np uk

NPUK NEWS

Autumn/Winter 2020

NPUK GOES DIGITAL!

Our first ever digital Annual Family Conference & Interactive Workshop

Dedicated Dave

We catch up with long serving NPUK Trustee and Former Chair David Roberts

UPCOMING COMMUNITY CAMPAIGNS

Look out for opportunities to get involved in many exciting campaigns in 2021

Don't be shy, get social with NPUK:

@NiemannPickUK @niemannpickuk @NiemannPickUK @NPUK 0191 415 06 93

REDUCE · REUSE · RECYCLE

NIEMANN-PICK UK AND CINEBITES PRESENT

go make memories!

PM SOCIETY DIGITAL AWARDS 2020 GOLD

PM SOCIETY DIGITAL AWARDS 2020 SILVER

BEST PICTURE Rare Film Festival 2020

BEST CHARITY FILM Rare Film Festival 2020

BEST ANIMATION BELIFFestival 2020

FINALIST Los Angeles Diversity Film Festival 2020

DISORDER The Rare Disease Film Festival official selection 2019

BEST SHORT FAMTA 2020

BEST ANIMATION UNOSGUARDORARO 2020

BEST SHORT AWARD Sällsynta Stories Festival Stockholm 2020

BEST EXPERIMENTAL Feel The Reel International Film Festival 2019

22-30yrs | 1st Prize chiyoko 2019

Niemann-Pick UK (NPUK)
Contact us:
info@npuk.org
0191 415 06 93

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STARRING LLEYTON COOMBES EMILY STRIDE CHRISTOPHER HUGHES BODHI RAE BREATHNACH ANDREA HALL

MUSIC BY CHRIS KÖBKE CINEMATOGRAPHER JAMES WESTLAKE

PRODUCTION DESIGNER ISA SHAW-ABULAFIA POSTER DESIGNER ROB HOTCHKISS

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Supporting those affected by Niemann-Pick

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NPUK News is designed with you, the NPUK Community, in mind...

So if you have any suggestions or submissions then we would love to hear from you!

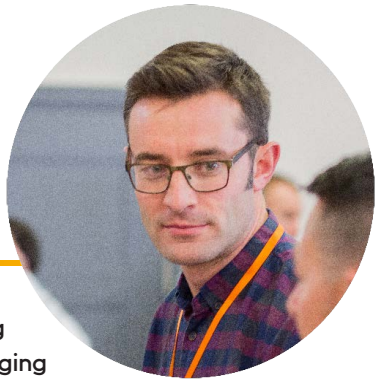
With thanks to Aztec Colour Print:

Many thanks to our printers Aztec Colour Print for yet again printing this vibrant magazine on behalf of NPUK!

Aztec have also handled the printing for all other publications for the past couple of years, ranging from leaflets, to patient passports, medical cards to banners and so much more.

Find out more about them at: *azteccolourprint.co.uk*

Welcome!



Chairman's Chat with Will Evans

Welcome to another issue of NPUK News. Notwithstanding the challenges that 2020 has presented to us, it is encouraging to see positive progress reported within these pages, bringing hope for our organisation and community.

The ongoing COVID-19 pandemic has affected all areas of our work and family lives, with many of our usual activities and essential services resigned to virtual. Our team has worked non-stop to provide support and comfort to our community, whilst personally trying to deal with the many changes and challenges this crisis has brought. We managed to quickly adapt to the new ways of operating and to continue providing our work and services in innovative ways.

Despite the huge disruption and the frustration of not being able to meet in-person, digital methods of communication, such as Zoom, has enabled us to keep in touch and to keep spirits up during lockdown and beyond. Our Board of Trustees agreed provision of an Emergency Response fund which enabled instant financial and wellbeing support for activities and education at home, plus additional staff hours to support communications regarding COVID-19. This included increased online content, activities and daily updates, plus a structured ten-week schedule to support, inform, and entertain the NPUK Community, referred to as our "Social Not Distant" campaign.

Our first ever digital Conference was a new experience for our community, but it was also new for us to organise. I would like to say a particular thank you to our 52 amazing speakers from a number of different disciplines and backgrounds, some of whom got up at 3am to address our 225 global attendees. In addition, the many volunteers who supported our online children's activities and social events, including the now legendary Quiz Master Terry Colwell and singing duo Carl and Emma Burdon. Everyone involved helped to make it a memorable, informative and engaging weekend. I of course look forward to the days we can all meet together, in-person, however I do feel this was a much-needed, productive and positive weekend for many reasons.

I trust you will find this issue informative and entertaining; please do contact our team with any questions or comments you may have and remember to share this issue with a friend once you've read it...social media is great but sometimes the old methods are the best!

I would like to wish you all a peaceful winter break, and I hope that 2021 brings a return to social interaction along with renewed hope for the future.

Warmest wishes,

Will

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Niemann-Pick UK

We are a small charity dedicated to supporting those affected by the group of rare genetic conditions known as Niemann-Pick diseases

Editorial

Developed in-house by NPUK Communications & Campaigns Manager John Lee Taggart, for any and all queries please contact: john@npuk.org

Grant Funders

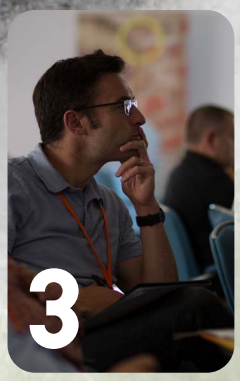
We would like to thank The National Lottery Community Fund, BBC Children in Need, and The Hollie Foundation for their invaluable sponsorship

Contents:

STAFF UPDATES

OUR CONFERENCE

Go Make Memories
We've updated our (now) multi-award winning short film poster - check it out!



Chair's Chat
Welcome and well wishes from our Chair Dr. William Evans

What progress has been made thanks to the "Shaping Our Future Together" project?

What's going on in the field of Niemann-Pick research? Find out on these pages

COMMUNITY STORIES

PARTNERSHIP WORKING

KID'S CORNER

COVER ARTICLE

COLLABORATIVE CAMPAIGNS

EDITOR'S NOTES

"Our Nigel" by mum, Christine Willman

Brilliant artwork by the very talented James Keeble! Learn more about the upcoming NPUK campaign, Invisible Illnesses, on page 38

Pages 44-45 are dedicated to our fantastic fundraisers...we have tried to include as many of you as we can, as you mean the world to us.

It's difficult to include everyone, so if you have had or are having a fundraiser and haven't been featured, get in touch with us at info@npuk.org and we will be happy to feature you in the next issue!

IN LOVING MEMORY

We remember all of those in the NPUK Community we have lost with the short but powerful poem, Never Forgotten

LAURA BELL
CLINICAL NURSE SPECIALIST

Hi Everyone,

I hope everyone is doing as well as can be? 2020 has definitely been a very different year for us all so far, and I know for us at NPUK the past few months have certainly seen us face many challenges as we adapt the way that we work, support and communicate with our community.

At the beginning of spring when we first faced the pandemic and lock-down we quickly adapted to working in new and different ways to address the needs of our patient community and continued to provide care and support in a hope to reduce the pressure on the NHS by answering queries and providing advice and information at the point of contact, as well as trying to reduce the need for patients and families to contact their metabolic teams. I am truly proud of our response!

Throughout the past 7 months I have continued to work closely with the specialist centres and along with all of my colleagues at NPUK - ensuring that we keep ourselves up to date with the latest government guidance to keep patients well informed by providing the most up to date information. It was also during this time we continued to support patients and families individually to help alleviate their anxieties and questions around COVID-19, social distancing, self-isolation and shielding.

What is Laura's role here at Niemann-Pick UK?

