



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

### **FAMILY NEWS BULLETIN**

**FEBRUARY 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) Website**

The NPDG (UK) website has recently been updated and now includes up-to-date research information, new fund raising stories and much more. If you would like to contribute to the site in any way please do get in touch via email [niemann-pick@zetnet.co.uk](mailto:niemann-pick@zetnet.co.uk), or you could write to us at the Central Office (address at the end of this bulletin). We would also welcome your feedback on both the website and this news bulletin. You can view the website at [www.niemannpick.org.uk](http://www.niemannpick.org.uk).

## **Niemann-Pick News**

If you would like your child/children's school head teacher, school nurse, doctor, or any one who works with your child/children to receive a copy of the Niemann-Pick News either by post or email (next one due to be published in April) then please let us know by contacting us by phone **0191 415 0693**, email [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk) or by post to the address at the end of this bulletin.

## **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](mailto:niemann-pick@zetnet.co.uk), or to Liz direct [Elizabeth.Jacklin@cmft.nhs.uk](mailto:Elizabeth.Jacklin@cmft.nhs.uk) (please note new email address).

Liz writes:

*Well it certainly feels like we have hit 2009 running! There is a lot going on in the world of Niemann-Pick research. I am presenting at the WORLD (We're organizing research on Lysosomal Diseases) Symposium in San Diego in mid-February. The work I am presenting is entitled "Crohn's Disease and NPC - is there a correlation?" The abstract has been published in the journal Molecular Genetics and Metabolism — a link to this abstract will appear on our website very soon. It is very exciting to present at meetings such as this, not only because it is a great honour, but it is also a great way of getting to meet eminent physicians and scientists from all around the world and to find out about the work that they are doing. I will be writing a full report about my trip to WORLD for the next issue of Niemann-Pick News.*

*An overview of the hot topics at the moment has recently been added to our website, so if you have not had a look recently then it is worth a visit - [www.niemannpick.org.uk](http://www.niemannpick.org.uk). If you do not have access to the internet, or if you find it difficult to use, then please do not hesitate to contact me and I will post the information out to you.*

*Chaperone Therapy is one area which is causing much excitement at the moment. This work is being carried out by Dr Dan Ory, from Washington University School of Medicine in the USA. It is believed that to overcome the genetic defect in Niemann-Pick Disease, chemical compounds could potentially "chaperone" mutant protein molecules through the cell's quality control mechanism. The first step, which is to screen for suitable compounds, can be done quite quickly. The compounds then have to be developed by pharmaceutical companies, tested on animals, and then healthy volunteers. Clinical Trials have to be devised and patients recruited. All of this is a*

*lengthy process, however, work is well underway and there is sure to be much more said about this in the coming months - as always I will keep you posted.*

*If there is anything you would like further clarification on, or if there is something in the research field that you have heard about, then please do get in touch, I am always very keen to hear your views and to receive your feedback. You can email me at **Elizabeth.Jacklin@cmft.nhs.uk**.*

## **Calling All Families Affected by ASMD Niemann-Pick Disease Type A and B**



Coming Soon – A New Booklet “Acid Sphingomyelinase Deficiency Niemann-Pick Disease and its potential treatment” – Can you help? As reported in a previous Bulletin, 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 Type A and B. The booklet will be entitled “Acid Sphingomyelinase Deficiency Niemann-Pick Disease and its potential treatment.” Work is now well underway and Jackie would like to ask for your help, for whilst it is very important to include up to date scientific and research information, it is equally important to include the real life experiences of families and how this disease has affected them or their child. Therefore, Jackie would be grateful if you would consider contributing to the booklet—your input would very much be appreciated. Perhaps you could tell us how your life/that of your child’s has been affected, for instance; has a large abdomen had an impact physically or psychologically? Is life adversely affected by bone pain? Is bleeding and bruising a problem? Do you find the slowness of research frustrating?

Please send your contributions to Jackie as soon as possible, or if you have any questions or would like to know more, contact Jackie on 0161 922 2414 or email **Jackie.imrie@cmft.nhs.uk**

## **Contact a Family Produces a New Podcast on Special Educational Needs**

A new podcast from Contact a Family has been produced to help explain the meaning of Special Educational Needs (SEN) and other related terms like SENCO (Special Educational Needs Co-ordinator). The podcast also has more information about what to do if you are concerned your child has SEN, what schools and the local authority can do to help, how to request a 'statutory assessment' and the 'statementing' process. To listen to the podcast visit **[www.cafamily.org/sen\\_england.mp3](http://www.cafamily.org/sen_england.mp3)**.



