, than in scientific matchmaker exchange. It is good that data has been shared all around the world to further research and knowledge in this programme.

**Game Changers in Care Provision**

Matt Johnson, Healthcare & Research Director at EURORDIS, in his summary told the audience that it was now 10 years since cross border initiative was set up. It had changed the landscape and the rules, in particular, clinical competition is replaced by clinical collaboration. A lot of rare diseases are multi-need. Regional and European connections could expand further. Centres the expertise will hopefully be known as European Reference Networks (ERNs’). These would become “magnets” for people to get a diagnosis. It would be hoped that they would establish a culture of learning based on trust and competency where standards of care would be used.

---

**Consensus Clinical Management Guidelines for Friedreich’s ataxia**

Guidelines for clinicians, patients and research to ensure better outcomes today, and for the future.

---

ERNs’ will hopefully break the isolation of care and build valuable relationships. A word of caution was given by Matt, hoping that they would not get lost in administration.

Dr. Till Voigtlander, Neurologist, Vienna felt that they would be centres which would accessible for Doctors but I think this point may be open for discussion again.

- Accessible for doctors – data samples – further drive for research
- Accessible for patients – National health care provider
- Assurance for all that there was trust between the experts. The patient would be able to know that the best was being done for them and the “payers” would know that these were expert centres. It was decided that they would be no a difference between adult and paediatric care as the transition from one area to the other had proved problematic over the years.

He also added a word of caution that he hoped that patients would not have false expectations in ERNs’.

**Summary from Friedreich’s Ataxia viewpoint:**

The conference was for all rare diseases, looking at it from a FA approach, there was much to learn. In particular, Dr Carl Morris, Solid Biosciences, Cambridge, USA is working on a treatment for Duchenne Muscular dystrophy after a family raised millions of dollars for him.

In reviewing European Reference Networks (ERN), from FA perspective, they would encourage clinicians to see standard guidelines in the care of FA which were established for FA (known as Concensus Clinical Management). In the long term, ERNS’s will be seen as areas of expertise and possibly areas where research can be done. Mr Enrique Terol, EU Commission presented his perspective, which was that ERN would be run by co-ordinator, all member states would be linked by an IT platform. After his presentation, Ms. Samantha Parker, from the pharmaceutical company, Lysogene, then joined the panel. She told the group that she looked forward to using the patient registers associated with the ERNs’ for research. However, Mr Enrique Terol, EU had said nothing about registers in his presentation. For me, this highlighted how far apart EU and industry were away from one another.

In a subsequent session entitled “How to structure Healthcare for better health outcomes”, Dr Muarizio Scarpa, Metabolic disease, Germany and
Prof Hanns Lochmuller, Neuromuscular disease, Newcastle, UK, did express the hope that they could use databases, previously set up by the EU in ERN’s. In reality, the Spinocerebellar ataxia group had their last meeting in December 2007. Would that database be relevant to today 2017. The European Friedreich’s Ataxia Consortium for Translational Studies (EFACTS) database comprises of only 20 of the 112 people who have FA in Ireland, It has just over 100 people from UK out of a possible 1,100. The small numbers in the ERN will be insufficient to attract pharmaceutical companies. In private conservation after the session, Dr Maurizio said the establishment of a useful patient register would be a significant challenge for ERNs’. Trying to use previously collected data would also present a significant challenge. He went on to say that compiling a database is not an initial step, and would be “way down the line”.

Editor needs feedback from other rare disease patient groups:

The devil is in the detail. I welcome feedback from other neurology groups on where they see the way forward. If your rare disease has a care plan, patient registry or any other relevant information please let me know so I can bring it to the attention of those in the rare disease Neurology ERN.

ERA-Net for Research Programmes on Rare Diseases (E-Rare)

The E-Rare consortium was built to link responsible funding organizations and ministries that combine the scarce resources for rare disease research and thus enable the participation of many researchers to transnational projects via Joint Transnational Calls (JTCs). The successful linking of research funding organizations in E-Rare-1 and the subsequent exemplary joint funding activities have attested the need of, and the acknowledgment from, the research community for transnational funding of collaborative, multidisciplinary and ambitious projects on rare diseases. It has leveraged funding for rare disease research in countries without specific programmes for rare diseases and thus enabled the participation of researchers in these countries to transnational projects.