Laura is NPUK's Clinical Nurse Specialist. She is based in Manchester at Salford Royal Foundation Trust but is available to support all patients and families across the UK, wherever they may be.

Through the wonders of technology we were able to reach out to many of our patients and families through the long period of increased isolation via virtual coffee mornings, weekly webinars and of course more recently our first Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases. It really was so important for us all to come together and support each other during this time and I know it was always so lovely to see you all (virtually!) in person as I have really missed the face to face aspect of my role over the course of the pandemic.

As the situation evolves we will continue to create and adapt new ways of working and so we can overcome any challenges we face. I will continue to work with the medical professionals both within the hospitals and community

settings and learn from experiences of our patients and families to help the wider community and to ensure that you receive the care and support you and your family need.

I would like to take this final opportunity just to remind you that as Clinical Nurse Specialist for NPUK I am here to offer practical advice and support on all aspects of Niemann Pick diseases from diagnosis and beyond. I can also help you navigate through your health and social care teams and treatment as well as offering support in times of difficulty.

If you need any help, advice or just a chat I am just at the end of the phone and you are always most welcome to contact me at any time. As you know I am always happy to hear from you or to accompany you to appointments or meetings or visit you at home (whether this be in person or virtually!).

Here for you always,

Laura

Email: laura@npuk.org
Phone: 07791 499 555

HOW LAURA CAN HELP YOU, AND YOUR FAMILY

- Practical advice related to Niemann-Pick disease and symptom management

- Support at clinics and appointments, as well as home visits where necessary (all COVID-19 related safety procedures are followed)

- Phone consultations and regular "check ins" whenever necessary

COVID-19: NPUK SITE
information for families

Coronavirus (COVID-19) has dominated the news since the outbreak began at the end of December 2019. This virus has affected people around the world, including those in the UK, and has changed how we live and work dramatically.

Naturally this has been a source of concern for our community and many others, including specific groups of patients who may be more at risk, including those affected by Niemann-Pick diseases. Consequently we developed a dedicated COVID-19 page on our website (www.npuk.org) in tandem with our Social Not Distant Campaign. Every article or recommendation included on this page had been "vetted" by the NPUK Team and/or related specialists, so you can be sure it is up to date and accurate.

Our dedicated page contains:
- The latest recommendations, restrictions, and guidance as set out by the UK Government
- The latest advice from NHS England, and associated organisations/specialist centres
- Other helpful articles, website links, or resources which may be of use to our community.

You can find the page at the following link: npuk.org/coronavirus-covid-19/

KAREN THOMAS
FAMILIES OFFICER

Hello Everyone,

First and foremost, I hope you and your loved ones are safe and well.

I have now been in post for a little over a year and boy what a year it has been! These are trying and uncertain times for us all and we've seen some big changes to our lives. We have all faced different challenges such as financial hardship (brought on by loss or reduction of income), the difficulties of home schooling, not seeing and hugging our loved ones, being at home more and feeling isolated or a combination of all of these things and more.

I am writing this from my home in Lancashire but regardless of where I, or my colleagues are based, I want to assure you that NPUK is 100% operational and we continue to be here for you all during these difficult times. We are here for emotional support, so don't hesitate to call any of the NPUK staff if you need to talk. Talking can ease stress and reduce anxiety and not everyone has someone to talk to, or their loved ones could be busy at work or struggling themselves.

What is Karen's role here at NPUK?

Karen provides non-clinical advice, information and support to all affected families across the UK. She can link in to your local social services or health care teams to ensure you are receiving the level of support appropriate for you, liaise with schools, offer non-clinical advice, and much much more!

You may be experiencing difficulties paying bills or essentials such as food. If you are experiencing any of these, or other difficulties, please give me a call as I can help via phone, video chat and/or email.

Speaking of video chats, like many of us I'm spending a lot of my time on video calls at the moment (mainly Zoom). I am someone who has spent years actively avoiding this stuff, preferring the intimacy of a face-to-face conversation, without the intrusion of a computer screen! However, I have dragged myself out of my comfort zone, and into all the new technology, so I can promise you if I can do it anyone can! It certainly has opened up new opportunities for increasing communication with families across the UK and it's great for me as the newest member of the NPUK team to put faces to names. If you too struggle with technology, please get in touch with the NPUK team and we can help you to overcome these

difficulties and in doing so give you the opportunity to join any online events we run, or just have a face to face chat with any of the team.

Our Digital Annual Family Conference in September could not have taken place without all this technology and it was heart-warming to see so many families connecting over that particular weekend. Terry's quiz had such a good turn out and I'm sure you'll all agree he did a fantastic job. We also heard our patients and families talk about their experiences in their 'patient voice' videos and I was so moved and proud of each and every one of them for coming forward and sharing their stories with us.

Since June I have hosted a monthly Queen Bees group (via Zoom) and it has been fantastic for mothers and partners of those with Niemann-Pick Disease to get together for an informal chat. Here is what one member of the Queen Bees said about the group:

"...The Queen Bees is such a fab idea, and I for one love catching up with an incredible bunch of women each time. Everyone's stories, experience, empathy and above all chat. Love a good catch up!

It's a relaxed safe environment where a laugh and a glass of wine, or a tear and a cry are equally welcomed.

Been a great way to meet new people and keep in touch with the wider NPUK community..."

If you would like to join the Queen Bees, please get in touch with me as you would be more than welcome.... the more the merrier! If you have any questions about this, or other ways in which I may be able to help you, please get in touch at any time.

With warm wishes,

Karen

Email: karen@npuk.org
Phone: 07423 106 595

NPUK CARE & SUPPORT GOES DIGITAL!

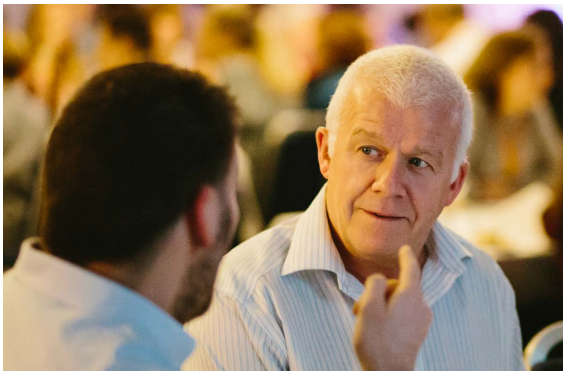
Like most individuals and organisations in 2020 we have had to quickly look into ways to fast-track the use of digital devices and applications to keep both our communication and support services open and running. Thankfully this is an area in which we are already familiar in, so we were well placed to make this adjustment! Here are just a few of the steps we have taken:

- Creation of the NPUK Social Not Distant campaign running across our for our social media accounts / platforms in a bid to support, inform, and entertain our community through lockdown(s)

- Development of our first ever Digital Annual Family Conference & Interactive Workshop hosted on the WorkCast platform

- Offering regular Zoom check ins/group meetings, as well as updating our support service to include a lot more digital "face to face" time

STEVE NEAL
PROJECT FAMILIES OFFICER



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

I hope you're all keeping well in what have been very testing times indeed. Times where our support services in particular have been more needed than ever – as a result throughout the last six months I have been working intensely with the NPUK Community to help support these demands, and ease any stress that the pandemic may have created for our families. These responsibilities have worked in tandem with my core duties trying to encourage the development of new positive wellbeing routines, and as ever assisting with benefits/grants to help reduce anxiety around these areas.

I like to think of my role as being led by you. An ever expanding area at this time has been additional support surrounding mental health needs and wellbeing, which is something I feel is a very important aspect to every one of our lives – we all need to know we're not alone, we all need to be aware of help and support that is available. From an NPUK standpoint we have begun to bolster the wellbeing of our patients, parents, siblings, and young people by focusing primarily on developing emotional resilience and self care. Whether this has been by working on skills like mindfulness, personal development, or simply turning up to Zoom coffee mornings, they are all positive steps forward.

