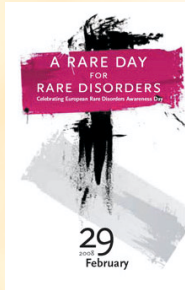


## A RARE DAY FOR RARE DISORDERS



The Genetic and Rare Disorders Organisation (GRDO) is an Irish alliance of voluntary groups, involved with genetics and other rare disorders. HDAI is an active member of GRDO

and has a representative on it's board. GRDO organised an information event in the Mansion House Dublin to celebrate the first European Awareness Day on 29th February.

A rare disorder is a disorder affecting less than 1 in 2000 people. It is estimated that there are 140,000 people in Ireland with a rare disorder.

The meeting was addressed by Professor Andrew Green, Director of the National Centre for Medical Genetics (NCMG) in Crumlin who spoke about the work of NCMG and the need for greater awareness and support for genetic conditions. Two people affected by a

genetic condition spoke about how their condition affects their lives.

Judy Windle of GRDO said "the low profile and public awareness of rare disorders in Ireland means that Irish people are facing tremendous difficulties. Scarcity of information and expertise and a lack of specific health policies translate into delayed diagnosis and difficulties in accessing care. Often people with rare conditions cannot find a relevant support network resulting in a feeling of vulnerability and isolation for them and their families". She added "When you look at the incentives in Europe and what is happening in the different member states of the EU, you see almost nothing happening in Ireland. To-day we hope will show those affected that they are not alone and that there is someone who can help point them in the right direction."

"GRDO acts as a primary source of information and support for Irish people affected by rare disorders and an important part of the Awareness Day is to make such people aware that the

support is available to them." she said. GRDO was joined by the Medical Charities Research Board, by RehabCare the health and social care division of Rehab Group and by the Irish Platform for Patients, Science and Industry (IPPOSI)

This was the first European wide awareness campaign for rare conditions. The 29th February was designated and promoted as Rare Disorders Day by the European Organisation for Rare Diseases (EURORDIS) which represents 300 organisations in 34 countries, covering more than 1,000 rare disorders.

The European Parliament held a Public Hearing in Brussels on 4 March to present the issue of rare disorders as a public priority and to promote national plans for rare disorders in member states. At present, France is the only EU country to have a rare disorders plan in place.

 For further information visit:  
[www.grdo.ie](http://www.grdo.ie)  
[www.eurordis.org](http://www.eurordis.org)

## LEISURE DAY

Would you like to attend a therapeutic leisure day in the Phoenix Park? This one day service allows a patient to use therapeutic facilities such as a Jacuzzi, massage, or music therapy etc. St Mary's run an enjoyable leisure day at their therapeutic centre which is located in a very scenic area of the Phoenix Park. Contact the office for more information.

## HDAI LIBRARY

We have a stock of relevant books in our library. These are available to our members on request.

## MEDICAL ID CARD

A free Huntington's ID card is available. Please send your photograph, address, phone number and an emergency contact person's phone number to the HDAL office and we will take it from there.

## DONATIONS

- Sincere thanks to The Teachers Union of Ireland Credit Union who recently donated €200 to HDAL. We are very grateful for your continued support.
- Thank you to Jack and Marie for their kind wishes and donation in February.

We are very grateful to all our members and friends for your support.

## COFFEE MORNINGS

You are very welcome to join us at our Coffee Mornings/Support Meetings in Cork from 11.00am to 1.00pm on the first Saturday of each month. The West of Ireland meeting is

held on a quarterly basis. In Dublin, meetings have relocated to a city centre hotel. We hope to host a carers workshop every third month in Dublin and Cork. Our Family Support Officer or

a committee member will host the meetings. Tea, coffee and scones are provided. Contact the HDAL office on 1800 393939 for more information.