### **Health and Well Being Questionnaire**

The British Market Research Bureau will be sending out thousands of 'Health and Well Being' questionnaires to families on behalf of the Department for Children, Schools and Families (DCSF). Information from the questionnaire will be used to find families to take part in a further survey about disabled children's services. Parents' answers will be used to develop a National Indicator for disabled children, which will measure the performance of each local authority's disabled children's services. Families chosen to complete the survey will be spread across each local area and have children with a range of disabilities. This is the first time there has been a specific indicator for disabled children's services. Local areas will

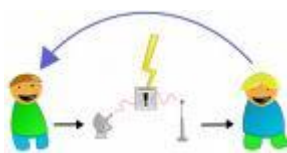
be able see how they are performing and compare with others. Areas that perform badly will be expected to improve their services and are likely to channel money and resources into meeting National Indicators where they are underperforming. The Disabled Children's Services National Indicator is a key tool to ensure services improve in your local area. It will focus on education, health and social care's attention on services for disabled children in a way that hasn't been seen before. The survey will initially be sent to some parents in the Spring and then to a larger sample in the Summer. After that it will be sent out on a regular basis once a year.

### **New Measures to Improve Services for Disabled Children**

The Government has announced £38 million funding to improve outcomes for children with Special Educational Needs (SEN). The funding was announced as part of The Children's Plan One Year On, a review of what has been done since its launch. There will be £31 million allocated to support pilot programmes to improve outcomes for pupils with SEN. There will be a further £7 million for other measures including strengthening school leadership and improving online reporting for parents.

For more information about the Children's Plan One Year On: a progress report, visit [www.dcsf.gov.uk/oneyearon](http://www.dcsf.gov.uk/oneyearon).

### **Action for Children with Speech, Language and Communication Needs**



The Government have published an action plan for children with speech, language and communication needs (SLCN). The action plan follows on from The Bercow Review, a recent review of services for children with SLCN by John Bercow MP. The government has announced it would invest up to £12 million to take forward its recommendations. The action plan, called *'Better communication: an action plan to improve services for children and young people with speech, language and communication needs'* has a range of initiatives to improve services for children and young people with SLCN and includes: up to 20 local area pathfinders to develop good practice guidance and a supplemental joint commissioning framework on SLCN; up to £1.5 million invested in grants to the alternative and augmentative communication sector; up to £1.5 million invested in a research programme looking at SLCN over the next three years. The Government will form a Communication Council and appoint a Communication Champion early in 2009, to give ongoing advice and support on developing effective services for children with SLCN. There will also be a National Year of Speech, Language and Communication in 2011-2012.

For more information visit [www.dcsf.gov.uk/bercowreview](http://www.dcsf.gov.uk/bercowreview).

### **Charity Launch Song**



Include Me Too, a charity supporting disabled children and young people and their families, have launched a song to raise awareness of the rights of disabled children. The song was created in April 2005 when disabled children and young people got together to raise awareness of the National Charter of Rights for Disabled Children and Young People, and now the song (Include Me Too) has been officially released. The video of the song can be viewed on the Children's Commissioner's website at

<http://tinyurl.com/59raby>. A CD of the song is available at £2.99 and includes three versions of the song, the new 2008 version, the original 2005 version and an instrumental. A video CD includes a signed version of the song and is available at £4.99. The song can be bought directly from the Charity, Include Me Too, and all proceeds will go to ensuring the rights of disabled children are supported and promoted. The Charity hopes the song will help promote the rights of disabled children and reach everyone, not just those who are aware of disability issues.

For more information on Include Me Too call **01902 711604** or visit **[www.includemetoo.org.uk](http://www.includemetoo.org.uk)**.

## **Physiotherapy for Children with Neurological Conditions**

Children with conditions such as cerebral palsy, cancer, stroke, brain damage and other neurological conditions could be taking their first steps with the help of an Oxfordshire local charity Footsteps Foundation. Private therapy is difficult for most families to afford, so this Charity has been launched to help children by paying for physiotherapy at the Footsteps Centre, which provides intensive physiotherapy for disabled children with neurological conditions. Families from all over the UK can attend daily physiotherapy for three week programmes at a time. Equipment at the centre includes the Spider, which enables therapists to implement a full course of exercises in any chosen position and to align the body by using a series of elastic ropes. The charity aims to raise £500,000 and needs the support of local individuals, clubs, groups and businesses to help even more children learn to walk. Visit **[www.footstepsfoundation.com](http://www.footstepsfoundation.com)**, contact via e-mail: **[pip@footstepsfoundation.com](mailto:pip@footstepsfoundation.com)** or call **01865 858 382** for more information.



