



## **NIEMANN-PICK DISEASE GROUP (UK)**

### **FAMILY NEWS BULLETIN**

*July 2009*

Hello and welcome to the latest Niemann-Pick Disease Group (UK) Family News Bulletin. In this month's edition:

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## The Niemann-Pick Disease Group (UK) Annual Family Conference



The NPDG (UK) Family Conference and AGM was this year held at the Holiday Inn, Ironbridge, Telford over the weekend of June 19th-21st. We welcomed several new families to the Conference and were re-united with many old friends; we were joined by delegates from France, the USA and Slovenia. We are very grateful to those families who shared their experiences of Niemann-Pick with us and of course to the professionals who gave up their time to share their knowledge and expertise. The Children's Programme was once again a great success with trips to Enginuity and the bowling alley, plus arts and crafts and fun in the swimming pool. On the Saturday evening the children, as well as a few adults, were entertained by the "Animal Man". Pictures from Conference 2009 can be seen by going to <http://www.facebook.com/album.php?aid=2021897&id=1541266703&l=f9b369fe0d>.

The Annual General Meeting of the NPDG (UK) took place at the Conference, with members choosing to re-elect the current serving Trustees, as well as new Trustee, Helen Carter. Roger Franklin and Caroline McDonald stepped down from the Board this year; the Group would like to thank them both for generously giving their support and time to the Charity.

Plans for Conference 2010 are already underway; we would welcome any suggestions as to how we can tailor the conference to meet your needs. So, if you attended this year and have yet to submit your feedback, or are thinking about attending and would like to see a particular speaker or subject covered, please email your comments to [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

### NPDG (UK) On Facebook



We now have a dedicated page on Facebook, so if you would like to add your support, comments, photos etc please go to [www.facebook.com/group.php?gid=125434951250](http://www.facebook.com/group.php?gid=125434951250).

### Biomarker Validation for Niemann-Pick Disease, Type C Study

A new therapeutic trial has been approved by the NICHD IRB (National Institute of Child Health and Human Development Institutional Review Board). The name of this study is Biomarker Validation for Niemann-Pick Disease, type C: Safety and Efficacy of N-Acetyl Cysteine. This study still has to be reviewed by the FDA (Food and Drug Administration), but the NIH (National Institutes of Health) are hoping to start enrolling patients this September. You can read further details on this study by visiting [www.niemannpick.org.uk/documents/NACtrialdescription.pdf](http://www.niemannpick.org.uk/documents/NACtrialdescription.pdf).

### NPDG (UK) Clinical Research Nurse – Liz Jacklin

Liz Jacklin, our Clinical Research Nurse, has provided the following update of her work for the Family News Bulletin. If you have any specific research questions that you would like to see addressed in future issues, please do forward them to us at the Central Office by email at [niemannpick@zetnet.co.uk](mailto:niemannpick@zetnet.co.uk), or to Liz direct [Elizabeth.Jacklin@cmft.nhs.uk](mailto:Elizabeth.Jacklin@cmft.nhs.uk).

Liz writes....

**Phase 2 trial of rhASM in Niemann-Pick B patients:** an announcement about the next stage of this study will be made at the ICIEM (International Congress of Inborn Errors of Metabolism) meeting in San-Diego in August. It is anticipated that there will be a UK centre for this phase of the study but as yet we do not know where. The study will be for patients over the age of 18 years only.

**NIH - Severity Scoring Study:** we continue to liaise closely with Dr. Denny Porter and his Research Nurse Nicole Yanjanin. The study has recruited 44 patients out of their initial target of 50; for this reason Nicole and Denny have decided to slow down on the recruitment of new patients in order to concentrate on longitudinal data, which will be collected over 5 years. However, they are still keen to enrol adult patients, as this is an area which they have low numbers for.

**Biomarker Validation for Niemann-Pick Disease, type C: Safety and efficacy of N-Acetyl Cysteine. ( NAC ):** an outline of this study has been posted on the Niemann-Pick website, so please see this for more information. We are currently looking at the feasibility of running the study here, as soon as we have some news on this we will let you know.

**CHORI-Study:** some outline information about this has recently been posted on the "Hope for Hollie" Forum. I am currently waiting to hear from the UK contact - a scientist at University College Hospital London to get more information about the study. I will pass this on as soon as possible.

**NP-C Registry:** a registry to capture information on all NPC patients around the world has now been developed. The aim of the registry is to describe the natural history/disease course and clinical outcomes of NPC. All centres around the world are being encouraged to participate. I am in the process of sending the necessary documentation through the Ethical review panels and learning about the on-line data forms. All patients and their families will receive more information about this through their local centres; it will then be necessary for them to give written consent before their information is entered. More information will be available shortly.