### WEST

Jun. 7th 2008 - Cuisle  
Sept. 13th - Castlebar

### CORK

May 3rd 2008 - Support Meeting  
June - No meeting (Cuisle Weekend)  
July 5th - Carers Workshop

### DUBLIN

May 17th 2008 - Support Meeting  
June 21st 2008 - Support Meeting  
July 19th 2008 - Support Meeting



Carmichael Centre,  
North Brunswick Street,  
Dublin 7.  
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Web: [www.huntingtons.ie](http://www.huntingtons.ie)

**DO YOU HAVE ANY ARTICLES TO  
CONTRIBUTE OR TOPICS YOU WOULD  
LIKE DISCUSSED? THIS IS YOUR  
NEWSLETTER**

The articles which appear in this newsletter may not necessarily represent the views of HDAL.



# NEWSLETTER OF THE HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND

# Hope

IRELAND

ISSUE NO. 34, Q2 08  
HDNL 2008

CHECK OUT OUR  
WEB ADDRESS

[www.huntingtons.ie](http://www.huntingtons.ie)

SUPPORT MEETINGS  
SEE DATES ON PAGE 4

## GENETICS

THE NATIONAL CENTRE FOR  
MEDICAL GENETICS HOLDS  
GENETIC CLINICS IN DUBLIN,  
CORK, GALWAY AND LIMERICK.  
CONTACT DETAILS -

TEL: 01 409 6902  
AND [WWW.GENETICS.IE](http://WWW.GENETICS.IE)

THIS IS YOUR  
NEWSLETTER. PLEASE  
SEND US YOUR VIEWS

## MEMBERS MEETING & AGM

JUNE 7TH 2008

Our AGM and Members Respite weekend takes place from the 6th - 8th June in Cuisle Respite Centre, Co. Roscommon. All facilities at Cuisle are fully accessible to people with disabilities. Please mark these dates in your diary. We will be sending out information and booking forms shortly.

If you can not attend due to travel difficulties we will try to work something out. We provide a return bus service from Dublin to Roscommon. The bus leaves Dublin on Friday evening and returns Sunday afternoon. We also hope to provide a bus from Cork again this year if there is sufficient interest.

## SUCCESSFUL FUNDING APPLICATION

The Department of Community, Rural and Gaeltacht Affairs, have allocated funding of €18m over the coming three years for 64 national community and voluntary organisations. This funding is part of a new Scheme to Support National Organisations in the Community and Voluntary Sector.

HDAI were successful in their recent funding application and will receive €25,000 per annum over a three year period towards our core costs such as rent, staff, administration and ongoing running costs. HDAL are very grateful to the Department of Community, Rural and Gaeltacht Affairs for this funding.

## SEA SAFARI



## ACTIVITIES FOR Young People

**CALLING ALL YOUNG PEOPLE.** We are going to have a day out at the Sea Safari during the next school break. If you are interested call me so we can arrange a day that suits all of us.

Elizabeth,  
Family Support Officer, Tel: 1800 39 39 39 or 087 051 7772.

## ANNUAL MAGAZINE

We'll be putting together our Annual Magazine for 2008 in the coming weeks so please send in your contributions.

If you have any personal stories, recipes, useful information, photos or jokes etc. we would be glad to hear from you.

## SUPPORT FOR YOUNG PEOPLE

HDAI RECEIVE FUNDING FROM THE COMMUNITY FOUNDATION FOR IRELAND

HDAI were successful in our application to the Community Foundation for Ireland to provide support for young people from HD families. We know that children from HD families often cope with anxiety and stress or have to help care for a parent. Funding will help us provide supervised fun activities for young people. For more information please contact:

Liz O'Sullivan, Family Support Officer, Tel: 1800 393939 or 087 0517772

## SWALLOWING DIFFICULTIES IN HD

HD patients often experience difficulty with chewing, drinking and swallowing. Dysphagia is the term used to describe this. Difficulties can occur in the mouth and throat and create a risk of food and liquid going into the lungs instead of the stomach. This in turn can lead to chest infections or pneumonia.

Dysphagia results from impaired voluntary control of the mouth and tongue, impaired respiratory control due to chorea, and impaired judgment, resulting in eating too rapidly, or taking overly large bites of food and gulps of liquid.

Dry mouth, which can be brought on by neuroleptics, antidepressants, and anticholinergics, may worsen the problem.

