FRIEDREICHS ATAXIA SOCIETY IRELAND
(Incorporating other Genetic Ataxias)
San Martino, Mart Lane, Foxrock, Dublin 18.
Tel: 01 - 289 4788  Fax: 01 - 289 8845  Email: ataxia@eircom.net
Website: www.ataxia.ie
Chy. No: 7076.

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Friedreichs Ataxia - Society Ireland
Progressive Disability from Childhood
Incorporating other Genetic Ataxias
EDITORIAL

EXCITING BREAKTHROUGH IN RESEARCH INTO FRIEDREICH'S ATAXIA

The news that was received at the FASI office on the 24th August was the most exciting and the most encouraging in relation to the Friedreich's Ataxia research programme to-date. It is almost unbelievable that a breakthrough appears to be a real possibility at last. Professor Gottesfeld and his team at the Department of Molecular Biology in the U.S.A. have achieved extraordinary success in restoring the missing Frataxin in the blood cells taken from Friedreich's Ataxia patients. This, of course, is only the first step towards finding a suitable treatment for the disorder but it is a huge giant step that brings us much, much nearer a solution to the ravages of F.A. We can only hope and pray that this research will progress successfully over the coming year and that soon we will be able to have a treatment for all our members.

We print below, a copy of Professor Gottesfeld's email and, as further information is received by us it will be put on our website so that you will be kept up-to-date on any progress. Please keep in touch with our website.

Editor

A Copy of Professor Gottesfeld’s email on the breakthrough

Dear All,

As you can imagine, with the press release from Nature, FARA, and other sources, and the article in the Daily Telegraph, I have received numerous emails and phone calls from Friedreich's patients and their relatives. So I apologise for this bulk mailing.

Just to let you know where our research stands, what we have reported in our paper in Nature Chemical Biology is that we have identified a class of compounds called histone deacetylase inhibitors that reverse the silencing of the frataxin FXN gene in white blood cells from Friedreich's ataxia patients and their carrier relatives. As many of you know, the genetic defect in FRDA is an expansion of a simple repeated DNA sequence in this gene. This expanded region of the gene causes the FXN gene to be silenced. So our laboratory looked for drug-like compounds that would reactivate the FXN gene in cells taken from FRDA patients. We found a compound that is only available for research purposes that did show some effect in these cells, then a chemist in my lab improved on this molecule. What we now have is a small molecule that increases the output of frataxin RNA from the previously silenced gene.

This is good news, but this does not necessarily mean that a treatment is at hand. We now need to show that the molecules do get into the brain of laboratory mice and function as expected. What we have to do next is to show that the molecules are safe in animals, and we need to determine the properties of the molecules in animals and finally figure out the appropriate dosing regimens for increasing RNA output from the frataxin gene. All of this takes time, and I estimate if everything were to go well, we might be able to have these molecules in human clinical trials in about 18 months or so. However, this may take longer.

I encourage you to check on the Friedreich's Ataxia Research Alliance web site for any future developments, and if a clinical trial is announced, such information will certainly appear on the FARA web site. The site is http://www.faresearchalliance.org

Best wishes to you and your families,

Sincerely,

Joel M. Gottesfeld, Ph.D., Professor, Department of Molecular Biology,
The Scripps Research Institute, California.
It is 25 years since the formation of the Friedreichs Ataxia Society Ireland, yet, when we were researching and investigating the various events and activities throughout the years we were surprised to realise we had come such a long way. The vast changes we encountered, the advances we have made and the numerous contacts which have been built up with government agencies, corporate bodies and indeed the friendships with members, their families, friends and also all the voluntary helpers.

The night of celebration was a huge success and indeed most enjoyable. We endeavoured to contact as many people as we could who were with us from the very outset of the foundation of the Society. Their immense contribution ensured that the best structures were put in place and that the most suitable ethos was adopted for the benefit of the members and their families.

Everyone who attended the celebration had a common bond and this was very obvious throughout the evening as the whole ‘FASI family’ became as one. The venue was absolutely ideal, it being Jury’s Hotel in Ballsbridge, Dublin, where the ambience was a real treat, the special meal was beautiful, the friendly and attentive service great and the facilities for us all most adequate. Nobody was forgotten. The music enticed the dancers onto the floor and the Special Guest, Aengus McAnnally, entertained us with his songs and stories.

