



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.

INTERNATIONAL RANDOMISED DOUBLE BLIND CLINICAL TRIAL FOR FRIEDREICH'S ATAXIA SUFFERS

Official Title: *A Six-Month Double-Blind, Randomized, Placebo-Controlled Study Investigating the Safety and Tolerability of **Deferiprone** in Patients with Friedreich's Ataxia*

Detailed Description:

This will be a multi-centre, double-blind, randomized, placebo-controlled clinical trial. A total of 80 patients with Friedreich's ataxia will be enrolled. Eligible patients will receive deferiprone oral solution or placebo at a total daily dose of 20 mg/kg/day, 40 mg/kg/day or 60 mg/kg/day, divided into two-daily doses for 6 months.

As usual there will eligibility criteria and in this case,

1. Patient must have a diagnosis of Friedreich's ataxia, with confirmed mutation (excludes point mutation) in the frataxin (FXN) gene and GAA repeats ≥ 400 on the shorter allele
2. Patients must be between the ages of 7 to 35 years.
3. For at least 1 month prior to start of treatment and during the study, patients cannot have idebenone, coenzyme Q10, vitamin C, vitamin E or other antioxidants as a supplement or as a drug therapy.
4. Patients must not have an iron deficiency.
5. Patient must not have received any investigational drug products within 30 days prior to enrollment into this study.
6. Patients must not have previously taken deferiprone. The doctors involved include Dr. Martin Delatycki, Murdoch Children's Research Institute, Victoria, Australia, Dr Franco Taroni Milan, Prof Pandolfo Brussels and Dr Munnich Paris



L to R: Dr Julie Greenfield, Dr Richard Festenstein, Sue Millman - Ataxia UK, Dr Elizabeth Harison - Ataxia UK, Barbara Flynn - FASI, Tom Kelleher - FASI, Dr. Laura Rooke - Ataxia UK, Dr Joel Gottesfeld, Dr Barry Hunt, Professor Massimo Pandolfo

Prof Pandolfo (pictured above at the 2008 Ataxia research conference) is one of the driving forces behind the deferiprone clinical trial.

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Spino-cerebellar Ataxias? The latest in science

On 5 & 6 December 2008 the last scientific meeting of the EUROSCA project was held in Mallorca at which there was great exchange of views and information. Thirty two researcher and/or clinicians from 22 groups from all over Europe attended and a summary of the conference by Dr Antonio Matilla can be read in this newsletter (see page 11). The outstanding achievements of the EUROSCA group have been:

1. A new clinical scale which is used by experts worldwide for dominant ataxias;
2. Identification and characteristics of cellular and animal models of molecular biomarkers that trigger ataxia; and
3. Identification of potential treatments for spino-cerebellar ataxias(SCAs,).

Projects like EUROSCA have demonstrated to the EU that best results can be obtained with collaboration between the best European scientists. Scientists shared budget, patients, information and knowledge. It is clear that to continue this valuable work further EU funding is needed. Another example of the value of the EUROSCA project is the recent article (the abstract printed below) on **Early symptoms in Spinocerebellar Ataxia Types 1, 2, 3 and 6**. Dr Luger Schöls has given us his permission to use the abstracts for our readers. This article involved 18 author/contributors from 8 different countries which would not have been possible without the EUROSCA project. The article was published in Movement Disorder December 2008

Early symptoms in Spinocerebellar Ataxia Type 1, 2, 3 and 6.

The onset of genetically determined neurodegenerative diseases is difficult to specify because of their insidious and slowly progressive nature. This is especially true for spino-cerebellar ataxia (SCA) because it has varying affects on many parts of the nervous system and the huge variability of symptoms.

We investigated early symptoms in 287 patients with SCA1, SCA2, SCA3 or SCA6 and calculated the influence of CAG repeat length on:

- (1) the age of disease onset,
- (2) people defining onset, and
- (3) duration of symptoms.

Gait difficulty was the initial symptom in two-thirds of patients. Double vision, dysarthria, impaired hand writing, and episodic vertigo preceded ataxia in 4% of patients, respectively. Frequency of other early symptoms did not differ from controls and was regarded non-specific.

Data about disease onset varied between patients and relatives by 1 year or more in 44% of cases. The influence of repeat length on age of onset was maximum when onset was defined as beginning with permanent gait disturbance. Cases with symptoms for more than 10 years were excluded. Under these conditions, CAG repeat length determined 64% of onset variability in SCA1, 67% in SCA2, 46% in SCA3 and 41% in SCA6 demonstrating substantial influence of nonrepeat

factors on disease onset in all SCA subtypes. Identification of the factors which can predict onset of disease is of interest as potential targets for modifying the disease become available. If disease modulating compounds become available, they may be most efficient when introduced to patients in early stages of the disease. On the other hand, treatment before disease onset may not be advisable if drugs have potential side effects. Thus, reliable diagnosis of disease onset is a major challenge and recognition of non-ataxia symptoms might be a promising approach.

This systematic study on disease onset in SCA demonstrated that gait ataxia is the initial complaint in only two-thirds of patients. When rather unspecific symptoms like cramps, restless legs and sleep disturbance were excluded, 16% of SCA patients report other problems than gait as the earliest symptoms. None of the early clinical signs had specificity for a certain SCA subtype but episodic vertigo was more common in SCA6.

The data confirms that CAG repeat length can only partially explain variability in age of onset of SCA. The data revealed the closest correlation with CAG repeat length when onset is defined by beginning of permanent gait disturbance and when patients with longstanding disease were excluded.

However, the data shows that onset variability in all SCA subtypes is driven by other genetic or environmental factors that remain to be identified.

Editorial

It was with great regret that *euro-ATAXIA* learnt last February that Dagmar Kroebel, formal secretary general of *euro-ATAXIA* died in December 2008. Dagmar was instrumental in forming *euro-ATAXIA* in 1987 in conjunction with Belgian, French, German, Irish and English patient organisations. She represented *euro-ATAXIA* in Cuba in December 1992 where Dr Alberto Nodarse, Neurologist in Havana, felt she represented the voice of thousand of ataxic people their claims and their hopes. We hope we are still honouring her memory as we continue to do that for ataxic people. A report from Cuba's most recent conference is available (see page 13) in this newsletter.

In the EURORDIS report (page 4) we can see that information and access to treatment for rare disease can be a significant problem for sufferers. You will see from the article on the availability of idebenone internationally (page 6), it is difficult for patient organisations to keep informed of the treatments that are available and where.

We are fortunate in *euro-ATAXIA* to have such wonderful scientists, clinicians to inform us about drug development so we can share the information with other patient organisations. Dr Mariotti, Carlo Besto Neurological Institute, Milan recently informed an ataxia conference that it is now possible to have prenatal diagnosis for Friedreich Ataxia carried out at the in Milan.