There are many warning signs which may suggest swallowing problems. The following are some indicators to look out for:

- Coughing or choking on food and drink
- Difficulty chewing food in the mouth
- Spillage of food or liquid from the mouth
- Food or liquid left in the mouth after swallow
- Gurgly voice especially after eating / drinking
- Frequent throat clearing
- Recurrent chest infections / pneumonia
- Frequent temperatures

**In order to avoid problems the following tips should be considered:**

- Eat slowly and without distractions.
- Prepare foods with appropriate size and texture.
- If a patient has problems their carer should supervise at eating times.
- Carers should know the Heimlich manoeuvre.

Early referral to a speech-language therapist will help identify swallowing difficulties, and periodic reassessment can identify changes in swallowing ability and suggest appropriate non-pharmacologic interventions such as a change in food consistency.

### How the Speech and Language Therapist can help

The Speech and Language Therapist (SLT) is involved at all stages of the disease. The SLT firstly assesses the swallow. There are a number of assessment options available. Frequently, the SLT will trial the person with a number of different food and liquid consistencies and observe for clinical signs of aspiration. Another assessment tool that is used regularly is known as video fluoroscopy. This procedure involves adding barium to various food and liquid consistencies. A moving x-ray is then taken while the person is eating and drinking. This helps determine if aspiration is occurring on a particular consistency. It can also be a useful tool in developing strategies for safe swallowing.

The SLT will then make a number of recommendations based on the assessment results for example introducing techniques to ensure safe swallowing or giving advice on optimal positioning.

It may be necessary to make food and / or liquid consistency changes. Dry, crumbly and chewy foods can be difficult to swallow safely so the SLT may recommend a softer smoother option. Similarly, drinks can be difficult to swallow safely as liquid moves very fast through the mouth and throat. The SLT may recommend thicker drinks which are slow moving. As there are many different grades of thickened drinks, the SLT will advise on the liquid consistency most suited to the swallow abilities. Changing food and liquid consistency can be an effective way of reducing the risk of aspiration. Each person is assessed on an individual basis and the SLT will therefore recommend a tailored plan.

Depending on the severity of the dysphagia, it may not be safe to take any food or drink orally. In these instances, the SLT may advise non-oral methods of feeding (e.g. a tube may be placed in the stomach to supplement what you eat). This decision is made in conjunction with the patient, family, dieticians and medical team.

Swallowing difficulties range from mild to severe. At all times, patient dignity and comfort are central to all decisions made regarding management.

*Thanks to Eimear Collins, Speech and Language Therapist, South Infirmary, Victoria University Hospital.*



*Congratulations to Denis and Ann who celebrated their 40th Wedding Anniversary by visiting their Granddaughter in the US.*

## TRIBUTE TO MEMBERS

We would like to pay tribute, in our newsletter, to members who bravely cope with HD. Do you have a family member or friend you want to pay a tribute to? Someone who copes with HD while getting on with their daily life? If so please let us know.

### THANK YOU

#### A NEIGHBOUR, A FRIEND AND A LOT MORE...

My partner was diagnosed with HD in 2002. I remember ringing my neighbour and saying "have you time for a chat?" I told her our devastating news.

Since then she has supported us unobtrusively in so many ways. Her delivery of homemade brown bread or scones lifts the day. She has also supported us by inviting us to dinner, so many times and has also given generous donations and support for HDAI. Also, offers to take the kids just when they were needed. Our annual trip to Cuisle would not be possible without Sandra Cannings help. I could go on.....

To know there is someone out there you can call on, in good and bad times, keeps us going.

Sandra is the best friend and neighbour we could ask for. She deserves many a bouquet of flowers.

**Thank you from Bernie & family**

*Sandra Canning will receive a bouquet of flowers from HDAI on behalf of Bernie & family.*

## BRAIN AWARENESS WEEK

**National Brain Awareness Week took place from March 10th to 16th, 2008. To mark the event the Neurological Alliance of Ireland held a series of events nationwide, calling for a major report into neurology services in Ireland to be published and implemented in full by the HSE. The national review of neurology services, chaired by Dr Marie Laffoy, was submitted to the National Hospitals in December 2007.**

In Dublin, a conference entitled "Neurological Conditions: The Health Care Challenge of the Next Decade" was held on 11th March in the Royal College of Surgeons to highlight the difficulties people with neurological conditions currently face in accessing services and how this is set to worsen as the numbers of people with these conditions increases as the Irish population ages.