What is Steve's role here at NPUK?

As our Project Families Officer, Steve is on hand to offer non-clinical advice and support regarding a range of aspects of Niemann-Pick related care issues. He is available to attend clinics, meetings, and other events with families, and can arrange home visits where necessary.

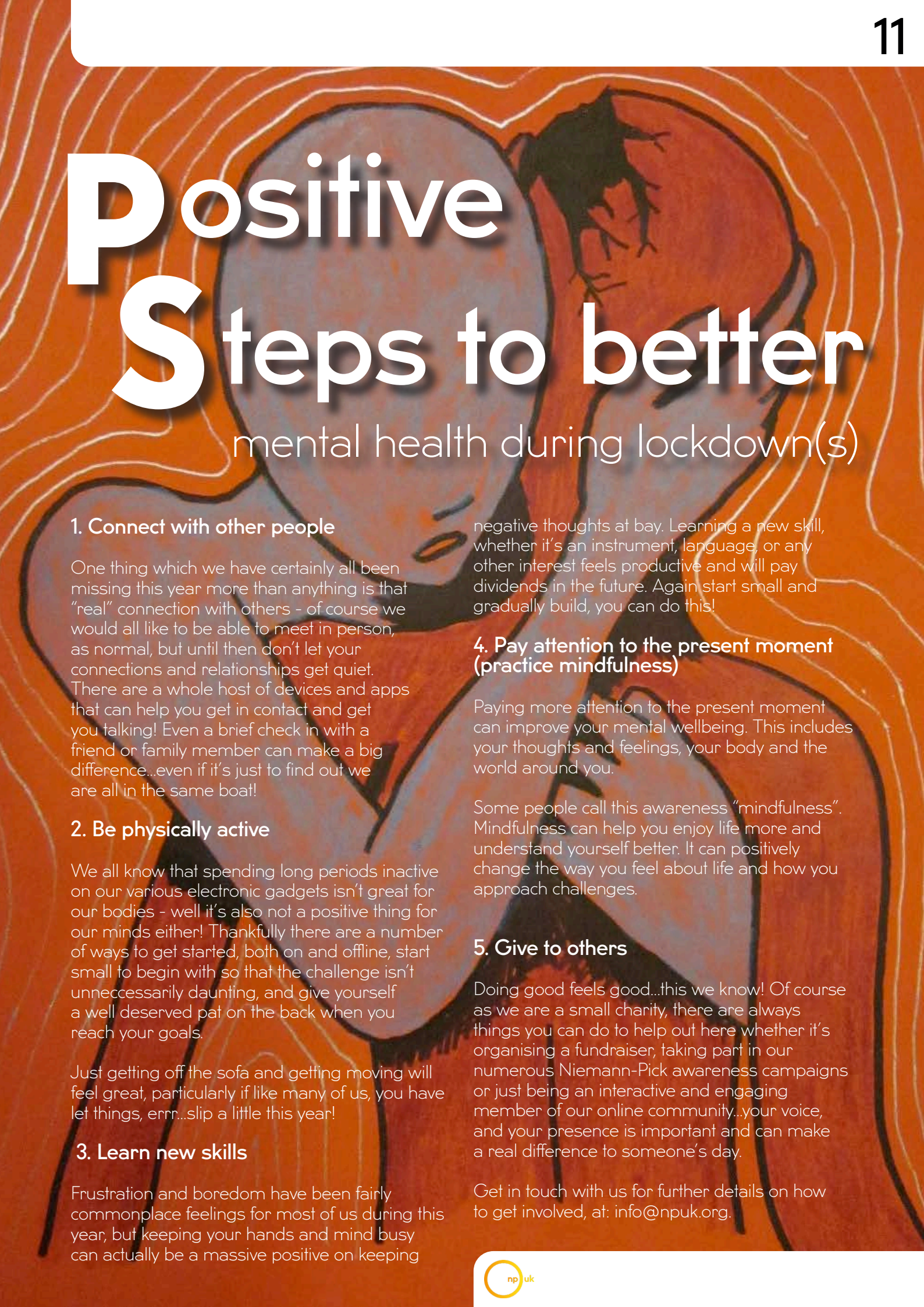
One thing which we have found to be a glimmer of positivity throughout this time is the development of a culture of support online, that's; patients, families, friends, staff, and more coming together to support one another through these challenging times. We want to work on further developing this support culture and to make it even more accessible through our social media and broader communications.

I want the whole of the NPUK Community to feel empowered, to have a voice, and to feel they can always reach out safe in the knowledge that there will always be a supportive ear there to listen no matter what.

Take care everyone,

Steve

Email: steve@npuk.org
Phone: 07787 818 885



Positive Steps to better

mental health during lockdown(s)

1. Connect with other people

One thing which we have certainly all been missing this year more than anything is that "real" connection with others - of course we would all like to be able to meet in person, as normal, but until then don't let your connections and relationships get quiet. There are a whole host of devices and apps that can help you get in contact and get you talking! Even a brief check in with a friend or family member can make a big difference...even if it's just to find out we are all in the same boat!

2. Be physically active

We all know that spending long periods inactive on our various electronic gadgets isn't great for our bodies - well it's also not a positive thing for our minds either! Thankfully there are a number of ways to get started, both on and offline, start small to begin with so that the challenge isn't unneccessarily daunting, and give yourself a well deserved pat on the back when you reach your goals.

Just getting off the sofa and getting moving will feel great, particularly if like many of us, you have let things, errr...slip a little this year!

3. Learn new skills

Frustration and boredom have been fairly commonplace feelings for most of us during this year, but keeping your hands and mind busy can actually be a massive positive on keeping negative thoughts at bay. Learning a new skill, whether it's an instrument, language, or any other interest feels productive and will pay dividends in the future. Again start small and gradually build, you can do this!

4. Pay attention to the present moment (practice mindfulness)

Paying more attention to the present moment can improve your mental wellbeing. This includes your thoughts and feelings, your body and the world around you.

Some people call this awareness "mindfulness". Mindfulness can help you enjoy life more and understand yourself better. It can positively change the way you feel about life and how you approach challenges.

5. Give to others

Doing good feels good...this we know! Of course as we are a small charity, there are always things you can do to help out here whether it's organising a fundraiser, taking part in our numerous Niemann-Pick awareness campaigns or just being an interactive and engaging member of our online community...your voice, and your presence is important and can make a real difference to someone's day.

Get in touch with us for further details on how to get involved, at: info@npuk.org.

SHAPING OUR FUTURE TOGETHER

The National Lottery Community Fund

Our five-year National Lottery funded project "Shaping Our Future Together" has been truly transformative and has enabled us to strengthen our community support and services in a number of ways. Our small team has worked across the UK and Ireland to ensure all those affected by or connected to Niemann-Pick disease can access support as and when it is needed. Although the project comes to an end early in the New Year, the legacy of this generous grant will continue through the services it has enabled, which are firmly embedded into our practice.

Awareness raising efforts plus advances in science and diagnostic techniques have increased diagnosis rates and helped us to reach a growing number of new patients. National Lottery grant funding allowed us to expand our staff capacity and services in order to meet additional demand and to continue providing a high level of practical, emotional and clinical support to new and existing patients and their families. In addition, we were able to develop new specific support initiatives for our BAME community, for Lads, Dads, and Carers, a group specifically for mums (The Queen Bees) and much more.