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

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EXPLANATION OF ABBREVIATIONS

| | |
|----------|---|
| AGM | Annual General Meeting |
| DIA | Drug Information Association |
| EFGCP | European Forum for Good Clinical Practice |
| EURORDIS | European Organisation for Rare Diseases |
| FARA | Friedreich Ataxia research Alliance |
| FEDER | Spanish Federation for rare diseases |
| FEDAES | Federación de Ataxias de España |
| GoFAR | Go Friedreich's Ataxia Research |
| SCA | Spino-cerebellar Ataxia |

European plans for rare disease – France takes the lead

Many thanks to EURORDIS for this summary on France's plans to tackle rare diseases

AFAF representative Claudia Baleyrier has been keeping a close eye on how the French government kept to its 10 point plan over the last 10 years and felt this EURORDIS report was an accurate reflection of what is happening in France

Heralded as a model by countries throughout Europe and the world, the French National Plan for Rare Diseases underwent an audit earlier this month. The comprehensive scheme reached the end of its four-year term in December 2008. In October 2008, a collective sigh of relief was heard upon the announcement of French President Nicolas Sarkozy that the plan would be renewed following a period of appraisal and reprioritisation designed to enhance the optimisation of resources and services for the benefit of the Fifth Republic's rare disease patients.

Analysing achievements over last 10 years

The New Year celebrations had barely drawn to an end before French experts were gathering to review and analyse the achievements and bottlenecks of the ten-pronged plan and to start the debate over which elements to include in the new version, scheduled to appear in 2010. (Ongoing projects will continue to be funded through the one-year interim period of 2009). Of the plan's ten strategic priorities, some elements have succeeded better than others. While some efforts moved forward to yield stunning results, a few initiatives barely got off the ground for a variety of reasons.

The ten major priorities of the French plan are as follow:

- 1: Increase knowledge of the epidemiology of rare diseases
- 2: Recognise the specificity of rare diseases
- 3: Develop information for patients, health professionals and the general public concerning rare diseases
- 4: Train health professionals to better identify rare diseases
- 5: Organise screening and access to diagnostic tests
- 6: Improve access to treatment and the quality of patient care

- 7: Continue efforts in favour of orphan drugs
- 8: Respond to the specific accompaniment needs of people suffering from rare diseases
- 9: Promote research on rare diseases
- 10: Develop National and European partnerships

Of these ten elements, the third, centred on developing and disseminating information, has been one of the most successful. Two entities that have contributed to the efficacious distribution of information are

- i) the centres of reference that France has designated for specific rare diseases and
- ii) the informational services developed by Orphanet – of which the many services offered, including a database of over 5000 rare diseases available in five European languages, the creation of a nomenclature and medical/scientific classification for rare disorders is moving forward the process of establishing harmonisation between regions and countries.

Other accomplishments of the French plan include the recognition of the profession of genetic counsellor fulfilled under the fourth priority and the designation of over 130 centres of reference for rare disorders under priority six, "Improve access to treatment and the quality of patient care".

Priority seven, "Continue efforts in favour of orphan drugs" has been achieved due in part to the forward-thinking reimbursement policies France has established that guarantee equity to its rare disease patients.

The ninth priority, "Promote research on rare diseases" has also enjoyed strong results. The sums invested in fundamental and clinical research have doubled in France.

Amongst the areas that didn't get very far, under priority one, "Increase knowledge of the epidemiology of rare diseases", the committee to designate rare disease registries was established late in the game, resulting in the designation of

only six rare disease registers by the end of the term of the plan.

Priority two “Recognise the specificity of rare diseases” saw the development of some 30 National Diagnosis and Treatment Protocols that have had positive consequences for patients (all products mentioned in these official guidelines are subject to reimbursement, including non-medical products such as nutritional supplements and special creams), but with over 6000 recognised rare diseases, there is clearly some distance yet to go.

Other points that did not get out of the starting gate include priority five: “Organise screening and access to diagnostic tests” and priority eight: “Respond to the specific accompaniment needs of people suffering from rare diseases” – although other initiatives within the French government are meeting some of these objectives. The next step will be the release of a report from the Public Health Council, based on the findings of an official evaluation committee and other key players. In the meantime, a French-language website created by the evaluation committee is available from which to monitor the ongoing assessment process.

National Plans in other EU countries

Spain has had a national plan for rare diseases since 2005. Isabel Campos has been very involved in the Spanish Federation for rare diseases (FEDER). The Federation came up with a proposal for a plan along ten strategic lines (similar to the plan adopted by France early 2005). It was complicated as Spain has seventeen different health systems. The 1978 Spanish Constitution established seventeen autonomous regions, in which the management of health services is decentralised. As a consequence, FEDER is also decentralised. Dr Francesc Palau, vice president *euro-ATAXIA* is also involved in FEDER.

Ireland had a meeting in January 2009 on how it should approach rare diseases organised by the Irish Platform for Patient Organisations, Science and Industry as to date it has no specific plan for rare diseases. Dr Christal Nourisser, EURORDIS, whom you will all remember from meeting her at the euro-ATAXIA AGM in Paris, November 2007 discussed the priorities above. She told us that diagnosis is still difficult to make for some patients, a fact that some ataxia sufferers will be

quite familiar with. She has also found that it is not uncommon for patient organisations to be relied upon to inform sufferers about what treatment is available. This is an unsatisfactory situation particularly if there is no patient organisation for a particular condition. We are fortunate in euro-ATAXIA to have such a good network.

Drug Information Association (DIA)

AFAF representative, Claudia Baleyrier has attended the EURORDIS summer school course which trains rare disease patient advocates about clinical trials and drug development. It took place in June 2008 in Barcelona. As a result of her involvement there she was chosen to attend the DIA European meeting held in April 2009 in Berlin.

DIA is a non-profit making organisation with 18,000 members worldwide. It funds itself from meeting and membership fees. It encourages and supports the exchange of information in an educational rather than commercial way. The purpose of the conference was to identify the problematic issue from the perspective of patients, health professional, industry and regulators and how to address these issues

DIA supports patient representatives, usually from rare disease organisations, to attend this meeting either offering full support or waiving the registration fee of over €1,000. This meeting had representatives from all over Europe including Bulgaria, Romania, Poland and Armenia. Specific sessions were organised for representatives of patient organisations. During these sessions it soon became obvious that even though the symptoms of the disease varied, they had similar problems in everyday life.