**Swine Flu:** I am sure you will be aware of the concern around swine flu. In most cases this is a mild and self limiting disease and we hope that you have not been unduly alarmed by the recent media publicity about this disease. Some individuals are at slightly increased risk of developing more significant illness than the general population should they catch the virus. These include pregnant women, children under the age of 5 years and patients with underlying diseases. For that reason, individuals with Niemann-Pick Diseases are considered to be of higher risk. There currently isn't a vaccine for swine flu but it is hoped that there will be one available later in the year. When it does become available it is recommended that those affected with NPD have the vaccine. If the individual affected with NPD comes into close or direct contact (usually meaning a household contact) with a case of probable or confirmed swine flu (diagnosed by a health professional) we would recommend that they receive a 10 day course of Tamiflu (as a preventative measure). If the individual with NPD develops symptoms of swine flu, Tamiflu is then prescribed for 5 days. The symptoms of swine flu are: high fever (over 38°C] or a history of fever, influenza-like illness (TWO OR MORE of the following symptoms: cough; sore throat; runny nose, limb or joint pain; headache; vomiting or diarrhoea) OR severe and/or life-threatening illness suggestive of an infectious process. Guidelines are constantly changing. To view the latest official information from the NHS regarding Swine Flu please visit [www.niemannpick.org.uk/documents/SwineFluInfoforWebsite.doc](http://www.niemannpick.org.uk/documents/SwineFluInfoforWebsite.doc).

## Parents' Tips / Ideas / Recipes



Following several requests from our Niemann-Pick families, we are looking to set up a page on our website for parents to pass on recipes, particularly ones that liquidise well, useful tips and ideas that may help other families living with Niemann-Pick Disease. So, if you have any ideas, tips or recipes, please do pass them on to [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

## ACT Family Companion

An invaluable booklet, "A Family Companion to the ACT Care Pathway for Children with life-limiting and life-threatening conditions" is now available free to families. You can order this direct from the ACT website or by calling **0117 916 6422** or emailing [Susannah@act.org.uk](mailto:Susannah@act.org.uk).

## Simple Rules on Backdating Extra Tax Credit Payments

If your child is awarded Disability Living Allowance (DLA) or has an existing DLA award increased to the highest rate, this can lead to extra child tax credit payments, known as the disability elements. Following recent lobbying, the rules on backdating these payments have been simplified. Prior to April 2009, any parent who wanted to make sure these disability elements were fully backdated had to notify tax credits both when they claimed DLA and when they got a DLA award. Many parents lost out because they were unaware of the need to tell tax credits that a claim was being made. However, since April 2009 the rules have been simplified. Now you only need to notify DLA once - when you get an award. If you are awarded DLA for your child (or have an existing award increased to the high rate for personal care) let tax credits know within three months of the DLA decision. So long as you do this, any extra tax credit payments will be backdated in line with the DLA award. You can access the new rules in full at [www.opsi.gov.uk/si/si2009/pdf/uksi\\_20090697\\_en.pdf](http://www.opsi.gov.uk/si/si2009/pdf/uksi_20090697_en.pdf).

## Increase in Family Fund Age Limits



Recent changes in the age limits mean that the Family Fund can now give grants for severely disabled children aged up to 18 in England, Northern Ireland, Scotland and Wales. To qualify, eligible families must have a gross income of no more than £23,000 pa and savings of £18,000 or less (figures current from 2008, reviewed periodically). The

fund can help families caring for a disabled child up to the age of 18 providing the application is received before the young person's 18th birthday. For information Tel: **01904 550037** or visit [www.familyfund.org.uk](http://www.familyfund.org.uk).

## Need Help to Pay for Essential Items?

Save the Children, in partnership with UK charity Family Action, is distributing cash crisis grants to around a thousand families who are struggling in the recession. They aim to deliver £150,000 to those families most in need. The money will be given to help people pay for basic essentials.

Help is for low income families with children up to 18 years old, including families with disabled children. It is particularly aimed at people living on benefits or who have just lost their jobs. Grants are available in England, Scotland and Wales. To find out how to apply for a crisis grant please visit Family Action at [www.family-action.org.uk](http://www.family-action.org.uk).

### **Budget Boost for Disabled Children and their Families**

The NPDG (UK) welcomes the significant investment announcement in the budget to provide additional financial support for disabled children and their families through the Child Trust Fund. The government will pay £100 (£200 for severely disabled children) per year into a disabled child's trust fund. Entitlement to these payments will be linked to receipt of Disability Living Allowance (DLA) and payments will start in April 2010.

