NPUK NEWS

Autumn 2018

IN THIS ISSUE

Annual Family Conference / Research and Clinical Trials / Fundraising News / BBC Children in Need & much more!

www.npuk.org / 0191 415 0693

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Chairman's Chat

Dr William Evans, NPUK Chair

Welcome to the Autumn newsletter, I hope you find the information and updates interesting and informative. Since the last issue of NPUK News, there's been much to get excited about...

I must start by mentioning what is the highlight of NPUK's year - our Annual Family Conference and Interactive Workshop. This September we were welcomed back to Wyboston lakes for another successful three day event which is now a truly international meeting with attendees from all corners of the world. The Conference was also our largest to date with record numbers of both family and professional delegates in attendance. I find the Conference weekend a truly invaluable opportunity to catch up with old friends and form new friendships, the environment is unique, a rare chance to chat with others who really do understand the challenges we face as individuals and families affected by Niemann-Pick disease(s).

The agenda for this year's Conference was packed full and as ever, the breadth of topics and advances across so many areas was exciting and encouraging to hear. If we reflect on where we were as a community just a mere ten years ago, these advances would seem almost unfathomable. However, despite all this promise we mustn't forget there is still so much to do. At present we still have only one licensed drug for Niemann-Pick disease(s) and although things are moving forward relatively quickly they are never quite as fast as we would hope or need.

Since the Conference NPUK remains as busy as ever, doing what it always does: helping and advising patients and professionals; supporting research and advocating for our community. For the latest news and updates from our dedicated Care and Support team make sure you read pages 6 to 11.

I am sure you were all excited to see Josh Cullip (age 12, NP-C), his Mum Jodie and our very own Clinical Nurse Specialist Laura Bell, featured on BBC One's Countryfile as they took on the challenge of the BBC Children in Need Ramble on the Isle of Skye. We are all so proud of Josh for taking on this huge challenge, and bringing attention and awareness to the amazing work of Children in Need, our charity, and Niemann-Pick disease type C.

When I see young people like Josh and Shona Beveridge (age 18, NP-C), who took on the Rickshaw challenge last year, putting themselves forward for events like this, I am in total awe. It is a very brave thing to do, not only the physical challenge, but perhaps even more so sharing to the nation their very personal accounts of how the disease impacts them and their family. I know this is not something that many of us would be able to do...so well done Josh, you truly made us proud! (For more on this please read pages 46-47).

I'd like to wish each and every one of you a wonderful Autumn, Christmas and New Year.

Warmest wishes,

Will

Chair, NPUK

"...if we reflect on where we were as a community just a mere ten years ago, these advances would seem almost unfathomable..."

Clinical Nurse Specialist: Laura Bell

Hi All,

Greetings from sunny Manchester!

Welcome to the autumn newsletter, it's hard to believe that autumn is upon us once more. I have been kept very busy over the past few months particularly in September and October as the children start back at school for the new term. I am always keen to hear how the children are settling into their new class, so attending a school meeting or providing a teaching session to the staff on the condition and how they can support the child in school is the perfect opportunity to do this.

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In September as many of you will know we held our 25th Annual NPUK family conference at Wyboston Lakes in Bedfordshire. We were joined over the weekend by many professionals and of course our amazing families from all over the UK and the world. We welcomed some new families this year for the first time to conference as well as families who have been attending for many years. This year we heard a lot of our patients and families talk about their experiences of living with Niemann-Pick and these were truly inspirational and fantastic talks. I was so proud of each and every one of you who got up to talk and share your experiences with us all.

During September, myself and the Care and Support team were invited to attend clinic days at Great Ormond St, Birmingham and Manchester Children's Hospital(s) where we got to spend time and talk with families to see if we can offer our support and help as well as spending time with the great teams that work at these specialist centres.

Again its that time of year for BBC Children In Need and as they very kindly part fund my position for NPUK we were delighted when they approached one of our families to see if they would be interested in taking part in this year's Children in Need Countryfile Ramble. We hope you enjoyed watching Joshua take part in his ramble last month!

As always we continue to travel around the UK and Ireland to wherever we are needed to support you at meetings, clinic appointments and home visits. As Elizabeth and I always say: "there is nowhere that we can't get to without a bit of planning!" So please if we can be of any help or support to you just get in touch.

Please remember we are here for you!

With warm regards,

Laura

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07791 499 555

Senior Families Advocate:

Elizabeth Davenport

Hello Everyone,

I hope that you are all feeling well and looking forward to a positive and peaceful winter. As ever, I am dedicated to my role here at NPUK and continue to support individuals and families affected by Niemann-Pick disease through my non-clinical advice and guidance.

But it doesn't stop there - I can also help refer members of our community to a counsellor (including finding funding to make this possible) if you feel that this would be of benefit to you or a loved one. This can be be for parents, family members such as siblings and others close to those affected by NPD. Having someone to talk to, and work through any and all difficult issues you may be facing is so very important to maintaining positive mental health. If this interests you please don't hesitate to get in touch with me and I can begin to make the arrangements necessary.

I have been quite busy with benefit reviews over the last few months, doing all I can to make sure that each and every member of the NPUK community is getting all of the benefits that they are entitled to. If you are currently looking at your review form for either Personal Independence Payment or Disability Living Allowance, and are a little lost then give me a call and we can go through the process together.

Universal Credit is now active in most parts of the country, and in some way or another it will affect everyone connected to NPUK. Any change like this can be both daunting and confusing at the best of times - so if you have been asked to move onto Universal Credit and need some assistance then as always I am on hand to help.

Preparations are underway for our Christmas Party, which as ever will be an amazing opportunity for our families to get together for some festive fun! More information can be found at page 74. Santa Claus has already got in touch with me and said he will be in attendance - I hope to see as many as you as possible there too!

With warm wishes,

Elizabeth

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NPUK PROJECT FAMILIES OFFICER

STEVE NEAL

Our NPUK Project Families Officer, Steve Neal, continues to develop links with individuals and families within the NPUK community as a part of our Big Lottery funded project "Shaping Our Future Together". Steve writes:

"...it's an exciting time to be a part of the NPUK community, as a lot of our initiatives start to gain momentum. Namely the NPUK Youth Council, which provides a vital opportunity for the younger members of NPUK to have their valued voices heard and to make a positive impact on the future of our organisation (for further information go to page 36). I am also extremely proud of the ever-growing Lads, Dads, and Carers group which is going from strength to strength (more information on page 9).

This year's Conference was, as ever, an emotional time for the families we help to support - but one thing we always have, is each other! As both a member of the NPUK Care and Support staff team and a family member of someone affected by Niemann-Pick disease type C (my mum Gloria has NP-C) I feel I am well placed to provide both a personal and informed ear when it comes to talking through any issues relating to a Niemann-Pick disease

Conference isn't the only time that I am available however, far from it! I am constantly busy travelling up and down the country visiting the lovely individuals and families of NPUK. I am dedicated to providing emotional health and overall wellbeing support - if you feel you would benefit from a chat or a visit. If there is anything that I can help with, then please don't hesitate to get in touch...after all, that's why I'm here!

This year has been a defining one for me personally, as I have really enjoyed working alongside our families at group events such as the All Ireland Meeting, Clinic Days, as well as trying to glean new approaches to care and support from other charitable organisations similar to NPUK.

I wish each and every one of you the best on the run up to the festive period, I hope you have a positive and peaceful Christmas and New Year.

With best wishes,

Steve

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07787 818 885

Lads, Dads and Carers

NPUK's 'Lads, Dads and Carers' Group was an initiative launched by Project Families Officer Steve Neal in 2017.

'Lads, Dads & Carers' is a lot more than just a closed Facebook group - it's an invaluable opportunity for the the male contingent of NPUK (the "Lads, Dads and Carers") to get together and share everything from experiences, struggles, frustrations, to positive coping strategies members may have stumbled upon, creating a strong support network in which we can help one another.

Despite there being many positives to interacting in the Facebook group and through messenger or texts, nothing beats being together face-to-face! Which is why we try to take advantage of being under one roof thanks to the Annual Family Conference - this year Steve provided a workshop where members could share everything from ideas to fears and we also had experienced counsellors (and Lads, Dads, and Carers group members) Tony Somers and Gaz Anderson join us to provide helpful insights.

Steve is currently working hard behind the scenes to try and facilitate activities for the Lads, Dads, and Carers group so that there can be more than one annual meet-up...stay tuned via social media for any and all updates! Please also get in touch with Steve if there are any topics or issues you would like to see discussed/receive advice on during these meetings.

If you would like to join the Lads, Dads & Carers' Group, please don't hesitate to get in touch with our Project Families Officer, Steve Neal (Email: steve@npuk.org / Tel: 07787 818 885) or our Communications Officer, John Lee Taggart (Email: john@npuk.org / Tel: 07984 366 334) who will be pleased to help you to get involved.

NEED ADDITIONAL SUPPORT?

There are a number of dedicated charities and support services that are in place to help men who are experiencing difficulties in their everyday lives, whether this be through stress, anxiety, relationships, and anything in between.

For a list of organisations which may be able to help, please visit: https://www.avoiceformen.com/men/mens-issues/uk-charities-that-help-men/

NPUK Project Team Leader:
Louise Metcalfe

Shaping our Future Together

In February 2016, NPUK was awarded a generous grant from the Big Lottery Fund to support a five-year project called 'Shaping our Future Together'. The project, now in its third year, aims to help the growing number of individuals diagnosed with Niemann-Pick disease, and their families. As well as all our on-going practical and clinical work with families, we aim, through this project, to provide more support for newly diagnosed patients and their families, and to reach out to more members of our BME community, siblings, and male community members.

Just where has the time gone? Unbelievably we are now into Year 3 of this project. Years 1 and 2 were very busy years and now in Year 3 our team is working with more people than ever before. This increase is partly due to improvements in diagnostic techniques and increased awareness, leading to more people receiving a Niemann-Pick diagnosis.

Through this project we want to reach out to as many newly-diagnosed patients and their families as possible, so that they know we are there, right from the start, to help and support them through difficult times. We know that being diagnosed with something so rare as Niemann-Pick disease can be a very isolating experience and as a team, we work hard to alleviate what can be a very lonely and frightening experience. Our community lives across the UK and we know that it is not always possible to find someone local to you who understands what it is like to live with Niemann-Pick disease. When families feel they are ready to do so, we encourage them to get involved in our Niemann-Pick UK community. This may be by joining our ever growing Facebook community, signing up for our regular newsletters or coming along to one of our events, where we aim to bring Niemann-Pick families together in a friendly and supportive environment.

Coming along to our Annual Family Conference is a great way to meet other families affected by a Niemann-Pick disease as well as learn all the latest developments in treatments, clinical trials and research. You can read more about this year's amazing Conference at Wyboston Lakes on pages 24-37.

One of the aims of our Big Lottery project is to help families to become more emotionally resilient and better able to manage the impact of disease progression. Sometimes people need a lot of very regular support from us when times are tough and others tend to get in touch every now and then when they need a bit of extra help or advice. We are always there to help you – be it a query with benefits, housing or helping your child get the right support at school. Read more about Elizabeth (Senior Families Advocate) and Steve's (Project Families Officer) work on pages 7-10.

BIG LOTTERY FUND

We want our members to have a better understanding of Niemann-Pick disease and to be confident when dealing with healthcare providers or statutory services by the end of this project. Our regular newsletters are packed full of information on events, fundraisers, treatments, clinical trials and research and our Facebook page and website are updated regulary with information that we hope is useful to you. Each year we revise and add to our publications and videos on the various aspects of Niemann-Pick disease to try and improve families and professionals' understanding of this rare condition. For example, this year we developed Patient Passports and Medical Alert Cards for ASMD and NP-C patients – if you would like one of these, please get in touch with us at the NPUK office.

If you haven't already, please take a look at our website www.npuk.org and our many videos on our You Tube channel by searching @npuk. Hospital appointments and clinic days are a good way to meet other patients and NPUK team members as well as learn more about your condition and how to manage it from expert clinicians. See more on page 13 about our Niemann-Pick Clinic Days.

iPad Project

One of the ways we hope to help more people is through our iPad project. Our Big Lottery Project includes funding to offer a number of iPads each year to adult Niemann-Pick patients and their families. So far, we have distributed 19 iPads to adults affected by a NP diagnosis. The aim of the project is to see if technology can help our members manage the many different aspects of living with Niemann-Pick disease:

- You could use your iPad to communicate with and send videos to our Clinical Nurse Specialist Laura Bell or other healthcare professionals involved in your care?
- Or perhaps you would get in touch with Elizabeth, our Senior Families Advocate, for advice, help or maybe a chat?
- You might want to join our NPUK Facebook community? For example, you could connect with our Dads, Lads and Carers group on Facebook?
- Use your iPad to find information on Niemann-Pick disease and read about the latest research?
- You might download apps that help you manage your condition?
- Take photos of family members and events?
- And of course you can use your iPad to watch films, catch up on TV shows, listen to music or play games with your family! It is entirely up to you.

We are now looking for more adults to take part in this project over the next 2 years. If you would like to take part in this project or would just like to know a little more at this stage, please contact NPUK on 0191 415 06 93 or by email at: louise@npuk.org.

Breaking Down Barriers

For the last two years, NPUK has been involved in the Breaking Down Barriers project, where a number of charities have come together to share information and good practice on developing supportive and inclusive services for families affected by genetic conditions. Research has shown that patients living with a genetic disorder can experience problems accessing services and information and this is particularly so for those from minority ethnic groups and for those with close cousin marriages. By working with a range of charities from across the UK, we have been learning about how best to engage with and develop services for all of our patients and how best to improve the awareness of the role of genetics in rare conditions. For more information on Breaking Down Barriers, please visit www.breaking-down-barriers.org.uk.

We are always interested in hearing from you about how Niemann-Pick affects you, your family and how we, as a charity, can help and make a difference to you. You can contact me at the NPUK office on 0191 415 06 93 or by email louise@npuk.org

Louise

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All IRELAND MEETING
Dublin, 2018

Earlier this year we were pleased to welcome families from across Ireland to meet the NPUK Care & Support Team.

On a bright July morning families from across Ireland met in Dublin with Clinical Nurse Specialist Laura, Senior Families Advocate Elizabeth, Project Families Officer Steve and Project Team Leader Louise...and what a fantastic time it was! It was for the most part an informal affair, giving both the community and staff team the chance to get to know each other better whilst chatting over lunch. Louise commented:

"...it was really interesting to share everyone's experiences of diagnoses, clinics and treatments in the Republic of Ireland, Northern Ireland and the UK. It also helped to find out what type of information and support our families wanted going forward. We all agreed that it should become a regular event and that we would try and get a medical professional in attendance at any future meeting..."

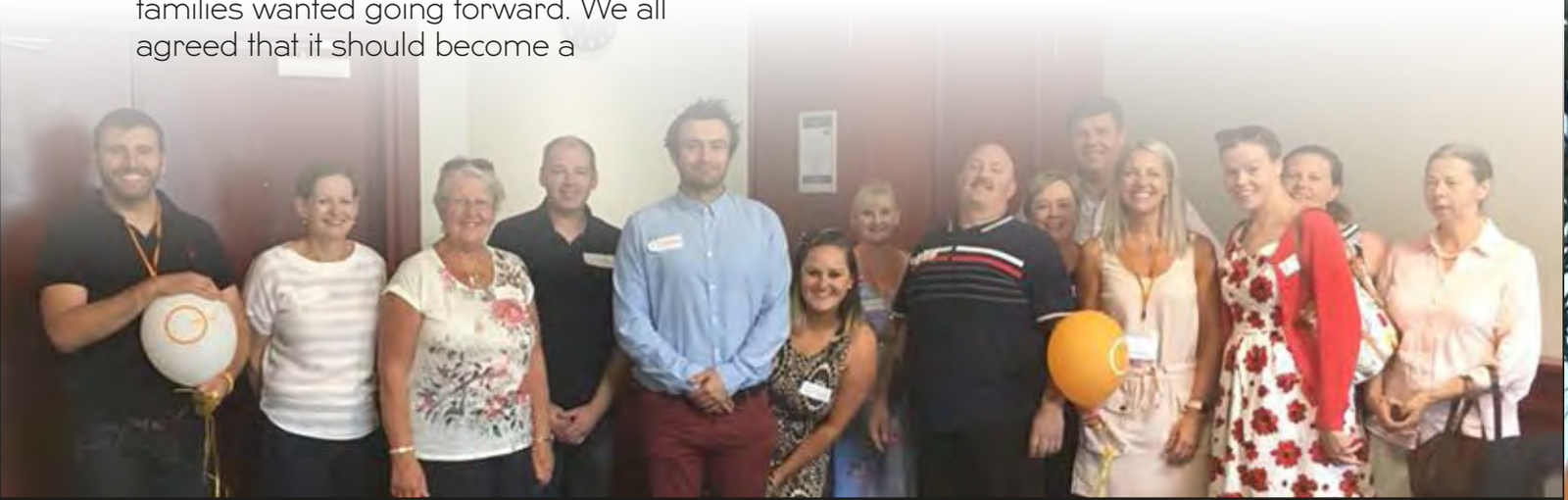
Particular topics of interest that were shared at the meeting included diet, exercise, how to avoid and/or minimise stress, medications and clinical trials. It will be helpful moving forward to know which issues affect our families in both Northern Ireland and the Republic of Ireland so that we can better tailor our service to support them!

