



NIEMANN-PICK DISEASE GROUP (UK)

FAMILY NEWS BULLETIN

NOVEMBER 2008

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

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Niemann-Pick Christmas Cards



We would like to thank all those who have purchased NPDG (UK) Christmas Cards this year, so far a total of almost £600 has been raised. There are still a few packs of Niemann-Pick Christmas cards available and these can be ordered by calling **0191 4150693** or by downloading a form from our website **www.niemannpick.org**

NPDG (UK) Family Directory 2009

We will shortly be in the process of updating the Family Directory, as you know, this Directory is confidential and is only sent out to those families who agree to share their details with other families. Would you please check your entry in the 2008 Directory and let us know if there are any changes or additions, such as email address, that need to be made. Please also let us know if you no longer wish your details to be included in the Family Directory. Details of any changes should be emailed to **sue.npdg@tiscali.co.uk** or posted to Sue at The Central Office address, (which can be found at the end of this bulletin) by 31st December 2008.

Family Conference 2009—Would You Like to be a “Family Voice”?



The 16th NPDG (UK) Annual Family Conference 2009 will be held over the weekend of 19th-21st June at The Holiday Inn Hotel in Telford. Further details and a booking form will be available on our website **www.niemannpick.org.uk** in due course. Each year the Conference has a special section called “Family Voices” which gives the chance for families to share their experiences of living with Niemann-Pick disease. If you would like to share your story and experience in the Family Voice session, please contact The Central Office on **0191 415 0693** or email **niemann-pick@zetnet.com**

NPDG (UK) Clinical Research Nurse – Liz Jacklin

Liz Jacklin, our Clinical Research Nurse, has kindly agreed to provide a regular 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 **niemann-pick@zetnet.co.uk**, or to Liz direct **Elizabeth.Jacklin@CMMC.nhs.uk**.

Liz writes:

What's happening with Research?

Well, although it might not feel like it for you as patients and families living with Niemann-Pick disease every day, there is quite a lot going on.

As reported in our latest newsletter, we are in the process of putting together all the paperwork for the NPC Natural History Study which we will be running in conjunction with the NIH. A number

of our families have decided to travel to Maryland to take part in the study over in the USA, they are being helped financially to do this by the "Hope For Hollie campaign" please see details of the Hope For Hollie Travel Fund included in this newsletter.

We are continuing to look at the various aspects of NP Type C disease. Currently we are studying at seizure activity and also the incidence of Crohn's Disease in Niemann-Pick Type C patients. I have been accepted to present the data from this work at the WORLD (We're Organizing Research on Lysosomal Diseases) Symposium in San Diego, next year. Hopefully this will open up discussions with some of the Worlds leading scientists' and clinicians who will be attending this meeting, and this will lead to further Niemann-Pick Research.

The Audit for ASMD Niemann-Pick patients (NPA and NPB patients) is now underway. This involves collecting data on the Natural History of the disease using both current and historical information. As soon as we have any data, we will share this with you.

*This is a very brief overview of my work at present, if you have any questions about this or anything regarding Niemann-Pick research - please do not hesitate to contact me at **Elizabeth.Jacklin@CMMC.nhs.uk** or telephone **0161 922 29667**.*

Niemann-Pick Type A and B - Why Change the Name?

Over the last decade it has become apparent that Niemann-Pick Types A & B, rather than being two separate 'types' of the disease, actually represent the opposite ends of a spectrum of the same disease, both caused by a deficiency of the enzyme Acid Sphingomyelinase (ASM). Many variations exist within this spectrum, in terms of clinical symptoms and rate of progression.

In the USA, scientists are beginning to refer to NP Types A & B under the name "Acid Sphingomyelinase Deficiency disease (ASMD)" rather than "Niemann-Pick disease" as this more accurately describes the condition. This is not in fact a new term of reference - Niemann-Pick diseases Types A and B were generally known as Acid Sphingomyelinase Deficiency disease (ASMD) until the early 1960's.