Tip -EURORDIS representatives in Berlin were eager to help other patient organisations. In particular they informed Claudia that if European organisation represented 11 patient organisations or more, it could apply for EU funding to help with its objectives

Availability & Reimbursement of Idebenone Internationally

In our last edition of euro-ataxia newsletter, Sept 2008, we reported that Santhera, the Swiss pharmaceutical company, had sought EMEA (European Medicines Agency) approval for idebenone. In November 2008 marketing authorisation was refused by the EMEA. In view of this I thought it was time to update readers about the availability of idebenone internationally.

In Europe each country is governed by its own individual laws. However there is EU guidance that there should be equal access to medicine all over Europe. The situation in America and Australia is not governed by each state but is controlled by central government. Idebenone costs are not reimbursed.

Australia: Idebenone is not a prescribed medication so it is not possible for it to be supplied to Friedreich ataxia sufferers. There is no contribution from health insurers. Some FA sufferers in Australia import it from USA in bulk form.

Finland: Idebenone is not available under their health care service. Patients have to import it and buy it themselves.

France: Since 2001 idebenone has been available through a scheme referred to as 'temporary use authorisation' method without charge to patients. The sufferer needs to obtain a prescription through a recognised hospital and it is stocked only in specific pharmacies. It is not available in a 'regular' pharmacy.

Holland: Idebenone has been freely available in drugstores in Holland but FA sufferers have to pay for it. Some insurance companies did pay for idebenone. This situation has also changed since December 2008 most likely as a result of Santhera unsuccessful application for marketing authorisation from the European Medicines Agency (EMA).

Ireland: Idebenone is not an approved treatment. It is occasionally prescribed in a hospital setting and refund of the cost is via a scheme called the "hardship scheme" re-imburement is dependent on

the area of the country you reside. It is not usually reimbursed by a private insurance company.

Italy: In 2004 the Italian Agency for Drugs (AIFA) approved the use of idebenone for Friedreich's Ataxia. This decision was reversed in Dec 2008 probably as a result of the decision of EMEA to refuse marketing authorisation. Now, Friedreich's ataxia sufferers must pay for idebenone. The decision to charge for the drug is being opposed by the Italian patients group.

Spain: Idebenone is prescribed for "compassionate use" and is paid for by the hospitals. When contacted by the editor, Isabel Campos said that she "believed that the patients, governed by EMEA across the Europe should try, to get idebenone approved for the cardiomyopathy, which accompanies Friedreich's ataxia." She hoped that euro-ATAXIA Board of Directors would organise this by writing to http://www.emea.europa.eu/htms/general/contacts/COMP/COMP_members.html

Sweden: Although idebenone is not an approved drug in Sweden, licensed doctors can issue prescriptions which will then cost a maximum of €180 per year for an adult or €180 per year per children (regardless of the number of children). Other medicines can be included in this cost.

UK: Idebenone is not a prescribed medication and not available under their Health insurance scheme.

USA: Idebenone is not a prescribed medicine in USA. It cannot be prescribed for compassionate use. FA sufferers who wish to take it have to pay for it themselves. Some buy idebenone 150mgs capsules while other buy idebenone powder and have it made into capsules by using a \$40 do-it-yourself (DIY) kit or getting help from their local pharmacist.

Editors note Many thanks for above contributions. We would be delighted to learn and share experience of the situation in other countries.

Predictors of Progression in Patients with Friedreich Ataxia (US Study)

LaPean A, Jefferies N., Grow C., Ravina B., DiProspero N
National Institute of Neurological Disorders and Stroke
Bethesda, Maryland; Mississippi, Rochester, New York

Summary of findings:

Friedreich ataxia is an inherited, progressive, neurodegenerative disorder that is clinically heterogeneous. Clinical features including the

- development of cardiomyopathy
- scoliosis
- disease progression including loss of ambulation
- interference with activities of daily living (ADL) relative to the length of the GAA repeat, age of onset, and age of diagnosis was examined in a retrospective cohort study of 61 genetically confirmed patients, three of whom had to be excluded as they did not know the results of their GAA repeat size. The size of GAA allele, use of antioxidants such as vitamins, dietary supplements, and idebenone was also examined.

One guanine and two adenine amino acids make up each triple repeat expansion that is referred to as a GAA repeat expansion or GAA allele. In a Friedreich ataxia sufferer, two of the 46 genes which have this triple repeat expansions. The size of this triplet repeat expansion usually differs on both genes and thence one refers to the shorter GAA allele (GAA1) and the longer GAA allele expansion.

The shorter GAA allele accounted for part of the variability in the age of diagnosis (46%) but had less influence on the age of onset (27%). Multivariate analysis demonstrated that age at diagnosis, which may incorporate other genetic and environmental factors, is more important than GAA length in predicting cardiomyopathy, scoliosis, and disease progression.

Discussion

They examined, retrospectively, the ages at which Friedreich Ataxia patients first had difficulty with ambulation, used assistance devices, and had interference with ADLs' as related to the smaller GAA (GAA1) repeat length. Their results indicate that which GAA1 length is significantly correlated to several clinical milestones in a univariate model. Age at the time of disease diagnosis is more predictive of disease progression to events such as loss of ambulation and interference with ADL than GAA repeat length.

The average age of first wheelchair use (20.3 years) is slightly lower in this study than in previous studies although "use" in this study was not defined as wheelchair dependence. Thus, many subjects in the study population may have opted for wheelchair use for convenience and safety over the risks associated with independent ambulation. The results are thus consistent with the studies that report the age at being wheelchair-bound to range in the early to mid 20s.

Although initial symptoms may be subtle, timely genetic diagnosis is important not only to alleviate uncertainty in the face of physical decline but also to facilitate participation in clinical therapeutic studies to potentially delay the progression or mitigate disease manifestations, particularly where earlier treatment(s) may be more likely to be effective.

There were a considerable number of sufferers taking antioxidants. 36% (mean age 20) of all participants had taken idebenone independent of trial participation, only 23% (mean age 13) were taking idebenone at the time of the interview, consistent with a previous study.

Foetal stem cell injections trigger tumours in Israeli boy with Ataxia

Provided by: Associated Press Written by: Lauran Neergaard

On the 17th February Feb. 17, 2009 as reported in the Washington press - A family desperate to save a child from a lethal brain disease sought highly experimental injections of fetal stem cells - injections that triggered tumours in the boy's brain and spinal cord, Israeli scientists reported.

In Israel, a 13 year old boy suffered from Ataxia Telangiectasis (A-T) which is severe enough to require that he use a wheelchair began complaining of headaches. Tests at Sheba Medical Centre in Tel Aviv uncovered a growth pushing on his brain stem and a second on his spinal cord. Surgeons removed the spinal cord mass when the boy was 14, in 2006 and they say his general condition has remained stable since then.