Our grant has helped us to achieve four main outcomes on behalf of our community:

• To increase knowledge and understanding within our community, to ensure that patients and their families can confidently interact with health and statutory services and make informed decisions about the care and services they receive.

• To ensure patients and families have increased access to health and statutory services through expert and timely support from NPUK.

What is The National Lottery Community Fund?

The National Lottery Fund raises money for good causes. People use this funding to do extraordinary things, taking the lead to improve their lives and communities. Every time you buy a National Lottery ticket...you help make this happen! Niemann-Pick UK is one of many organisations which have benefitted from their fantastic grant scheme.

• To improve the overall mental health and wellbeing of our patient community, with a particular focus on those newly diagnosed, adult patients and those that might be harder to reach.

• To increase emotional resilience amongst patients and their families, helping them to manage challenging symptoms and improve daily life and relationships.

In addition, to support communication with our team and interaction with our community, the grant enables us to provide iPads to patients and families. The aim of this is to better understand how technology can help patients and families to manage their condition, access

support and information and stay connected. Of course, you can also use them to watch films, listen to music and play games! We simply ask that those participating connect with us on a regular basis and tell us the ways in which their iPad improves or assists daily life. We still have a few iPads available, if you would be interested in learning more, please get in touch with any member of our team.

This year has brought new and unexpected issues for our small team, we have however, adapted to the restrictions COVID-19 has placed upon us and have focused our efforts on providing support and services in new and innovative ways. Our Team took to Zoom, Microsoft Teams, WhatsApp, Facebook and any other means to ensure our community felt supported and connected. Our usual in-person Annual Family Conference, which is a key event in our calendar and brings together the largest gathering of Niemann-Pick patients, families and professionals in the UK, went digital for the first time. The event presented many of the same opportunities for interaction, for catching up with the latest in research and clinical developments and for having a little fun with friends old and new.

We regularly update our website with information relevant to our community, and have even set up a dedicated COVID-19 page to ensure that trustworthy and reliable information is presented to members of our community in a timely manner. Along with this we have increased our social media presence, and began a series of

"...National Lottery grant funding allowed us to expand our staff capacity and services..."

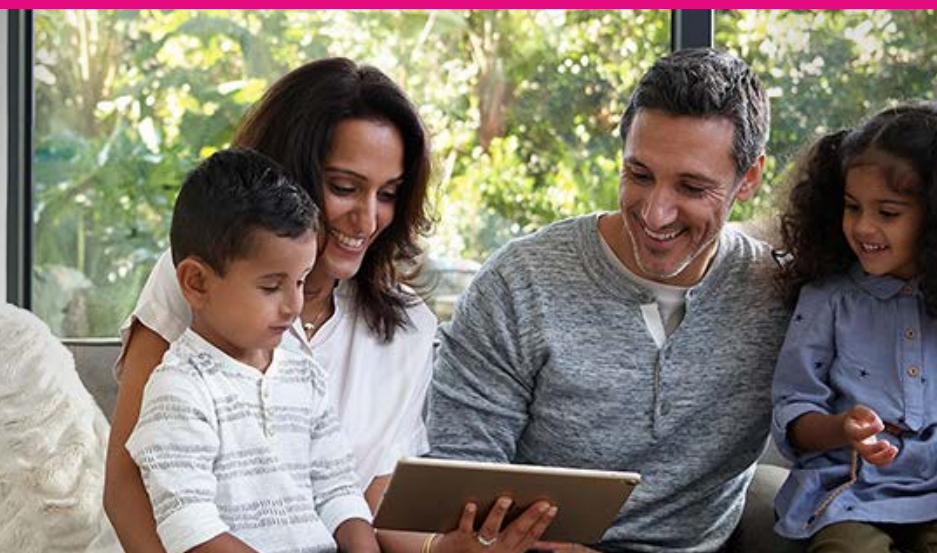
monthly e-newsletters to keep people updated in between issues of our larger "NPUK News" magazine, (which you are currently reading!) which is packed full of information relevant to patients, families, and those with links to our charity, including; news on the latest clinical trials and research, fundraisers, support tips, and fun light-hearted articles, which we all need this year more than any before!

If you would like to learn more about our Big Lottery project, please contact us directly at the NPUK Central Office on 0191 415 06 93 or info@npuk.org

COMMUNITY FUND

Did you know that NPUK still have iPads available for adult patients and their families as part of our National Lottery funded project,"Shaping Our Future Together"?

The deadline for this initiative is January 2021, so there is no time to waste!
For further information please contact us at the Central Office on 0191 415 06 93 or by email at: info@npuk.org



NPUK Communications & Campaigns Manager, John Lee Taggart, pictured here with Lleyton Coombes (age 13, NP-C) takes us through how we developed this year's first digital Conference...

How the NPUK Conference went digital...

Our Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases (don't worry that's the last time I will use the full title in this article), is in many ways a perfect illustration of everything that NPUK is about. It encompasses everything that our small charity represents, has to offer, and is proud of, all packed into a single busy weekend every year; a gathering of families, friends, scientists, and professionals working in the field of Niemann-Pick disease in one place, to not only hear what exciting developments may have been made and what the future may potentially hold, but also to laugh, cry, and reminisce safe in the knowledge that in this place, at that time, there are people who truly understand.

Well that was lovely, even if I do say so myself. But as we are all more than painfully aware, 2020 had other ideas when it came to gatherings of just about any description; you could barely pop out for a cuppa, never mind a BBQ at your Auntie Maureen's or, gasp, a larger get-together such as our Annual Family Conference!

So because of this like so many other organisations, we were faced with some difficult choices. We had to firstly address how we would adjust to the new unchartered territory we found ourselves in...a world filled with more Zoom calls and breaking news headlines than mankind had ever witnessed before. What alternatives could we make the most of, and how could we (temporarily) adjust our usual service so that our families and wider community would still feel supported?

During the first lockdown we developed the "Social Not Distant" campaign, an expansive online weekly programme designed to see the families and friends of NPUK through the frustration of these new and challenging restrictions; it not only kept people informed and entertained in equal measure, but actually seemed to bring us all closer together. There were more connections and conversations than ever before...sure, it wasn't something any of us wanted forever, and it certainly doesn't beat meeting in person, but there were undoubtedly huge positives to take away from it

WE SIMPLY MISS OUT

and some lovely moments to treasure. This is likely when the idea of "digital versions" of our existing services and events began to truly take root, after which we knew certain choices would have to be made...

As our Chief Executive Toni Mathieson explained back in September, when it came to Conference "we had two clear decisions – to cancel/postpone until 2021 in the hope that the landscape would have improved, or to just simply go for it and try to deliver the best online version of our milestone event that we could".

Increasingly our Conference caters not only to those in the UK, but also to the international audience; friends, family members, and advocates of Niemann-Pick from all corners of the world. It makes for an empowering atmosphere to see that under one banner, real progress can be made - that culture, languages, and other differences are second to the joint cause we all rally behind. So, what right then do we have to deprive everyone from these connections and conversations? How could we possibly just tear away the chance to experience Terry's Quiz and his impossible Only Fools and Horses' round, or to learn about gene therapy from Charles Vite with support from his special assistant (a massive Bart Simpson stuffed toy!) or, or, or...have a sing-a-long during Carl

COULDN'T

and Emma's kitchen concert?! No way...it simply had to go ahead, we simply couldn't miss out!

So that's what we did, we put everything into preparing the best Digital Conference we could... and approximately 49 nervous breakdowns later...we had something which we felt reflected most of the things which make our multi-varied event the special weekend it is; we had some fantastic expert speakers from a wide range of disciplines who would offer great value to our community members, we had presentations on subjects that were both relevant and current to both the Niemann-Pick community and beyond, and also through the amazing efforts of our Staff and Volunteer team, had lined up some wonderful leisure activities to alleviate the potential stiff necks and digital exhaustion that can occur from days spent glued to our screens!