### **New National Representative Body for Special Schools**

The Department for Education and Skills has provided £150,000 in start up funds to create a national representative body for special schools. The new body is called the "Federation of Leaders in Special Education" (FLSE). It aims to shape and deliver SEN policy to ensure every child's needs are met. For more information visit [www.flse.org.uk](http://www.flse.org.uk).

### **Special Educational Needs: A Guide for Parents and Carers**

A new edition of the Guide (for England) is now available free from the Department for Children, Schools and Families. To get your copy call **0845 6022260** or visit [www.tinyurl.com/qowlpg](http://www.tinyurl.com/qowlpg).

### **Walks With Wheelchairs**



For descriptions of leisure routes in Britain, with maps, that are accessible to children's vehicles visit [www.walkswithwheelchairs.com](http://www.walkswithwheelchairs.com) and [www.walkswithbuggies.com](http://www.walkswithbuggies.com). Other useful information can also be found on these sites, such as whether dogs are allowed on the routes and what facilities are available.

### **KIDS Direct Short Breaks Service**

KIDS, a national charity for disabled children and young people have received funding from the Department for Children School and Families (DCSF) to develop a new short breaks service. KIDS Direct Short Breaks will use internet and text messaging technology to link families with disabled children to short break workers in their local area who are trained and managed by KIDS. The service aims to put families in control of their short breaks so they can manage them to fit around family life and be personalised to the interests and aspirations of the child or young person (up to the age of 25). KIDS would like to hear from parents about how the service would

support their family and get an idea of the local demand. You can use their online form at <http://tinyurl.com/cc7300> to register your interest and they will then notify your local authority to provide them with information on Direct Short Breaks and how it could help your family. For more information call **08454531000** or visit [www.directshortbreaks.org.uk](http://www.directshortbreaks.org.uk).

### **Free Holidays for Children With Life Limiting Illnesses**



Camp Quality UK is a charity that takes children with a potential life limiting illness on holidays, giving them the chance to be themselves and have fun. The holiday programme is structured to cater for all abilities and disabilities and there is a medical team on hand. Every holiday is funded entirely by Camp Quality UK and is provided free of charge to each child. For more information visit [www.campquality.org.uk](http://www.campquality.org.uk).

### **Online Advice about Equipment for Disabled Children**



Disability charity, the Disabled Living Foundation has launched an advice and information website on equipment for children and young people with disabilities. The site, Living Made Easy for Children, has been created by occupational therapists and aims to bring together information on every product for children and young people with disabilities into one place. The site includes a star rating system which enables users to review and discuss products with other parents. Users can also ask specific advice from occupational therapists. Their website is [www.livingmadeeasy.org.uk/children](http://www.livingmadeeasy.org.uk/children).

### **New Website for Every Child Matters**

The Department for Children, Schools and Families has re-developed the Every Child Matters (EMC) website. The new site incorporates three former sites: Every Child Matters, Local Authorities and Sure Start.

It is aimed to be a single channel for information aimed at professionals within the children's workforce, including information on Aiming High for Disabled Children, special educational needs and disability. You can visit their website at [www.dcsf.gov.uk/everychildmatters](http://www.dcsf.gov.uk/everychildmatters).

### **My Future Choices**

My Future Choices is the magazine of the Transition Information Network, with articles designed to help young people who will continue to need some support as they go into adulthood, to grow in independence. To receive this magazine a copy of this magazine call **020 7843 6006**. The magazine can also be downloaded by going to [www.transitioninfonetwork.org.uk](http://www.transitioninfonetwork.org.uk).

### **New Guide to Help Healthcare Professionals "Get it Right"**

Prominent health organisations, including the Royal College of Nursing and the Royal Society of

Medicine, have joined forces with Mencap to produce a best practice guide for health professionals. "Getting it Right" aims to stop discrimination and focuses on improving healthcare for people with a learning disability in the NHS. It is accompanied by an interactive website. To view the report go to [www.mencap.org.uk/displaypagedoc.asp?id=6770](http://www.mencap.org.uk/displaypagedoc.asp?id=6770).

## **Free to a Good Home**

We have been kindly offered the following items, which are in good condition but no longer needed, free to a good home: lift up hand rails for a toilet; one collapsible shower chair (ideal for holidays); a Telford chemical toilet which fits under a chair; a hydraulic bath chair; an AEG wheel-chair motor. If you are interested in any of these items, please contact the Central Office on **0191 415 0693** or by email at [niemann-pick@zetnet.co.uk](mailto:niemann-pick@zetnet.co.uk).