Many thanks to each and everyone for making this a wonderfully enjoyable and memorable night – it certainly was a fitting way to remember 25 years of progress and achievements.

The response from all the members following the 25th celebration was that Jury’s Hotel should be the venue for future FASI functions – as long as possible. This was mainly because the facilities were excellent for wheelchair users. Unfortunately Jury’s Hotel Ballsbridge will be demolished in the not too distant future.
Swallowing Safely with Ataxia

By Julie Regan, Senior Speech & Language Therapist, Adelaide and Meath Hospital, Tallaght, Dublin 24.

1. Swallowing for eating and drinking

Swallowing involves a finely co-ordinated sequence of events to ensure the food and drinks pass safely from the mouth to the stomach and not into the wind pipe which leads to the lungs.

Several different groups of muscles and nerves are involved in both the mouth and the throat area to ensure the following:

• The food must be chewed and made ready for swallowing.
• The tongue must move the food backwards.
• The airway must close and lift up out of the way so food cannot get in.
• The oesophagus must open to let the food in.
• The oesophagus must close again and the airway opens up once more.

2. What is a swallowing problem?

Ataxia can cause both weakness and lack of co-ordination in the muscles involved in swallowing. This can cause the swallow to be impaired. The medical term for a swallowing problem is dysphagia.

3. Signs and symptoms of a swallow problem

There are many different signs and symptoms associated with a swallowing problem. If any of the signs below match what happens to you, try and talk to someone about it and ask your health care team for advice.

- You swallow repeatedly
- You cough and splutter frequently when eating or drinking or experience choking during meals
- Your voice has a “wet gurgly” quality during or after meals and you often need to clear your throat
- When you try to eat you dribble. Food and saliva escape from your mouth or can go up into your nose and cause sneezing
- You find it easier to eat slowly
- You are avoiding certain foods which catch in your throat (e.g. dry crumbly brown bread, cereal, peanuts)
- You quite often keep old food in your mouth, particularly when you have not had a chance to get rid of it unseen
- You feel “chesty” after meals and are losing weight

Symptoms

If eating and drinking causes you that sort of distress, you have sometimes avoided eating and drinking altogether. You may become dehydrated and begin to lose weight. This in turn can lead to the development of malnutrition. The most serious hazard is when food and drink leaks into the airway (the medical term for this is aspiration). This can lead to the development of chest infections or pneumonia.
4. What to do?

Contact your GP/local hospital to arrange a swallow assessment by a trained Speech and Language Therapist (SLT). Identification of a swallow problem is very important to reduce the risk of food and drinks going down the wrong way and to prevent dehydration or malnutrition. SLT’s also work closely with nurses and dieticians to make sure you are getting enough nourishment.

A swallowing impairment resulting from ataxia will not usually be cured, but there are many ways to make eating and drinking easier, so you should not hesitate to seek professional advice.

The SLT provide various forms of treatment including:

- Diet modification (e.g. thickening drinks to prevent them from entering the windpipe)
- Postural changes (e.g. chin down posture while swallowing can reduce the risk of fluids/foods entering the windpipe)
- Exercises (e.g. tongue strengthening exercises / palatal exercises)