At present only a few European countries fund research on rare diseases through specific dedicated programmes. Therefore, the funding of transnational collaborative research is the most effective joint activity to enhance the cooperation between scientists working on rare diseases in Europe and beyond and thus reducing fragmentation of research in this field.

The calls performed in the E-Rare-1 (2006-2010) and E-Rare-2 (2010-2014) programmes have shown that funding of projects on rare disease research in a coordinated way is clearly possible and needed as there is a significant interest for collaboration between rare disease researchers in Europe.

E-Rare aims to:

1) Fund transnational collaborative research through yearly Joint Transnational Calls (JTC) contributing to International Rare Disease Research Consortium (IRDiRC) objectives

2) Develop a strategic research agenda
3) Strengthen the collaboration with European Research infrastructure

4) Involve patient organisations in research findings,

5) Transform E-Rare into a sustainable network

6) Ensure that all stakeholders are actively engaged in and informed about E-Rare activities

Since 2007 the E-Rare Consortium implemented 6 Joint Transnational Calls (JCT) for collaborative multidisciplinary research projects open for any rare disease (except rare cancers, rare infectious diseases and adverse reactions to drugs), with a wide range of possible topics and approaches. In seven years €56.4 million was invested to fund 79 research projects involving 347 research teams.

The highly competitive nature of the Joint Transnational Calls resulted in funding of very high quality projects. A large proportion of submitting researchers have outstanding track records with publications in the best-ranking journals. The assessment of the E-Rare funding programme achievements based on the analysis of the final project reports of JTC2007 and JTC2009 confirmed also that E-Rare funded projects largely contribute to reducing fragmentation of resources and achieving critical mass of data and samples for research projects. All funded consortia initiated new infrastructures (databases, registries and biobanks) with which they achieved the critical mass of samples/data necessary for the development of the project. E-RARE funding facilitated the academic training of a substantial number of young researchers: 58 MSc and 76 PhD students were trained in the context of the 29 analyzed funded projects.

Finally, E-Rare was recognized as a catalyst for new collaborations but also for cooperation sustainability. 77 % of consortia established new collaborations thanks to the E-Rare funding and more than half of them succeeded in obtaining subsequent funding for their project. The importance of E-Rare as a collaboration “stimulator” was also confirmed by an inquiry in Spring 2013 among researchers that applied to E-Rare calls JTC2007 up to JTC2012 but did not succeed in obtaining funds. The response rate to this survey was more than 20%. Despite the fact that these applicants were not funded by E-Rare, 50% of the responders confirmed that applying to the E-Rare calls triggered the establishment of new collaborations and most of them pursued this collaboration even without E-Rare funding.

Editors note E-Rare does seem to have opened doors for some research diseases.
Kids Care may suffer when parents clash with Medical Staff

From a study published in Journal Paediatrics

- Rude parents can rattle medical staff enough to compromise the quality of care their critically ill child receives, a new study suggests. Medical teams in a neonatal intensive care unit made worse decisions during simulated emergency scenarios if they had been treated rudely by an actress playing the role of an angry family member, the researchers found. Exposure to rudeness helped explain about 40 percent of the variance in good medical decision-making between different teams in the study, said co-author Amir Erez. He is a professor with the University of Florida Warrington College of Business.

"There is a lot of concern about medical errors, but the medical field is not paying attention at all to the effect that social interactions can have on performance," Erez said. "They need to pay attention to this, because this could potentially save lives."

But, the researchers also found that doctors and nurses could "inoculate" themselves against potential rudeness by taking part in computer training that decreased their emotional sensitivity, Erez said.

In the study, four medical teams at an Israeli teaching hospital had to perform five emergency scenarios during the day. Three of the teams started their day confronted by a "mother" who accused them of misdiagnosing her child. The fourth team served as a "control" group, and was not exposed to rudeness.