Unfortunately the day seemed to fly by, and before they knew it our Care and Support team were back on a plane to the UK, however they were pleased with what was a positive and productive meeting.

We were excited that one of our families from the Republic of Ireland were then able to attend our Annual Family Conference this year, giving us another chance to have a nice catch up!

We hope that more families from across Ireland will be able to join us in 2019 - watch this space for more updates!

For further information please contact NPUK directly by either email at: info@npuk.org or by phone on: 0191 415 06 93.



Clinic Day Fun!

A big part of our work at NPUK is attending Clinic Days and hospital appointments along with many of our Niemann-Pick patients and their families.

Clinic Days are held regularly across the UK at Specialist Centres in Manchester, Salford, Birmingham and London. So far this year we have held 7 Clinic Days and have seen 73 patients in either a specialist Niemann-Pick clinic or hospital appointment.

Clinic Days try to bring together, where possible, all the professionals that may be involved in your care, such as the doctors, nurses, physiotherapists, dieticians, speech and language therapists and our own Clinical Nurse Specialist, Laura Bell.

It is a great opportunity to have everyone in the same place, at the same time, to help you with any issues you may have in managing your condition. Elizabeth Davenport, our Senior Families Advocate and Steve Neal, our Project Families Officer, attend Clinic Days, whenever possible, to meet patients, new and old and help with any non-clinical issues you might have.

We know that coming to hospital can be a stressful time, especially so for our children. We try to make Clinics as relaxed, stress free and informative as possible. For example, at Royal Manchester Children's Hospital, we aim to organise our clinics so families can meet each other in an informal setting, enjoy a bit of lunch and even some entertainment. Louby Lou dropped in on a September clinic to meet some of our fantastic children and young people. In September we were also thrilled to attend a Clinic Day in the brand new Rare Disease Centre at Birmingham Women and Children's Centre where we were able to use the wonderful new chill out and sensory rooms to meet privately with families before and after their appointment. We are working hard to organise similar clinics at other hospitals. In the pipeline are plans for a dedicated Toddler Clinic for Niemann-Pick patients and a Transitions Clinic to support our young people as they move into Adult Services. We would love to hear your thoughts on Clinic Days and how we can make sure they work well for you.

For more information please contact NPUK Clinical Nurse Specialist by email at: laura@npuk.org.

Research Report:

Bill Owen, NPUK Trustee & Research Coordinator gives an update on the latest in Niemann-Pick research:

Introduction

This report provides an overview of medical science presentations made at our Annual Family Conference and Interactive Workshop. The majority of the presentations were given on the Friday at the Interactive Workshop where the target audience comprised of professionals from healthcare services, universities, the biotech industry and patient group representatives with an interest in the Niemann-Pick diseases. Certain presentations were also given at the Family Conference during the Saturday sessions.

Research pathways

The programme was comprehensive in that all aspects of research were included. A brief review of the main areas under which research is conducted is provided below with presentations given under the main research headings outlined, further on.

Basic/diagnostic research

Topics addressed basic science/ diagnostics that seek to understand the operation of the cell under normal and failure conditions. Although our understanding of the cell has increased dramatically over the previous few decades there remains much to learn and, in this respect, research into diseases is at the forefront of medical science discovery. Our interest in this basic research is in the identification of targets for therapy and developing new ideas to the point where the proof of principle is established. Basic scientific research into cellular function provides the raw material for the development of both treatment and preventive strategies with the three areas being interactive and overlapping. A significant contribution to funding these studies has been made by patient organisations in Europe and the USA.

Treatment research

Where studies achieve success and demonstrate potential for affecting the course of the disease new research is required that will enable a particular treatment to be developed to the point where it may be proposed for clinical trials. This is usually when much higher budgets are required to move the concept studies into clinical therapies. These activities are conducted by the pharmaceutical industry and start-up companies funded by venture capitalists. A considerable body of evidence must be complied to satisfy the medicinal regulatory bodies such as the EMA and FDA and this evidence must show that safety is foremost, that the medicine can be tolerated and is capable of altering the course of the disease in a meaning-ful way. If the medicine is approved by the regulators then it must be submitted to the authorities for evaluation of efficacy against price.

Preventative research

This area of research remains much neglected by the health authorities and many of the major patient organisations but the landscape is slowly changing. Prevention of diseases such as the Niemann-Pick relies on data generated from basic/diagnostic research such as metabolomics and genetics. It differs from the treatment approach in that instead of being applied to symptomatic individuals its application


"...research into diseases is at the forefront of medical science discovery..."

is to large populations who, in general will be healthy. It represents a step change in thinking but has great potential to prevent many children being born with rare genetic diseases.

Patient Group involvement

Whilst we, as a patient group continue to pursue the treatment approach we also are concerned to prevent families of the future from having children with the diseases. Our main initiative at present is to build a case for newborn screening for NPC and presentations were given on this from the UK perspective and from the Firefly Fund in the USA.

NPUK Peter Carlton-Jones Award

Each year since 2006, NPUK have presented an award to a student or young researcher who has carried out research into the Niemann-Pick diseases and has submitted an abstract of the study to the charity. This year we were pleased to make the award to Ecem Kaya, a PhD student from Professor Platt's laboratory at the University of Oxford and Dr Bernado Barrios-Crispi studying at Dr Emyr Lloyd-Evans's lab at the University of Cardiff.

Each candidate gave a presentation on their work covering the development of symptomatic treatment for ataxia in NPC and a new approach to screening for therapeutic compounds.

Patient Registry – INPDR

A presentation on the development of the International Niemann-Pick Disease Registry (INPDR) was given covering the background to the project, its initial development phase and the work to be carried out in the near future in relation to populating the registry with data from patients all over the world and its use in research and clinical trials. The USA patient group, NNPDF has recently announced the launch of the registry and advertised its presence with a Webinar in mid October.

(Left to right) Bill Owen, NPUK Communications Officer John Taggart and INPDA Vice-President Sandy Cowie share a laugh at the NPUK Annual Family Conference & Interactive Workshop 2018.

Basic Research / Diagnostics Studies

Professor Platt introduced the first session that addressed the links between Niemann-Pick type C disease, the NPC1 protein,
disease causing pathogens and also malfunctions of the cell's metabolism that lead to other diseases such as Alzheimer's and Parkinson's.

NPC1 and pathogens. Pathogens such as viruses and bacteria have co-existed with humans and other species for millions of years and have evolved to overcome our immune system which, in turn, has evolved to resist their parasitic invasion - the battle goes on! Studies are being undertaken to discover how the pathogens gain entry to cells and make us ill or worse, kill us.

Some of these such as Ebola and mTB have been found to utilise the endocytic pathway into the cell but need to avoid being ingested by lysosomes where enzymes would dismantle them for recycling of their molecular content. By targeting and disabling the NPC1 protein that resides in the limiting membrane of the lysosome the pathogens are able to survive and multiply. The task of the scientists is to identify exactly how the interaction with NPC1 takes place and devise therapies to block the pathogenic action.

NPC1 and gut ailments. The immune system associated with the gut is highly complex as it must coexist with millions of bacteria of different types - part of the microbiome that inhabit our bodies and are essential for our health. In NPC disease the cells of the gut and the immune cells are compromised and this can lead to illness as a result of bacteria getting out of control. An increase in irritable bowel syndromes (IBS) has been reported in patients with NPC. Small scale trials using treatments already in use for IBS conditions to reduce the inflammation arising from infection by the bacteria are in progress with NPC patients.

NPC and Huntington. Diseases of the central nervous system present a major challenge to investigators due to the complexity of the CNS and the lack of understanding as to how the various sub systems integrate. Similarity between diseases is a useful way of understanding some of the mechanisms of action that result is diseases. It has been known for some time that there are pathological similarities between NPC disease and Alzheimer's that related to the debris within the brain such as tau defective neurofibrillary tangles and amyloid beta plaques. Studies have also identified possible links with Huntington's disease, which is another devastating neurological disorder. A study in a Huntington animal model with the Huntington protein removed showed a reduction in lysosomes in cells that led to storage such as found in NPC. The study also found that the Huntington protein does target lysosomes and is active at this site and may act as a chaperone to NPC1.

Neuronal synapses. - NPC is well known to affect the central nervous system but working with CNS cells, especially neurons brings additional challenges. Connections between neurons are made by junctions called synapses and signalling molecules between neurons must pass over a gap. Cholesterol is part of the transporter for the signalling molecules but as is well known defects in the NPC1 protein reduce the availability of cholesterol leading to reduced signal traffic. Studies have shown that the cholesterol can be redistributed by activation of cyp46, a cholesterol transporter in the CNS and this improves cognitive and behaviour activities in mice.

Novel 9bp mutation. - The capability to edit genes using a tool named CRISPR/Cas9 and to introduce novel mutations into animal models has given researchers greater scope for studying the function of proteins. Studies at the US NIH have done this with new animal models and one such study was presented. One discovery from the study is that the new mutation caused over 50% neonatal lethality within 24hours mostly

due to inability to breath. It was not clear if the 9 base pair deletion was similar to the null mutation but in this case, the mice live longer. The study also raises the question as to whether a similar situation applies to humans where death of a baby would not be captured by statistics distorting the true incidence of the disease.

Lysenin study - The study of molecular location and trafficking within cells is necessary to achieve an understanding of cellular function. Tracking may be achieved using a variety of tools such as dyes, fluorescent markers or naturally occurring molecules that can attach to the molecule of interest. Sphingomyelin is an important component of cell membranes and is recycled through lysosomes. An efficient tracker of sphingomyelin is the worm derived molecule, lysenin that has been available for some years but is now ceasing to be produced. Plans to carry on production at Cardiff University are now almost complete. In outline the process requires extracting the DNA from specific types of 'super-worm' and inserting this into bacteria for replication. Various modifications take place including a tag for antibody binding and recognition. The work on production of lysenin is well advanced and requires to be published.

Screening for therapeutic molecules – Current methods of screening drugs for therapeutic use seem to underperform and although many thousands of compounds have been screened results are disappointing. The methods employed did not identify drugs already known to be beneficial to NPC – eg miglustat and arimoclomal and perhaps this is not surprising as the process is arduous requiring microscopy study which is labour intensive and slow. Using an approach based on lysotracker, an indicator of lysosomal expansion and screening the compounds over a longer period – over 5 days, for shape and charge distribution is proposed as a way of improving candidate identification. To date a study using this approach has, retrospectively, validated current NPC therapies as capable of reducing lysosomal storage and further work using over 150 compounds with similar characteristics to current drugs in use and some already FDA approved is nearing completion. Next steps are to further test a group of 10 molecules on all NPC cellular phenotypes and publish standard guidelines for drug screening of all lysosomal storage diseases.

Disease Treatment and Therapy Development

Gene therapy development

This area of therapeutic development holds great promise although it has a history of serious problems. Our charity first became involved in this topic soon after the discovery of the NPC1 gene in 1997. It all seemed so simple as a naturally occurring mouse model was available, the gene was now known and a viral vector, HSV2 had shown promising results in delivery of a reporter gene in animal models. All we needed was funding and this was obtained after a successful application to the National Lottery. To keep it short it was in retrospect doomed to failure but, silver linings do exist. We learned a great deal from the project and best of all we attracted Fran Platt to the disease. Fran has been our star performer in the UK ever since. Sadly and because some researchers in the US became too ambitious a patient died in a gene therapy experiment and the FDA sin-binned all studies for the next 10 years.

In the last few years the science has moved on considerably and a generation of viral vectors based on Adeno Associated Virus (AAV) are showing great success in application to a number of diseases, even in humans. Like all therapies, early intervention remains important and this requires early diagnosis. A further problem recognised is that first treatment success is vital as the immune system will respond to subsequent administrations of the vector. There remains a great deal to learn and there are things that we will not know until the experiments are conducted. In this session presentations were given on studies that described some of the problems such as type of animal model, method of administration,

For information about current clinical trials for Niemann-Pick disease:

Visit www.clinicaltrials.gov, a database of privately and publicly funded clinical studies conducted around the world.

distribution of the vector in complex organs such as brain, transfecting a range of cell types – neurons various glial populations, getting the therapeutic gene to express, packing the therapeutic gene into the viral genome – and many more.

Studies underway for NPA and NPC were presented. Type A requires the introduction of the ASMD gene that codes for a soluble enzyme. This enzyme will spread to adjacent cells from the ones that are transfected and the vector used, an AAV9 variant, will transfect both neurons and glia cells. The study focussed on the CNS of a knockout model of the ASMD mouse and a benefit of using AAV9 is that it is able to cross the blood-brain barrier (BBB) although intra-cisternal administration is being used to better target the virus to CNS. The US National Institutes for Health (NIH) in collaboration with others is looking at different vectors aimed at improving transfection capability, types of promoters to improve gene expression, payload and other aspects of this experimental therapy. Studies in mice show a new variant, the AAV9.php.b to have considerably greater transfection efficiency in the CNS – greater than x40. Videos showing the performance of mice with various treatments showed impressive results in terms of performance and longevity of the PHP.9 treated animals and improvements at cellular level such as reduction of cholesterol storage. Further developments are awaited with interested.

A study taking place in the UK using a variant of AAV9 was presented, the problem areas mentioned above discussed together with measures to improve vector performance. Although there were initial difficulties of packing the large NPC1 gene into the viral genome studies are progressing well with no inflammatory response detected. The study employs a cohort treated with miglustat for comparison in addition to untreated controls. Video evidence was shown which was impressive. Tests on the mice such as rearing walking gait and tremor were all normalised and life span was increased and, further increased with dose escalation. In the CNS neurodegeneration was reduced significantly as was reduction of cells in the cerebellum. A factor limiting the improvement is that the therapy is targeted to the CNS and the remainder of the body is not treated. Liver disease becomes a dominant factor in clinical deterioration. A major factor in the successful results is that the mice are given therapy at the new born stage allowing the transfected gene to spread as the mouse pup grows. Optimisation of the vector is playing an important part in the work and treated animals show highly extended survival using these vectors. There is considerable cause for optimism for this therapy indicated by the work to date - but the challenge of getting it to the patient early enough remains to be resolved.

Clinical Trials

Dr Marc Patterson introduced this session with a review of the need for, and, the development of clinical trials over time and the progress made in the last 10 years. Dr Patterson added a caution that clinical trials are experiments and although every precaution is taken to ensure that medicines are safe and can be tolerated, efficacy remains to be shown in humans with a disease. Currently, there are five new trials in progress to treat NPC, and NPB. These are:-

- The NPC intravenous Trappsol trial being conducted by CTD Holdings
- The NPC intrathecal VTS270 trial being c onducted by Mallinckrodt (previously Sucampo and Vtesse). Concern was raised about the development of a port device for administration which is understood to have come to a halt.
- The NPC1 Arimoclomal orally administered trial being conducted by Orphazyme.
- The Olipudase Alfa NPB (ASMD) trials being carried out by Sanofi Genzyme on adults and children.
- The small scale trial for ataxia symptomatic relief using N-Acetyl-L-Leucine by Intra Bio. This new trial utilises a drug with 60 years use in patients with ataxia and is reported to improve motor function and cognition in NPC.