5. Tips when eating and drinking

- Direct treatment (e.g. providing sensation to back of mouth to speed up swallow)
- Look out for signs of swallowing problem/aspiration as outlined.
- Look for patterns during meals - are drinks causing you to cough? Or are dry crumbly foods catching in your throat?
- Sit upright for all eating and swallowing. Never eat or drink when lying down
- Take small amounts (i.e. sips & small mouthfuls) of food and liquid at a time.
- Take a small sip of liquid, hold it in your mouth until you feel ready to swallow and then make an effortful swallow.
- Drink liquids and eat food separately. Make sure that you have swallowed down all the food before the next mouthful.
- Give a dry swallow after each mouthful of food or liquid to ensure it is safely swallowed.
- Eat small meals often as larger meals result in the swallow tiring.
- Avoid dry crumbly foods such as digestive biscuits. Alternatively, moisten them with jam/butter.
- Avoid tough chewy foods such as toffee's, fatty meat etc.
- Avoid solids in a liquid base such as minestrone soup etc. Try to keep food the same consistency.
- Try not to talk while eating or drinking. This can upset your breathing pattern for swallowing!
- Stay upright for 30 minutes after eating, do not lie down.
- Try putting your chin to your chest (see below) when swallowing to stop drinks/food from catching in your throat. Also, try turning your head to the left or right to clear food or drinks that are stuck in your throat (see illustrations).
People are more aware of what is healthy and unhealthy in the food that they eat today. There are good and bad fats, bad fats are saturated fats which are found in full fat dairy products and fat meats, they are also found in cakes, biscuits, pastries and packet goods, in other words, processed foods. We do need some fat but the fat we need is unsaturated fat. Nuts and seeds are a very good source of unsaturated fats—Pumpkin Seeds, Flax Seed, Linseeds and nuts are all high in unsaturated fats.

Why are unsaturated fats so essential? The body cannot produce some of the essential fatty acids that we need and these can only be got from certain foods mainly seeds and nuts.

The most important fatty acids are Omega 3 and 6. They ensure proper brain development and have shown to give significant help with hand eye co-ordination and they greatly benefit skin conditions such as eczema, psoriasis and acne. They help women suffering from P.M.T. and going through the menopause as they regulate hormones. They reduce pain and discomfort in joints where arthritis is a problem and improve mobility. They also help the kidneys get rid of excess fluid. Depression and mood swings can also be helped and they play an important role in the care of hair and nails and are a very good source of energy and vitality. They give us an all over good feeling.

I am not saying that if you take Omega 3 and 6 you will become Superman overnight but they do help in the areas mentioned.

I take flax seed oil capsules and I find them very beneficial but everyone is different. It is available in granule capsules and liquid form and can be got from any good Health Food Shop or Chemists. Some Flax Seeds don’t contain Omega 3 and 6 so check the contents.

Aine Reilly

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SOME SUITABLE EXERCISES WHILE SITTING

Exercise Programme

The exercises should be done for short periods several times a day. Use a firm chair with arms and sit well back in the chair. Perhaps, you and the person you are caring for should do these exercises together to suitable music.

Head
- SLOWLY bring your chin forward towards your chest, then lift your head backwards to look up to the ceiling.
- SLOWLY turn your head to look over your right shoulder and then over your left shoulder.

Hands
- Make a tight fist and then stretch fingers out straight.

Arms
- With elbows straight, lift both your arms sideways and upwards to shoulder level, and then lower.

Legs
- Lift one knee up to your chest and then lower. Repeat with the other knee.
- Straighten out one knee, hold for four seconds, and then lower. Repeat with the other knee.

Breathing
- Take deep breaths in through your nose and out through your mouth aiming to get the air down to the bottom of your chest. Take 3 deep breaths, then pause before taking another 3.

Feet
- Foot circling - one foot at a time.
- With knee straight, point foot up and down. Do the same with the other leg.

If you feel worse after a particular exercise leave it out for the present. Learn to listen to the needs of your own body - note what is helpful and note what is less helpful.

These exercises may not be suitable for some medical conditions, check with your doctor if you are in doubt.

A MEMBERS DIETARY TIP

Some suitable exercises while sitting
**Quick and easy Roast Chicken.**

There is a Roast Chicken you buy in Marks & Spencer’s and it comes in two bags. Just before placing on the turntable in the microwave, take the outer bag only off. Microwave on full power for 25 minutes. Just before the last 5 minutes turn-back sides of bag and leave to cook for last five minutes. *Hey presto, a delicious roast chicken.*

**Stuffing simplified.**

You can buy stuffing in a box, the packet of Country Kitchen Stuffing is very good. Place contents of pack into a microwave bowl. Add 50g (2 oz) of butter and quarter pint of boiling water and stir well. Allow to stand for 10 mins. Put cover on bowl and cook on full power for 4 minutes. Leave to stand for 2 mins. *Serve chicken and stuffing with mashed potato and vegetables.*

**Crunchy Fruit Surprise**

*by Brenda (Office)*

1 tub of Crème Fraiche

1 tub of Mascarpone Cheese

1/2 pt carton of Cream

Frozen Fruits of the Forest (fresh if available)

A packet of Amaretti Biscuits.