The actress told the teams, "I knew we should have gone to a better hospital where they don't practice Third World medicine!" and threatened to move the child to another hospital. One team received no preparation for this encounter. But, the second team took part in a 20-minute computer game beforehand that exposed them to angry and happy faces, providing feedback that made them less sensitive to hostile emotions. The members of the third team were asked to write a narrative about the rude event after it had occurred, to possibly diminish any lasting effect it might have on them.

Earlier studies have shown that rudeness from an authority figure can affect a medical team's performance, and this study revealed that rudeness from a parent can also cause doctors and nurses to make poor decisions, Erez said. However, the computerized training beforehand erased this effect, by subconsciously raising the team's tolerance for negative emotions, he added. "When we raised the threshold of people's sensitivity to anger, they didn't perform less well than the control group," Erez said.

Writing a narrative about the rude event had no benefit on performance, possibly because the exercise affected participants on a conscious rather than subconscious level, Erez suggested. The study was published online Jan. 10 in the journal Pediatrics.

These findings show that doctors and nurses are human beings vulnerable to the effects of harsh emotions, said Dr. Brian Alverson, chairman of the American Academy of Pediatrics' section on hospital medicine. "The reality is when we as humans are emotional, logical cognitive thought is a lot more difficult," said Alverson, an associate professor of pediatrics at Brown University in Providence, R.I. "When you're being emotionally attacked, it's harder to sit there and crunch the numbers quick."

Dr. Jessica Madden, a neonatologist with the Cleveland Clinic, added that the problem is made worse by the fact that intensive care unit teams often work in high-stress environments with colleagues they barely know. "We're continually working with teams who come together who really haven't worked together before," Madden said of neonatal intensive care units. "They can literally be meeting for the first time to take care of a sick baby."
Training currently focuses on communication within the newly formed team, "but it doesn't factor in that with the patient-centered care model, we do have the parents with us," Madden said. "That's another layer of stress and worry that's going on as we try to focus."

However, Alverson is concerned that extrapolating the study findings -- for example, by applying them to other units not faced with split-second life-and-death decisions -- could lead doctors and administrators to shrug off real and lasting problems in each hospital's system. "When families come in and act rude, the majority of the time it's because of something we've done. [For example,] we left them five hours in an ER waiting room without talking to them," Alverson said. "It's an opportunity lost where we could be looking at ourselves and asking how we can conduct our practice so people are happier."

Editors comment: My Gran always said, it is nice to important but it is more important to be nice

---

**Abbreviations**

EFACTS  
European Friedreich’s Ataxia Consortium for Translational Studies

ePAG  
European Patient Advocate

ERN  
European Reference Network

EURORDIS  
European Organisation for rare diseases

FA  
Friedreich’s Ataxia

FAer  
Person with Friedreich’s Ataxia
European Reference Networks (ERN) - What are they all about?

European reference networks (ERN)s’ are centres of expertise and healthcare providers that are organized across borders. It is planned that the patient does not move in this network but expertise is shared over a virtual consultation platform.

It can be a challenge to provide highly specialised treatment or care for patients who have complex conditions. This is especially true when the prevalence of such conditions is low, as is the case for rare diseases. This challenge is due to both the scarcity of expertise and to the scattering of small patient populations across the EU, sometimes in isolated locations where expertise does not exist or cannot be accessed. In 2017, European Reference Networks (ERNs) will provide for the first time a unique opportunity for clinicians to work cross border in Europe in healthcare in order to tackle this challenge.

European Reference Networks (ERNs) create a clear governance structure for knowledge sharing and care coordination across the EU to improve access to diagnosis and treatment, as well as the provision of high-quality healthcare for patients.

This initiative of the European Commission, supported by all Member States, aims to address common challenges faced by professionals when diagnosing and providing highly specialised healthcare in complex, rare or low prevalence diseases. It does not interfere with already existing networks. ERNs are part of the legal framework of the EU Directive on Patients' Rights in Cross-Border Healthcare Directive.