Was the boy prone to tumours anyway or were the foetal stem cells to blame? A Tel Aviv University team extensively tested the tumour tissue and concluded it was the foetal cells that caused the brain tumour. Among other evidence, some of the cells were female and had two normal copies of the gene that causes A-T. A feature of A-T is that sufferers have an underlying poor immune function could have allowed the growths to take hold.

Using stem cells from multiple foetuses that also were mixed with growth-spurring compounds "may have created a high-risk situation where abnormal growth of more than one cell occurred," wrote lead researcher Dr. Ninette Amariglio of Sheba Medical.

She urged better research to "maximize the potential benefits of regenerative medicine (i.e. stem cell) while minimizing the risks."

A-T is not helped by stem cell therapy in the first place, said stem cell specialist Dr. Marius Wernig of Stanford University, who said it is unclear exactly what was implanted.

"Stem cell transplantations have a huge potential," Wernig said. But "if people rush out there without really knowing what they're doing ... that really backfires and can bring this whole field to a halt."

This newsletter reports that Scientists are furiously trying to harness different types of stem cells – the building blocks for other cells in the body – to regrow damaged tissues and thus treat devastating diseases. But for all the promise, researchers have long warned that they must learn to control newly injected stem cells so they don't grow where they shouldn't, and small studies in people are only just beginning.

Over the years with my involvement with ataxia we hear of several families who have travelled long distances, often to Russia and China, to avail of different treatments. To date this is the first reported case of serious side effects but it emphasises the need for careful research and use of stem cells.

Source: http://ca.news.yahoo.com/s/capress/090217/health/health_stem_cells_tumour

Health Service typos

Here are a small selection of typos which were actually typed. Since we all at times have to put up with the occasional lack of bedside manner from the medical profession, I thought we could all appreciate this medical humour:

The patient had no previous history of suicides,

Patient has left her white blood cells at another hospital,

This patient of mine has been depressed since she started seeing me in 1993'

Sex and Personal Relationships

Reproduced from Target MD, August 2008 issue, with the kind permission of the Muscular Dystrophy Campaign.

Being diagnosed with a neuromuscular condition in adulthood, or attempting to come to terms with the progression of an existing condition, is emotionally challenging and raises many questions for those affected. Sex (actual or desired) and personal relationships (again, actual or desired) are a very important part of many people's lives but this is an area often ignored by professionals leaving individuals with many unanswered questions. In this article we attempt to address these questions and help people feel better able to discuss their concerns with those close to them and with any professionals supporting them.

Most (but not all) people with a neuromuscular condition have normal sexual function and desire – a fact often not appreciated by others. Disability caused by weakness may make having sex more difficult and some people may find they have additional psychological issues to contend with.

Louise Hastings, Principal Genetics Counsellor at the Centre for Life in Newcastle has worked with hundreds of people with a neuromuscular condition. She talked about sex and relationships in a presentation at a Limb Girdle muscular dystrophy conference back in 2003. Louise spoke about neuromuscular conditions causing changes in body shape and weight, changes to the appearance of the muscles and changes to walking style and gait and she looked at how this can alter a person's "body image" affecting self confidence and raising questions such as: "Does my partner still fancy me?"

We all try to cope with difficult situations differently and often in a relationship one person wants to talk things through and the other doesn't feel able to. This can cause enormous stress. There is no right or wrong way to feel – just the way you do feel. Take your time, respect one another and try to show your care through actions if words are hard. If necessary seek professional help (with or without your partner) to find a way forward you feel comfortable with. Your GP or an organisation like "Relate" can be very helpful.

It is important to recognise that whilst we are

shaped by our experiences you are the same person you were before your diagnosis – and those close to you are likely to continue to love you for the reasons they always have even if they too struggle with the challenges of the situation.

Changing role because of disability

Sometimes disability can impact on the role you feel you have traditionally held within your relationship or family. For example, some may see part of the role of a husband as that of a provider (more so in some societies and cultures than others) and if disability impacts on the ability to work this can undermine the "role" held and affect feelings of self esteem and sexual performance. Try to recognise that roles can, and do change over time. There are no rights or wrongs in this and each of us has to find the role that works best for us in our individual circumstances and relationships.

Care needs are often a concern – both for the person with the disability and for their partner. Not everyone has the capacity or desire to be a "carer" and open, non-judgemental discussion can be enormously helpful here. Different people bring different strengths to their relationships – someone who is not good at the practical side of care provision may be a great organiser and can use these talents to arrange a care package that works.

Some people will be keen to separate their care needs from their needs as a lover/partner and doing so may help maintain sexual mystery and desire as well as lessen feelings of vulnerability. For other couples the provision of personal care support from the able bodied partner may bring added closeness. Try to work out what is right for the two of you and if necessary involve outside agencies to reach workable solutions and to obtain the support required.

As one man in his 30s with FSH muscular dystrophy said "My wife is my wife, not my carer and this can be difficult. We do have emotional conflicts but we also have a strong marriage. The main thing we have to deal with is change but we put our heads together to overcome problems."

Society's view on disability and sex

The fact that society often sees adults with disabilities as asexual is reinforced when professionals suggest housing adaptations or equipment that don't take this side of life into account. One gentleman with Becker muscular dystrophy married a lady with two teenagers and the couple went on to have a child together. Prior to his marriage he had lived in a small adapted bungalow. On requesting a housing transfer he was told that he was not a priority as he "hadn't warned them he might get married and have a family." The bungalow had been allocated to him fifteen years prior to his marriage!

Another gentleman, who was in his late 70s and had been married over 50 years, said the thing he missed most now that he had to sleep alone downstairs in a single, electrically adjustable bed was "the marital bedroom and sex – or at least that night time kiss and cuddle."

Of course, not everybody with a neuromuscular condition is in a relationship. Finding love and a sexual partner can be challenging for us all and may seem more so to someone with a disability. All the usual advice about taking a positive approach applies here – it is a quality which attracts others. Keep active and try to maintain a social life – preferably one where you meet new people. One website with lots of advice about finding a partner (and about sex and relationships and the impact a disability can have on them) is www.outsiders.org.uk.

Some people will be concerned about how their disability might impact on their ability to have a sexual relationship. Muscle weakness, inability to move the body easily and contractures can all make the physical act of intercourse problematic. As always, good communication is the key. Bob Mauro, man in his late 50s with polio is quoted on www.accessibility.com.au in their set of articles on "sex and disability." He points out that engaging in sexual activity when you have a disability requires the four Ts "Time, Trust, Trying and Talk". Another tip is to "focus on the process not the outcome".

Medication

Medication can affect sexual function and desire. Dr. David Hilton-Jones, Director of the Oxford Muscle Centre noted in the past that many drugs, even at appropriate therapeutic doses, can have an

adverse effect on aspects of normal sexual function. It is impossible to be comprehensive and if you think that there is any possibility of a connection you should discuss it with the relevant doctor. Some times the association is obvious, for example loss of the ability to gain or maintain an erection shortly after starting a new drug, but the effect may be more insidious.