Gently whisk the cream in a bowl and add the tub of Crème Fraiche, keep mixing for a little while, and then add the tub of mascarpone cheese. Roughly break up the amaretti biscuits and put them in a bowl. Pour the defrosted frozen fruits (or fresh if available) on top. And on top of this place the cream mixture. Sprinkle with chocolate flake or fresh fruit on top. Chill in fridge and serve.

**Pork in Sweet and Sour Sauce**

*by Clare Louise Creedon*

**Cooking time: about 17 mins**

227g can pineapple cubes in fruit juice

Chicken or vegetable stock

5ml (1tsp) oil

1 medium onion, finely sliced

1 garlic clove, crushed

2 celery sticks, finely sliced

450g (1lb) lean pork, finely sliced

30ml (2 tbsp) tomato puree

45ml (3 tbsp) red wine vinegar

45ml (3 tbsp) soy sauce

45ml (3 tbsp) brown sugar

30ml (2tbsp) cornflour

Salt and freshly ground black pepper

1 green pepper, seeds removed and finely sliced

1. Drain the pineapple and make the juice to 300ml (?pt) with chicken or vegetable stock.

2. Put the oil, onion, garlic and celery in a large bowl, cover and cook in microwave for 5 mins, stirring once.

3. Stir in the pork and pineapple juice mixture.

4. Whisk together the tomato puree, vinegar, soy sauce, sugar and cornflour, then stir into the pork mixture. Season with salt and pepper.

5. Cover and cook for 5 mins. Stirring once or until boiling. Continue cooking, covered, for about 5 mins, stirring twice or until the pork is tender.

6. Stir in the pepper and pineapple, cover and cook for about 2 mins until hot. Serve with rice – try Uncle Ben’s Express Rice it takes only 2 minutes in the microwave and there’s no wash-up after!

*If anyone has any tasty recipes they’d like to share with us all, please let us know in the office and we will put them in the next issue*
THE FASI HOLIDAY 2006

Cuisle 2006 was once again a raving success! To start with the weather was fantastic which made all the events so much more enjoyable. Unfortunately we were unable to go to the Races this year as they had been postponed till the following week but we didn’t let that stop us. Peter Finn organised pantomime horses and we had our own races and even took bets on the horses! It was a great laugh. Peter also organised “It’s a Knockout” and we had great fun with the large wellies and the buckets with the holes. A big THANK YOU to Peter for all his work - it was super fun.

The bowling and swimming were as popular as ever and this year the new bowling alley in Roscommon town made it easier to attend so we went twice. The Beautician and Masseuse were kept really busy all week and on the last night the hairdresser was there to beautify us all for the final night. The costumes were super and everyone made a great effort to dress as their favourite “Nursery Rhyme” character. The Holiday would not have been the success that it was without the staff in Cuisle who made us all feel very welcome and also our own helpers who we couldn’t have done without. The food, as usual, was fantastic with all our tastes being catered for. The photos below will confirm what a great time was had by all.
Dublin City Marathon

Gareth McKeever ran the Marathon last year in aid of the Society and has decided to give it another go again this year so we hope you will all show your support by getting sponsorship for him taking on this challenge again this year. The best of GOOD LUCK Gareth from all of us!

The Ladies Mini Marathon

Many thanks to all the ladies who ran, walked or crawled(!) in the ladies Mini Marathon this year. It was a wonderful achievement and by all accounts it was a great day – the sun even came out!

Parachute Jump

Attracta Lafferty undertook a Parachute Jump to raise funds for the Society. There is a photo to prove she did it!!

