



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

### *FAMILY NEWS BULLETIN*

*May 2010*

Hello and welcome to the Niemann-Pick Disease Group (UK) Family News Bulletin.

In this month's edition:

- **Save the Date! The NPDG (UK) Family Conference 2010**
- **NPDG (UK) Clinical Research Nurse—Liz Jacklin**
- **2011 London Marathon**
- **Baby Congratulations**
- **RADAR—The Disability Network, There to Help**
- **Cash Counts—New On-Line Benefits Calculator**
- **Family Fund—More Money—Get Applying!**
- **Study For Life-Limited Children and Their Families**
- **Extra Support For Families With Disabled Children Outlined in Green Paper**
- **Parent Participation Works! Find Out How**
- **Families With Disabled Children Lack Mental Health Support**
- **New Local Powers to Investigate Complaints About Special Educational Needs (SEN)**
- **Fair Access to Care Services (FACS)**
- **Training For Families From Early Support**
- **Care For The Family**
- **WellChild Awards 2010**
- **New Projects for Young People**
- **Travel Insurance**
- **Fundraising/Donations**



## Save The Date! The NPDG (UK) Family Conference 2010



This year, the NPDG (UK) Annual Family Conference is moving from our usual spot in June to the weekend of 10-12th September. The venue of the Conference will be The Robinson Executive Centre, Wyboston Lakes on the Bedfordshire/Cambridgeshire border. Further details will be available by the end of May.

### 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:

#### ***Biomarker Validation for Niemann-Pick Disease, Type C—Safety and Efficacy of N-Acetyl Cysteine (NAC)***

*A number of families have shown interest in the above pilot study, currently underway at the National Institutes of Health, in Maryland, USA. I have recently heard from Dr Denny Porter, the Principle Investigator for the study: he has informed me that recruitment is now complete, and the study is due to conclude in August this year. Once Dr Porter has evaluated the data, we will hear if the study is to be extended. If this is the case, there is a possibility that the next phase of the study will be multi-centre. I will of course keep you posted.*

#### ***Update from Genzyme on Acid Sphingomyelinase Deficiency (ASMD) Clinical Trials***

*In April 2009 Genzyme announced the completion of the Phase 1 study to evaluate the safety of single dose administration of recombinant human acid sphingomyelinase (rhASM) as a treatment for ASMD Niemann-Pick Disease types A&B. We have recently heard that Genzyme are making plans for the Phase 2 trial, including engaging regulatory agencies in discussion about the available preclinical and clinical data and conducting further preclinical research. Genzyme also plan to publish results from an international, retrospective natural history study of ASMD Niemann-Pick Disease types A&B. We look forward to receiving further information in due course.*

#### ***Zavesca - FDA does not approve use for NPC patients in the USA***

*In my recent Research Report for **Niemann-Pick News Spring 2010**, I regretfully informed you that the Food and Drug Administration (FDA) in the USA had not given approval for the use of Zavesca in NPC patients, at their hearing in March. This was a devastating blow for the US NPC community. Actelion Pharmaceuticals, manufacturers of Zavesca, has now received a complete response from the FDA in regard to this application. They have been asked to supply further clinical information, and will work diligently with the FDA to address the points raised. On a positive note, Zavesca is now approved for use in the United Kingdom, Australia, Brazil, Russia, Korea and Canada.*

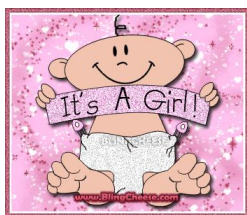


### **2011 London Marathon**

As previously reported in this e-bulletin and Niemann-Pick News, the NPDG (UK) has secured a place in the 2011 London Marathon. If you or anyone you know, are interested in competing in the 2011 Marathon on behalf of the Group,

please contact either Toni or Sue at the Central Office to register your interest before 30th May 2010. We will be holding a ballot on June 1st to decide who the lucky person will be!!

## Baby Congratulations



The NPDG (UK) would like to offer congratulations to Jackie Imrie, our Clinical Nurse Specialist, on the safe arrival of her second granddaughter. Jackie's son Ian, and his wife Vicki, had a baby girl, Hannah Rose, on 13th April weighing 8 pounds.

## RADAR - The Disability Network, There to Help

RADAR, the Royal Association for Disability Rights, have published two new books which support freedom and independence for families living with disability: **NKS (National Key Scheme) Guide 2010**—this is a complete guide to accessible disabled toilets around the UK. ***"If Only I'd known that a Year Ago - Everything you need to know about living with ill-health, injury or disability"***, contains up-to-date useful practical advice on essential services and contains a section on disabled children and support to help them have full and independent lives pre and beyond school. If you are interested in either publication visit RADAR's website [www.radar-shop.org.uk](http://www.radar-shop.org.uk). You can also contact them on **020 7250 3222**.

## Cash Counts—New On-Line Benefits Calculator



Contact a Family has launched **Cash Counts**, a new online service dedicated to ensuring families with disabled children are getting every penny they are entitled to. This new service includes an on-line benefit calculator, allowing families to work out how much they are entitled to; top tips on the benefits available to families with disabled children and a frequently asked questions page. This new service can be accessed via the Contact a Family website [www.cafamily.org.uk](http://www.cafamily.org.uk).