Funding will be provided by Member States and the European Commission to run these networks.

It is not feasible to create a separate ERN for every one of the over 6000 rare diseases that exist; the clinical community therefore organised ERNs according to disease groupings. This grouping of diseases does not prevent a patient from being able to go to a disease-specific centre of expertise.

Friedreich Ataxia is part of the Neurological ERN. It is based in 23 countries and had 38 health care providers involved. It is the largest of the ERNs' as Neurological illness makes up 23% of the total of all rare diseases.

Services provided by the ERN members (health care providers) will include delivery of specialist advice on diagnosis, care and treatment of rare and complex cases. This specialist advice will be based on collective experience, knowledge and expertise generated in the network. Other services will include patient referral, specialist care planning advice to local services for complex cases, multidisciplinary disease teams case reviews, highly specialised surgery, and treatment planning, review follow-up and discharge. ERN’s have to demonstrate that they are patient centred. Patients and patient organisations play a crucial role due to their expertise in rare diseases.
European Patient advocate groups – ePAG

What are they? What will they do?

EURORDIS has worked hard to ensure that patient involvement is a key element of ERNs and their governance. With this in mind, they have organised European patient advocate groups which have become known as ePAGs’. This initiative is supported by the European Commission Expert Group for Rare Diseases. EURORDIS have organized that patient advocates be involved in all of the 23 ERN’s. The Neurological ERN has asked EURORDIS to provide them with 6 patient advocates (ePAGs’) of which I am one. ePAG membership is open to ALL patient organisations, including non-members of EURORDIS.

**ePAG – European Patient Advocacy Groups**

When I first became involved in this process, I was unsure what I was taking on. There are so many neurological rare disease, I felt one ERN was not enough for them. However, I soon realised that it was important to work within the structure provided. We would all like if our own individual disease could have its own ERN but as that is not financially possible.

Our initial task as ePAG was to review the EU application which the ERN co-ordinator was making. Having some idea of the EU involvement in rare disease for the last 15 years, it has helped me assess their current programme. The ERN hopes to use existing patient databases. I pointed out to the ERN co-ordinator and publicly at the EURORDIS meeting in Edinburgh that given some of the databases date back to 2007 (Spinocerebellar ataxia project) I felt they would not be up to date enough to incorporate into the new ERN structure. Hopefully, the Friedreich’s Ataxia, EFACTS project, whose funding ended in 2015 will soon be too outdated to use.

ePAGs’ plan to:

1) advocate for patient care which is respectful of patients rights and choices.
2) Ensure that processes to address all ethical issues and concerns for patients are in place, balancing patient and clinical needs appropriately
3) Advice on transparency in quality of care, safety standards, clinical outcomes and treatment options
4) Develop ePAG feedback and evaluation framework across all ERNs’ to provide patient experience feedback of ERN and healthcare providers experience
5) Contribute to the development and dissemination of patient information, policy, good practice, care pathways and guidelines
6) Contribute to research e.g. defining research areas important to patients and their families and disseminating research related information and
7) Identifying expert centre to join the ERN as a full member or affiliated partner

In essence, ePAG will hopefully contribute to the ERN board to provide the perspective of patients on all relevant aspects of the ERN strategy, policy and organisational processes.

Examples of my contributions:

At the September 2016 meeting on data sharing (see more details on page 12) Prof Helen Cross, childhood Neurologist, gave a demonstration on how the pilot e-lipsy ERN had worked over the previous year see (http://www.elipsy.eu). To me, it seemed like she ran a very effective ERN. Currently, she allows patients access to the site and they could see what document were needed prior to their clinical case being presented to the ERN. To date, it has not been agreed how access to the ERN will work. In the subsequent discussion, I asked that patients be allowed to continue to have this access. As it is agreed that rare disease patients are often expert in their own illness, they will be able to contribute significantly to the referral process.

I am a member of the sub-committee on care co-ordination within the ERN. At a webinar in late November 2016, I suggested a change in emphasis
in the care co-ordination plan which was adopted by the committee. While these changes may seem subtle, I do feel that in future meetings of this group a more patient centred approach will take place.