• Miglustat - this drug remains very much in use and the only one to be licensed for use in many counties although not in the USA. It is generally considered to reduce the rate of disease progression in NPC.

The trials are each at different stages of progress and presentations were given on the background to each and the current situation. Non were able to present results although Orphazyme indicated that they are very close to issuing a report on their initial results. Any further updates will be issued under the authority of each individual company and we will make these available to our community. In the case of the Sanofi Genzyme trial which has been very much delayed concern was expressed as to why the trial continues when the evidence from Phase 1 patients is that the ERT works very well, is well tolerated and toxicity issues have been resolved. A trial review is urgently needed to resolve the delay in making this medicine available to NPB patients.

Design and management of clinical trials

Clinical trials for rare diseases raises many new issues and calls into question the validity of trials design used for large scale trials. One problem is that it is often difficult to recruit sufficient patients for participation and this is compounded by the need for exclusion criteria based on a range of factors. Whist there are patients excluded because of advanced disease state the main selection criteria is based on age with little attention being paid to the state or rate of disease progression. A more useful approach is to select patients based on their assessed rate of disease progression to arrive at an annual severity increment score. This is simply the total rate of progression divided by age. This approach, ASIS, provides for exclusion of patients whose rate of progression is very slow or fast. The trial medication is unlikely to have any noticeable effect on these groups and the efficacy of the treatment will be distorted.

The ASIS approach also provides a predictive measure of patient disease progression once the initial rate has been determined from one of the various severity scales in use. It has been established that each patient has their own rate of progression once they become symptomatic and that this rate is constant throughout most of the disease course. The major severity scale devised by the NIH has many main domains and many sub domains. Studies have established that these could be much reduced without significant loss of accuracy. A reduction of 15 domains to 5 has shown 80-90% concurrence with further reduction to 3 domains being possible. The ASIS approach is being used in association with the N-acetyl L-leucine trial. Discussions are needed with regulators to look at the approach which will assist in trials recruitment and selection.

NBS: An Approach to Prevention

Newborn screening (NBS)

This session was introduced with a presentation from Professor Paul Gissen on the complexities of genetics in the Niemann-Pick diseases and the need for classification of novel DNA sequences identified.

Developments in the UK and USA

A major problem facing diseases of the CNS, and perhaps other non-dividing cell populations is that clinical diagnosis is not possible until the patient is symptomatic. At this time irreversible damage has been sustained – dead cells cannot be treated! There is usually a significant diagnostic delay until a disease is identified which does not help. Studies on animal models have shown that the earlier therapy can be administered, the better the outcome. Using one of the preventative strategies – NBS, it may be possible to identify the disease pre-symptomatically, at least at cell level. New discoveries of biomarkers using LC-MS/MS combined with a progressive library of pathogenic mutations provides an opportunity to predict clinical disease at birth. This will allow early administration of treatment and the ability to plan an ongoing clinical management programme, social and educational needs, financial implications and

reproductive choices for the immediate and wider family.

If a NBS test had been available in the UK over the previous 10 years it is estimated that well over 50% of babies could have avoided being born with NPC. The requirements for NBS are complex and NPUK is collecting information to ascertain whether sufficient data of the appropriate quality is available and what research is required to fill any gaps. Central to implementing NBS is the need for a test and progress in developing this was described by US and UK scientists at our last Conference. Some issues raised then still remain to be resolved.

The USA has different problems in that each state makes its own decisions on what NBS tests to adopt. Adoption would usually be based on recommendations from the Recommended Uniform Screening Panel (RUSP) that currently recommends NBS for 35 different diseases but some states test for many more than this, New York for example screens for over 60 conditions. NBS for Niemann-Pick type B ASMD was submitted to the RUSP in 2008 but was rejected because of the lack of evidence, a problem common to most rare diseases. NBS pilot schemes would help in generating the data needed and such a study was conducted for a group of lysosomal storage diseases (LSDs) over a period of 4 years on nearly 70,000 newborns and included ethical approval. Lessons learned included the identification of milder forms of some of the diseases, later onset groups and population specific groups. Although positive screening resulted in diagnostic follow up, a significant proportion of babies remain in undetermined disease status, a situation for concern. Encouragingly, the infrastructure and the processes worked well which bodes well for further NBS programmes.

As there is currently no provision for a NBS test for NPC in the USA, a group of diverse stakeholders have formed a Working Group to progress the inclusion of this test by the RUSP.

The Firefly Fund is leading this effort and is joined by the Ara Parseghian Medical Research Fund and leading professionals. The Working Party was formed in 2017 and has developed a NPC1 Newborn Screening Strategic Plan. This requires the collection of considerable pilot data and the intention is to screen between 150,000-250,000 newborns to demonstrate the ability of the test to identify NPC affected babies. The pilot scheme is expected to begin shortly.

Research into NPC conducted over many years has resulted in the generation and publication of a great deal of data on the disease natural history, disease diagnosis and clinical management. The information, although derived from patients who already have the disease and, primarily aimed at the design of therapies will be of great value to a newborn screening application or other preventative measures. Examples of the tools that have been generated as a result of the research include a range of therapeutic interventions, Niemann-Pick Disease International Registry that includes a library of disease causing mutations, Consensus Guidelines for the Management of NP diseases.

The role of genetics

Whilst the emergence of biomarkers in blood is suited to the concept and practice of NBS the subsequent diagnostic confirmation must take account of the individual's genetic sequences. A library of disease causing mutations has been established and this is extended by the discovery of new mutations in patients presenting with the disease. In addition there are genetic changes termed polymorphisms that are benign and do not impair protein function. A third category of changes to the DNA sequence also exists whereby the clinical significance of these changes is not known and difficult to predict in terms of effect. A presentation was given on this by Centogene, a company set up to identify and monitor large cohorts of NPC patients worldwide with the intent to improve early diagnosis but data produced will also be of value in NBS.

Traditionally sequencing of genes has been expensive and time consuming but advances in technology such as the development of nanopore sequences mean that whole genome or exome sequencing can be carried out rapidly and at low price. Whilst problems remain to be resolved attention is now focussed on the interpretation of the DNA readouts obtained in terms of their effects on the resulting protein and importantly, the health of the person. Following biomarker identification in NBS in a large population screen novel genetic sequences will be identified and will raise questions about potential disease presence. Mass screening is needed to identify the variants to be found in a population such as the UK and although projects are in existence such as the 100,000 Genome Project, this is unlikely to be a major contributor to Niemann-Pick diseases.

Summary

Progress continues to be made on all research fronts and we are hopeful that the current trials results will give us confidence that we are doing the right things to combat the Niemann-Pick diseases. Very recent announcements by some of the companies give cause for optimism but those of us directly involved in these trials will form our own views based on our experience. The ASMD/NP type B is in need of rapid action to make the medicine available to patients but it remains unclear how this will happen. For Type C we will need to wait a little longer.

Bill Owen, Trustee and Research Co-ordinator for NPUK

If you have any questions for Bill, you can contact him in the following ways:

Email - info@npuk.org
Phone - 0191 415 0693

(Left to right) one of the Peter Carlton Jones 2018 winners, Bernardo Barrios Crispi, sits alongside Dr Emyr Lloyd-Evans of Cardiff University at this year's Interactive Workshop

25th Annual Family Conference & 9th Interactive Workshop on Niemann-Pick disease(s)

This year's Annual Family Conference was a special milestone marking the 25th year of this important and valued event...in the same breadth, a lot and a little has changed.

This year we returned to the familiar surroundings of Wyboston Lakes, Bedfordshire, which even despite it being half-way through an extensive facelift was as warm and welcoming as ever.

Throughout the year we strive to work on behalf of individuals and families affected by Niemann-Pick disease (NPD). We work to ensure access to optimum care, whilst attempting to increase our shared knowledge of the condition and accelerate the research taking place in the field - this hasn't changed! But what has changed dramatically is the size and scope of our charity, and the impact we now have on a global scale, since that first Family Conference 25 years ago. For some of those who were there in the early days the difference will be staggering, from a few families to over 250 attendees this year...making it the largest NPUK Conference ever!

With each passing year we are joined by an ever-more diverse group of both professional and family delegates, and thanks to a jam-packed programme there was something to interest everyone in attendance, whether you were a visiting family, professional, or friend of the NPUK community. There was the opportunity to hear the most recent developments regarding clinical trials and research for Niemann-Pick disease(s) (more information on pages 14-23), a fantastic Children and Young Person's ActivityProgramme (read more at page 35), an abundance of workshops and breakout sessions, and entertainment to allow our community to unwind and have fun!

We were pleased to see so many people getting involved with our recently produced "Selfie Frames" throughout the weekend! They're a fun and novel way to show unity in the face of Niemann-Pick disease... after all, as we grow in number so does the chance to raise further awareness whenever we get together.

In fact, there are some fantastic photos of our community from this event, only a percentage of which will be used in this issue of NPUK News...so if you spot yourself, or a friend/family member in a photo and wish to have a copy simply get in touch with our Communications Officer, John, at john@npuk.org.

For those in attendance this year, you may have already filled out a feedback form (either online or offline) - but for those that haven't, we want to hear your views! This is why after every Conference we ask you, our community, for your opinion of the weekend and the service that was provided, so that we can gain a fuller understanding of how you feel about Conference and how we may be able to improve it in future years.
You see, each and every year we want to make the experience better for you and your family, and that starts with open and honest feedback.

To request a feedback form please contact us at the NPUK Central Office by either email at: info@npuk.org or by phone on: 0191 415 0693, and we will arrange one to be sent to you.

"...could not wish for a better team. Every year the team amaze me by how they bring the event together. They are all amazing – big thank you..."

"...so glad we went to the NPUK conference, no matter how hard and emotional it was..."

"...was lovely catching up with old friends and meeting new ones..."

Jenny Charman: My First Conference

Jenny Charman, co-founder of fundraising super team 'Harvey and the Brave Little Soldiers', shares her story of her first NPUK Annual Family Conference & Interactive Workshop:

What is your connection to NPUK?

I helped co-found the group Harvey and the Brave Little Soldiers (HBLS) in honour of my nephew, Harvey. We are here to raise vital funds for NPUK's ongoing research and for the incredible support they provide to families. I initially opened a JustGiving page telling our story in July 2017 with the aim of raising £1,000 for NPUK. The support was overwhelming and we smashed that target - our total now stands at close to £20,000! This has been achieved by climbing up mountains, holding a clubbercise night, sponsored runs, bike rides and lots of other fantastic fundraising events. We want to put the FUN in FUNdraising (cheesiness intended - ha).

Please visit our website for more info and to see pictures of these events:

www.harveyandthebravelittlesoldiers.com

What was your first impression of the Conference weekend?

I really was not sure what to expect at the NPUK conference but the whole weekend was amazing. I came away completely in awe and inspired by the many people I had been lucky enough to meet and I couldn't wait to share my experience with my family and friends. A resounding theme of the weekend and what I took away was hope. Hope for the future and hope for now. Families in similar situations were able to share stories and experiences. The weekend was able to act as a sanctuary and enable people to get away from what they have to deal with from day to day.
I was also amazed by the number of researchers and scientists that were at the conference and their willingness to share their knowledge not only during their official talks but just in normal conversation. During my first evening at the conference a very kind family introduced themselves and told me that by the end of the weekend it would feel like I have known everybody for ages. This sounded a cliché but certainly rang true after the two days.

Our community often refer to NPUK as a "family"...why do you think this is?

At times like these and events like the Conference, NPUK could not do any more for their families. Everybody is made to feel

welcome and there is a real family feel. Many people during the weekend actually referred to everyone there as their "NP Family". Day to day, NPUK provide emotional, as well as practical support and have developed a strong family support network, helping to reduce feelings of isolation and despair.

What was your Conference highlight?

On the Saturday I was lucky enough to go to Wicksteed Theme Park with the Children and Young Person's Activity Programme, and meet lots of the amazing children and volunteers, many of whom have attended the Conference for many years. As they kept saying "once you come to one, you come back". It really felt like I was made to feel like one of the family and we all had so much fun, despite the rain! The children didn't stop smiling all day, and neither did I. I will definitely be coming back next year and I look forward to seeing everybody again.

What would you say to someone considering joining us at our next Conference?

If you are questioning whether or not to attend next years conference what I would say is ask yourself this: Are you affected by Niemann- Pick directly or indirectly? Would you like to meet people who understand (REALLY understand) what you are going through? Would you like to hear about all the latest Niemann- Pick research happening right now? Well if you answered yes to any of these questions then I wholeheartedly recommend you attend as you will not be disappointed.

Together, we can help NPUK to continue their amazing and holistic charity, which so many families would be lost without.

Thanks for sharing your story Jenny!

SHONA BEVERIDGE:
My Conference Weekend

Shona Beveridge shares her thoughts on this year's Conference and explores her story with NPUK so far.

I spoke in the "My Story" section of the NPUK conference in 2017. It was hard reliving my diagnosis, but I was very happy to share it with everyone! I spoke about what lead to me being diagnosed with NPC, at such a late age. My parents only noticed I was different from all my friends at 13. I was taken to the GP at 14, who referred me to my local hospital for tests, I was initially going to be diagnosed with dyspraxia, but another consultant thought there was ataxia and something else going on as well, This resulted in me having blood taken and tests ran on it for all the common genetic ataxia diseases but the results all came back negative and I had more blood taken, which they ran tests on it for the rarer genetic ataxia diseases via an ataxia genetic panel test. Niemann Pick type C came back as positive – I was almost 16 years old.

The 23rd NPUK Annual Family Conference (2016) was just after I had been diagnosed, but my parents felt that it was far too soon after my diagnosis. My paediatric consultant had seen NPC before thankfully, because a boy who is currently on Miglustat has it - he was diagnosed at 10 and lives in Embo just north of Inverness. Anyway it was a quick diagnosis because we went to the GP's in August, and had a diagnosis by July the next year. It was at Conference 2017 that I told everyone I was in selection for the Rickshaw Challenge during my "My Story" speech.

This year (2018) at Conference I spoke in front of specialist doctors, many of whom never get to hear from patients and the stories behind them. I did this with my. younger sister Kelsie who also has NPC, but is not as bad as me. Fortunately I'm on the VTS-270 Cyclodextrin trial, (I started in March 2017), and I think this has helped me in more ways than one. I think my walking has improved because of the drug as well as my speech. I believe that I now have less tremors than before I started the drug trial. Lots of the specialist doctors wanted to speak to my sister and I, and ask us both questions, which we would try to answer if we could.

I also gave a speech about the Rickshaw Challenge. I felt honoured to be giving this speech, as Toni and Laura could have chosen anyone else to represent NPUK in the Rickshaw Challenge. I felt confident

about giving this speech, as everyone knew me from my appearances on "The One Show". I explained to the audience who exactly my team mates were and why they had been chosen, then I said a big thank you to everyone that came out to support me at the stops along the route from London to Glasgow. I explained about our chaperones and why they were there to the audience of NPUK families, who were patiently waiting to hear my legendary adventures from the Rickshaw Challenge.

I couldn't wait for Conference this year as I was a volunteer on the kid's day out to Wicksteed Park. I was with all the younger kids, and mostly stayed with Lleyton doing everything that he did. I helped Lleyton make his chocolate mug brownie on the Sunday in the Junior Bake Off, which was good as I wanted to make one, but couldn't take it home as I flew.

I enjoyed this year's Conference, but it was very different to last year's Conference. Last year's Conference (2017) was my first, so I didn't know what to expect from it. I believe Conference is a place that families can get together, because we all understand Niemann-Pick and how it affects everyone in different ways. I believe that even if a child has died due to Niemann-Pick, you can still keep coming to Conference because I consider the whole Niemann-Pick community as a big family who understand that Niemann-Pick affects individuals in different ways and those affected by Niemann-Pick types A, B and C no matter their age, love living life to the fullest!

By Shona Beveridge

SUPER NPD SCIENTISTS CYCLE FROM OXFORD TO CAMBRIDGE!

We are pleased to announce that the brilliant (and likely exhausted!) individuals below, who are currently working towards a cure for Niemann-Pick disease(s), managed to complete their cycling challenge...raising a whopping £3,255.50 in the process!