Well, we were certainly proud of how it came together, and appreciate each and every person who made it possible - it was an NPUK Family effort.

Although, I would like to take this opportunity, on behalf of the rest of the NPUK Staff and Volunteer Team, to thank coffee and other caffeinated products for their unwavering support during the entire Conference process. We couldn't have done it without you.

Joking aside we do hope you enjoyed the Digital Conference experience and found it to be useful, informative, and entertaining. I for one personally look forward to when we can all handshake, high-five, and hug as normal in the future...a future which I hope arrives sooner rather than later.

But until that day meeting by these digital means will have to do, and interacting through social media or Zoom ("Can you see me? Can you... all hear me?" **YOU'RE ON MUTE!!**) will ensure we keep chatting(ish), and keep supporting one another.

Take care everyone, and as ever,
stay in touch!

John

Bill Owen, NPUK Trustee Research Report

This report provides an overview of the NPUK Conference held this year on a digital platform with presentations from medical science and clinical professionals working in the field of Niemann-Pick diseases.

The Conference Programme, entitled Digital Annual Conference & Interactive Workshop on Niemann-Pick Diseases 2020 can be found on the NPUK website. The presentations given have been recorded and are available to view on-line. Further information on this can be obtained from the NPUK Central Office by email at: info@npuk.org.

I have listed the presentations given on clinical trials but have not summarised these as press releases are available from each company and I wish to avoid potential conflict of reporting. Updates on these activities can be obtained from the NPUK website or from the websites of each company.

COVID-19 & Niemann-Pick Disease

This year has seen major changes throughout the world as to how we have needed to adapt in conducting our affairs. Our Charity is no exception and the need to move to an on-line conference soon became evident. Pandemics are inconvenient but cannot be ignored!

The opening session of our event was dedicated to COVID-19 and its possible relation to the NPC1 pathway within cells. The session was introduced by Dr Fran Platt and Dr Will Evans with presentations from Dr Jonathan Ball of University of Nottingham, and Dr Stephen Sturley of Columbia University, New York. The background to the COVID types of virus emerging over previous decades was described and the danger of viruses jumping between species explained. Although this event is comparatively rare it can have devastating consequences for humans especially as with COVID-19, it has an environment to spread throughout the world very quickly. This rapid spread is facilitated by the mobility of individuals for trade, holidays, migration and other reasons.

Studies are proceeding apace throughout the world to learn about COVID characteristics and behaviour within and without the body with the aim of identifying targets for intervention. The virus has a large RNA genome and has a number of novel sophisticated methods to thwart the immune system. There are plenty of targets for intervention but development and testing is needed and time is of the essence. The ability to use repurposed drugs will hopefully reduce the time needed for testing but a vaccine may still be delayed in reaching the population. The concept of herd immunity will probably progress in one form or another of its own volition, whether or not it is health care policy. The main point is that for most countries their resources in healthcare systems, academics and pharma-biotech industries are all pointing in about the same direction.

A study is taking place to probe the resistance to COVID-19 virus in NPC cells. There is good reason to look at this as lysosomes and their precursors are on the internalisation pathway for many viruses and bacteria infecting cells and some have been found to interact with NPC1 protein by various means. The interaction can disable NPC1 and prevent entry of the pathogen into the lysosome where it would normally be destroyed. It is known that carriers of rare genetic conditions

Panel of speakers at the NPUK Interactive Workshop on Niemann-Pick Diseases 2019 at Wyboston Lakes, Bedfordshire. (Bill Owen - far right)

sometimes exhibit what is termed heterozygote advantage giving some degree of resistance to a disease. Sickle cell disease in malaria being a classic example. The study aims to identify whether any advantage by way of virus resistance is apparent in NPC carriers.

Basic Science Updates

Understanding Niemann-Pick disease

Prof. Frances Platt from the University of Oxford introduced her presentation by describing the general anatomy of a cell with a focus on lysosomal compartments that act as recycling centres for the vast variety of complex molecules that contribute to the life and function of a cell. Typical lysosomal diseases are the result of defects in one or more of 70 enzymes that carry out the recycling function. NPC1 is different in this respect as it is one of about 200 proteins located in the lysosomal membrane. The functions of these proteins are largely unknown but the NPC1 proteins is known to be involved in the movement of lipids, various other molecules and regulation of cholesterol levels within the cell. Defects in NPC1 result in lysosomal storage and subsequent pathology.

From an inheritance and evolutionary perspective, our genes are shared with many other creatures such as mice – 93%; yeast – 13%. The gene is also present in soya beans. One line of investigation being followed relates to identifying how various proteins interact in the trafficking pathways. Some success has been achieved in this with studies into NPC2 interaction with NPC1 within the lysosome, but investigations need to be widened to identify interactions external to the lysosome. This work is ongoing and may lead to a better understanding of the cholesterol regulatory pathway. Mice and yeast provide good animal models for research due to the similarity of genes with humans.

Studies of Enzymes in Lysosomal Storage diseases (LSD)

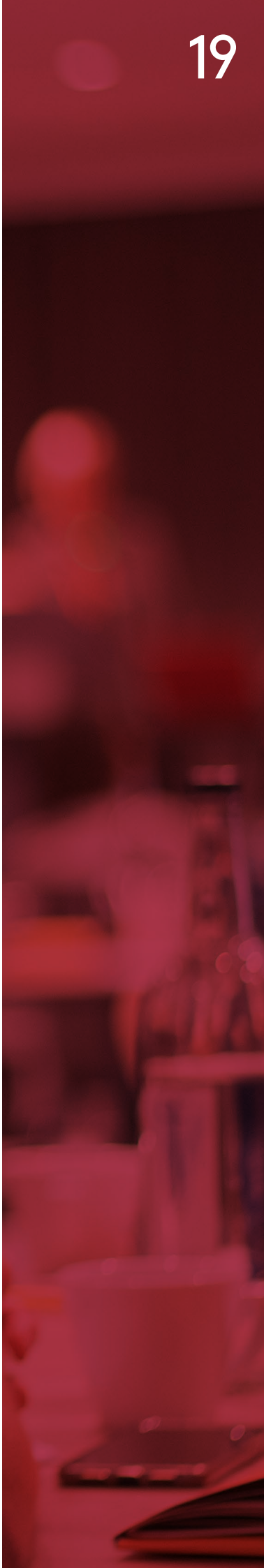
An update on this complex topic was provided by Dr Emyr Lloyd-Evans from Cardiff University. A recent study undertaken utilised 20 known assays and 3 new to examine the level of enzyme activity in a range of LSDs. Depending on the enzyme defect/disease some showed increased activity whilst others were reduced. It was noted in various cell populations – brain and liver, that Gaucher and alpha-mannosidoses have similar profiles to NPC1. It may be possible to utilise these profiles to identify and diagnose some LSDs. A further finding was that lysosomal acid lipase (LAL) is much reduced in some LSDs and when reintroduced to cell cultures then reduction of cholesterol storage occurred. It may be that LAL activity could be a new disease biomarker.

Pharmacological enhancement of the endocannabinoid (ECB) system to treat brain pathology in ASMD and NPC

Dr Lola Ledesma from Universidad Autonoma, Madrid is conducting studies on the ECB receptor, CB1, and its ability to promote sphingomyelin (SM) storge reduction in NPC1 knock out cultured neurons. SM storage being a consequence of ASMD (NPA/B) defect and is known to be linked to storage arising in NPC. Studies in symptomatic mice using an oral treatment, FAAH, that inhibits ECB degradation demonstrated a reduction in storage in a range of tissues. Further studies are ongoing.