The significant disadvantage of being involved as ePAG is that the position is not reimbursed in terms of time spent working. To date travel expenses have not been reimbursed. No ERN has received money to date from the EU either (February 2016).

The diagram below gives you some idea of what is expected of me as an ePAG. It gives a clear strategic message that ERNs should involve patient representatives to play an active role in the governance structures of ERNs.

The diagram below gives you some idea of what is expected of me as an ePAG. It gives a clear strategic message that ERNs should involve patient representatives to play an active role in the governance structures of ERNs.

How ePAGs’ will engage in ERN

- Teleconferences
- Workshops
- RareConnect
- Webinars
- Online surveys
- 1) information dissemination,
- 2) improved access to expertise centres,
- 3) introduction of an external quality assessment scheme for genetic diagnostics,
- 4) virtual multidisciplinary consultation,
- 5) participation in major diagnostic research projects including Rare Disease Connect platform that the percentage undiagnosed will improve.

What is up and coming?

The neurological ERN will be launched in Vilnius, Lithuania on 9&10th March. A further meeting is organised for the network in mid-May ‘17. ePAGs’ have been given to understand that ERN hope to increase diagnosis by at least 20% in those people with a rare neurological disease. Therefore, one of the first tasks that I hope to become involved in, is to see what criteria are needed for these undiagnosed patients, so that they can be discussed by the ERN.

The Neurological ERN hopes that through
- 1) information dissemination,
- 2) improved access to expertise centres,
- 3) introduction of an external quality assessment scheme for genetic diagnostics,
- 4) virtual multidisciplinary consultation,
- 5) participation in major diagnostic research projects including Rare Disease Connect platform that the percentage undiagnosed will improve.

I need feedback from the Neurological organisations that I represent

To date, in my capacity as ePAG I have tried to use what I have learnt from EU funded ataxia projects that I was involved in over the last 14 years. However my knowledge is limited to Ataxias and in the last few years Friedreich’s Ataxia so I urgently need to know your concerns?

Please contact me preferably by email marykearney@gmail.com
Or telephone +353 87 2653065
Data sharing in ERN

Inherent in the work of any ERN is data sharing. No work can take place without it. As ePAGs’, EURORDIS invited newly elected ePAGs’ to a 2-day conference on data sharing in Brussels. About 10 of the 84 ePAGs’ volunteered to go. I was fortunate to be able to attend. EURORDIS was keen for us to attend to represent the patient’s opinion since it is their data that is being shared. As the patient does not move in the ERN, the data is shared between the professionals, it was decided the data sharing topic, merited a conference itself.

The slide (by kind permission of EURORDIS) summaries many of the issues that were agreed that day

Prior to the conference 11am start, an early morning meeting of members of those countries already involved in ERNs’ and the EU commission took place. These EU employees, the ERN co-ordinators, representatives from member state, attended our meeting on data sharing.

The main aspects to data sharing are highlighted above, protection of privacy, securing consent, robust governance and feedback on use. Only clinical and genetic data relevant to the clinical presentation is shared, not complete case notes, tests and scans.

If the ERN recommended further tests, these results would also be shared. Final diagnosis and or specialist advice is also shared.

Usually the more severe the illness the person has, the less worried they are about people knowing their medical details.

It is planned that there will be a common regulatory framework which will facilitate data sharing and harmonise protection of sensitive data. The governance should be adaptive as advances in technology, needs and expectations of all the stakeholders will be liable to change.
Levels of engagement relevant to Friedreich’s Ataxia

Engaging with government agencies is an emerging area for rare diseases. It came up for discussion at the European rare disease patient conference, EURORDIS, in Edinburgh and Friedreich’s Ataxia American patient conference in Philadelphia. A considerable amount of time was devoted to the topic with special emphasis on engaging with the European Medicines Agency (EMA) and the Food and Drug administration (FDA).