In the build up to September's Annual Family Conference & Interactive Workshop a team compromised of the Platt Lab at the University of Oxford and the Rahim Lab at UCL, set out on a two day, seventy mile cycle challenge from Oxford all the way to Wyboston Lakes, Bedfordshire in time to join our event!

The group included Fran Platt, Ahad Rahim, Kerri Wallom, Issi Platt , Michael Hughes, Claire Fletcher, Gokhan Yilmaz, Ecem Kaya, Raffaele Pastore, David Priestman, Hsin Chen, Maria Fernandez-Suarez and Dave Smith cycled and Ernesto Artaza (Senior and Junior) drove the support van!

The scientists came up with this new challenge in a bid to raise awareness of these devastating conditions and raise funds for two charities that support those affected by Niemann-Pick diseases, Niemann-Pick UK and the Niemann-Pick Research Foundation, as well as funds for research into these condition(s).

The team completed their challenge on the Thursday afternoon, a day before the Interactive Workshop, to cheers from the NPUK Staff Team and other onlookers - there was no squad more deserving of a sit down and a nice cold drink.

Well done everyone! Thank you for your fantastic efforts - those funds raised will make a huge difference!

Do you have Instagram? Make sure you follow the @plattlabchronicles to see the photo updates surrounding this event, and any news regarding future fundraisers - there is talk of a potential walk and canoe challenge next year, so watch this space!

MY STORY...

Dave Smith, Research Assistant at the University of Oxford and intrepid organiser of Platt Lab's fundraising challenges, shares the story of his connection to NPUK:

When did you first hear about Niemann-Pick disease?

I first heard about Niemann-Pick disease type C when we got the NP-C1 null mouse strain in the lab in 2000. The mouse has a severe phenotype.

What is your role within the Niemann-Pick community?

I am a Research Assistant working in Fran Platt's lab at the University of Oxford. I am responsible for all of the mouse studies in the lab. So I see at first-hand what treatments are coming through and which ones look the most promising.

What, in your opinion is the outlook for Niemann-Pick disease research and the potential future benefits for patients?

Hopefully some of the results that we get in the mouse studies will suggest that clinical studies are the next step. However to transfer the results to humans may take quite a while.

You're a familiar face at our Annual Family Conference & Interactive Workshop – what do you think makes it so special for families and health professionals alike?

I do like all the friendliness. Everyone is focused on the same goal: to make life better for the patients.

You took part in a 70 mile bike ride from Oxford to our Conference this year, along with other scientists working in the Niemann-Pick field. What was your motivation?

After going to my first NPUK Annual Family Conference and Interactive Workshop on Niemann-Pick Disease(s), and seeing all of the brave and inspiring people I met, they are all the motivation I needed. If I can suffer for a few days then it is all worth it!

Thank you Dave for sharing your story with us, as ever we commend you and the rest of the Platt Lab for the fantastic work you do year after year!

Peter Carlton Jones Award

This annual award of up to £1,000 is made available to individuals who are engaged in either research, teaching, treatment or care in the field of Niemann-Pick disease, within the public or private sectors in the UK.

Bernardo Barrios Crispi, Cardiff University:

Well done on your Peter Carlton Jones Award win! Can you tell us a little bit more about the project you have been working on?

Firstly, I want to thank everyone at Niemann-Pick UK. The Peter Carlton Jones award means a lot to me and is the first award in my career in the UK. Considering the advances that have been made in this disease, it is an honour to be given this award from this community.

This project is an advance on a previous UK NPUK funded summer studentship, in which the student developed an early drug screening assay for Niemann-Pick. My project took this assay, converted it and confirmed it as a high throughput drug screening system.

The work was a collaboration between our lab and a Spanish company with the aim of identifying new potential medicinal drugs for NPC. We made a few surprising findings along the way, in particular, that monitoring changes in the cholesterol levels in Niemann-Pick cells is not a good way of doing a drug screen for this disease, as you would not identify miglustat. This left us wondering whether other drugs which could be beneficial like miglustat are not being identified because of the cholesterol screening strategy. We devised a new method that measures the size of lysosomes in NPC (they are swollen with fats) and used this for our drug screen. This method has worked very well, allowing us to have identified several new potential NPC medicines which we will be reporting back to the company. Ultimately, we hope that the NPC community will consider our findings and adapt our method.

How does your project seek to help those affected with Niemann-Pick disease?

The long-term benefits of this project are twofold. Firstly, it is possible that the company may take some of the new medicines we have identified forward to the clinic. Second, our findings help other researchers working on drug development on NPC to have an improved research tool to screen for new medicines which in the long term has the potential to lead to clinical benefits for NPC patients.

Did you enjoy this year's NPUK Annual Family Conference & Interactive Workshop? What are the positives of attending, and meeting the NPUK community face-to-face?

I really enjoyed the experience of being part of this year's NPUK conference. This is the second time I have been at this conference. I was inspired last year in the first one I attended which lead me to work on this disease. Having the possibility of sharing with the families and people affected by the disease is what motivated me to continue working hard. In particular, hearing the talk given by the two teenage sisters was inspirational, one of the most moving moments of my weekend. Dr Emyr Lloyd-Evans gave me the opportunity to get involved and I will take this experience into account when deciding my future work as a doctor.

What are your hopes for the future of Niemann-Pick disease research?

I hope the outcomes of various clinical trials in NPC will be successful and that the disease-modifying therapies continue to be developed for everyone affected by this condition. Personally, I aspire to continue working on this disease during my career in the NHS.

Ecem Kaya, The University of Oxford:

Well done on your Peter Carlton Jones Award win! Can you tell us a little bit more about the project you have been working on?

Thank you very much! Within the scope of my PhD project, I am trying to find novel drug-based approaches for treating Niemann-Pick Type C, mainly focusing on Acetyl Leucine.

How does your project seek to help those affected with Niemann-Pick disease?

Acetyl Leucine has already been trialled in observational studies in Niemann Pick Type C patients, so we know that it is improving some aspects of the disease, particularly ataxia. The aim of my project is to use animal and cellular models of NPC to determine which aspects of the disease this drug can improve and investigate its mechanism of action. We are also combining acetyl leucine with known disease modifiers, like Miglustat, to see if they are synergistic when used together in NPC mice.

Did you enjoy this year's NPUK Annual Family Conference & Interactive Workshop? What are the positives of attending, and meeting the NPUK community face-to-face?

It was my third NPUK conference and I learn a lot every time I attend! The conference is a great opportunity to catch up with the most recent developments in Niemann-Pick Disease research and I am always inspired by the amazing NP community. I love to see that it is becoming more and more international every time I attend, we get to meet amazing people and broaden our perspectives at both a professional and personal level.

What are your hopes for the future of Niemann-Pick disease research?

NPC is a challenging disease to treat but the latest developments on gene therapy and effective combination therapy with various pharmaceutical tools look very promising. I am hopeful that rapid developing technologies and scientific advances will make a huge impact on NP treatment in the near future.

How to apply

We are now inviting applicants to submit for the Peter Carlton Jones Memorial Award 2019...if you are involved in a research project which could make an original contribution to the scientific or public understanding of Niemann-Pick disease, it's treatment or cure.

Submissions should be in the form of an abstract outline, on one side of A4 paper (approximately 600 words), which includes clear statements on the work undertaken, methodology, findings, and any principal features in regard to the nature of the project. Submission should be made to:

Toni Mathieson (Chief Executive), by email to: toni@npuk.org

Closing date for submissions: 31st July 2019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CHILDREN & YOUNG PERSON'S Activity Programme

Each and every year we endeavour to put together an exciting Children and Young Person's Activity Programme to ensure that the younger members of our community also enjoy their conference experience.

The Children and Young Persons' Activity Programme runs alongside the main Annual Family Conference agenda, allowing for the younger generation of NPUK to enjoy being together to unwind and have fun, whilst their parents and/or family members take part in the scientific presentations. Every year the programme provides an off-site visit to a local attraction, which makes for an exciting day in which those in attendance can make priceless memories. This year we visited Wicksteed Park...there were bumper cars, amusement park rides, and much much more - not even the bad weather could stop the youth of NPUK having a good time!

NPUK Chief Executive Toni Mathieson commented: "...the Activity Programme enables family members to participate in the main Conference programme with the knowledge that their loved ones are being properly cared for and appropriately entertained by our team of dedicated volunteers - some of whom have been with the Programme for over 15 years, showing just how much they enjoy their role! We hope that everyone enjoyed the weekend, and look forward to seeing you all next year!..."

We received much-needed grant funding from The Hollie Foundation which helped to cover the costs of the Children and Young Persons' Activity Programme. For more information on The Hollie Foundation, who also provide individual grants to families please visit: www.theholliefoundation.com.

Last but by no means least we want to say a huge thank you to our amazing team of volunteers who return year after year to work with the children and young people attending this event. In doing so they enable the children and young person's parents and carers to take time out to listen to the presentations, attend workshops and enjoy social time together. It's a true team effort and we are so thankful to each and every one of you for your hard work, and tireless dedication.

NPUK YOUTH COUNCIL

The NPYC is specifically for the young people of the NPUK community (aged 11-20), which includes those affected, their siblings, and friends.

The NPYC was formally launched at our Annual Family Conference held in September 2018 and aims to provide an opportunity for young people to feed into the work of NPUK, to share their thoughts about the activities and services we provide now and to contribute to the design and development of our future work. It is also a great way for young people to get involved, to learn about the condition that affects their family and help to raise awareness.

NPUK Project Families Officer, Steve Neal is taking the lead in the development of the NPYC, with support from the wider NPUK team. Steve facilitated a workshop at our Annual Family Conference to discuss the Youth Council and enable young people to tell us what they would like their youth council to look like. Their feedback included the introduction of key roles to help ensure the NPYC can be effectively managed. This will include the creation of the following:

• A communications team to make sure that written and digital information for children, young people and professionals is available and accessible, via social media and other mediums.

• A Chair, who will help to plan and manage meetings (we will take it in turns so that everyone gets the chance to chair a meeting, if they wish!)

• A Secretary, who will help to take the minutes/notes of each meeting so that we don't forget anything.

Why did we introduce the NPUK Youth Council?

- We wanted to ensure that the voices of our young people are heard, that their views, concerns, and ideas are listened to and that they can contribute to the future work of NPUK, including the development of new services that may have an impact on them or their family members.

- To provide an opportunity to raise and campaign on issues that are important to the children and young people within the NPUK community, such as; social aspects, mental health and cyber bullying.

- To ensure professionals (e.g. scientists, doctors, etc.) understand and take notice of the needs and experiences of all children and young people affected by Niemann-Pick diseases.

- We also want to help and support the young people of our community to increase their skills, knowledge and confidence.

We hope to hold regular meetings, possibly three times a year, with additional meetings if the NPYC is working on specific projects, planning special events, or require community input. For further information please get in touch with Steve Neal by either email at: steve@npuk.org or by phone on: 07787 818 885.

CARL MASON

We were happy to welcome director and producer Carl Mason to our Annual Family Conference & Interactive Workshop this year to capture our community in action throughout the jam-packed weekend!

You may have already noticed that in recent years we have been trying to boost our visual presence in the digital arena, with an extra focus on our videos...after all it is through these mediums that we are able to tell the stories of the individuals and families affected by Niemann-Pick disease(s) which are central to everything we do as an organisation, as they are both our reason for being and reason for continuing to fight for further awareness of these rare conditions.

We were fortunate enough to reconnect with director and producer Carl over the Conference weekend. Carl worked tirelessly to create short YouTube style video clips which we will share via social media throughout the year to highlight what it is like to live with Niemann-Pick disease and why care, support, and research are so important!

NPUK first worked with Carl back in 2015 on the IMAGINE short film (more info to the right) which has helped catapult him into a number of exciting and interesting projects over the years since.

Subscribe to our YouTube channel @NiemannPickUK.

IMAGINE is a short film which has been hugely influential in raising awareness of Niemann-Pick disease on a global scale.

Supported by Niemann Pick UK, the International Niemann Pick Disease Alliance and the Hope for Hollie Foundation. Maisy Nixon plays the lead role in IMAGINE, which was produced and directed by filmmaker Carl Mason. The ten minute film follows Millie, a young girl who is diagnosed with Niemann Pick Type C and highlights the devastating impact it has on her and her family. The moving short was inspired by Hollie Carter, who was diagnosed with NP-C age 2, and aims to raise awareness of the disease.

The film currently has over 1.5 million views on YouTube... please watch and share to help continue the spread of awareness! Link below:

www.imagineshort.com/

Fundraising FOCUS

Our fundraisers are central to everything that we do here at NPUK, as without voluntary donations and grants we would not be able to continue our work supporting individuals and families affected by Niemann-Pick disease.

Great North Run

We were fortunate to have not just one, but five Niemann-Pick UK runners taking part in the BIGGEST Great North Run EVER this year.

Our fantastic fundraisers included; Jacob Buchanan Mathieson, Lynder Mathieson, Alex Sabrina Dektereff, Stephanie May Wilson, and Katie McGrath - all of whom should be incredibly proud of themselves!

By taking part in in this extremely challenging event, they have not only raised vital funds which will enable us to continue to support those individuals and families affected by Niemann-Pick disease, they have also raised awareness on a huge platform as local and international press have a heavy presence throughout the day.

Elaine Fisher - Fun Day Stand

Massive well done to Elaine Fisher and friends who made the most of a sunny day and raised a fantastic amount in support of Niemann-Pick UK.. Speaking on the event, Elaine commented:

"...what an amazing day - England playing took away some of the customers but we still raised an amazing £685.00 at today's fun day stall with money still coming in. Thank you to everyone who came and everyone who donated, you're all wonderful..."

Elaine's daughter, Holly, has Niemann-Pick disease type C, and was the motivation behind this fundraiser...friends, family members, and people interested in supporting NPUK banded together in her honour and raised funds in the process. Just look at this squad - so inspirational!

Jane Owen and Paula Perkins working hard at the crafts and tombola tables at this year's Annual Family Conference

BECOME AN NPUK VOLUNTEER

Want to become part of the amazing NPUK volunteer team? As part of the dedicated group you could:

- Make a difference to those affected by Niemann-Pick
- Help raise vital funds and awareness
- Make new friends
- Gain new skills
- Have fun!

Make 2018 your year by getting involved: email us at info@npuk.org for further details

Chris and Michael Charman, along with Darren Kok and Dan Bracknall took on the challenge of the Prudential Ride London this year...and absolutely smashed it! Dressed as the Teenage Mutant Ninja Turtles - they were always sure to turn heads, but when it comes to raising funds and awareness of Niemann-Pick disease(s), that's exactly what we want!

The fundraising total for our very own Teenage Mutant Ninja Turtles is currently sitting at over £1,500...an incredible amount that the group should be very proud of. We want to say a huge thank you for Chris, Michael, Darren, and Dan's dedication - you really are superheroes!

Despicable Us: Minions Cycle

Simon and Chris, also known as the fundraising duo 'Despicable Us', smashed their 300 mile cycling challenge and have raised well over their original target in the process...so much so that they were in the top 1% of JustGiving's August fundraisers - that's out of 23,190 people - unbelievable! To date the dynamic duo have raise over £5,000 for NPUK. Simon commented:

"...we completed the challenge pretty much in one piece. As did our fantastic tandem complete with Minion Trev on the back. We were sore and tired but your unrelenting support means we've far exceeded our target. We simply couldn't have done it without you..."

Thank you guys, we feel very fortunate to have such dedicated fundraisers!

Ben Nevis Climb

Peter Jones, along with his sister Nicola Murrow and other family members and friends, climbed Ben Nevis in July in memory of their dear friend Sally Hibbert who sadly passed away from NP-C.

Following donations from the good people at Calor Gas Saxham Depot, and funds raised as a result of the event, a grand total of £500 was achieved - such an amazing way to keep Sally's memory alive!

Nick Vakis-Lowe

You will be hard pushed to find a more dedicated fundraiser than Nick Vakis-Lowe, who ran six marathons in six months this year...raising over £1,000 in the process! Nick took on this fundraising challenge as part of the Harvey and The Brave Little Soldiers, a fundraising super team co-founded by NPUK Volunteer Jenny Charman in honour of her nephew, Harvey, who has Niemann-Pick disease type C (more information on pages 26-27).