The mechanisms of sexual dysfunction are varied. Loss of penile erection can be caused by a wide range of drugs, including those used for treating high blood pressure and other heart conditions (such as cardiomyopathy in muscular dystrophy). Many drugs can affect libido including those used for treating psychiatric issues (which may, of course, affect sexual function) and epilepsy. If in doubt, ask.

Level of energy needed for sexual intercourse

If you are concerned about heart or respiratory issues talk to your specialist and ask about the affect these may have (or not have) on your sex life. Everybody's situation will be different so personal advice is essential. The American Muscular Dystrophy Association says:

"Sexual intercourse generally uses about as much energy as walking three miles an hour, and that can be a strain for people with weakened respiratory or cardiac muscles. Some positions require less energy than others so experiment. Many people find a side lying position easier. Of course, respiratory and cardiac problems should be evaluated and treated by a doctor – for the sake of your sex life – and your life."

All sexual choices are personal. As in the able bodied population some people will seek same sex relationships much or all of the time. For many people sex outside a loving relationship is not an option whilst this may be something others seek or decide to pay for.

The important thing is to ensure that your choices are right for you. As Bob Mauro who was cited earlier says: "If you are in a relationship that puts you at odds with your sexual identity you should look elsewhere for someone to love and to love you."

Young people

Their thoughts and feelings need to be respected and listened to and a positive attitude towards their concerns taken. Work together to seek answers whilst accepting that sometimes there are no easy answers. Respect the act that your child may choose (as all teenagers often will) to seek advice, information and support (often without your knowledge) from their friends or from professionals rather than from within the family. As a parent you can help by ensuring that open discussion is encouraged and sources of professional help identified.

Young people who have grown up with a progressive disability or who have a disability acquired in childhood, face very different

challenges to adults who were able bodied in childhood.

There are challenges around parents and other adults accepting that sexual issues are relevant for them and that sexual desire will exist. Advising a young person “not to think about it” or to “concentrate on other things” will not be helpful.

Editor's Comment:

This is a very sensitive subject for everyone irrespective of disability. I would like to compliment Muscular Dystrophy for prioritising this most important topic and I hope it will help our readers with difficulties in this area. Further email addresses are available from the editor if help is required

Report of Ataxia conferences - worldwide

euro-ATAXIA – Dublin Sept 2008

A very successful conference in Dublin in September 2008 was organised as a joint venture between Friedreich Ataxia Society Ireland and Ataxia UK. Full report from the conference is available on the Irish and Ataxia UK websites. See www.ataxia.ie and www.ataxia.org.uk. In announcing the upcoming **euro-ATAXIA** annual general meeting, **euro-ATAXIA** also published these objectives:

Objectives of euro-ATAXIA:

- Announce the actual state in the investigation of recessive and dominant ataxias in Europe and all over the world.
- Keep in touch those Scientifics who work with ataxias in Europe with specialists who can organize medical trials.
- Incorporate researchers and doctors (with great experience in different areas of investigation) into different investigation projects on ataxia.
- Show the advances of the different groups and set up new investigation strategies.
- Plan strategies to the recovery of patients.
- Establish collaboration strategies among all European patients' organizations.
- Spread scientific advances in ataxia among

researchers and members of European and North American associations.

-Establish collaboration mechanisms to apply for the financial support of the VII Marco Program of investigation

EUROSCA - Dec 2008 Spain:

It has been a busy few months for the Spanish patient organisation (FEDAES) as they were involved in hosting EUROSCA conference Dec 2008. They have their own annual meeting in June 2009 closely followed by the euro-ATAXIA annual meeting in Valladolid, Spain (see page 18 & 19 for details).

The final meeting of EUROSCA held 5-6 December in Mallorca, Spain. Prof Antonio Matilla, Barcelona gave details of the conference in the FEDAES January newsletter and kindly translated them for **euro-ATAXIA**.

EUROSCA, as you all know, is a multidisciplinary project funded by the VI Framework Program of the European Commission for the period 2004-2009 to investigate the different aspects of dominant Spinocerebellar ataxias (SCAs), including clinical, genetic, translational scientific research which has

the ultimate goal of designing and establishing effective therapeutic strategies for ataxia patients. Fifty members of the 22 groups forming EUROSCA attended from all over Europe, including Austria, Belgium, France, Germany, Italy, Poland, Spain and United Kingdom.

The meeting was opened by Mr. Holm Graessner, EUROSCA Manager, followed by the welcoming words of Ms. Isabel Campos, Vice-president of (FEDAES) who thanked EUROSCA for choosing Palma to hold the meeting.

EUROSCA today



Among the research activities are

- 1) the completion of the world's largest registry of clinical data and the largest collection of DNAs from all over Europe,
- 2) the definition of a new Clinical scale becoming a reference to clinical neurologists throughout the world for dominant ataxias,
- 3) the identification of 3 new genes for dominant ataxias as well as the characterisation of their molecular defects,
- 4) the identification and characterisation in cell and animal models of new molecular routes and biomarkers associated with ataxia, and
- 5) the identification of new potential drug targets that could be possibly exploited in therapeutic strategy, including those using Lithium, histone acetylation inhibitors,

interferon beta, and drugs stimulating the autophagic process, to promote cellular degradation of mutant proteins, among others.

In addition to the high scientific quality, the meeting also reflected the excellent relationships among all EUROSCA members that are so important and needed in order to have fruitful collaborations. Projects such as EUROSCA are an example of how pan European funding should be invested and would not have been possible without EU money.

Dr Antonio Matilla in his own presentation on SCA1 outlined his hopes for the future. He hopes to continue work with the animal models which have been constructed for some of the therapies and that this will verify that they are effective at preventing or at least delaying the neurodegenerative process associated with SCA.

Cuba – October 2008

At the 3rd International Congress on Hereditary Ataxias October 2008, in Holguin, Cuba, world scientists who attended the event at the centre for research and rehabilitation had the opportunity of hearing about the use of Zinc in SCA 2. It was thought that zinc, being the most abundant trace element in the nerve tissue after iron, could have an impact on the progressive development of this disease, with molecular basis associated with neuro-toxicity processes. Clinical trials were therefore carried out on its effectiveness by giving zinc for a period of one year. Some improvement was noted.

Cuba and Mexico have the highest incidence of SCA 2. The conference demonstrated the achievements in the research on the modifier genes and molecular biology from the genome level. It showed how Cuba aimed at improving the quality of life for patients suffering from ataxia and people with risks of contracting the disease.