Whole Genome Sequencing (WGS) in Adult Onset NPC1

Dr Andrew Munkacsi of the Victoria University of Wellington, New Zealand has studied NPC1 disease over many years with much of his work conducted using a yeast model of the disease. Genetic studies have shown that there is considerable variation in the NPC1 gene sequence and protein that appear to translate



into a wide variation of clinical presentation and progression even in individuals with the same mutation. A possible explanation for this is that each individual, person or animal has a different genetic makeup/background. This has been demonstrated in studies of disease progression in mice with different genetic backgrounds but all having a common NPC1 mutation. Studies in yeast have identified genes that are able to modify the disease severity by partially correcting downstream cell pathology, but the number of genes/interactions make studies difficult and statistical methods need to be employed linked to Artificial Intelligence (AI) machines with deep learning software.

In association with colleagues in Australia a study is being conducted to identify differences in genetic interaction between sibling pairs presenting with NPC1 disease. Six sibling pairs are involved in the initial study – one of each pair having paediatric onset and one adult onset. Potential genes to target have been identified and early results using a drug intervention have shown reduction in cholesterol storage. It is now intended to prioritise adult onset patients but recruiting sufficient numbers is difficult. To date 13 adult, 12 paediatric and 100 healthy controls have been enrolled and will be studied to identify what is unique in adult onset. Using AI, it is hoped to identify differences in patterns of genetic activity to inform approaches to new interventions.

Therapeutic Studies

Medicine Discovery Institute (MDI)

Dr Helen Waller-Evans has worked on NP disease research for many years at Cardiff University. She now heads up a laboratory in the recently established MDI, a small company within the university, whose mission is to discover medicines for diseases that are either difficult or cannot be treated such as the NP diseases. The MDI is an academic organisation, rather than industrial and combines chemistry and pharmacology to carry out studies. Various approaches to identifying suitable drug candidates are used including X-ray crystallography to study drug interaction with the target protein. Drugs that appear promising may require modification prior to testing in cell-based disease models to establish safety and demonstrate effective drug interaction. The drug discovery process is usually prolonged and expensive with safety being a main driver but repurposing existing drugs with a known safety profile can result in major benefits. The use of new technology microscopic techniques are used for high throughput drug screening. NPC is one of the diseases being studied by staff and students.

Miglustat improves swallowing in children and adolescents with NPC1

Miglustat improves swallowing in children and adolescents with NPC1. This Observational Study at the National Institute of Health (NIH) was presented by Beth Solomon. Although no drug treatments have been approved by the FDA for NPC treatment many patients have off label access to the drug but are required to fund this treatment themselves.

A number of studies into the use of miglustat have been conducted over past years in relation to swallowing (dysphagia) as it is a distressing feature of NPC but this latest study used measured parameters in the swallowing process. A major finding from this work was the identification of 'silent aspiration' which results in food or drink

entering the bronchial pathway but without apparent discomfort to the patient and so is not ejected by coughing. This finding was not previously reported.

The overall conclusion of the study was that miglustat treatment has resulted in stabilisation of the swallowing in most patients, reduced their risk of silent aspiration and its consequences and generally improved their quality of life.

Focus on Gene Therapy

We were treated to a number of highly informative presentations on gene therapy and its potential application to NPC. Although progress has been encouraging in recent years there remain many technical issues to be overcome and we shouldn't hold our breath in anticipation that this therapy will arrive in clinic any time soon. Caution is needed when reading about recent highly successful interventions in diseases such as spinal muscular atrophy (SMA). The NPC1 protein is not excreted by cells as are soluble proteins requiring that a much greater dose of the viral vector is needed in order to reach each cell.

The laboratories in the UK and the USA are pursuing their different approaches to applying this vector for NPC1 therapy and in using different animal disease models. These approaches were presented by Dr Charles Vite, University of Pennsylvania, Dr Cristin Davidson SOAR-NPC a US patient group and Dr William Pavan of the US NIH. Dr Vite described the GT development process using a coffee cup analogy relating some of the problems to size of cup and additives such as milk and sugar to 'activate' the contents of the cup.

Dr Ahad Rahim from UCL referred to the history of GT and the FDA moratorium on its use following ambitious studies resulting in patient death in the USA. Recent developments in vector design have resulted in the Adeno Associated Virus (AAV) which is proving to be a major tool in the medicine bag with an excellent safety profile. Major issues being addressed include, packing the gene into the vector; delivery across the blood-brain barrier, delivering sufficient vector to have a clinically noticeable effect, delivery to the defined target areas, consideration of peripheral organs and tissues, avoiding immune system response – the list goes on.

Longer term issues will present further problems. GT is a one-off treatment, it cannot be undone or re-done and it will be expensive. In order to realise the full benefit of GT it is essential to identify patients at the earliest stage, preferably at the pre-symptomatic stage. This is not currently possible but progress in new-born screening will help to ensure success of a clinical trial.

NPC Clinical Severity Scales - A Delphi Study

This presentation was given by Dr Will Evans, Chair of NPUK. A problem facing those conducting clinical trials on diseases such as NPC is that of measuring disease progression and changes to progression following administration of a drug intervention during a clinical trial. A number of severity scales have been developed some more comprehensive than others but more demanding on the patient and on the clinical trials team. This study has looked at how best to use these scales or whether a new scale was needed.

The approach taken was to use the Delphi methodology to arrive at a consensus from clinicians expert in NPC treatment and currently utilising one of more of the existing scales. The study process comprised obtaining and analysing responses to questionnaires over three phases of refinement and included the use of the scales in different settings such as clinical monitoring, trials enrolment, trials monitoring and assessment and international use.

A paper on the study will be published in 2021 and although consensus was not achieved a majority of opinion favoured the current 5 domain severity scale applicable to the majority of patients but not accounting for serious symptoms in some patients such as seizures.

Orphazyme Survey and Interviews: Understanding the Impact of NPC.

This presentation was given by Ms Claire Burbridge of Orphazyme. Although conducting clinical trials concerns many factors such as safety and efficacy, there are less visible factors that have significant influence on the lives of the patients involved and their families. This study set out to learn more about these factors using web-based communications and telephone contact. It is the progressive symptoms of the disease that give rise to additional problems such as stress, care giving, usually by parents, and financial difficulties.

The study contacted families mainly from the USA with some from the UK. Knowing which symptoms require the most attention and cause the greatest difficulties was a main goal of the study. As maybe expected the main symptoms of concern were the 5 domains in the clinical severity scale such as swallowing. It is from these symptoms that the greatest burden is imposed on the care givers. This study is important as it looks behind what is seen and recorded in the clinic where only the tip of the iceberg is apparent.

The International Niemann-Pick Disease Registry (INPDR)

An update on this most valuable development in recording information on the Niemann-Pick diseases was presented by Dr Conan Donnelly, Registry Manager, Mr Shaun Bolton, Operations Consultant and Mrs Jackie Imrie, Recruitment Consultant and NPUK Trustee. The background to the international project was explained and the important aspect of facilitating both professional and family/patient contributed data emphasised. The importance of patient reported/inputted data has been recognised by medical professionals, the pharmaceutical industry and the regulators of medicinal products.

The registry data will contribute to the natural history and contain longitudinal data, assist disease research and post marketing studies of the medicines being trialled. In addition, the registry will capture aspects such as the disease burden as measured in Quality of Life studies and socio/economic impacts. There are many problems being addressed including design of patient input pages having the need to cater for many languages and meeting the regulatory requirements of the nations taking part. Recruitment is progressing and it is aimed to enrol 1000 patients by the end of 2022. The need to achieve EMA qualification will also be pursued.