Often Niemann-Pick Types A and B do not get the attention they deserve, as compared to the more widely known Niemann-Pick Type C. With this in mind, scientists worldwide would prefer to give Niemann-Pick Types A and B their correct name, to avoid confusion and to assist in raising the profile of this condition.

Patients at the severe end of the ASMD spectrum, Niemann-Pick Type A, have a very rapidly progressive neurodegenerative disease usually resulting in death before the age of 4. In contrast, patients at the opposite end of the spectrum, Niemann-Pick Type B, have a more variable range of severity and symptom involvement. This condition is usually diagnosed in childhood, due to the presence of an enlarged liver and/or spleen. The majority of NPB patients survive well into adulthood with little or no neurological involvement, but respiratory problems can cause disease complications.

The NPDG (UK)'s Clinical Nurse Specialist, Jackie Imrie, is currently working with Dr Ed Schuchman, Professor and Vice Chairman for Research at Mount Sinai School of Medicine, to produce an information booklet about ASMD Niemann-Pick Disease. The booklet will be entitled "Acid Sphingomyelinase Deficiency Niemann-Pick Disease and its potential treatment." If you would like to contribute to the booklet, perhaps by providing a story about how the disease has

affected you or your family, or if you would like to know more about the booklet, please do contact Jackie on **0161 922 2414** or email Jackie.imrie@ccmc.nhs.uk.

Free Epilepsy Alarms

The Muir Maxwell Trust is offering vouchers for a free epilepsy alarm to families of children with severe epilepsy. Parents can apply directly to the Muir Maxwell Trust for an epilepsy alarm, or go through their health visitor, community nurse, epilepsy nurse, GP or consultant. They will receive a voucher from the Trust, which requires countersigning by a medical professional who can confirm the diagnosis of epilepsy. The voucher is then sent directly to the manufacturer and the alarm will be delivered within a week. To request your epilepsy alarm voucher, email their Family Liaison Manager, Ann-Marie Nelson at info@muirmaxwelltrust.com with the following information: your child's name and date of birth; your name, full postal address and contact telephone number and a short description of how your child's seizures manifest themselves, alternatively you can call the Trust Office direct on **0131 273 5256**.

The Trust may also be able to help with other items of equipment needed by parents for the home, these range from electronic beds through to large car seats, sensory equipment or special sunglasses. For further details go to www.muirmaxwelltrust.com.

Caudwell Children Helps Make Ben's Dream Come True



Caudwell Children, a charity which makes dreams come true for very special children throughout the UK, will be sending 8 year old Ben Scott, who has NP type C, and his family to Disney World, Florida to meet Mickey Mouse, just before Christmas. We understand from Ben's mum, Lyndsay, that the Charity still has funds available to provide specialist equipment, days out, holidays etc. If you would like further information visit their website at www.caudwellchildren.com

Hope For Hollie—Travel Fund



At a recent meeting of the NPDG (UK) Trustees and Helen and Pete Carter, founders of the Hope For Hollie Campaign, it was agreed that some of the money raised through the "Hope For Hollie" Campaign be used to set up the "Hope For Hollie Travel Fund". This Fund will cover the travelling costs to the USA for one adult and one child to enable them to take part in the NIH observational study into NPC, or will assist families in accessing any future research studies in the US. To apply to the Fund, or for further information, please contact Toni Mathieson at the Central Office on **0191 415 0693**.

Say It With Flowers

The NPDG (UK) has recently teamed up with a company called Charity Flowers Direct based in Guernsey. Every time you order flowers through Charity Flowers Direct 15% of the total price will

be donated to the NPDG (UK). So whether it's Happy Birthday, Happy Anniversary, Happy Christmas, Happy Easter, Congratulations, a present for Mother's Day or just to show you care, order from Charity Flowers Direct. Their website is www.charityflowers.co.uk; their Order Hot-line is **08705300600** and we are listed as **The Niemann-Pick Disease Group (UK)** and the code to quote **TNPD**.

The London Marathon



Following numerous suggestions from families and friends wishing to compete in the London Marathon on behalf of the NPDG (UK), but unable to gain a place in their own right, we are in the process of trying to secure a Silver Bond Place which would guarantee ONE place every FIVE years in the London Marathon starting in 2011.