Ataxia UK have recently published a link from their website which give details of www.ataxia.org.uk/publications_and_pictures/cuba

Friedreich Ataxia Research Alliance (FARA) (www.curefa.org)

FARA presented a one-day symposium at The Children's Hospital of Philadelphia, USA on December 6, 2008, for patients and families to give up to date clinical information, therapeutic approaches and current research being conducted in the field of Friedreich's Ataxia.

The Children's Hospital of Philadelphia also welcomed guest speakers include Sam Bode, Kyle Bryant, and Chelsea Chamberlain. These young people, all FA sufferers highlight their accomplishments despite living with Friedreich's Ataxia.

A visit to FARA web site is a must for any FA sufferer or relative. In particular it is encouraging to see their presentation on clinical trials (www.curefa.org/pipeline.html) which shows 5 different clinical trials in FA in 2009. They also have a section on family resources which is very helpful for any sufferer and their family.

National Ataxia Foundation (NAF) Conference

NAF held its annual meeting in March 2009 in Seattle, Washington DC organised by the local Seattle ataxia support group and British Columbia ataxia support group. This was the first year that the conference has been co-hosted by an international ataxia support group. Nearly 500 people attended the very successful event with representatives from Australia, Canada, Hong Kong and Switzerland.

Kyle Bryant, an ataxia sufferer himself rode for 193.56 miles over 4 days, 3 of them were in wet, cold and windy conditions with log trucks blowing grit into the wind and worsening the weather conditions. Over \$220,000 was raised for ataxia research in the effort and well done to all who took part.

On the medical presentation Dr Thomas Bird, Washington talked about the common ataxia in Seattle area. He gave details of the multi-disciplinary approach to assessing a patient with ataxia and finished by telling his audience that the future is bright for ataxia.

Dr Gregory Carter talked about the rehabilitation aspects of ataxia. In particular he emphasised how important it was to treat the whole patient including telling patients about their rights. Patient can direct their care, clinicians should provide information, gave the patient options and be supportive not directive. Rehab programmes need to be based on sufferers goals and expectations. He informed the group that there is a growing body of evidence that shows exercise is beneficial in ataxia. Pool therapy is ideal therapy as the heat of the water treats spasticity.

Speech and swallowing techniques were Krisite Spence chosen subject. She emphasised that it is important to close down speech. This is difficult to do and the sufferer may feel the speech is already too slow. Slowing down speech may prevent “overshoot” and increase the clarity of speech. It also helps to co-ordinate speech components (lungs, larynx, lip, tongue, jaw, palate) and it also allows the listener to process what the sufferer is saying. She also gave strategies for sufferers to use to improve communications and interactions.....



Picture by kind permission of Dr Michael Wilensky

Dr Michael Wilensky gave general guidelines about medications that are used for ataxia patients. He did talk about side-effects and I think the cartoon above illustrates the point very well. He finished his presentation advising about alternative medicines

- No scientific evidence to prove efficacy
- Beware of side effects
- Do not overdose
- Take only recommended doses
- Good nutrition can contribute significantly to wellness

Saturday after noon saw Marek Napierala present his study of targeting DNA structure of Friedreich Ataxia patient in an effort to find a drug treatment which would help to raise the level of Frataxin in the sufferer. It was an interesting but very scientific lecture.

Dr Henry Paulson talked about the Poly-glutamine ataxias. Glutamine codons are CAA and CAG. In ataxia it is CAG that is repeated. Hence the name polyglutamine ataxias for SCA1, SCA2, SCA 3, SCA 6 SCA7 and SCA 17 which have a repeat of CAG but on different chromosomes. The nature of these ataxias is that they have highly varied disease manifestations and the phenomenon of "anticipation" i.e. successive generation develop at an earlier age of onset. In such cases the child of an affected parent may even present before the parent and will have a greatly expanded repeat. Despite the similarities in disease process different protein in the body are affected causing different disease processes.

Ralph Miller demonstrated wheelchair yoga and Dr Sid Gilman, University of Michigan reviewed sporadic ataxias. He concluded by saying 50% of adult onset sporadic ataxia could not be diagnosed initially despite full evaluation. Most cases followed over time can be diagnosed. He pointed out there still is a confusing amount of terminology to describe undiagnosed ataxia.

ON Sunday morning Dr Sue Perlman discussed how to manage your ataxia and your neurologist. She said that initial frustration with diagnosis may stem from the fact that the true nature of ataxia may take a year or two to reveal itself. Therefore the first time you visit a neurologist they may not be able to nail the diagnosis.

Sufferers are frustrated at how few physician are familiar with ataxia, how long it takes to be diagnosed, how little the neurologist has to offer after diagnosis.

Dr George (Chip) Wilmot gave the closing presentation and summed up the conference by saying:

- take control of what you can,
- supplements are not drugs,
- there are exciting new therapies all around us and
- never underestimate the value of dedication and inspiration.

Editors comment:

Further details of National ataxia foundation can be found at www.ataxia.org/events/2009-amm-presentations.aspx

GoFAR www.fa-petition.org

Bernardo Ruggeri the European representative for Friedreich Ataxia met Repligen pharmaceutical company in Washington DC on Saturday November 15th 2008. Repligen informed the group that it was evaluating several HDAC inhibitors in both laboratory tests and preclinical trials. Pre clinical tests and good manufacturing practices are required to establish if an experimental drug is safe for human clinical testing. As a result of progress in this area Repligen plan another meeting in USA in July 2009 with Friedreich ataxia researchers and patient representatives. GoFAR organised a meeting in Turin 5th June 2009 for the European Friedreich ataxia clinical network of researchers in advance of USA meeting.

Researchers, neurologists and patient representatives from all over Europe attended and there was a valuable exchange of views. Dr David Jacob illustrated Repligen's plans for development of HDAC inhibitors.

Professor Festenstein (Imperial College, London) presented data on a new compound which increases levels of frataxin in patient cells and in mouse models. This is a HDAC inhibitor which is commercially available, thus fewer safety studies would be needed in advance of clinical trials.

There are at least ten centres of excellence in Europe with expertise to run trials in HDAC inhibitors. Repligen is interested in running trials in Europe and recognises the need to enter into discussions with EMEA this year. There is a need for orphan drug designation status for the drugs to be tested in trials. The European network is committed to continue meeting and to prepare baseline data for Repligen phase II trials.

Netherlands

The Dutch have recently had the guidelines for the treatment of Neuromuscular scoliosis translated into English and is available on the web at www.scoliosisjournal.com/content/3/1/14 Jon Buning (VSN) told me their annual meeting in Holland on 26th September 2009.