Nick's son, Corey (age 9), also chose to do something amazing this year by asking for donations to NPUK in lieu of gifts for his birthday...and raised an awesome £100. Thank you Corey, your kindness is an inspiration to us all!

Charity Acoustic Night

Jenny Charman and the Harvey and the Brave Little Soldiers team organised a night of fun entertainment with fantastic musical talent at the Flapper, Birmingham. Many thanks to the featured artists; Ellie Jones, Scribble Victory, Aesthetics, Hadleigh Ford, and Marc Halls as well all of those who worked behind the scenes to help make it happen!

Along with a later quiz night, the grand total of both fundraisers came to approximately £600 - well done!

In Awe of Helen Beveridge

In an absolutely staggering feat of endurance Helen Beveridge swam the entire 25 mile length of Loch Awe, the longest loch in Scotland, to both raise awareness of Niemann-Pick disease type C and funds for Niemann-Pick UK...a challenge that lasted over 23 hours (wow!). At the time of writing Helen's JustGiving page has raised a grand total of £4,380...almost double her original target!

March for Matthew

Rebekah Louise Cayzer was inspired to take on a five mile march in honour of her cousin Matthew who has NP-C. After seeing the struggles that he goes through, but keeps on fighting with a smile on his face, she was inspired to raise funds and awareness for NPUK, the charity which supports him and his family.

Rebekah has raised £50, with her JustGiving page still open for those wishing to add to her total. Get in touch with John, our Communications Officer, to find out more information on how to donate towards this fundraiser by email at: john@npuk.org.

Belfast Tough Mudder

Just look at the dedication of these two fab fundraisers - before and after the Belfast Tough Mudder and smiling all the way!

We want to say a huge thank you to Alana and Karen who took on the challenge of the infamous Tough Mudder challenge with the aim to raise as much funds for NPUK as possible whilst pushing themselves to their physical limits! Well, they came through on top and managed to raise a eye-watering £1,450. Don't worry about the t-shirts girls, you can keep 'em!

Jenny Fisher/ Chloe Bellis Foam Run

Jenny Fisher and Chloe Bellis absolutely smashed the Birmingham Gung-Ho tackling the inflatable obstacles and bubbles with a smile on their faces! The duo raised £75, which will go towards helping support those affected by Niemann-Pick and their families.

The Gung-Ho event certainly looks like a lot of fun - 5km of foam and we are sure, laughter, and all for a good cause too! We are sure that many of our community will want to have a try of this...to find out when the next Gung-Ho event near you is happening visit: www.begung-ho.co.uk.

Fairhurst Accountants

We want to say show our appreciation to everyone from Fairhurst Accountants for taking part in their charity walk, and event which got the whole gang together and helped to raise almost £1,500 in the process!

The funds raised will be equally split between two very deserving organisations, The Brain Tumour Charity and Niemann-Pick UK.

Tom's Red Dead Redemption

In line with the release of Red Dead Redemption 2 Tom Marwood sat down to do what he loves best - gaming! But there was a deeper reason to this event as Tom was hoping to raise funds for Niemann-Pick UK in memory of his friend's mother who had been supported by the charity in the past. Tom managed to raise almost £700 and counting...brilliant!

13th Annual Niemann-Pick Charity GOLF Day

On a sunny June day, the 13th Annual Niemann-Pick Charity Golf Day took place, bringing together supporting businesses for a day of fun that raises vital funds in the process.

The much anticipated event organised yet again by Craig Mathieson, took place at the beautiful surroundings of the Ramside Hall Golf Club. It is held every year in memory of Craig's niece Lucy Mathieson, who sadly lost her fight with Niemann-Pick type C in 2007...this serves as the motivation to both raise funds for Niemann-Pick UK and awareness of Niemann-Pick disease(s) to the wider public.

The NPUK Central Office Team were on hand to help set up the day, and then after a shotgun start the fun commenced! There were many donated prizes up for grabs, including wine, free golf day passes, health spa passes and much much more...we hope that everyone enjoyed their day, and we look forward to the next one!

So many individuals and companies came together to help make this day one of the best ones yet, but special thanks must of course first go to Craig Mathieson, as well as Ramside Hall Golf Club, Aztec Colour Print, and sponsors Pulman SEAT, Pulman Volkswagen, and Pulman ŠKODA for their kind support and continued help with funding.

Erith

ENABLING THE CONSTRUCTION INDUSTRY FOR 50 YEARS

We have been absolutely staggered by the donations made by Erith Contractors and the Institute of Demolition Engineers, which currently sit at a whopping £165,000.

Back in June Erith Contractors held an unforgettable fundraising evening entitled "Nancy's Night Out"...which was a charity event held in support of Niemann-Pick UK. Well...the event went ahead as planned and was an incredible success! NPUK Chair William Evans and NPUK Co-opted Trustee Fran Platt were pleased to be in attendance, and graciously accepted the tremendous donations that were made.

David Darsey (pictured on the right), Institute of Demolition Engineers (IDE) President, had this to say regarding the motivation behind setting up what is sure to be a fantastic black tie event. A story which is sure to resonate with the NPUK community:

"...my family and I were delighted to welcome my granddaughter Nancy into the world on 4th November 2016. However, in the days after her birth it became clear that Nancy was suffering with underlying health problems.

After numerous test and consultations, our family was given the heart-breaking news that Nancy had Niemann-Pick Type C.

They are, however, a small charity whose true strength comes from the dedication of families, benefactors and volunteers. [---] We want to shine a light on this rare and ruinous disease and raise support for the efforts of NPUK in helping Nancy and others like her..."

Funds from both this event and other connected donations have led to a huge overall fundraising total. We want to say a massive thank you to David Darsey, Erith Contractors and the Institute of Demolition Engineers - these funds will make ahuge difference to the individuals and families affected by Niemann-Pick disease(s) in our community, as they will allow us to continue our support service, push for greater awareness, and further research into this condition.

BIRTHDAYS

ON FACEBOOK

We have been blown away by the kindness of our wider community who have jumped on the new Facebook donate feature with typical NPUK enthusiasm!

A recent small, but hugely useful Facebook feature, is doing a lot of good as it allows users to request donations to support Niemann-Pick UK in lieu of gifts. This has equated to a surge in donations as our community members grasp the new opportunity with both hands and encourage their friends and family to give what they can for their special day!

CEO Mark Zuckerberg wrote in a post that birthday fundraisers, despite the fact the service only launched last year, have helped raise over $300 million for more than 750,000 non-profit organisations. He said those charitable groups range from food banks, to animal shelters, to Alzheimer's research.

This isn't the only fundraising option Facebook users have, you can also set up custom fundraisers for any occasion, you may have found that when you type @NiemannPickUK, an option for a donate button pops up...allowing you to turn a status or photo update into an opportunity to raise vital funds for our charity. An example of this in action is provided by this year's Annual Family Conference & Interactive Workshop photographer, Charlie Müller, who posted about the positive experience of his first Conference, added a donate button...and raised £110 in the process!

We want to say a massive thank you to each and every birthday boy or birthday girl who has chosen to support NPUK on the build up to their special day! At the time of writing you have helped raised a grand total of almost £4,000...incredible!

If it's coming up to your birthday and you would like to get involved with an NPUK fundraiser, but are not sure where to get started...then we can help you! Simply get in touch with John, our Communications Officer, by email at: john@npuk.org or by phone on: 07984366334 and he will be happy to help you.

BBC Children in Need

This year is geared up to be another huge year for NPUK alongside BBC Children in Need, as we push for further awareness of Niemann-Pick disease(s) and the people affected by these conditions.

Last year we were fortunate enough to be represented by not just one, but two NPUK families, thanks to the Hitchens family sharing their personal story as part of "Pudsey Rocks" and Shona Beveridge participating in The One Show's Rickshaw Challenge. This year the forever inspiring Josh Cullip (age 12, NP-C) is acting as an ambassador for both NPUK and Niemann-Pick disease as he takes on the Countryfile Ramble.

Josh has Niemann-Pick Type C, a rare degenerative life-limiting disease which causes symptoms similar to dementia and affects around 500 people worldwide. Josh's condition has meant that he's had to re-learn how to do basic tasks like tying shoelaces; he sometimes struggles with his balance and can get tired quite easily. This is why it was so inspiring to see Josh, head out to the Isle of Skye with his parents to take on one of Countryfile's toughest ever rambles alongside presenter Matt Baker and NPUK Clinical Nurse Specialist Laura Bell...simply put, he's an absolute fighter!

Josh is supported by Laura and Niemann-Pick UK which is assisted by a 3 year BBC Children in Need grant which funds Laura's position supporting children and young people affected by the disease. Laura's work helps to build children and young people's emotional resilience and confidence, as well as their ability to cope with the physical symptoms of the disease.

Josh loves being outside so was really looking forward to his ramble, it was truly a dream come true - made even better, when Children in Need veteran Shona (pictured on the right with Laura) madea special trip to the airport to wish him all the best! Throughout the ramble, as was clear from the final programme, Josh kept up his spirits even when his body began to tire; he sang songs and cracked jokes, spurred on by inspirational messages from superstar Ed Sheeran and WWE wrestlers Dean Ambrose, Roman Reigns, and Seth Rollins - he made it to the top, he made it to "Neverland"!

Since Josh's feature on the Countryfile episodes there has been an outpouring of positive responses on social media, as well as an increased number of people getting in touch with NPUK to either ask for more information on Niemann-Pick disease(s) or make donations...this is all down to the awareness raised by one very special boy and his doting family!

How does BBC Children in Need help NPUK?

BBC Children in Need is very important to us here at NPUK; their generous grant funding programme has provided part-funding for our Clinical Nurse Specialist since 1999. This has enabled us to continue providing much-needed support to children and families affected by Niemann-Pick diseases – support that is not available elsewhere and would not exist without the kindness and generosity of all those donating to the BBC Children in Need Appeal each year.

Our superstar Josh, turns 12!

October 28th was a special day for two reasons, firstly it was the day that we all tuned in to watch Josh complete his ramble with Matt Baker in the Isle of Skye, and secondly because it was the day that he turned 12 years old! His loving mum, Jodie, wrote at the time:

"...well Sunday is a huggeeee day, our Joshua has reached a massive milestone - he will be turning 12 years old!

That's a whole 12 years of being told varying things that didn't make sense. Like when we first got the diagnosis at 8 months old due to him having such severe liver disease...we were were told that there was a high chance that he would not reach 5 years of age. We were told if he did survive he would be severely disabled...to just go away and make memories.

Well we have done exactly that, and although we do have Niemann-Pick disease type C symptoms, but nothing that majorly affects his quality of life at this moment...we are still here making memories and Joshua continues to do us proud.

Although birthdays are a great days, I can't help but think how many more birthdays we will have. I now class Joshua's milestone as a good luck card...another hurdle, another milestone reached. Most parents worry how many presents to get, or what to do for their children's birthdays, but I can't help let NP-C's shadow take over my emotions, which makes me worry if this will be the last birthday we can enjoy.

Joshua shares his birthday with Zayn who also suffered from NP-C, but is sadly now celebrating his birthday in heaven.

Two very special boys..."

We welcome Arthur to the NPUK Family!

We were pleased to welcome baby Arthur to the Niemann-Pick UK family this year.

On the 16th August 2018 the world was made that little bit brighter by the arrival of the beautiful Arthur Buckingham weighing in at 7lb 8oz.

Arthur is pictured here on the left with doting new mother Sherry Buckingham. Sherry has ASMD Niemann-Pick disease type B which makes the happy and healthy delivery of Arthur all the more reason for celebration!

We wish Sherry, Arthur, and the rest of the family all the best - keep us posted with more pics!

We want to help share your story...

Are you an individual or family member affected by Niemann-Pick disease(s)? Would you like to share your story to spread further awareness of Niemann-Pick disease to the wider public? If you are interested on being featured in articles, videos and short films on behalf of NPUK then we want to hear from you!

Remember that by telling your story, you can provide a personal insight into the lives of those affected by Niemann-Pick disease, which can lead those outside of the community to get involved and join the fight for change.

For further details of how to be involved please contact our Communications Officer John by either phone on: 07984366334 or by email at: john@npuk.org.

NIEMANN-PICK TYPE C DISEASE (NP-C) IS A

RARE PROGRESSIVE IRREVERSIBLE CHRONICALLY DEBILITATING LYSOSOMAL STORAGE DISEASE[1–3]

NP-C affects all ages[1]

Incidence of NP-C is 1 in 90,000 live births[4]

Likely an underestimate due to lack of clinical awareness[1]

NP-C takes on average 5 YEARS to diagnose[5]

That's...

43,824 HOURS

...waiting for an answer, watching a loved one get worse

THINK AGAIN THINK NP-C

THINK AGAIN. THINK NP-C aims to support healthcare professionals unfamiliar with NP-C to recognise the key signs and symptoms of NP-C and reduce the time to diagnosis

Individual symptoms are non-specific to the disease[1,3]

If you are a:

Paediatrician

LOOK FOR ATAXIA, DEVELOPMENTAL DELAY, HEPATOSPLENOMEGALY

Paediatric hepatologist/neonatologist

LOOK FOR HEPATO/SPLENOMEGALY, NEONATAL CHOLESTATIC JAUNDICE, NEONATAL LIVER DYSFUNCTION

Adult neurologist/psychiatrist

LOOK FOR COGNITIVE DECLINE, ORGANIC PSYCHOSIS, PROGRESSIVE ATAXIA

Have you checked for eye movement abnormalities?

Vertical supranuclear gaze palsy (VSGP) is present in virtually all patients[1,3]

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To help reduce the time to diagnosis visit www.think-npc.com today

THINK AGAIN THINK NP-C

This is a project co-ordinated by the International Niemann-Pick Disease Alliance with the collaboration and financial support of Actelion Pharmaceuticals Ltd

PROGRESS TOGETHER
Inpda
International Niemann-Pick Disease Alliance

SAME BUT DIFFERENT

As part of Same but Different's Rare Project, which hopes to introduce more people to the individuals behind rare conditions, Niemann-Pick UK Chief Executive Toni Mathieson was asked to tell her story from the first diagnosis of her daughter Lucy, to present day:

"I first heard of Niemann-Pick disease when my daughter, Lucy, was about 5 weeks old. She had been unwell from birth as she had a very large tummy due to an enlarged liver and spleen. She underwent a lot of tests and we were told she may have Niemann-Pick disease. I went away, and at that point we didn't have a computer, so I asked my friend to look it up and find out what it was. She brought reams of documents and struggled to tell us how devastating it was. We looked at our gorgeous little baby and just could not believe that she was so unwell. Of course, the blood tests came back, and it was confirmed that she did in fact have Niemann-Pick disease Type C. It took a long time to get used to the diagnosis and to even be able to say those words.

When we were given the initial diagnosis we were given the wrong information as he said she had Type A, which is a lot more severe. He said that we had 6 months, if that, with our child, and the words he used were, you can take her home to die. He also said some other things that will always stay with me forever. I would love to revisit that appointment with him and have the strength to tell him how much an impact his words had upon us. He destroyed us completely within that appointment. Delivering news like that is very, very difficult but I feel that there must be more sensitive ways.

However you deliver news like this it will be difficult to hear. I think it is important to consider how much information to give at the point of diagnosis. I don't think parents can take it in. They will hear the words you utter, that is, they have this condition and they will hear no more. At subsequent appointments, and with support from patient groups, is perhaps a better time to discuss more detailed information. I definitely think that when you're speaking to parents for the first time and delivering awful news that they must have somebody there with them, who can support them, and who can listen for them.

We spent a lot of time crying after the diagnosis. I had this tiny little bundle just here, and I think, in the past if something was broken you would just put it down and get a new one but you can't with a baby. It's broken, it's yours and you've got these feelings of I want to fight for her for everything.