Whickham Villa Health Club, Chase Park Rehabilitation Centre

Chase Park Rehabilitation Centre, based in Whickham, Tyne and Wear is a centre which supports adults with neurological conditions and other complex needs. The Centre's aim is to empower people with disabilities to fulfil their potential by providing high quality, physical and social rehabilitation with holistic care to promote independence. Within the facility there is an award winning health club that has been designed specifically as a truly accessible space for people of all ages and abilities within an extremely high quality and relaxing inclusive environment. Facilities include an Internet Café, gym, flotation tank and hydro pool; as well as a therapy room offering a wide range of beauty treatments and aromatherapy massages. If you would like to find out more, visit their website www.whickhamvilla.co.uk or call **0191 488 7351**.

Direct Payments



Direct payments are a way of getting services that allows you more choice and control over how your disabled child's needs are met. However, many parents are confused about what direct payments are and how they work in practice. If your local council agrees that your child needs care services, you can choose to get money to buy these services yourself, this is called a direct payment. You can use a direct payment to buy services from a private care agency, voluntary organisation or employ someone yourself. You can ask for a direct payment if you are already getting help from your local Department of Children's Services because your child has a disability. If you are not currently receiving help, you will first need to ask for an assessment of your child's needs. Contact a Family produce a free detailed guide, "Getting Direct Payments for your Disabled Child". To get your copy, call their helpline on **0808 808 3555**.

ContactPoint—A new tool for children’s services

ContactPoint is a new online directory to help practitioners find out who else is working with the same child. This system is being introduced across England and aims to help children’s services to work together more effectively to support the well-being of a child. This will help to reduce the number of repeat assessments and referrals which can be distressing for a child and their family. By law, ContactPoint will only hold demographic information about a child, contact details for parents/carers and for service providers across health and social care, education and justice. ContactPoint will not hold any case information, medical notes or school records and strict security and access controls will be in place. All ContactPoint users must have a current CRB and have completed mandatory training. For further information, visit the Every Child Matters website at www.everychildmatters.gov.uk/deliveringservices/contactpoint.

Clinical Genetic Services—Measuring Outcomes

Do you live in the North West of England? Would you like to help improve services for families with genetic disease or conditions? If so, Marion McAllister, a Genetic Counsellor and MRC Research Fellow, is carrying out research to develop ways of measuring how people with genetic disease or conditions may benefit from seeing a geneticist/genetic counsellor or attending a genetics clinic. As part of this work, Marion is developing a questionnaire that will examine patients’ thoughts, feelings and attitudes about the family disease or conditions. She is looking for people from families with genetic diseases or conditions to help her with this. It would mean attending a meeting that will last for up to two hours to discuss the wording of the questionnaire, and to make sure that it addresses the issues that are really important to people from families with a genetic disease or condition. If you would like to know more, contact Marion on **0161 2758979** or by email at marion.mcallister@manchester.ac.uk.

Disabled Children Should Have the Same Opportunities for Play and Fun



The NPDG (UK) welcomes “Going Places”, a report by the campaign Every Disabled Child Matters launched by Schools Secretary, Ed Balls. This report highlights the barriers faced by disabled youngsters when accessing fun and activities, including negative attitudes from other children, lack of access to play equipment and lack of training in disability equality for staff in council play schemes and leisure facilities.

Every Disabled Child Matters is calling on the government to deliver on promises made in its Play and Youth strategies and to ensure disabled children are given the same opportunities for play and leisure as their non-disabled peers. For more information about the campaign visit www.edcm.org.uk/Page.asp.

Free Courses to Help You Care With Confidence

Caring with Confidence, part of the government’s ‘New Deal for Carers’ aims to help carers (including parent carers) make a positive difference to their life and that of the person they care for. You will be able to mix and match how you develop your knowledge and skills, by taking part

in one or more free, local group sessions, by using self learning packs or by accessing online resources. The first face-to-face sessions are already taking place in Sunderland and Leicester. Sessions will then be rolled out across England from January 2009 over the next three years. Self-study packs and online resources will be available from January 2009. For further information visit www.caringwithconfidence.net.