As patient organisations, it has been recommended that we are ready for such engagement. You can see from the diagram that initial patient engagement only concerns their own care. When one becomes involved in a patient organisation, those with whom you engage change.

It is not uncommon for patient organisations to be involved in the care management plans, clinical trial protocol, and now ERNs’. In recent months we have witnessed a treatment being approved for the neuro-muscular degenerative condition of Spinal muscular atrophy. One must hope that some treatment will come soon for Friedreich’s Ataxia.
Are there risks being involved in Clinical trials?

While all clinical trials carry risk, the risk of doing nothing is greater. The WHO had defined what is an acceptable risk in a clinical trial. Different diseases have different symptoms. In any rare disease, one may have a huge range of symptoms, some worse than others. Some people may be able to live with some of them and some may not.

Representing those with different neurological illness presents a significant challenge as I cannot be familiar with every rare neurological illness. I need you to inform me, as your patient advocate, what is acceptable for a particular illness.

In a slowly progressive illness, people are less likely to accept the risk of being involved in a clinical trial. In a more rapidly progressive illness like Friedreich’s Ataxia, those with it are more likely to accept a greater risk of being involved in a clinical trial.

Within that context, usually, if there is a chance that a clinical trial could completely reverse FA, the patient would be liable to accept a more significant risk than if the treatment prevented progression.

Patient organisations can  
1) give valuable information to researchers about what they consider valuable endpoints for a clinical trial.  
2) be aware of where would be the most appropriate site for a clinical trial.  
3) know how best to appeal to people so they will participate in a clinical trial.

Editor’s note: I would love to hear from patient organisation about their interest in clinical trials so I can communicate it to the rare disease neurological ERN.

Accelerated Assessment status granted for drug for rare illness

Spinraza for spinal muscular atrophy

In October the European Medicines Agency (EMA) validated Biogen’s marketing authorisation application for Spinraza as a treatment for Spinal muscular atrophy, a devastating neuro-degenerative disorder and a leading cause of death in infants. In addition Biogen has submitted regulatory filings in Japan, Canada and Australia.

On Friday, 23rd December, FDA approved the first treatment for this condition as well - http://www.wallstreet-online.de/nachricht/9195011-uss-fda-approves-biogen-s-spinraza-nusinersen-the-first-treatment-for-spinal-muscular-atrophy Earlier this month, it became available in Ireland for the 2 patients who have this condition.

A clinical trial earlier in 2016 demonstrated that Spinraza showed improved survival, the drug allowed infants to sit, stand or walk when they would not otherwise have been expected to. One hopes that some treatment will come for Friedreich’s Ataxia patients. At EURORDIS, organised, Council of European Federation meeting in Paris, November 2016, EURORDIS spoke about the future presence European patient organisations at EMA meetings when drugs are submitted for approval for rare diseases.

When a drug is being approved or reviewed by the EMA, patient experience may be submitted as evidence of its value. Such patient experience needs to be factual and not overly emotional.

Editor’s note: From a Friedreich’s Ataxia viewpoint, if a drug was to be submitted to EMA for accelerated approval, it would be ideal if all the European organisations which represent those with FA had a common voice. One needs to keep in mind, that FA is significantly more progressive than most of the other ataxias.
Tips for Friedreich Ataxia (FAers’)

Friedreich’s Ataxia, when it is present initially can be mistaken for drunkenness and young people, most often recently diagnosed with ataxia often have to try and explain what Friedreich’s Ataxia means. This can prove quite difficult and embarrassing for these young people, who are just trying to cope with the fact that they have a progressive incurable condition. It may even stop these young people going out and meeting people in public places.

How to keep walking

These young people, usually do not want to feel different, they do not want people to know they have a disability yet, if they do not give the public some indication of their disability, the public often wrongly assume the worst, they are drunk or have taken drugs. If the FAer carries a stick, it shows disability. It is not uncommon for a FAer to stand by a rubbish bin, pretending to be talking on their phone to try and get their balance back. However, the stick may even trip up the FAer so why not try a walker – people will want to help you all the more!