Telling people was really difficult because we had this diagnosis of a progressive neurological condition. We didn't know when the symptoms would happen or how long Lucy would be with us. When they see your child start to grow normally and look normal and act normal then they forget. They forget that you as a mother are going through, what I would call a grieving process, because from the minute you're told your child is not going to live a full life, you start to grieve for the all the things they won't be able to do or the experiences that you won't have together. So whilst my friends were thinking ahead to university and marriage and all these lovely things, I knew they were never going to happen for Lucy and that was so very difficult to cope with and deal with. I would be out and have a lovely day with friends and their children and then go home and break my heart. That I suppose comes with the isolation of a rare disease and that's why I think having a community knowing other mums who are affected or whose children are affected by rare diseases is so important

For me, being involved in the patient group Niemann-Pick UK has been an absolute life saver. Honestly, I've said this before that I don't think I would be sitting here and being able to speak as coherently as I can without them. That community has supported me throughout and been there for me and helped me in so many ways, not just emotional and practical support but the fact that I can feel as if I've actually making a difference on behalf of my children

Lucy was born in 2003 and we quickly became pregnant again. We thought everything would be lovely and we would have another child, and we did, but unfortunately that child was also affected. Lucy's sister, Hannah, and she was born in January 2004. It was a devastating time when she passed away. We had so much going on in our lives anyway and I really wasn't very easy to live with at that point in time. My husband was the one who picked up the pieces and brought me back to reality. That and the fact I had to care for Lucy of course, because I had to get out of bed everyday to look after her and to be there to make those memories and I just had to get on with life.

We thought we would maybe try once more. This time we had a lovely little boy, Sam, and again, unfortunately, he had Niemann-Pick disease too and we lost him. It was at that point I think that I went really off the rails and needed a lot of help and support from my family and friends. Again my husband really pulled me through. It was the year after Sam passed away that I became more involved in Niemann-Pick UK and took on a role with them and that gave me a sense of purpose. I knew that sooner or later I was going to lose Lucy too and I needed to have something after that where I could make a difference.

Hannah and Sam passed away in 2004 and Lucy in 2007. I also lost my father in 2004 so I lost my father and two children. It was an incredibly difficult time.

If I was to speak directly to the science community the one thing I would say is, if you can, put yourself in the shoes of a mum or a dad with a child with a rare condition. Think how helpless you would feel if there was nothing that could help that child and that you were going to lose that child. The difference that you could make to families by finding something that could slow the progression of the disease long enough for something else to be found that can help a bit more is immeasurable. Patients need time with their families, parents need time with their children and starting at a very basic research level right through to patients, bringing the bench to the bedside is exactly what we need

To a family who'd just been given a diagnosis I would say take your time, listen to your own feelings and when you're ready, if you're ready, reach out to other patients, via a patient group or via your GP or treating consultant. Try and understand the experiences of others and learn from those and understand that usually, even though you're a rare disease patient, you aren't alone. The things that you are going through and feeling are very normal and someone out there will have been through them before you and will be able to help you

Following our experiences with our children I became involved with Niemann-Pick UK back in 2005 and I now have the position of Chief Executive with that organisation. I help to oversee the strategic and operational activities and we do so much now to support patients including supplying a clinical nurse specialist who works with paediatric and adult patients. She's based in Manchester but she travels across the UK to wherever a patient is. At any one time if they need a home visit or somebody to accompany them to the clinic appointments, Laura can go along and sit with them and help them to understand what they're being told about their condition, help them afterwards if they have any questions and make sure that they can access their local care and services that they need. We don't just look at the patient that is affected by the condition, we help their wider family too. There's lots of difference that we can make and I hope that being part of this community can help families to feel less isolated and alone with this rare condition

I didn't want anyone's pity and I still feel I don't need it, even though yes I get that reaction, if someone says to me have you got children, and then I tell them our story, I can see the pity in their eyes. I don't want that, I want them to listen and understand that I am very grateful for the experiences I've had and the time that we had with our children. It's very special to me and I feel very lucky. I know that sounds ridiculous, but I do, I feel really lucky and I want people, if I talk to them about my experiences to learn from me and pass it on and raise awareness and support and help, and that's what I want from them and not their pity."

Same but Different use the arts to create **greater awareness** of the person behind **rare disease** and disability. Recently the organisation released short video clips via. their YouTube channel which feature NPUK Chief Executive **Toni Mathieson** speaking on a variety of topics ranging from the importance of research to rare disease communities to the importance of support when receiving a rare disease diagnosis. You can watch these videos on the YouTube channel: **@samebutdifferentcic**.

For more information on Same But Different, including more content such as the above please visit: www.samebutdifferentcic.org.uk.

DEMENTIA STRIKES CHILDREN TOO

We are happy to announce that plans are underway for the continuation of the Dementia Strikes Children Too campaign. The community-driven campaign, which is led by a collaborative team of three patient organisations, Niemann-Pick UK, Batten's Disease Family Association, and the Society for Mucopolysaccharide Diseases, with support from biopharmaceutical company, BioMarin, will seek to raise awareness, advance understanding and improve care to those affected individuals and families.

Last year we flooded Westminster Tube Station with Dementia Strikes Children Too posters to raise awareness...what will we do next? *Stay tuned!*

RARE REVOLUTION MAGAZINE

Rare Revolution Magazine is a new digital magazine which seeks to give a voice to patients affected by rare conditions whilst supporting the charities that represent and support them.

We were fortunate enough to take part in a Rare Revolution Magazine promotion this year which allowed us to take over their social media channels for a full 24 hours...appropriately named the #tuesdaytakeover, it served as a great collaboration between two organisations focused on raising awareness of rare diseases.

Throughout our #tuesdaytakeover we created unique content such as fact file graphics on ASMD Niemann-Pick disease types A, and B, as well as Niemann-Pick disease type C for use on the Rare Revolution Magazine social media pages, and also broadcast the extensive work of NPUK and the support that we can offer individuals and families affected by NPD.

Huge thank you to the great people at Rare Revolution Magazine for the #tuesday takeover opportunity - it was such fun and allowed us to get our important message out to a wider group than ordinarily possible!

Make sure you give them a 'like' on their Facebook page so as to never miss an update - for more information please visit:

www.rarerevolutionmagazine.com

PROGRESS TOGETHER

Inpda

International Niemann-Pick Disease Alliance

Who is the International Niemann-Pick Disease Alliance (INPDA)?

The International Niemann-Pick Disease Alliance (INPDA) is a global network of non-profit organisations working in the field of Niemann-Pick disease (NPD). Founded in 2009, through the collaborative efforts of Niemann-Pick UK and the National Niemann-Pick Disease Foundation (USA), the INPDA now has 18 member groups in 15 countries; Argentina, Australia, Brazil, Canada, China, France, Germany, Italy, The Netherlands, Norway, Spain, Switzerland, Taiwan, United Kingdom, USA.INPDA members work together, overcoming language and cultural differences to facilitate progress in research and improve health and social care outcomes for all those affected by Niemann-Pick diseases.

INPDA Information Portal

The INPDA Information Portal is a shared space that offers access to information and resources relating to Niemann-Pick disease. It is a true reflection of the culture present in the INPDA, bringing together a wide range of helpful information for patients, their families and the professionals working in this field, in different languages and formats.

You can access the INPDA Information Portal at: www.inpda.org/inpda-information-portal

The INPDA Loire Valley Meeting 2018

This meeting, which takes place every two years, provides an opportunity for health professionals in the research field to discuss fundamental, translational and clinical research in the informal atmosphere of Château des Grotteaux.

It has been set up in line with INPDA's central aims, and seeks to:

- Encourage researchers to meet regularly.
- Share ideas and practice regarding research and clinical trials.

LVM 2018 served as a unique international scientific meeting as it was restricted to discussing fundamental, translational and clinical research in an informal atmosphere.

We want to say a huge thank you to NPSuisse - Schweizerische Niemann Pick Vereinigung and Niemann Pick Selbsthilfegruppe Deutschland who organised and jointly funded this event on behalf of the INPDA.

Huge thanks, of course, to all participating scientists (pictured below) that made this event possible.

PROGRESS TOGETHER

In 2016 the International Niemann-Pick Disease Alliance (INPDA) successfully completed a three year EU funded project to develop a patient registry for Niemann Pick Diseases.

The resulting International Niemann-Pick Disease Registry (INPDR) is a disease-specific registry is now owned and managed by the INPDA with support from professional partners involved in the care of patients affected by all types of Niemann-Pick disease (ASMD and NP-C).

The INPDR encourages global collaboration between patient groups, clinicians, scientists and researchers, all of whom wish to improve care and treatment options for Niemann-Pick patients everywhere. As well as influencing patient care, the INPDR will encourage efficient and timely diagnosis, improve understanding of disease progression and support global research efforts.

2018 has been a year of significant progress for the INPDR as work commences to transfer the registry its new home with OpenApp, a well-established, Dublin based, IT/Health Informatics Company, who have helped to develop and support a number of similar registries in the last few years, including for Cystic Fibrosis and Tay Sachs.

In tandem, the INPDR continues to actively recruit patients, with just over 300 currently enrolled in 7 countries. The INPDR will be the most valuable asset we have as a patient community and your help is needed to achieve a robust and complete resource. The active participation of patients from around the world will provide valuable natural history data on this rare disease, facilitating the development and recruitment of future clinical studies and supporting access to new and emerging therapies. .You can learn more about the INPDR and how to participate by visiting www.inpda.org

In a number of countries it is now possible for patients to participate in both the Clinical Registry and the Patient Reported Registry (where patients can sign up themselves), a pilot study is currently underway in the USA that will soon enable patients to join the Clinical Registry. The pilot will commence at the Mayo Clinic, and the INPDR team is working in close collaboration with our USA based colleagues at the National Niemann-Pick Disease Foundation (NNPDF) to facilitate this.

To keep up to date with news and information about the INPDR, please 'Like' and follow us on Facebook @INPDR to ensure you don't miss out! If you have any questions or if you would like further information regarding the INPDR, please contact us at info@inpda.org

For more information on the INPDR please visit: www.inpdr.org or email: info@inpdr.org

WEAR JEANS CHANGE LIVES

JEANS FOR DAY GENES

Friday September 21st, 2018, was not only special as it marked the first day of this year's NPUK Annual Family Conference & Interactive Workshop, it was also...Jeans for Genes Day!

Life-altering genetic disorders, including Niemann-Pick disease(s) affect approximately half a million children in the UK. Jeans for Genes Day is both a fundraising and awareness-raising day which seeks to shine the light on those affected with these genetic conditions - and help to make a tangible difference.

On Jeans for Genes day, participating schools and workplaces ask students and staff to wear jeans and casual wear to school and work instead of their usual uniforms and formal workwear...it's a fun way to raise some funds and make a statement that you support the fight against genetic disorders!

This is why if you were around Wyboston Lakes, Bedfordshire for this year's Interactive Workshop you may have spotted many of our professional speakers and attendees traded in their smarter trousers or skirts for something with a bit more denim!

Money raised on the day and over the weekend is used in an annual grant programme which seeks to provide funds to support groups and organisations Some of the outcomes of fundraising include holidays for families affected by genetic disorders, provision of communication equipment, sensory equipment, support groups, support websites, mobility aids, adaptations, education and training, and much more.

There is a dedicated website for Jeans for Jeans Day, where you can find out more. Donations can be made on the website all year round, and you can even begin to arrange your event for next year to get ahead on the planning! Please visit: www.jeansforgenesday.org

GLOBAL NIEMANN-PICK DISEASE AWARENESS MONTH

Ebony Jamie Samuda wrote this piece for Global Niemann-Pick Disease Awareness Month, which explores the story of her son Marii's battle with Niemann-Pick disease type C...

"...Marii was diagnosed with Niemann-Pick Disease type C at the age of four months old as he showed signs of an enlarged liver and spleen from birth.

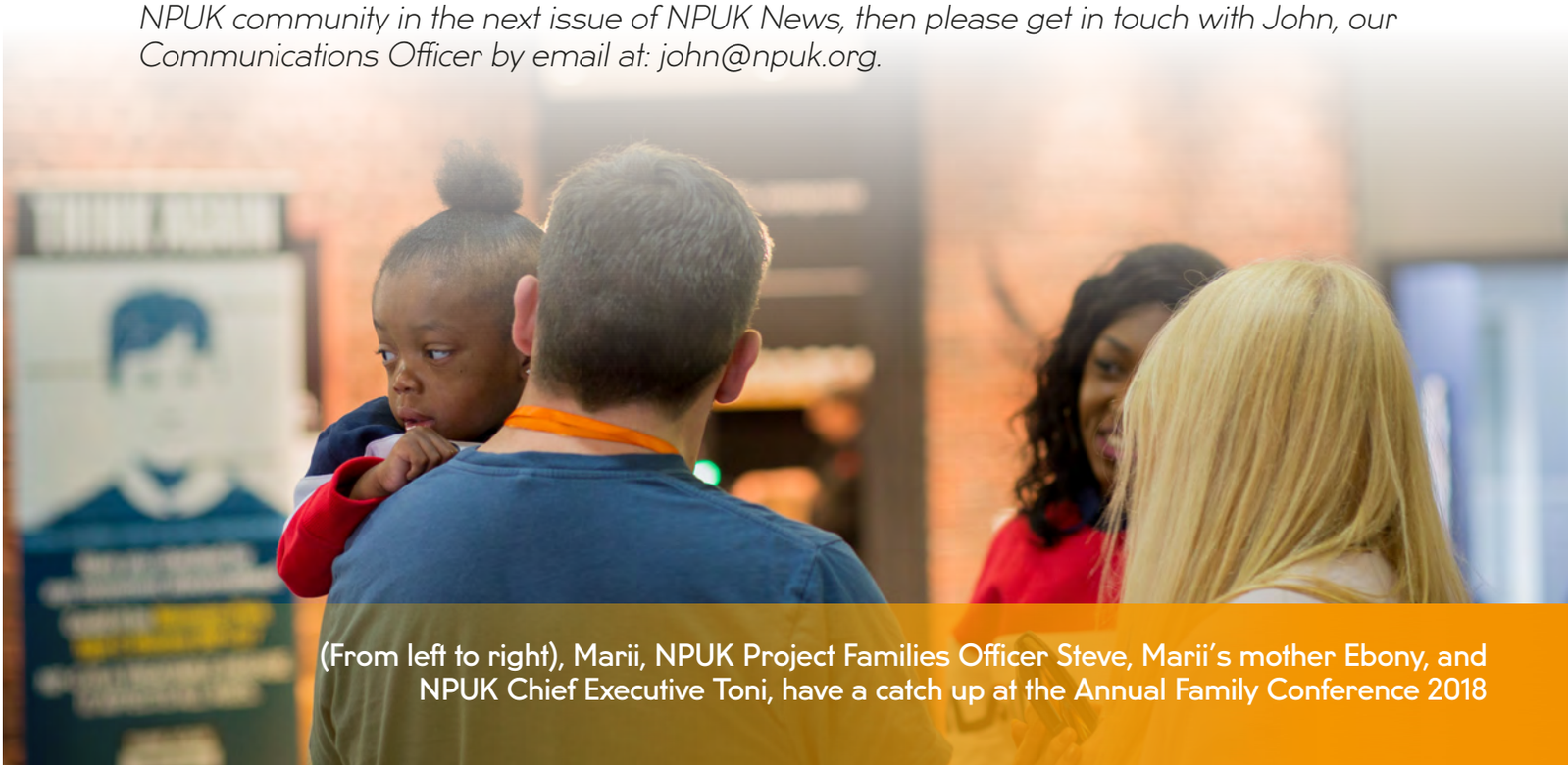
He is now almost four years old and has global developmental delay and difficulty walking but has made some improvements in those areas over the last few weeks. Although Marii has to go through so much he keeps smiling , laughing and just being happy. He continues to fight everyday and never gives up until he's made himself able to do something.

We are currently under the care at GOSH (Great Ormond Street Hospital) and Evelina London Children's Hospital (as Evelina is where our journey started, and Marii has a great relationship with Dr Vera and his nurse Emma). Both places offer great care and have been hugely helpful so far in our journey.

We have managed to come across some lovely people when meeting other parents, carers, etc. along the way. With having a child with a rare condition it's nice to know we are not alone on this journey.

Keep the fight up my boy, Mummy is so proud of you and how far you have come so far. The battle is far from over, as long as there is hope we will fight together hand in hand, side by side..."