Help Write a New Handbook

The authors of a new “Special Needs Handbook” are calling for contributions from parents and those working with disabled children. The handbook is intended as an information-sharing, daily problem-solving guide for all who work with, or care for, children with disabilities. The authors are looking for tried and tested tips on what works from as many different sources as possible—parents, teachers, occupational therapists, speech and language therapists and educational psychologists. When completed, the handbook will be available free of charge as a download on the internet and people will be able to print off and copy any material to use either personally, or to give freely to others, including support groups, schools and therapists. If you would like to contribute contact Helen or Deborah on **020 71832277**. You can also email at helen@specialneedshandbook.com or Deborah@specialneedshandbook.com.

Has Support for Parents Improved Over the Last Ten Years?

The Handsel Trust has launched a large-scale on-line survey to explore how the support for parents of children with disabilities has changed over the past decade. Ten years ago, the first SOFTY (Support Over the First Two Years) survey asked 500 parents about their experiences and the results were used widely to form national policy and local service development.

The Handsel Trust want to reach the parents of children with any disability or special need, as long as it was diagnosed when the child was aged under 6 years of age. If you are a parent and would like to take part in this survey, which is anonymous and will only take a few minutes to complete, then go to www.handseltrust.org. The results of the first SOFTY survey can also be downloaded from the site.

Fundraising



Lots of fundraising events have been held since our last Family News Bulletin.

A fantastic total of over £12,200.00 has been raised in the following ways

- Luke Peacock and friends participated in a sponsored bike ride
- The Marriott Hall Hotel Men’s Golf Team made the NPDG (UK) their chosen charity for the year and raised funds by holding tournaments, quiz nights and race meetings
- David Hirst competed in the London Triathlon

- UIA in Stevenage held a Dress Down Day
- The Garden Club, Radcliffe on Sea held several fundraising events
- Residents of Astral and Solar House in Sunderland took part in many fundraising activities, including a hamper raffle
- Inez Wedgbrow ran the Market Bosworth Sprint Triathlon
- A Mount Snowdon Climb was organised by Dave Williams
- Doxford Park Community Centre held a hamper raffle
- Sarah Coll competed in the Great North Run
- 1st Great Bowden Brownies held an evening in honour of Lois Peacock
- Kait Pyne organised a Bonfire Party
- Mrs G Clark held a coffee morning
- The Staff of OSP in Market Harborough held a raffle
- Abbey National held a Dress Down Day (this was match funded)
- Pats Pilates in Milton Keynes held a Charity Day
- The “Campaign for Calum” held a silent auction for Manchester United V Celtic tickets which was won by KMP in Scotland



General donations/contents of collection boxes have been received from Mr and Mrs Akooji, Daniel Broom of the Puffing Billy Restaurant in Exeter, JM and E Johnson of Firecroft Engineering in Cheshire, Mrs O’Neil, Myra Scott, Nick Swift. Jacqueline Colquhoun, Christine Scott, ANP Stores, The Barge Inn, Denise and Billy in Sunderland, One-Six-One bar in Cheshire, John Dyer and the Owens.

We have also received donations in loving memory of Beryl Woore, Sarah and Katie Forrester, Barb Webb and Bel Stallard.

The Niemann-Pick Disease Group (UK) would like to thank all of our families and friends who have worked so hard to raise money for the Group. Remember, it doesn’t matter how small or large the donation, every penny helps the NPDG (UK) to make a positive difference to the lives of those affected by Niemann-Pick disease.

If you would like information about holding a fundraising event, please contact Sue or Toni at the Central Office on **0191 4150693** or email **niemann-pick@zetnet.co.uk**

We hope you enjoyed reading this edition of the Family News Bulletin. If you have any articles, suggestions, announcements or comments for future issues of the Family News Bulletin which you would like included, please send them to **Sue Lowe, 11 Greenwood Close, The Pastures, Fatfield, Washington NE38 8PD** or by email to **sue.npdg@tiscali.co.uk**.