At the recent European ataxia conference, Dr Isabelle Husson recommend upright walkers, (Dolmite Alpha) similar to that pictured above. They are available in Belgium for €300-400. They help those with ataxia stay on their feet for longer. As we all know, when a person starts to spend more of their day in a wheelchair, their muscles deteriorate even more rapidly.

Some FAer’s find posterior walkers more useful than the more usual anterior walker - which goes in front of the person using it. Recently, Nimbo posterior walkers, without the arm supports, are available directly in Portlaoise, Ireland costing €150.

Unfortunately, teenagers find these walker have little sex appeal and usually will not use them in their teens. By the time they get to their 20’s they realise how “they have missed the boat” and cannot weight bear or walk even using them. By that stage, they find transfers more difficult and this brings increasing problems for them.

Writing skills

Even though, FAer’s will seldom write, if they lose the skill completely, it could be a loss to them as they may not be able to sign their name on important documents. Practice your writing, if you stop writing, Maybe do some colouring.
Speech
Unfortunately, it is also affected in FA. It is important that those doing oral exams inform their teachers of this problem. Speech therapists can often give a few hints that help, i.e. say your words slowly, use short sentences. If teachers are informed, it may be possible for the oral examination to take place first thing in the morning when the FAer is at their best.

With the passage of time, speech may deteriorate but it is recommended that FAers’ annunciate words out loud. If you are alone, i.e. driving, it will help in the long term with your speech – Use it or lose it. What might be useful in company is to annunciate the words of a song out loud.

Fatigue
This is a constant problem for many people but worse so in FA. Try to get a balance between doing some work yet not overdoing it. If you become very emotional it will take all your energy

Osteoporosis
Osteoporosis which is brittle or fragile bones is a real issue for people who cannot take weight bearing exercise. It is more common in those who use wheelchairs than in the general population. Maybe mention it when you attend for your regular ataxia appointment. A DEXA scan is used to assess your risk of developing osteoporosis. Remember that you need to expose your face and arms for 20-25 minutes on a daily basis so that your body can make enough vitamin D to keep your bones healthy. A diet with adequate calcium is also important for healthy bones.

Filling your car with petrol
For those who drive, they may find petrol stations where one can use a card beside the pumps a particular advantage. It saves getting out the wheelchair or walker every time one needs petrol.

Research - What is happening in Ireland

Ireland will hopefully join EFACTS:
I was delighted to hear that two of the Drs’ from the Ataxia clinic in Adelaide Meath Hospital in Tallaght have attended training in Germany in November so that they can become part of what was known as the EFACTS study.

EFACTS was a European funded initiative where by a medical database of those with Friedreich’s ataxia was complied. It had centres in several European countries including UK. People had to be over 18 years to be involved. Currently, this medical database is being expanded but its future viability is in doubt as it no longer has European funding. This funding is the problem which underlies all such European projects including the current ERNs’

Approximately 20 of the 112 people in Ireland who have FA were registered on the EFACTS database. Ataxia Ireland informed the European ataxia conference in Switzerland in May 2016 that they were aware of 112 people with FA in Ireland, Ataxia UK said there were over 800 in UK. On hearing this Prof Pandolfo, University Hospital, Brussels, Belgium told the group that given these numbers in Ireland and UK, it would be possible to run a clinical trial in either site. Given the fact that we all speak English, this makes Ireland or UK a very attractive option for a clinical trial.

Retinal Nerve Fibre Layer (RNFL) monitoring:
The ataxia clinic in Adelaide Meath hospital is now able to measure the eye (optic) nerve in a procedure called RNFL. If you are asked to participate in this project, it would be great if you could. Dr Petra is now able to do this in the Dr Raymond Murphy wing of Tallaght hospital. This eye nerve measurement is important as it is the only place in the body where one can assess the thickness of any nerve. Given that FA is a neurological condition, this is important. The plan is to measure the optic nerve over time (1-2 years). It is particularly important that you attend for your follow-up appointments. Dr Petra has been most helpful in facilitating measurements when a patient attends a regular appointment in the hospital.