Golf Classic

A very successful Golf Classic was held on June 15th in Glen of the Downs Golf Club. The Friedreich's Ataxia Society was the sole beneficiary of the event this year. The funds raised are a welcome addition to the Society’s

Damos’ interesting facts

• Butterflies taste with their feet.
• A duck’s quack doesn’t echo, and no one knows why.
• In 10 minutes, a hurricane releases more energy than all the world’s nuclear weapons combined.
• Thirty-five percent of the people who use personal ads for dating are already married.
• Elephants are the only animals that can’t jump.
• It’s possible to lead a cow upstairs, but not downstairs.
• Women blink nearly twice as much as men.
• A snail can sleep for three years.
• Our eyes are always the same size from birth, but our nose and ears never stop growing.
• All polar bears are left handed.
• An ostrich’s eye is bigger than its brain.
• A crocodile cannot stick its tongue out.
• African elephants only have four teeth to chew their food with.
• A house fly lives only 14 days.
• The oldest breed of dog is the Saluki.
• The bee hummingbird of Cuba is the smallest bird in the world.
• An ostrich can run up to 70 km/h (43mph).
• The world’s smallest dog is the Chihuahua, which means "tiny dog in the sky."
• Pea crabs (the size of a pea) are the smallest crabs in the world.
• 75% of wild birds die before they are 6 months old.
• Pork is the world’s most widely-eaten meat.
• The coyote is a member of the dog family and its scientific name, "canis latrans" means barking dog.
• A giraffe can clean its ears with its 50cm (20 in) tongue.
• The South American giant anteater eats more than 30,000 ants a day.
**HANDY INFORMATION**

**COSY FEET**

If you are having a problem looking for shoes and can’t find ones that fit comfortably try getting in touch with Medical Mobility in Killarney. They stock the Cosy Feet Brand and they can be supplied by mail order. There number is 086–2308788. They will forward you a brochure with all styles available.

**SAFETY TIP**

Every home should have at least one working smoke alarm, ideally one in every room.

A working smoke alarm can double the chance of survival. Remember to replace smoke alarm batteries twice a year to ensure that they are working.

**TRAVEL WEBSITES**

*Travel Websites - check these out to see if there is anything that might interest you!*

- www.wheelchairaccessibleeurope.com
- www.accessibletravel.co.uk
- www.worldonwheelz.com
- www.access-able.com
- www.disabilitytravel.com
- www.laspiedras.co.uk
- www.assisiaccessible.it – Italy Accessible Assisi
- www.eria-resort.gr – The Greek Island of Crete

**FREE DIRECTORY ENQUIRIES**

If you have a problem looking through the phone book for a number Eircom offer a Free Directory Enquiry Service for blind or disabled customers. Phone Eircom on 1800 574 574 for an application form.

**A USEFUL WEBSITE TO VISIT**

**www.assistireland.ie** – This site provides information on assistive technology and a directory of products available from Irish suppliers. It also has information on Entitlements which members might find useful to know.

**A PRAYER FOR THE STRESSED**

*By Peter Finn*

Grant me the sanity to accept the things I cannot change,
The courage to change the things I can
And the wisdom to hide the bodies of those I had to kill today because they got on my nerves;
And also help me to be careful of the toes I tread on today
as they may connect to the feet I have to kiss tomorrow!

Help me to give 100% at work:

12% on Monday
23% on Tuesday
40% on Wednesday
20% on Thursday
and 5% on Friday

and help me to remember, when I am having a bad day and it seems that people are trying to wind me up, it takes 42 muscles to frown, 28 to smile and only 4 to extend my arm and smack someone in the mouth.

**To be honest, sincere,**
**easy and pleasant to live with is of more value than much gold**
CONGRATULATIONS to Marian Goff

Who has achieved a BA (with honours) in Counselling and Psychotherapy By completing “The Impact of Friedreich's Ataxia and Other Hereditary Ataxia on Quality of Life – An Irish Study” Marion is most grateful to the members of FASI who co-operated in this study and to the Society for sending the questionnaires to each one.

WELL DONE MARIAN!

A Sad Farewell

It is with great sadness that we report the loss of 3 of our cherished members since the last edition of our Newsletter. Ciarán Logue departed this life in June 2005, Rosemary Halton-Smyth died in February 2006, and Stuart Walker sadly passed away in February 2006. Their untimely deaths were made more poignant by the fact they were all such young and lovely people. May they rest in peace. We pray that God will bring peace and comfort to their families and friends.

Regina and Richie have been helpers on our Annual Holiday for a number of years. They got married last year and returned from their honeymoon to come on the FASI Holiday! We would like to congratulate them both on the safe arrival of their son Jamie.