## **Jewish Health**

A new project - Jewish Health - is being carried out at the DIPEX Health Experiences Research Unit based at the University of Oxford. They are looking to interview Jewish people who have direct or indirect experience of health issues that are often related to their ethnic background. This includes certain cancers, conditions like Tay Sachs disease, Gaucher Disease, cystic fibrosis, Canavans Disease, Crohn's Disease, Niemann-Pick Disease, as well as broader experiences of being a carrier for genetic conditions. While these health issues are not uniquely Jewish, the focus of this project is the way in which they are experienced within a Jewish context. This study will explore how people manage health and illness as well as issues surrounding death and burial practices. The Jewish population in Britain incorporates a range of people with varying commitment to Jewish practices, observances, culture and identity from the ultra Orthodox community to people who define themselves as atheist. DIPEX would like to interview people across this spectrum to produce a comprehensive picture of personal experiences of health issues among Jewish people. Funding is also being sought to interview others who experience these health issues to get a fuller picture of people faced with any of these diseases. The outcome of the project will be a Jewish Health section on the award winning website, **[www.healthtalkonline.org.uk](http://www.healthtalkonline.org.uk)** which is a unique award-winning website of people's experiences of health and illness. Recorded and filmed interviews, combined with qualitative research led by researchers at the University of Oxford, enable patients, friends, family and health care professionals to find out more from the experiences of others. Interviews carried out by researchers in people's own homes are presented on the website, alongside information about illnesses and treatments. These are linked to other websites and support groups, and a forum for sharing experiences and thoughts. You can listen to or

watch videos of the interviews, read about people's experiences and find reliable information about conditions, treatment choices and support. With nearly 50 illnesses and conditions already covered, there are thousands of video interviews on the site, spread across the available topics, with many more in progress. If you would like to be involved in the project, or would like further details contact Sara Ryan on **01865 289374** or email [sara.ryan@dphpc.ox.ac.uk](mailto:sara.ryan@dphpc.ox.ac.uk).

### **Family Fund Extra Website Launched**

The Family Fund charity has launched a new online shopping site called Family Fund Extra. Any family with a disabled child, as well as disabled young people themselves, can join Family Fund Extra for free and receive discounts of up to 25% from leading online retailers, including Comet, Argos, BSM, Haven Holidays and Stone Computers. Other people can shop on line via the Extra site, which links to nearly 100 leading retailers including Tesco, Marks and Spencer, Amazon, Boots, HMV. Retailers will give Family Fund Extra commission on every purchase. Disabled children's families will save money directly and the Family Fund will receive new income to increase its support for families with severely disabled children. To register visit [www.familyfundextra.org.uk](http://www.familyfundextra.org.uk)



### **Aiming High Participation Fund**

Aiming High for Disabled Children is a three year project to transform services for disabled children and young people. A key aspect of this is increasing the amount of short break activities that are available which disabled children and young people can get involved in. The idea behind the fund is to enable disabled children to participate in activities that they would like to do, but where something is stopping them from joining in. Where a short break activity has been identified, but a barrier exists that is stopping a child joining in, an application can be made to the fund to have that barrier removed. You can apply for funding if your child is disabled, you know what activity they would like to do, you are clear what the barrier is that is stopping them from doing it, the activity will give you a break from caring and one of the people who works with your child supports your application. Applications to the fund can be made by downloading a simple application form from [www.sncwd.org.uk](http://www.sncwd.org.uk) or by contacting the Aim High Programme Manager, Karen Parry, on **0191 566 2190**.

### **Stay informed with The Project - The Children's Rare Disease Network**

The Project is creating an online community where children with rare diseases, their families, and the organisations that support them will have the opportunity to communicate, collaborate, become educated and tap existing resources, helping them with their daily challenges - The Children's Rare Disease Network. Children and their families need information, resources and a coherent voice to speak about issues common to children who suffer from a rare disease. The Network will seek to work collaboratively with other organisations and raise the profile of the problems faced by people with rare diseases so that more funding can be directed into this area of great need. The Network will work with skilled journalists to build and establish a central website with focused content about research, policy and other developments around rare disease initia-

tives for individuals and the organisations that support them. The Network will serve as an online home for people with rare diseases that will bring together families, organisations, physicians and researchers to exchange information and to help each other. For further information visit [www.theprojectcharity.org](http://www.theprojectcharity.org).