Whilst on the subject of benefits, the earnings threshold for qualifying for **Carer's Allowance**, the only state benefit specifically aimed at carers, was increased from £95.00 to £100.00 per week in April 2010. Parents with disabled children who are working and who would like to know if this change in the earnings threshold will affect them should call Contact a Family's freephone Helpline on **0808 808 3555**.

## Family Fund - More money - Get Applying!



The Family Fund helps families with severely disabled children to have choices and the opportunity to enjoy ordinary life. The Fund gives grants for things that make life easier and more enjoyable for the disabled child and their family, such as washing machines, driv-

ing lessons, hospital visiting costs, computers and holidays and will consider funding anything connected to bringing up a disabled child. If you have had funding from the Family Fund previously, you can re-apply using their on-line facility. First time applicants will need to call them as the initial application requires more details. For more information visit their website on [www.familyfund.org.uk](http://www.familyfund.org.uk)

### Study For Life-Limited Children and Their Families



ACT (the only UK organisation supporting families with palliative care needs) has been awarded a big lottery grant to fund a two year research project called 'The Big Study for Life-Limited children and their Families'. This research project will be based in the West Midlands and will be the first in-depth study in the UK to examine how well the needs of children with life-limiting conditions and their families are being met. The key elements of the research are: geographical mapping of services for life-limited children and the children and families that use them; analysis of families' met and unmet needs; analysis of the effectiveness and characteristics of professional networks, communication, co-operation and collaboration; an economic evaluation of service delivery, providing costs for various models of care to meet needs and user participation, which will be a feature across all the four strands of the research. For more information on ACT, see their website [www.act.org.uk](http://www.act.org.uk)

### Extra Support For Families With Disabled Children Outlined in Green Paper

Support for All: the Families and Relationships Green Paper, launched in February 2010 by the Secretary of State for Children, Schools and Families, proposes new measures to support families with disabled children to lead ordinary lives. The Paper proposes to extend the offer of key worker support to families with disabled children aged 0-19, initially with a testing phase in 2010-11 and with a view to creating a national programme. There is also a proposal to increase relationships support specifically for families with disabled children.

To view the Families and Relationships Green Paper go to <http://publications.dcsf.gov.uk:80/default.aspx?PageFunction=productdetails&PageMode=publications&ProductId=CM+7787>

### Parent Participation Works! Find Out How



Together for Disabled Children (TDC), a partnership between Serco and Contact a Family, has just published the **Parent Participation How to Guide**. The purpose of parent participation is to enable parents to get involved in service planning and decision making so that services meet the needs of families with disabled children. The aim is to ensure resources are not wasted on services which parents and families do not take up. The **How to Guide** is designed to help parents and professionals work together and is divided into 12 sections, so you can pick and choose the information you need. The **Parent Participation How to Guide** can be read or downloaded by going to <http://www.togetherfdc.org/Topics/ParentParticipationHowToGuide.aspx>

## Families With Disabled Children Lack Mental Health Support



The results of a survey by youth mental-health charity YoungMinds show that a third of parents with disabled children aren't receiving the support they need for their children's emotional or mental health problems.

87% of parents found barriers to getting help, including lack of available services, long waiting lists, being blamed for their child's problems and their child not fitting the criteria for support. 60% of those families who have received help questioned its worth.

The parents who responded to the survey suggested access to support urgently needed to be improved through: better education for GPs; fast tracking for families with very complex problems; more parent support groups and one point of access for all services. For more information visit the YoungMinds website [www.youngminds.org.uk](http://www.youngminds.org.uk)

## New Local Powers To Investigate Complaints About Special Educational Needs (SEN)

Following recommendations from the Lamb Inquiry, the Department for Children Schools and Families (DCSF) has given the Local Government Ombudsman (LGO) new powers to investigate complaints from parents unhappy with Special Educational Needs (SEN) support for their child. Anyone contacting the DCSF with complaints about their child's SEN will now be advised to contact their LGO. The remit of the LGO has been extended to consider complaints about the internal management of schools in respect of SEN. It also allows the LGO to investigate all parents complaints about provision for children with SEN. For more information go to [www.dcsf.gov.uk/lambinquiry/downloads/ed-balls-letter-to-brian-lamb.pdf](http://www.dcsf.gov.uk/lambinquiry/downloads/ed-balls-letter-to-brian-lamb.pdf).