FARA Australasia www.fara.org.au

The big news from “down under” is their involvement in the deferiprone Phase II double blind trial will commence shortly under Associate Professor Martin Delatycki in Melbourne. Participants will be aged 7 to 17 years olds (differs somewhat from European part of this trial) Australia wide. FARA Australasia strongly encourage the Australian FA community to support our involvement in this international trial. Contact Varlli Beetham our Executive Director on 03 8615 4808 or varlli@fara.org.au.

Association Française de l'Ataxie de Friedreich held their annual conference in Nouant on 21st March 2009. It was well attended and participants were able to hear of the latest developments in France on fundamental research, diabetes in Friedreich Ataxia, psychological problems and the pioglitazone trial (see www.clinicaltrials.gov.) as well as the other major clinical trials in Europe

Federación de Ataxias de España (FEDAES) held their annual meeting on 19-21 June 2009 in Villagarcía de Campos, Valladolid. Drs. David Genis, Antoni Matilla and Victor Volpini were invited to present their research. FEDAES are also hosting the **euro-ATAXIA** AGM in city of Valladolid on 25th & 26th September 2009, for programme see page 18 & 19.

Report from European Organisations**European Health Forum**

European Health Forum have organised a conference in Austria 30 September to 3rd October on financial crisis and Health Policy. One of the three hour workshops is on European Health information system and communication on diseases. The first part of the workshop will explain how the new EU health information and knowledge sharing system will be set up and what it means to citizens, stakeholders, experts and politicians.

The second part will concentrate on the current EU initiatives on chronic and rare disease, focusing on rare disease, cancer and Alzheimer's disease

European Forum for Good Clinical Practice (EFGCP)

EFGCP held their annual conference in Prague in January 2009. Dr Frank Wells, co-chair of the EFGCP ethics working party consolidated suggestions and ideas from the plenary sessions and developed a 12 point plan which EFGCP plan to take further. The following were part of the 12 point plan:

- Definitions of “fraud” and particularly “misconduct” are needed with demarcation between them, across Europe and the rest of the world.

- Every country conducting research should have a body to assess research integrity

- The importance of training stakeholders in clinical research projects in the principles of research integrity and the prevention of fraud and misconduct cannot be over emphasised
- Training of research ethics committee on their role in prevention of misconduct and its management
- Guidelines are needed on the encouragement for and protection of the genuine whistle blowers
- A system to monitor detection of misconduct

For a full report of the conference see [/www.efgcp.be/Downloads/confDocuments/EFGCP Annual Conference 2009 Report.pdf](http://www.efgcp.be/Downloads/confDocuments/EFGCP%20Annual%20Conference%202009%20Report.pdf)

What Determines Disease Severity in the Spinocerebellar Ataxias?

Pulst S, Professor Neurology Salt Lake City
Published in Journal Watch Neurology Dec 2008

Dr Stephan Pulst is professor of Neurology at Salt Lake City in USA. He studied medicine in Germany but then went to Harvard medical school in Boston. He still has close connections with Germany as seen when he presented at the GeNeMove conference in Bonn in May 2007. He found in this research that detailed analysis reveals different factors

- 1) affect disease severity and
 - 2) non-ataxia symptoms
- in the four most common SCA variants.

Spinocerebellar ataxias (SCAs) are caused by mutations in an ever-increasing number of identified genes. The most common types, SCA1, SCA2, SCA3, and SCA6, result from translated CAG-repeat expansion mutations. Despite great progress in understanding the molecular pathogenesis of these disorders, relatively little is known about factors that determine severity and clinical phenotype.

Now, researchers studied clinical severity of these four SCAs in relation to age of onset, repeat length, and disease duration. In 526 patients from 17 medical centres, these investigators measured disease severity by using the recently introduced Scale for the Assessment and Rating of Ataxia (SARA) and number of non-ataxia

symptoms by using the Inventory of Non-Ataxia Symptoms (INAS).

In SCA1, SCA2, and SCA3, the SARA score increased (from a worsening of symptoms) with increasing CAG-repeat length and with disease duration. SCA1 had the fastest progression after onset. SCA6 progression was influenced more by patient age than by disease-related factors (higher SARA score was associated with older age at onset and longer disease duration). The mean INAS score for SCA6 was lower than for other SCAs. Overall, INAS scores increased with disease progression.

This is the largest study to date examining clinical and genetic aspects of SCAs. The findings emphasize the importance of multicenter investigations for rare disorders. This study highlights the importance of non-ataxia features as markers of disease progression in SCA1, SCA2, and SCA3. The authors confirm that SCA6 has fewer non-ataxia features than do other SCAs. They did not measure progression prospectively, but the findings provide a foundation for prospective natural history studies, which will be essential for the design of clinical trials.

Charot-Marie-Tooth - What is it?

Charcot-Marie-Tooth disease is named after the three neurologists who first described the condition in 1886. Many different names have been used to describe Charcot-Marie-Tooth disease but the other commonly used name is **hereditary motor and sensory neuropathy**. This is an accurate term as it refers to the two primary features of this condition i.e. the condition is hereditary and it affects the motor and sensory peripheral nerves. The term 'neuropathy' refers to the fact that it is the peripheral nerves (which connect the spinal cord to the muscles, joints and skin, carrying messages in both directions), which do not function normally'

Charcot-Marie-Tooth disease is used to describe a group of conditions that give rise to weakness and wasting of the muscles below the knees. Symptoms usually begin in late childhood or early adulthood. The initial symptom is foot drop early in the course of the disease. This also causes "hammer toes" i.e. where the toes become curled. and often those of the hands. Many affected people also have loss of feeling in the hands and feet in later life as the disease progresses.

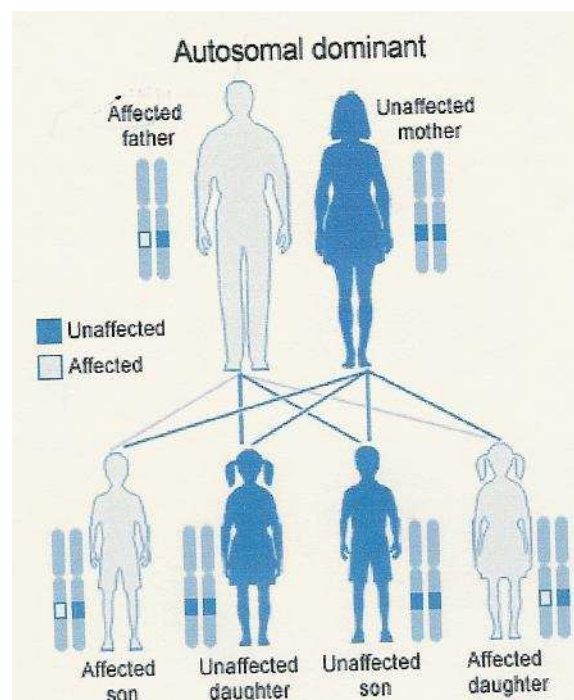
Charcot-Marie-Tooth disease is also referred to as peroneal muscular atrophy because the peroneal muscles on the outer side of the lower leg are usually the first to be affected. Other names for this disease include Dejerine-Sottas disease and hereditary hypertrophic neuropathy. However, the term Charcot-Marie-Tooth disease is now the favoured term and is most commonly used in the literature.