We thank Ebony for sharing her story with us - if you would like to share your story with the NPUK community in the next issue of NPUK News, then please get in touch with John, our Communications Officer by email at: john@npuk.org.



(From left to right), Marii, NPUK Project Families Officer Steve, Marii's mother Ebony, and NPUK Chief Executive Toni, have a catch up at the Annual Family Conference 2018

Help & INFO

Our Senior Families Advocate Elizabeth Davenport has hand-picked the following help and information from Contact - we hope you and your family find it both informative and useful!

* ***Tips on handling absences from school:***

If you are a parent and/or carer of a child affected by Niemann-Pick disease there will inevitably come a time when you have to learn to handle absences from school, for one reason or another. The following information is written with reference to the law in England, however general principles apply to all UK nations.

Absence for medical reasons:

All parents have a duty to make sure their child attends school regularly. Of course it is only natural that *all* children are likely to have short absences from school when they are under the weather, but children with additional needs, such as those with Niemann-Pick disease, are far more likely to have lengthy or repeated periods off school.

Lengthy absences could be for a variety of reasons, such as surgery, clinical trials or other treatments. Whereas short, repeated absences could be due to having frequent hospital appointments or because of intermittent crises between periods of being reasonably well.

It is important to keep closely in touch with the school about your child's health. You should inform them in advance of any medical appointments or planned treatment.

Your child might not be well enough to do much work, but continuity and maintaining a sense of normality is a positive thing and should be the goal!

For short absences, your child's school should keep in touch and send work home if they are off for more than a day or two. Some schools have a bank of activities available to download from their website. Others may provide some outreach support, for instance the child's support assistant coming and doing some work with them at home. While your child is off, the school should help them keep in touch with classmates, for example by writing, over the phone, through photos or class newsletters, over skype, and so on.

Please remember, you cannot be prosecuted or fined if your child off school with a genuine medical absence

If you are being fined or threatened with prosecution, please call: 0808 808 3555.

Universal Credit - *explained*

Universal Credit is gradually replacing Employment and Support Allowance (ESA) as the main benefit to claim if you can't work because of sickness or disability - this would affect those with Niemann-Pick disease. When you make a claim for Universal Credit, you may be asked to attend a Work Capability Assessment. This is designed to assess how your disability affects your ability to work. We are happy to guide you through the process!

For further information please contact Elizabeth at: elizabeth@npuk.org

Time Off Work for Parents or Carers of Disabled Children

The law is complicated. Below is only a general outline to some typical employee rights regarding time off work for family responsibilities, such as in emergency circumstances and when attending hospital visits and clinic days.

Parents of children under 18 can take unpaid parental leave if they have been with their employer at least a year. Unless you have a different agreement with your employer, you can be expected to give 21 days notice and be limited to four weeks a year. Parents can take up to a total of 18 weeks altogether for each of their children before the child is 18.

Parents taking parental leave for a disabled child (who is entitled to Disability Living Allowance or Personal Independence Payment) can take it in blocks of a day.

Your employer cannot penalise you for taking parental leave. There are circumstances in which employers can postpone parental leave. If you need further information on this please get in touch with use here at NPUK on our 24/7 helpline at: 0191 415 06 93.

Emergency Leave for Dependants:

Employees have the right to take unpaid time off work to deal with an unexpected event involving someone who depends on them.

Your employer cannot penalise you for taking time off work during an emergency, however you must inform your employer as soon as physically possible that you require time off and provide an estimate of how long you think that you will be absent. You also need to try to find alternative arrangements (maybe the grandparents can help? Or a childminder?) and keep a record of when you have tried them and why they are not possible solutions that would enable you to work.

There is no official upper limit on how much time off you can have for such an emergency, although it must be reasonable in the circumstances. If you are part of a couple or the other parent is active in the care of the child, the time off that is reasonable in the circumstances would take into account the fact that there is another person to share the care. Normally only one parent would be expected to have the time off at once, however where a child is very ill, has an accident or is having a major operation, it may be reasonable for both parents to have reasonable time off.

The legal right to take time off is normally for an emergency. Knowing in advance that something will happen does not mean that you definitively cannot have the time off for it. E.g. a childminder needing time off next week can count as an 'emergency' if you tried but were unable to make alternative arrangements for your children.

Adapted from www.workingfamilies.org.uk

Finding Support Groups

Support groups are fantastic places to get practical and emotional advice from other parents with children affected by Niemann-Pick disease(s) and other life-limiting conditions.

There are groups which are for specific conditions such as Niemann-Pick UK and other connected charities, however for some, general support groups can also be helpful to give the opportunity to talk through tough times.

As a starting point, the people listed below can often give you a helping hand in finding out what's on offer in your local area. They include:

- Local authorities must publish information about the support that's available in their area. This is called a 'local offer'. The quality of information varies, but there should be support groups and contact information.

- NPUK Clinical Nurse Specialist Laura Bell, Senior Families Advocate Elizabeth Davenport and Project Families Officer Steve Neal, may be able to point you in the right direction, when it comes to care and support.

- Netmums.com is a fantastic resource to find out local resources and services in your area.

- Gov.uk is also a decent resource to find local support groups.

Online support groups:

Make the most of all of the online opportunities out there by exploring online communities and forums, many of which will contain individuals going through many of the same struggles you may be battling with. For example:

- Netmums has an area to 'chat about life with special needs and disabilities' Mumsnet has a forum on 'special needs children'.

- Search the National Network of Parent Carer Forums for a local forum on a number of related care issues.

- The charity Contact's online community and parent carer forum list is full of parents talking through familiar problems - which helps you feel less isolated.

Remember that we are always here if you need us, feel free to phone on: 0191 415 06 93 or email: info@npuk.org if you need any form of support.

Adapted from www.scope.org.uk

UKY | AVON TYRRELL

Avon Tyrrell works to make the outdoors inclusive and accessible for all, including those with specialised needs and those who may require a little bit of extra assistance... adventure starts with Avon Tyrrell!

Avon Tyrell are completely committed to being a fully accessible centre and work to offer everyone the same opportunities to experience, learn and develop through adventure and discovery.

If you or a loved one has additional needs due to Niemann-Pick disease, with Avon Tyrrell there is still nothing to hold you back - their professional and friendly staff deliver high quality support in a safe, accessible environment. It is due to their hard work and excellent facilities that *everyone* can enjoy the outdoors and consequently create amazing memories that will remain long after the day of fun is over!

Avon Tryrrell operates in New Forest National Park, Hampshire, close to good transport links to make access to the site easy. The organisation offers:

- A wide range of accessible accommodation options and activity packages to suit every budget

- Additional equipment for use in accommodation and throughout the site

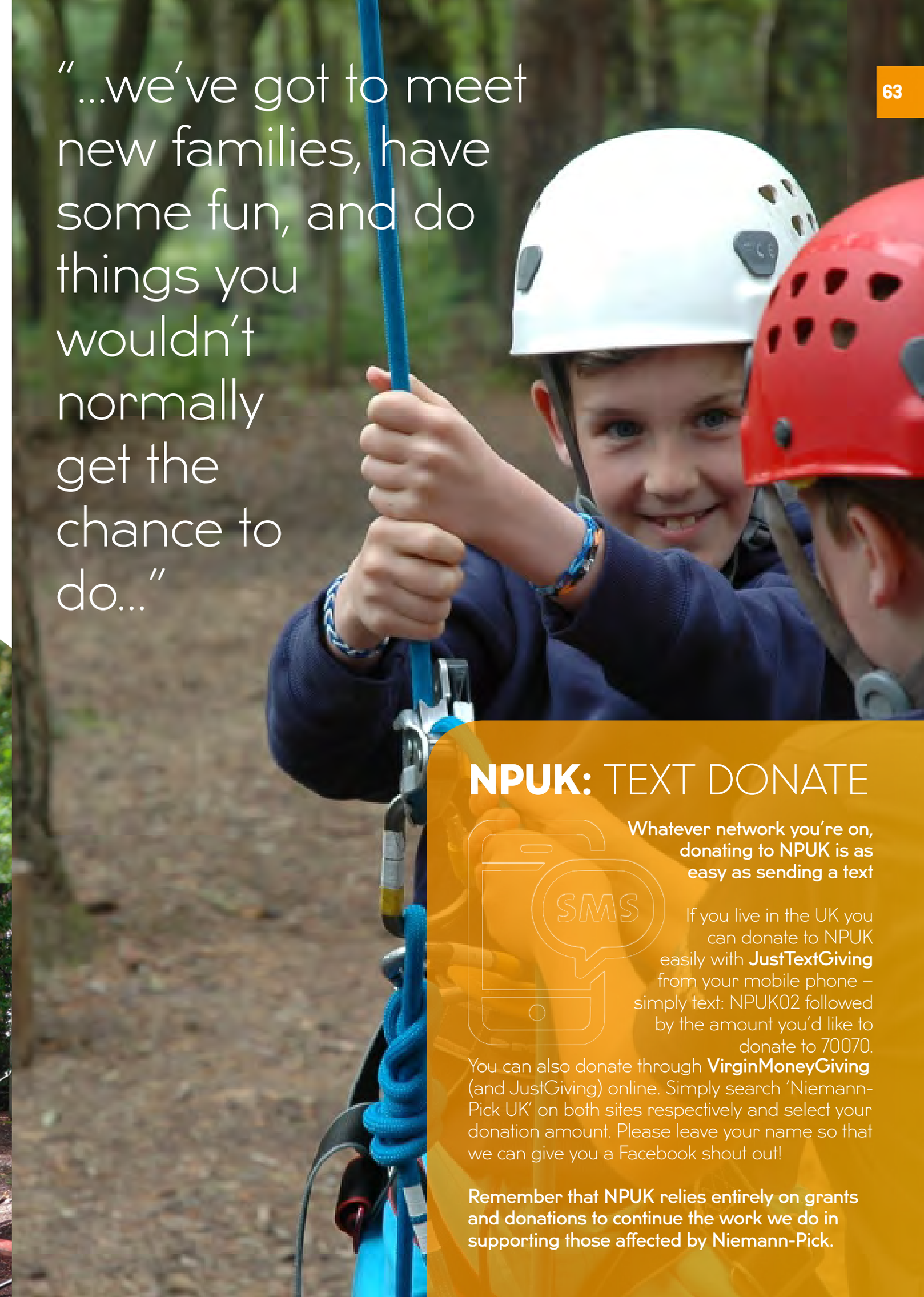
- A variety of exciting adventurous and environmental outdoor activity experiences

- Fully qualified and experienced instructors trained to adapt sessions to meet individual needs

- Family Inclusion events held throughout the year

If you require any additional equipment or have any queries or concerns regarding what Avon Tyrrell can provide and the services they offer, please contact their Guest Services Team on 01425 672347 or email info@ukyouth.org.

You can also find out more information at their dedicated website: www.avontyrrell.org.uk.



"...we've got to meet new families, have some fun, and do things you wouldn't normally get the chance to do..."

NPUK: TEXT DONATE

Whatever network you're on, donating to NPUK is as easy as sending a text

If you live in the UK you can donate to NPUK easily with **JustTextGiving** from your mobile phone – simply text: NPUK02 followed by the amount you'd like to donate to 70070. You can also donate through **VirginMoneyGiving** (and JustGiving) online. Simply search 'Niemann-Pick UK' on both sites respectively and select your donation amount. Please leave your name so that we can give you a Facebook shout out!

Remember that NPUK relies entirely on grants and donations to continue the work we do in supporting those affected by Niemann-Pick.

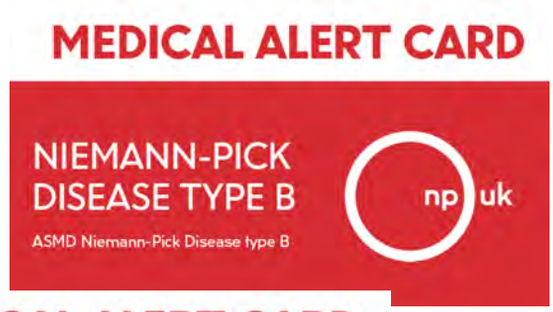
Patient Passports & Alert Cards

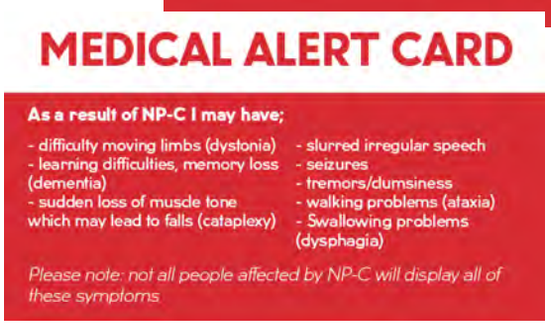
First debuted at this year's NPUK Annual Family & Interactive Workshop on Niemann-Pick Disease(s), we hope our new publications prove to be a huge benefit to our community.

We understand that for those affected by a rare and life-limiting condition such as Niemann-Pick disease, describing symptoms and potential challenges can often be a difficult process...things can be easily misunderstood, or certain symptoms could be forgotten. This is why we created these Patient Passports, in house, with the help and guidance of both our NPUK Care and Support Team and affected individuals and families in our close community.

The Patient Passports (currently available in ASMD Niemann-Pick disease type B and Niemann-Pick disease type C versions) provide an easy-to-follow template upon which those affected by NPD can list their details, contact information, details about their condition, symptoms, and even likes and dislikes. We hope that it can save many a repetitive conversation, and make things clearer for both the patient and connected health professional alike.

As an accompaniment to the Patient Passports we have also drafted Medical Alert Cards which act as first points of information in either emergency situations or when brevity is necessary. Approximately the same size as a credit card, and therefore easy to carry in a pocket, purse, or wallet, these cards provide details regarding Niemann-Pick disease symptoms, related care issues and difficulties, and contact information for both the patient and other individuals/organisations who may be able to provide further assistance.





Laura Bell, NPUK Clinical Nurse Specialist, commented:

"...we had a very positive response regarding both the Patient Passports and Alert Cards, when we introduced them to our community at this year's Annual Family Conference. We hope that they will prove to be effective tools in the future. Anyone who wants a copy please don't hesitate to get in touch! ..."

For further information on both of these publications, and for details on how to get your own copies please contact us by email at: info@npuk.org.

Gaz Anderson

COUNSELLOR / LIFE COACH / PUBLIC SPEAKER

We were pleased to welcome experienced counsellor and life coach Gaz Anderson to our Conference this year alongside NPUK Project Families Officer Steve Neal, and NPUK Volunteer Tony Somers.

Here's a little Q&A we had with him recently to get an insight into what led him into this role:

1. What's your background, and how did you get into counselling?

I didn't have the best start in life... struggled with mental health from as far back as I can remember, although I didn't realise that at the time. Was always in trouble as a kid and in young adulthood.

Counselling didn't help me and was referred to a clinical psychologist, after which two of my daughters died whilst I was still having treatment. I spent time in Libya, Uganda, and Afghanistan before returning home and studying counselling at college.

I have worked as a therapist since but not your stereotypical therapist! I get people on bikes, walks, on the focus pads, etc. My programme, Safeguarding Me, is now been rolled out in mainstream schools.

2. People can sometimes be apprehensive about receiving counselling – what would you say to these people?

I totally understand why people would be apprehensive about going into therapy as it took years for me dealing with the shame that I felt weak. But when I did go through the doors and to me this was the hardest battle of all for me looking inside myself, but I'm here now and I know if I hadn't walked through the psychologists door I would be dead, so it saved my life. I would say give it ago it's not for everyone but it could change your life for the better

3. You're working alongside Steve and Tony with Lads, Dads, and Carers – what are the advantages of such a support network?

I understand dads not talking and could talk forever about this! But I used to say there is no right/wrong way of grieving but there it *is* wrong to not grieve at all, like I did. Lads, Dads, and Carers is a good outlet to talk, and there's company to keep you safe. Having people who have had similar experiences, and therefore can relate to each other works wonders as it's easier to talk to someone who has been through the same things. I hold Steve and Tony in the highest regard, both amazing people.

4. What was your first perception of NPUK and our community at Conference?

One word, humbling! It's inspiring to see how people with similar experiences pull together as an organisation. When my children died I had no support, so to see the great support you all give each other is like I say, humbling. Despite the link being a connection you wouldn't wish for, if that makes sense, they are amazing people and there was plenty of love.