### What Makes My Family Stronger

Contact a Family want to hear from parents and other family members caring for a disabled child about their biggest priorities to strengthen their family practically, socially and emotionally. They have compiled an online survey to find out families' current experiences and to ask what they would wish for if more help and support were available. Claire Pimm, Director of Policy and Communications at Contact a Family, said: *"We know from our work with families with disabled children that they want to lead ordinary lives. Living without fear of debt and poverty, the opportunity to work, the chance to enjoy leisure activities as a family, getting an education to meet a child's needs and the importance of a good night's rest are essential to making any family stronger. The results from our What Makes My Family Stronger survey will form new research and will help us campaign for better lives for all families with disabled children. We will also use the findings to raise awareness of what life is like raising a disabled child."*

If you are a parent or other family member caring for a disabled child, please take the time to complete Contact a Family's What Makes My Family Stronger survey before Sunday, 15 March 2009. Visit [www.cafamily.org.uk/surveys/strongersurvey](http://www.cafamily.org.uk/surveys/strongersurvey).

### Second "Hope for Hollie" Charity Ball Arranged

After the amazing success of last year's ball, the Hope for Hollie campaign have arranged a "Hollie-Wood" Ball, again at the Ballroom at Stadium:MK. Tickets are priced at £65 per person or £600 per table of ten (larger tables are available on request. There is also an early bird discount of £10 off per person or £125 off per table of ten for all bookings made by **28th February**. The organisers will have a printable poster on their website soon, [www.hopeforhollie.co.uk](http://www.hopeforhollie.co.uk), together with a link to pay for your tickets via Paypal, and much more information on the night of entertainment in store. If you want to take advantage of the early bird offer, and get your tickets booked now, please send cheques made payable to "Hope for Hollie" to the booking office at 13 Peebles Place, Bletchley, Milton Keynes, MK3 7SX.

### Overhaul of the Blue Badge Scheme

An overhaul of the Blue Badge scheme has been announced by the Department for Transport. The Blue Badge scheme provides a range of parking concessions for people with severe mobility problems who have difficulty using public transport. The scheme operates throughout the UK. New proposals announced include extending Blue Badge entitlement to children under the age of three with specific medical conditions; people with the most severe mental impairments/extremely disruptive behavioural problems and specific individuals with temporary mobility problems lasting a minimum of one year. A new system of assessing eligibility for the Blue



Badge is also being developed with the aim to standardise assessments throughout the country. For advice on the current rules for Blue Badges ring the Contact a Family Helpline on **0808 808 3555** or visit the website **[www.dft.gov.uk/transportfor you/access/bluebadge](http://www.dft.gov.uk/transportfor you/access/bluebadge)**.

## Fundraising

Just to show how a little goes a long way towards raising funds for the NPDG(UK) - a raffle of various tins of unwanted Christmas sweets (unopened of course) was held at Val Ridley's community centre and raised over £50.



Other recent fundraising events include: a Charity Variety Night in Blackpool, organised by Carl and Emma Burdon, full details of the evening can be found on **<http://www.blackpoolgazette.co.uk/blackpoolnews/Stars-come-out-for-sick.4894053.jp>**; Carl Hitchens and Helen Roberts participation in the New York Marathon (you can read Carl's story on our website **<http://www.niemannpick.org.uk/articles/newyorkmarathon.html>**); Clyst Vale Community College held a non-uniform day; Mr and Mrs Sanderson organised a fundraising event at their local cricket club in Wakefield; Dereham Rapid Response Team raised funds in lieu of sending Christmas cards— these events raised just over £6,200.

We also received donations and the contents of collection boxes from Mrs Keogh, Mrs Pamela Thompson, Mrs Bromilow; Major Allan Duncan (Army Charity); Langport Motor Company; The Barge Inn and Gary Johnston —which totalled £515.31. Talking of collection boxes—we still have some yellow collection boxes and buckets in the office—if you would like one please contact Central Office at the usual address, email or telephone number.

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](mailto:sue.npdg@tiscali.co.uk)**.