How is Charot-Marie Tooth inherited?

There are different ways that Charcot-Marie-Tooth(CMT) can be inherited but the commonest forms of CMT disease are inherited in an autosomal dominant way. This type of inheritance is seen in Charcot-Marie-Tooth disease type 1 (CMT 1) and type 2. There are sub types of subtypes of Charot – Marie-Tooth type 1 and type 2 which share clinical symptoms. They affect different gene and for which there are currently gene test available.

Charcot-Marie-Tooth can also be inherited in an autosomal recessive type way which is similar to the way that Friedreich Ataxia is inherited. This is seen in Charcot-Marie-Tooth type 3 (CMT3), also know as Dejerine-Sottas disease. However this is an old classification and now Charot-Marie-Tooth type 3 disease is more commonly referred to as CMT4F. Charcot-Marie-Tooth type 4 has several different types for which gene testing is also available.

Charcot-Marie-Tooth disease can also be inherited in X linked dominant form and it is referred to as CMT X-linked. A woman with an X-linked dominant disorder has a 50% chance of having an affected child. The daughters of a man with an X-linked dominant disorder will all inherit the condition. CMTX affects approximately 10-20% of all CMT patients



Many thanks to National library of Medicine USA for diagram

There is no current standard treatment for CMT. Patients are advised to maintain a healthy weight as extra weight places additional stress on the joints. The chemotherapy drug vincristine has proven hazardous and should be avoided in CMT



euro-ATAXIA Scientific conference, Friday 25th September 2009 - Hotel Meliá Olid, Plaza San Miguil Valladolid, Spain

08:45 Registration & Welcome Isabel Campos FEDAES & Dr Francesc Palau, Vice president *euro-ATAXIA*

09.30-13.30 Session I – Recessive ataxias Chair: Dr. Francesc Palau

09.30 Current state of histone deacetylase inhibitors investigation''

- Prof. Massimo Pandolfo, Hôpital Erasme, Belgium).

Drosophila and frataxin: effects of its reduction and over expression

- Dr. José Vicente Llorens Department of Genetics, Valencia.

PPAR-gamma agonist - New Implications for the Friedreich's Ataxia Therapy.

-Dr. Daniele Marmolino, Hôpital Erasme, Brussels.

Response of Friedreich ataxia murine models to the IGF-1 "

-Dr. Ignacio Torres Alemán: Laboratory of Neuroendocrinology. Madrid

Friedreich Ataxia, computational dynamic Model of the proteins that form the complex Iron –sulphur in *Saccharomyces cerevisiae*

Dr. Isaac Amela Abellán. Laboratory of Bioinformatics Institute of Biotechnology and Biomedicine (IBB) Universitat Autònoma de Barcelona''

11.10-11.30h: Coffee break

11.30 Altered lipid metabolism in a *Drosophila* model of Friedreich Ataxia

-Dr. Dr. Juan Antonio Navarro, Universität Regensburg, Germany

Knock-down of frataxin gene expression in human neuron-like cells as a cell model for FA

-Dr. Javier Diaz Nido, Molecular Biology, University Autonoma, Madrid.

Human neuronal cell models and gene therapy strategies for Friedreich ataxia''

-Dr. Filip Lim. Faculty of sciences, University Autónoma of Madrid

Cellular therapy on ataxia: Possibilities of application

- Dr. Salvador Martínez Pérez, University Miguel Hernández. Campus de San Joan. Alicante''

13.30-15.10 lunch.

15.10-17.00 Session II – Dominant Ataxias Chair: Dr. Mercedes Pineda

15.10 Current state of the investigation on dominant ataxias

- Dr. David Genís. Servicio de neurology. General Hospital of Gerona. Spain

Spino-cerebellar ataxia type 1 molecular pathways, pathogenic mechanisms, and therapy.

Dr. Antoni Matilla Dueñas. Barcelona

RNA interference on Spino-cerebellar ataxia 3

Dr. Luís Pereira de Almeida. Dept of Neurosciences, University of Coimbra, Portugal.

Cell Therapy in a Mouse Model Purkinje Cell Degeneration

-Dr. Manuel Álvarez Dolado, Sevilla

Session III – Clinical Trials Chair: Dr. Antoni Matilla

- 17.00 Clinical trials in adults with Deferiprone, idebenone, Riboflavin, IGF-1**
-Dr. Francisco Javier Arpa. Department of Neurology. Hospital La Paz.- Madrid.
- Clinical trials with GH and EPO**
-Dr. José Luis Muñoz. Service de Neurology. Hospital Gregorio Marañón. Madrid.
- Ataxia with Co Q deficiency in children**
Dr. Mercedes Pineda Marfá & Dr. Asunción Aracil. Neuropediatrics, Barcelona.
- 18.00-18.30 Coffee break**
- 18.30 “Friedreich ataxia’s patients Monitorship**
-Dr. Rafael Artuch Iriberrí, Hospital Saint Joan de Deu. Department of clinical biochemistry
- “New therapeutic strategies for hereditary ataxias, in particular our results on EPO**
-Dr. Giuseppe De Michele, Università Federico II. Naples. Italy.
- "Pioglitazone in FRDA: Why and How?**
-Dr. Pierre Rustin, Robert Debre Hospital, Paris. France
- Results of the clinical trials with Fosfatil-B vitamin complex**
-Dr. Benedicta Catalán. Valladolid University Hospital.
- Results of the multicentre clinical trial with Idebenone –**
- AWAITING CONFIRMATION TAKEDA.

Discussion on results facilitated by Dr. Antoni Matilla

21.30h: Dinner

SATURDAY - 26th September 2009

10.00-13.00 - Session VI - Meeting of Patient Organisations

Coordinator: Sue Millman, Secretary-general *euro-ATAXIA*

- What activities could we develop to achieve European financing?
 - Meeting of the steering committee of *euro-ATAXIA*
-

SOCIAL PROGRAM - SATURDAY, 26th September 2009

10.00 to 13.00 A tour of places of interest in Valladolid for the participants who haven't to take part in Session IV.

Participants who have registered will be supplied with the conference documentation on Friday 25th September, 08.45h, where the conference will take place. You can find more information about the program and the social work of FEDAES in: www.fedaes.org

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