## **NPDG (UK) 2009 Christmas Card Competition**

We are pleased to announce the winners of our 2009 Christmas Card Competition, judged by attendees at the Annual Family Conference. All the pictures received were wonderful, with special thanks going to our joint winners:- Alice Byrne, Ellen Byrne and Annie Pyne. Full details as to how you can order these cards will be sent out with the Autumn edition of Niemann-Pick News, and an order form will also be available to download from our website in September.

## **Fundraising**



### ***Niemann-Pick Fundraising Items***

At Conference 2009, the NPDG (UK) launched their new shopping bags. These canvas bags measure 16 x 15 inches and feature the Group's logo on the front. They are available from the Central Office for a suggested donation of £3.00 which includes postage and packing. We also introduced a limited edition 10 inch Niemann-Pick Teddy Bear which proved very popular with delegates at Conference. We only have a few of the bears left which are available for a suggested donation of £10 which includes postage and packing. If you are interested in either a bag or a bear, please contact us here at the office on **0191 4150693** or email [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

## **General Fundraising News**

### ***The 'Hope for Hollie' Hollie-wood Ball 2009***

On the 16 May 2009 the "Hope for Hollie" campaign hosted its second Annual Charity Ball at the Stadium MK in Milton Keynes and, despite being up against the credit crunch, the organisers pulled off another amazing event with over 300 people attending and helping to raise over £13,000 for the NPDG (UK). Guests included NPC families, who had travelled a distance to show their support, Milton Keynes' local businesses, and a number of the MK Dons players who continue to give their full backing to the Hope for Hollie campaign and the NPDG (UK).

Guests were entertained by a superb charity auction hosted by the Littler Britain tribute act followed by a disco and finally, to close the event, a live performance by X Factor finalist Laura White, who travelled from another gig in Blackpool that night to show her support.

The "Hope for Hollie" 2009 campaign DVD was also launched on the night and had a real impact on everyone in the room, not only helping to raise awareness of Niemann Pick Disease but also helping to increase the sums of money donated on the night via the auction, raffle, casino and other fundraising opportunities. You can view this DVD online at [www.hopeforhollie.co.uk](http://www.hopeforhollie.co.uk).

### ***Hope for Hannah Charity Golf Day and Evening Masquerade Ball***

The Hope for Hannah campaign has organised a Charity Golf Day and Masquerade Ball on Monday, 31st August at Glossop Golf Club. If you are interested in attending either the golf day or Ball, please contact **01457863207** (day time) or **01457 762621** (evenings).

### ***General Fundraising Events***

During the months of June and July 2009, the NPDG (UK) received over £27,700 from donations, collection boxes and fundraising events organised by families and friends and we would like to thank all those involved.

Recent fundraising events were organised by The Elliott Family, The Jolly Potter's Football Club, Good News Pre-School, Hallaton Church of England Primary School, Jennifer Powell, Samuel Laycock School, Shani Miller Darlington, Pinner Bowling Club, Jon Tannock at Network Rail, Val and Rose and all residents of Astral House; the Group also raised over £680 at the Family Conference from the sale of raffle tickets, bags, books, toys and the usual tombola. Fundraising events were held in conjunction with the Hope for Hollie Campaign by Hachette Children's Books, Arizan Hair MK, Trailer Resources and, of course, the Hollie-Wood Ball which has already been mentioned in this e-bulletin. Two successful golf days were held, one by Carl and Emma Burdon for Campaign for Calum and the other by Craig Mathieson and friends. Several fundraising events were held by The 'Georgina's Gang' Appeal, set up to raise funds and awareness of NPD in Guernsey where Georgina Pearce, NPC aged 13, lives with her parents Tania and Ian and her sister Jessica.

General donations were received from Sarah Beaumont, Astral House, DJB Haulage, The Hospital Saturday Fund, Mr Vyas, Paul Lingard and Helen Brook, Mrs Ramachandron and Mrs Forrester.

The Barge Inn, Billy and Denise, Café des Fleurs, Loughton Sports and Social Club, Roy Green, Andy Cudlip, Martin and Gemma Owen, Mr Ross, Peter and Myra Scott and Wendy Wynne all donated the contents of their NPDG (UK) collection boxes.

We have also received generous donations "in loving memory" of Mr Walter Hurst and Mr Reevel Tye.

We hope you have enjoyed reading this edition of the Family News Bulletin. If you have any articles, suggestions or comments for future editions of the Bulletin, please send them to **Sue Lowe, 11 Greenwood Close, The Pastures, Fatfield, Washington NE38 8LR** or email [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

If you have received this Bulletin through the post and would now like to receive it by email please let Sue know at the above address or email her at [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

**Disclaimer: Information which appears in this e-bulletin is for the express purpose of raising awareness and does not necessarily reflect the views of the NPDG (UK). All medical information should be reviewed with your doctor before being acted upon.**