### NAI's Recommendations include:

Easier access and shorter referral times to review by a Consultant. This can only be achieved by substantially increasing the number of neurologists, geriatricians and rehabilitation physicians in Ireland.

- Co-operation between hospital-based and community-based services with emphasis on the needs of the patient.
- Automatic entitlement to a GMS card based on disability and NOT on income.
- Communication of diagnoses by specialists (neurologists, geriatricians or other specialists where appropriate) and not by junior doctors.
- Early post-diagnosis follow-up appointment and communication between hospital-based service and general practitioners.
- The promotion of multidisciplinary teams in specialist centres for the management of complex Neurological conditions.
- Immediate entitlement and early access to relevant clinical professionals (occupational therapy, physiotherapy, speech and language therapy etc.) within the community.
- Clear management and follow-up protocols on discharge from hospital.

**Further information on Brain Awareness Week and NAI can be obtained from the NAI website, [www.nai.ie](http://www.nai.ie).**

## Fundraising Events

### Turning Trash to Cash for HDAI

HDAI continue to collect old mobile phones. If you have any old mobiles you no longer need we would be happy to receive them. HDAI have received €966 from C & C Cellular so far in 2008 from recycling old phones. Please send us any old mobiles you no longer need or call the office if you would like to organise a collection in your area.

### Sloan Rangers Line Dancing

'Thank You' to Phil and her friends who continue to donate to HDAI, through their regular line dancing classes.

### Eve's Hair Salon

Thank you to Eve's Hair Salon in Milltown, Tuam, Co Galway for raffling an Easter Egg and raising €70 for HDAI.

### Gael Scoil Colmcille

Gael Scoil Colmcille, in Santry designated HDAI as their chosen charity for their 2008 Jersey Day and raised a marvellous €484.50 for HDAI. Many thanks to Principal Sandra Ni Gharbhain and to staff, pupils and parents who kindly supported the day.

### Fundraising Ceili

Thanks to Atracta Lynch, Sean Broderick, Tony Whelan and Mary Whelan of Craobh Sean Treacy CCE for their generous support in organising a fundraising ceili for HDAI. Thanks also to an excellent fear an ti, the Brian Boru Ceili band, the Mary Brogan and Mary Murphy set dancing classes, to those who bought cakes and refreshments, and to people who donated spot prizes and gave donations. It was a great fun afternoon with enjoyable tunes and dancing. The event raised a very impressive €925 for HDAI. Special thanks to Sean Broderick who has offered to host an annual fundraising ceili for HDAI.

### Women's Mini Marathon

Thanks to all of you who are participating for HDAI.

## Future Events

### Head Shave

Jennifer is planning to say goodbye to her lovely silky locks if she raises €500 in sponsorship for HDAI. Jennifer has very bravely offered to have her head shaved at Cuisle. If you would like to sponsor Jennifer contact the HDAI office.



### Are you a Golf Enthusiast

Peter Grogan, a friend of HDAI, has very kindly offered to do a 50/50 golf tour, in aid of HDAI and the Marie Keating Foundation. Peter will play 50 golf clubs in 50 days starting on May 23rd and ending 50 days later on 11th July. Peter will play on courses throughout Ireland so if you are a regular player you may like to support Peter at a golf course near you. You could also offer support by sponsoring a prize for a raffle. Contact the office for more information

### Cork City Marathon

A group of five friends from Cork have bravely agreed to participate in the Cork City Marathon on June 2nd to raise funds for HDAI. We wish Michael, Andrew, Owen, Anna and Michael all the best with their training and preparation. If you are a Cork resident perhaps you can cheer them along on June 2nd.



*Anna (right) in Co. Mayo to represent HDAI and accept proceeds from the Balla Walk from Margaret.*