5. What have you been working on recently?

I am writing a book called Daddy and the Two Bears which is based on my journey as a father. It wasn't until last year when my wife was diagnosed with cancer that everything came to a head and I realised I hadn't grieved for my daughters in 14 years...so I put pen to paper and started writing a father's journey. There's been a lot of interest and professors have wrote prefaces on it.

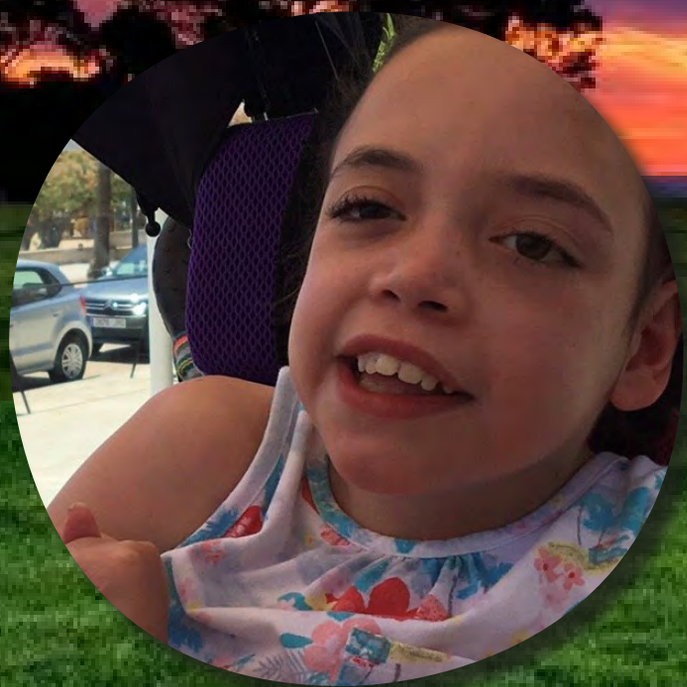
You can contact Gaz at: gary@securemindsolutions.com, for further information visit: www.securemindsolutions.com

In Loving Memory...

We dedicate the following few pages to the memory of beloved members of the NPUK community who are sadly no longer with us.

Grace Hitchens: 27/2/2005 - 15/3/2018

Our beautiful Grace was born on 27th February 2005, 10 minutes ahead of her twin sister Emily. When we first saw those beautiful big brown eyes we were in love. Grace also had a big brother Harry who quickly came to tease and adore his two little sisters in equal measure. There wasn't much that Grace didn't like, she loved her food (except eggs, she hated eggs!) cake being her favourite. Grace loved messy play, that was one of her favourite activities in school and we have a wonderful video at home of her painting in the garden with daddy, Emily and Harry, 3 little artists, it's priceless. She loved being around people and I'm sure everyone who knew her would agree she touched their lives in a special way, whether it was capturing their gaze with those beautiful big eyes and that beguiling smile or pinching your nose, glasses or anything nearby using her telescopic arms and peripheral vision - pure magic. She loved boys and didn't let her lack of speech affect her ability to get noticed! I could always tell when she was up to mischief, those eyes would widen, blink with those huge lashes and a big smile would spread over her face - eyes focused on her target. Her laugh was infectious and I can hear it in every photo of her that I see.



Grace's last days were spent at Martin House, where we were able to care for her and make final memories with the support of the wonderful care team who have become family over the years. Grace passed away peacefully, shortly after her 13th birthday. Grace's life may have been brief, her sunset came far too soon and we will miss her as long as we live. We will remember what Grace and Harry, taught us about true friendship, how to be truly happy and most of all what true love is. Piglet: "How do you spell love, Pooh?" Pooh Bear: "You don't spell it, you feel it" x

Tj Colwell: 11/8/2005 - 17/1/2018

Tj, it's been just over 7 months since you left us. Not a day has gone by when I don't think of you and smile or I think of you with tears in my eyes. These are just some of the memories I have:

I smile when I remember the day you were born, and how proud I was to have you as my first child, as soon as I saw you, you had my heart 100%, I knew then that my life would be all the better for having you in my life.
I remember how small you were, and feeling so scared to touch you or pick you up in case I hurt you, but as soon as I done it, I didn't want to put you down. I remember the first time I changed you, you peed all over me, as if to say welcome to fatherhood, I hope you're ready.

I smile because I remember the happy times, like you singing and dancing along to your mum's songs on ClubLand TV, giving it your all to songs like, Fredde Le Grand (Put your hands up for Detroit), Bruno Mars (The Lazy Song), Caramell (Caramell Dancing), The Black Out Crew (Put a Donk on it) to name just a few, but mainly JLS. I think everyone will remember you strutting your stuff and doing all the dance moves, and having everyone watching you. Now you're gone, how are we meant to carry on?

I've always been scared of dying because of the unknown, but the reality is, I'm now scared of living.

I saw the following poem (featured to the right) and wanted to share this with you, it's called If Roses Grow in Heaven.

Written lovingly by Terry Colwell, Tj's dad.

If Roses Grew in Heaven

If Roses grow in heaven

Please pick a bunch from me.

Place them in my Sons arms

And tell him they're from me.

Tell him that I love and miss him

And when he turns to smile,

Place a kiss upon his cheek

And hold him for a while from me

Because remembering his is easy

I do it every day

But there is an ache within my heart

That will never go away.

Sophira Clarke: 29/8/1985-24/6/2017

My daughter Sophira sadly passed away on the 24th of June 2017 at the age of 31. She had Niemann-Pick type C and was diagnosed when she was 4 years old - being one of the youngest patients at the time to be diagnosed with this awful disease. She was in the top 10 back in the 1990s to be asked to go on the clinical trials...she was not expected to live to her teens.

When she was born she was very jaundiced, the doctors thought she had a liver problem. She was under Professor Diedre Kelly and she had to have an operation to see if she had a problem, but there was not anything wrong with her liver. Professor Diedre Kelly said we should send a biopsy to a doctor in France, and it was this doctor who provided the diagnosis of Niemann-Pick disease type C.

Sophira was 11 when she wrote a poem at school about "Peace at Christmas" (right)

Peace at Christmas

Here I am, sad and alone,

Thinking of all the bad things done,

The news is full of doom and gloom,

I hope there will be joy everywhere soon.

People in the nations afar,

Will look upon the Christmas star,

To make their wish for peace on Earth,

And free them from the pain of war.

Will John Major and Bill Clinton help,

To make their wish come true,

Everyone would be happy,

Expecially me and you.

If all the world could be the same,

Far off lands would suggest no pain,

Children everywhere would laugh and play,

If only the world could be this way.

Tina, Sophira's mum who passed away in November 2016 got in touch with the media to highlight this disease. Noel Edmonds got to hear about it and decided to feature it on Noel's Christmas Presents which aired in 1996 on the BBC. Sophira was very lucky to be invited to 10 Downing Street to meet John Major, and also visited Disney World Florida. Whilst she was there with the family she was invited to meet President Bill Clinton and his wife Hillary, who signed the poem.

Sophira had such a great attitude and determination about herself, about her independence as much as she could, and lived life to the best of her ability. She was strong and full of life, not much would get her down - not even her stubbornness!

I just wanted her to be a "normal" daughter, and to be able to have children. I wanted grandchildren. I wanted her to be able to be in a relationship, to fall in love with someone. I wanted to set her up in a nice house. I wanted the best for her...I wanted the world for her.

Since Sophira's passing myself, my partner, and our son have been fundraising at our local pub and it is with much gratitude from our lovely friends that we have managed to raise £2 ,210.50. We have raised this by doing various things like; meat raffles, cricket events, dry October, our son selling his Xbox to name just a few.

So it is with our pleasure that we can give this cheque to Niemann-Pick UK.

Much love Pete, Emma & Tyler.

Matthew Lingard
12/3/1998 – 10/11/2017

Having received support from NPUK over the years, Matthew's mum Helen reflects on her most treasured memories of her son:

Matthew was born on 17th March 1998, St Patricks day, the eldest of our four children, a much wanted and very loved child who taught us all so much.
In the weeks after Matthew's death we heard so many lovely things about Matthew and been reminded of so many stories. The overwhelming theme being Matthew's sense of fun and the joy he took from life and the joy he gave to others.

Matthew was diagnosed at 6 weeks with Niemann-Pick Type C. We were told he might not make his first birthday but instead he enjoyed nearly two decades of a happy and fulfilled life and brought much fun and laughter to all who knew him.

One of the consequences of Matthew's diagnosis is that we celebrated every milestone he reached which otherwise we might have taken for granted. Obviously the first milestone was his first birthday which not only did he make but he was healthy and thriving and we celebrated with friends and family some of which had come from a long way away – I remember it as a hot and sunny day, which always seemed to be the case for Matthew's birthday. It has remained a very special memory and it was a turning point for Matthew's health. From this point Matthew was able to enjoy several years of healthy and normal childhood.

Matthew was always kind and willing to help others with a smile or just a touch. I recently asked friends and family to think of any words associated with Matthew and from so many people I heard about Matthews kindness, humour and happiness. I remember Matthew would always share his sweets and toys and especially loved and cared for younger children, he was always loving and generous with his sister Beth and brothers Tom and Oliver.
Despite Matthew's diagnosis we were always determined that we would not wrap him up in cotton wool but would encourage him to live life to the full – and he did, he was not terribly sporty but still managed a good game of tennis, could ski, ride a bike, go surfing at the beach and swim, he was a member of his school swim squad and I will always remember the time he got a speeding ticket in a competition once for swimming too fast!

When Matthew was a baby we used to hope that he would be have the chance to learn to talk and communicate as we felt that this would be how we would really get to know him.

Well we shouldn't have worried - not only did Matthew learn to talk he didn't ever stop and what's more he learned to sing. Matthew's love of music first started when at 6 months when he joined music classes and so began our years of children's car cassettes! From this point on Matthew sang his way through life – living with Matthew could be likened to being in an am-dram society – lots of singing, dressing up and constant plays and re-enacting. Not to mention Matthew's love of jokes – usually bad ones! Matthew did have a talent for making pretty much anything into a joke and he certainly did make people laugh with him. Matthew loved all things music and theatre and loved going to see musicals - Mama Mia was his favourite, he must have seen it hundreds of times and for his 18th birthday we all went up to London with him to see it on stage.

A few years ago, Matthew even stared in a film himself – the story of Ian Dury, called 'Sex and Drugs and Rock and Roll'. Unfortunately, he was too young at the time to go and see it at the cinema! As it is totally unsuitable for children. Matthew enjoyed being part of the community and going to the local nurseries and schools allowed him to do this.

Just after his 6th birthday Matthew joined the Beavers scouts and went on to enjoy many years of scouting activities. Matthew also enjoyed skiing and had his first lessons at the dry ski slope. Every Saturday Matthew, Beth, Tom and his cousin Laura would attend the Saturday morning kids ski sessions and he enjoyed several ski holidays.

In the middle of year 5 Matthew was finding life in main stream school difficult and it was time to start looking for alternatives to support Matthew in the long term, in what would be an uncertain future. We were lucky to be accepted and given funding for Treloar School where he spent 10 very happy years. They helped him adapt to his deteriorating condition, solving problems and keeping him as active and involved with all aspects of life. He especially enjoyed dressing up for the end of year prom.

Over the years Matthew has been lucky to be supported by several charities. We were able to go to Lapland to see Father Christmas one year with Starlight and then he was lucky enough to be selected for a trip to Disneyland in America with Dreamflight. He spent 10 days away from his family with a fantastic team of volunteers. I'm glad I didn't know but I should have guessed as he went on every ride in the parks - the higher and faster the better with lots of laughter and screaming.... He also enjoyed swimming with the dolphins and visiting all the other parks and ever since he regularly looked at the photos of that holiday with great pleasure.

Gradually Matthew began to lose many of his skills, but he remained positive and fun loving. He never lost his sense of humour. In November Matthew became ill with a chest infection which he could not fight. After two weeks in hospital we had to say goodbye to our beautiful son. Matthew taught us so much and we are very proud of him. He enriched all our lives and we miss him so much.

REFLECTIONS

The Reflections group was created by Niemann-Pick UK Senior Families Advocate Elizabeth Davenport, to provide care and support to those who have lost a loved one to Niemann-Pick disease.

This year's Annual Family Conference provided a wonderful opportunity for the Reflections group to get together - it not only serves as a safe space in which to remember our Niemann-Pick angels, it also allows our bereaved community members to support one another. Elizabeth comments:

"...the Reflections group at Conference this year was amazing. We had so many families joining us to celebrate their beloved family members.

We had a great selection of craft materials and everyone sat and talked while making some fantastic pictures. Quite a few new families attended this year and were a little apprehensive as they didn't know what to expect...they were pleasantly surprised by how relaxing Reflections is!

Put simply, Reflections is a place to talk and celebrate the lives of family members who have passed. Yes feel free to cry if you want, but feel free to laugh and share the good times too! I just want to thank everyone who came and made it such a special meeting..."

The group will meet agan at next year's Annual Family Conference & Interactive Workshop at Wyboston Lakes Bedfordshire, however if you prefer to meet and talk with Elizabeth individually, you can contact her throughout the year by email: elizabeth@npuk.org or phone on: 07896 197 576.

The Wisdom of the Reflections Tree

A tree, with branching arms that reach out to touch grieving souls,
is bedecked with carefully crafted thoughts,
written in aching love, and torn from within painful hearts.

The tree's knarled fingers interlock to shelter and protect those for whom it cares.
It knows their hurt and distress, and wishes them no further harm,
for the tree is laden with many words of love;
it is a rich harvest of stored memories,
growing ever brighter with the passing years,
reflecting a bestowed wisdom of many generations.

It has endured dark and dreary winter days, but is revived
when kind and gentle thoughts fill the flowered meadows of memory
with the warmth of happy summer words.

It is a home, a place to which we can return,
to read again the garland of gentle memories,
which tell of happy times,
enriched with golden laughter.

The memories grow with the passing of the years,
And are a presence stretching into our futures,
going even beyond the limit of our lives;
They are a tributes to lives unlived, cut short by faulty codes,
yet marked by courage, trusting love and great affection.

Their vulnerability demands our attention,
for they, as sons and daughters, believe that we as parents,
can conquer all that threatens their existence.

Now, forever children within our minds,
and each special in their own particular way,
they outlive us, as we grow old and are bent with age.

David Wray
26/09/2018

np uk

HOW TO PLAN YOUR NPUK FUNDRAISER...

Much of the work we do is funded entirely through fundraising activities and successful grant applications - that's why we value our fundraisers so much! By using these five steps, you will be well on your way to planning a successful event:

1 Choose something fun: The only limit to fundraisers is your imagination. There are so many events to choose from...check out our fundraising pages (page 38-45) for inspiration!

2 Set a date and a target: Setting a date is the first step towards making your fundraising goals happen - so set a date and target, and stick to it!

3 Get in touch with NPUK: Let us know what you have planned, so that we can share news of your event and provide information and materials to support you.

4 Spread the word: People need to know about your amazing event, so make sure you utilise social media to get the word out to everyone!

5 Recognise that you are an absolute hero: Your dedicated efforts keep NPUK going, and we appreciate this more than words can say! So never doubt - whether your event is large or small, raises a little or a lot - you are making a difference!

NPUK Volunteer Graham North speaking at an INPDA fundraising event this year

For further information on fundraisers including the NPUK Fundraising Guide please visit our website at: www.npuk.org

With grateful thanks to our grant providers:

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BBC Children in Need

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NPUK

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Note from the Editor...

I would like to thank all of those who have contributed to this issue of NPUK News. Please continue to submit articles, stories, poems, advice, children's contributions, fund-raising ideas, and anything else you feel may be of interest...especially photographs!

Send your contributions for the next edition of NPUK News to John, our Communications Officer, by email at john@npuk.org, or by post at: Niemann-Pick UK, Suite 2, Vermont House, Concord, Washington, Tyne and Wear, NE37 2SQ.

Disclaimer: Information which appears in this newsletter is for the express purpose of raising awareness and does not necessarily reflect the views of NPUK. All medical information should be reviewed with your doctor before being acted upon.

np uk

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