Ara Parseghian Medical Research Foundation

Dr Sean Kassen, Director of the Fund from the foundation provided an update on some of their activities by a review of the Foundation's formation and its progress over 20 years. The Foundation works closely with the University of Notre Dame IA and has been a major fundraiser in support of research into NPC science.

Recent activities have focused more widely on organising the Patient Focused Drug Development group to promote the patient voice in all aspects of drug development including regulatory aspects. A report on activities and progress – Voice of Patient was due out but has been delayed. More recently the Foundation has joined with the FireFly Fund and others to promote new-born screening for NPC throughout the USA, this in recognition that early diagnosis is essential for therapies to be optimum. The Foundation has recently teamed with others to accelerate the pace of therapeutic development in a new approach – Accelerate – Develop – Cure.

Disease Prevention Through Early Diagnosis

An update on New Born Screening (NBS) from the USA

Dr Melissa Wasserstein from the Children's Hospital, at Montefiore, Bronx NY provided an update on the Screen Plus initiative being undertaken in the USA. This programme of work is a continuation from a previous initiative to introduce a pilot research programme and improve the detection of lysosomal storage diseases (LSDs) at birth thereby providing an option for early intervention for babies testing positive for a condition.

Having established that the hospital infrastructure could accommodate the introduction of an extended NBS programme the recently approved Screen Plus study aims to recruit 175,000 families for a 5 year study of 14 disorders, although this aspect has been delayed due to the Covid pandemic. Disorders may be included or removed as the research unfolds and the criteria for inclusion must meet the RUSP requirements such as availability of a validated test, significant health risk if not tested, benefits to outweigh burdens, availability of a therapy.

Screen Plus is a research programme aimed at improving the accuracy of screening. The approach being used is to employ multi-tiered assays for each condition which will reduce false positives and negatives. Other features of the study include long term follow up to determine the impact of NBS on health and to evaluate the validity of ELSI factors eg ethics, legal in the NBS process. The study is the largest NBS programme in the USA and includes NPC and ASMD (NPA/B). Patient groups such as the Firefly Foundation, AMPRF and other US groups are part of an integrated team progressing this important programme.

In addition to the NBS pilot other studies are planned to look at affected siblings for comparison of genetic factors and to form a

Clinical Round Table to address problems arising and monitor the progress of the study. Topics of interest include determination of the timing for the introduction of treatment following a positive test. The planned programme is subject to re-scheduling as a result of Corona pandemic.

An Update on NBS in the UK

This update was provided by Mrs Georgina Morton from the MLD Trust. Her interest in NBS derived from personal family experience with her daughter being diagnosed with MLD after presenting with symptoms. Bone marrow gene therapy treatment was obtained in Italy with daughter responding well to treatment. It is essential in MLD and many other conditions that therapy is administered early in the disease progress otherwise it is too late. This realisation has inspired her to form the Archangel Trust to promote early diagnosis of MLD but realistically this needs to be at the new born stage. She has recognised that the problem exists for many other diseases and has set up a forum for collaboration.

Most countries adhere to the World Health Organisation criteria for approving a NBS programme into the healthcare system. Different nations interpret the criteria in different ways and in the UK a strict interpretation is adopted by the National Screening Committee. This has led to the UK falling behind most of the advanced nations in introducing NBS testing for serious diseases. Many of the patient organisations feel that the current system of approval is in need of overhaul and NBS testing should not be dealt with under the same authority as is responsible for general population screening. The Archangel Trust will pursue change in the system by enrolling the support of politicians, medial and scientific professionals and patient support organisations. Progress is being made but COVID intervention is making matters difficult.

Clinical Trials

Presentations were given as follows. More detail of presenters can be found in the Conference Programme online:

• Dr Robin Lachmann, National Hospital for Neurology, London. Clinical Development of Olipudase Alpha for ASMD Niemann-Pick disease type B.

• Update on Cyclo Therapeutics Inc. Clinical Trials for NPC. Dr Caroline Hastings, Children's Hospital and Research Center, Oakland, CA, USA.

A UK update from Dr Reena Sharma, Salford Royal Foundation NHS Trust and Dr Sharon Hrynkow on development of the Trappsol treatment from a company perspective.

• Dr Fran Platt, University of Oxford and Dr Tatiana Bremova University Hospital, Bern, Switzerland gave an update on the new treatment, IB1001-201 N-acetyl-L-leucine aimed at symptomatic and neuroprotective aspects of NPC.

• Dr Elizabeth Berry-Travis from Rush University, Chicago, USA provided an update on trials with Adrabetadex (formerly VTS270).

• Dr Marc Patterson of Mayo Clinic, Minnesota, USA described treatment of NPC with an HSP amplifier similar to the Orphazyme approach.

Comment on the therapeutic development and clinical trials landscape

Many years passed before the first therapy became available for NPC. This was miglustat and over a long period has been shown to slow the rate of disease progression and improve distressing symptoms such as swallowing. Other therapies are either in development or undergoing clinical trials as reported in this conference.

The basic principle of each therapy varies considerably as does the method of administration. These developments have been shown to provide effective intervention in pre-clinical studies on cell and animal models but in all instances, treatment has so far shown to be truly effective if administered early, ideally at the pre-symptomatic stage. Thereafter results show diminishing returns and at best disease stabilisation.

Those families involved in clinical trials will be the best judges of the efficacy of a trial therapy they are on. The trial outcomes will be reported by each company for their therapy and the results will be assessed by the regulators and these results will inform the Marketing Authorisation that the companies seek in order to receive reimbursement for the significant funds and effort that has been invested in their drug development programme.

We have seen from experience using miglustat that many years may go by before the beneficial effects of the drug become apparent. Not all regulatory bodies are convinced on the question of efficacy and miglustat remains unlicensed in the USA. Even when licencing is achieved the question of 'who pays' is an increasing problem given the high price of the drug and limited market.

Looking to the future, adoption of early disease detection measures such as new-born screening will provide a better starting point for therapies to demonstrate how good they really are and provide the major benefit of warning families of the danger of a disease when considering adding to the family. The UK needs to adopt greater use of pilot schemes such as the US Screen Plus programme. Every year that goes by means we lose more people to these diseases. We need action and investment as a matter of urgency. The Archangel Trust initiative is a good starting point.

Many thanks, as ever, to NPUK Trustee and Research Co-ordinator Bill Owen for this in-depth overview of our Interactive Workshop and the current landscape of Niemann-Pick research.

If you have a question for Bill related to this article, please get in touch at: bill@npuk.org

SAVE THE DATE

15th - 17th October 2021

Wyboston Lakes, Bedfordshire

Niemann-Pick UK (NPUK) would like to cordially invite you to our 28th Annual Family Conference & 12th Interactive Workshop on Niemann-Pick Diseases - which we hope will be a return to an in-person meeting and provide an opportunity to celebrate our 30th birthday!

CLINICAL TRIAL UPDATES:

When Niemann-Pick UK (NPUK) was founded in 1991 we could have only dreamed of so much activity in the field of Niemann-Pick disease research! Following is a brief summary of recent press releases regarding current clinical trials, to learn more please visit: npuk.org.

Cyclo Therapeutics Inc

Cyclo Therapeutics Phase 3 Pivotal Program Can Begin Enrollment per US FDA: "We are very pleased to receive the FDA's notification that we can proceed with the Phase 3 clinical trial. This announcement signifies the most significant milestone for our company but also for the patients, families and caregivers in the NPC Community as well." said company CEO N Scott Fine. "We will continue to collaborate with the NPC community as we drive this study forward towards market authorization as rapidly as possible."