## Fair Access to Care Services (FACS)



The Social Care Institute for Excellence (SCIE) has published updated guidance on the Fair Access to Care Services (FACS), first introduced in 2003. FACS is a framework to help local authorities determine eligibility for adult social care services, but contains guidance on transition for children with disabilities to adult services as well as the rights and needs of young carers as children and young people. The 'Carers' section in FACS 2010 identifies the need to take account of support from carers, including parents caring for a disabled child, and the sustainability of their caring role. To read the guidance and information booklets visit: [www.scie.org.uk/publications/guides/guide33/index.asp](http://www.scie.org.uk/publications/guides/guide33/index.asp)

## Training For Families From Early Support



For many families, the birth of a child with a special educational need or a disability is a life-changing experience. Parents will have a lot of questions and needs but may not know where to ask for help - or what to ask for. They may be faced with practical difficulties, or may have to make major decisions about their child which they may not feel able to make. Early Support training gives families the knowledge they need to manage in their new situation and make informed decisions and to help them understand and

get the most out of support services. To find out more about the different training courses that are available by going to [www.direct.gov.uk/en/CaringForSomeone/CaringForADisabledChild/DG\\_180194](http://www.direct.gov.uk/en/CaringForSomeone/CaringForADisabledChild/DG_180194)

## Care For The Family



Care for the Family is a national charity helping to support families. Part of that work includes **Another Way**, a telephone network for families with children with additional needs, headed up by Richard and Maggie Stapleton, who themselves have a child with additional needs. Another Way provides a telephone befriending service which offers understanding, empathy and support giving you the opportunity to say what you really feel and talk openly on the phone to a trained befriender, who is in a similar situation to you and can offer insights from their personal experiences. You can also receive encouragement by signing up for their email newsletters. To find out more about their work visit their website [www.careforthefamily.org.uk/anotherway](http://www.careforthefamily.org.uk/anotherway). You can read their latest newsletter at [www.careforthefamily.org.uk/anotherway/newsletter](http://www.careforthefamily.org.uk/anotherway/newsletter).

## WellChild Awards 2010



Do you know a sick child whose bravery is an inspiration, or perhaps you want to recognise the work of an amazing healthcare professional who goes the extra mile for sick children? Celebrate their achievement by nominating them for a WellChild Award and let the rest of the world know about them. Categories include: bravest child, doctor, nurse, research, volunteer and team. For more information go to [www.wellchild.org.uk](http://www.wellchild.org.uk); email [awards@wellchild.org.uk](mailto:awards@wellchild.org.uk) or phone **0845 458 8171**

## New Projects for Young People



Skillz For Life, working alongside National Star College and Speak, an organisation made up of young disabled people, are organising fun sessions for 13-18 year olds across England looking to learn new skills and build confidence. To find out if there is a project near you, Contact Philippa Gillespie on **0207 798 6125** or [p.gillespie@whizz-kids.org.uk](mailto:p.gillespie@whizz-kids.org.uk)

## Travel Insurance



We have recently had several enquiries from our families regarding travel insurance; our website has a list of those companies offering travel insurance for people with disabilities. For more information, go to [www.niemannpick.org.uk/care\\_and\\_support/travelinsurance.html](http://www.niemannpick.org.uk/care_and_support/travelinsurance.html)

## Fundraising/Donations

Since our last e-bulletin in January the NPDG (UK) has received just under £18,500 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 Nat West MK South England Corporate Service Centre and Nat West Private Banking Social Club (Hope for Hollie Campaign); Southwater Event Group; Denton West End Primary School; Clyst College; Will and William Woodhouse; Emma Potts; Charlotte Metters; Leslie and Tony Tifford; Gary Draper; Jodie O'Grady; Members of King's Heath Lodge; Carlos Gomes; Tony and Liskin Jellings; Dawnay School; Bishop's Stortford College; Tim Cullington; Christine and Joe Scott and Doxford Park Community Centre.

General donations were received from Paula and Mark Perkins; Janice Brooks; Sally Tungate; Mr Davison; Mary Cousins; Mrs V J Campbell; Dawson Group; Simon Grant; Mr and Mrs Loughran; Dr Backhouse; Marie McGreevy; Inger Smale; St George's Church & St Anne's Trust; Miss Nan Smith; Evelyn Mathieson; Brenda Keogh; Billinge Residents Association; Mrs Carol Yates; Tharsini and Nesan Rajanayagam; South Hylton Working Mens' Club; Mr and Mrs Hulme; Mr and Mrs Peacock; Lorraine Bishop; Wendy Peart, Sheila Peacock and anonymous Charities Aid Foundation (CAF) cheques to the value of £800.

We also received generous donations in remembrance of Matt McAdam and further donations in loving memory of Bob Merrick and Mary Christine Bradley

Val Ridley; John, Blaikie and Carol, Langport Motors and Genzyme emptied the contents of their collection boxes.

Finally, we received a very generous donation from Dr Heti Davies towards the running costs of the Children's' Programme at our Annual Family Conference.

Information about holding a fundraising event on behalf of the NPDG (UK) can be found on our website [www.niemannpick.org.uk/get\\_involved/get\\_involved.html](http://www.niemannpick.org.uk/get_involved/get_involved.html). You can also contact either Sue or Toni at the Central Office for more details or a fundraising pack.

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, or have received this e-bulletin by post and would now like it by email, please contact **Sue Lowe, 11 Greenwood Close, Fatfield, Washington, Tyne and Wear NE38 8LR**, or email at [sue.npdg@tiscali.co.uk](mailto:sue.npdg@tiscali.co.uk).

**Disclaimer: Information which appears in this 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.**