Full press release available, here: npuk.org/cyclo-therapeutics-phase-3-pivotal-program-can-begin-enrollment-per-us-fda/

Orphazyme:

Orphazyme Submits European Marketing Authorisation Application for Arimocolmol for Treatment of Niemann-Pick disease type C: Kim Stratton, Chief Executive Officer, said, "This filing in Europe is a significant milestone for Orphazyme as we work toward our first potential approvals of arimoclomol in major markets. There are few options today that can address the devastating effects of NPC, and we are hopeful we can address an important need for this community. We look forward to working with EMA as they complete their review of our application."

Full press release available, here: .npuk.org/orphazyme-submits-european-marketing-authorisation-application-for-arimocolmol-for-treatment-of-niemann-pick-disease-type-c/

IntraBio Inc

IntraBio Reports Further Detail on Positive Data from IB1001 Multinational Clinical Trial for the Treatment of Niemann-Pick disease Type C: "IntraBio Inc today announced positive results from the full data set for its multinational clinical trial of IB1001 (N-acetyl-L-leucine) for the treatment of Niemann-Pick disease Type C (NPC). In total, 33 subjects aged 7 to 64 years with a confirmed diagnosis of NPC were enrolled across 9 clinical trial sites in the United States, United Kingdom and Europe. [---]

A total of 33 patients were recruited into the study and 32 patients were included in the mITT analysis set. IB1001 demonstrated a statistically significant and clear clinically meaningful improvement in symptoms, functioning, and quality of life for pediatric and adult patients with NPC.

Full press release available, here: npuk.org/intrabio-reports-further-detail-on-positive-data-from-ib1001-multinational-clinical-trial-for-the-treatment-of-niemann-pick-disease-type-c/

NOTE: clinical trial updates are current at the time of printing, for the latest articles please visit our website at npuk.org and look in the News section.

Graham Kirk's

My name is Graham Kirk, I'm 35 years of age and I've got Niemann-Pick disease type C. I want to tell you a little about my story, spanning from 2010 (the year of my diagnosis) until now, today, 2020. So what am I? I'm a Dad, a brother, a son, a husband, a friend, an uncle, and a godfather – the picture on the right is of my three children; Emily, Alannah, and Jamie, and the one below is a picture of myself and my wife (Ciara) on our wedding day along with the rest of the Kirk family, so my Mam and Dad, and my sister Paula sitting down – in this photograph there's two people with NP-C, and unfortunately one isn't with us today. It was taken two years after my original diagnosis.

So September is a special month for me as there are a lot of milestones in the month; I received my diagnosis in September 2010, days later we flew to attend our first NPUK Annual Family Conference, sadly September is the month my elder sister passed away at age 30 (2012), and it's also her birthday month.

Now, believe it or not but this photo was taken a week before my diagnosis. I've always been an active child; I've played Gaelic Football all the way up to adult senior level, I play golf twice a week and currently play off 9... it's creeping up a bit but I can't blame Niemann-Pick for that, and during the first lockdown I started to run 5Ks just to keep the mind active and fresh.

I don't work anymore - this is a decision we took three years ago after we were married.

story

So far...

I've always had a strong work ethic; I worked on local farms as a teenager, had shifts at the local SPAR shop all the way throughout college, which helped get me through, and after that I finally landed my dream job as a media advertising graduate. I had a big secret though...being so well, but having NP-C, it was not something that I wanted to disclose to colleagues. So when I had appointments I used to just bunk off rather than tell them, as I knew the appointment was more important than the work. But it also came apparent to me after a number of years that despite moving to three different agencies, I was stuck at a certain level – I just couldn't move forward.

Our decision was to start a family after we were married. We always knew we wanted to have a family, but now that Niemann-Pick was involved Ciara went for a genetic test and found out she wasn't a carrier...and that's when we had Allanah in October 2013, followed by twins 18 months later, Emily and Jamie. My family is my focus, it is my everything. They drive me mental, but I love them to bits!

So I've been living with NP-C for ten years now. It all started with my sister having it, having been diagnosed in early 2010, which led me to go see her Doctor, Professor Healy, in Dublin. He was confident from his initial assessment I had it too, and unfortunately the blood test confirmed this...it was a total shock to me personally; I was healthy, active, working, in a relationship and independent in every way. This was far, far away from the typical lifestyle of a person affected by this disease.

"...my family is my focus, it is my everything. They drive me mental, but I love them to bits..."

When I went on my first date with Ciara she found it strange that I flicked my head up and down, but she put it down to first date nerves... little did we know it was vertical gaze palsy. As time progressed I became more aware of my symptoms and recognised when I had new ones; my eye movements are still the same, I've began suffering from cramps usually after a long day of activity (walking, golfing, running), I find the strength in my hands is also reducing (opening jars, lifting kettles etc.), on rare occasions in the early days I did used to have panic attacks which would come

on unexpectedly - I've learned to control these and it's been years since I've had one. Recently I joined the Mater Hospital and they've given me hearing aids following my latest check up, which are great things to have!

So what have I learned from living with Niemann-Pick disease type C? Well, NPUK is a lifeline, a massive community network for me which means I don't feel isolated! I'm not sure of the exact numbers but you can probably count on one hand the amount of patients with NP-C in Ireland so to have this connectivity with the UK is something which I feel really passionate about.

Myself and Ciara make the most of every moment, in 2018 we went on a trip to Salford in Manchester Hospital to the Mark Holland Metabolic team...but it conveniently coincided with the Christmas market! So Ciara and I, in both 2018 and 2019, enjoyed the Christmas market and had a lot of fun...of course 2020 is to be confirmed at this point! I've learned to lead a positive and active life and to not allow NP-C to dictate my journey.

I do have questions though...would a simple screening with an eye test have caught my condition earlier in my life? On reflection should my enlarged spleen and issues surrounding my birth have been investigated further at the time? We'll never know, but it's something that annoys me. Looking ahead, dealing with Niemann-Pick is one thing, but dealing with it during a pandemic is completely different...I find it frustrating, obviously, with the way people view me. They don't know I have an underlying condition, but I planned to stay safe and well; I cut down my social contacts, I also self isolate if I hear there's an outbreak close to the area.

I'm looking forward to 2021 for starting a new journey with the Dublin clinicians at the Mater Hospital. I will continue to make memories with my family and my friends, and I hope that next year will continue to go the way it has been for me in my life to date. I know that maintenance of my fitness, and making good lifestyle choices, is another thing which is really important - I know I've mentioned jogging since the first lockdown came in...but it's something I know not every Niemann-Pick disease type C patient can do, and I feel blessed to be doing it. I think about this every time I run.

Graham originally shared his story in a presentation at the Digital NPUK Annual Family Conference & Interactive Workshop 2020

OUR NIGEL

Originally presented by the wonders of a Zoom recording at our Digital Annual Family Conference, Christine Willman's story of her family's journey with her son's Niemann-Pick diagnosis just had to be shared again on these pages...it's a warm, funny, emotional, and above all else, *real*, insight into her life...

Hello to anyone who happens to be out there! I'm Christine, long overdue for retirement Mum to Nigel who has been diagnosed with Niemann-Pick disease type C (NP-C), something I'm not sure if I've quite come to terms with yet.

What was the road to diagnosis like for you?

The route to diagnosis is probably best described as a Northern Rail network journey because it started in Clitheroe and we ended up in Salford. But we went via Blackburn, Preston, St. Mary's at Manchester and then back to Salford where we finally received his diagnosis.

Depending on what reference point you use, this took between 2 and 17 years...it became an epic journey even by current rail standards!

When did you receive the diagnosis? How did it affect your lives?

I suppose our lives became a car crash when Nigel, who is now 41 this year, suffered a psychotic episode at the age of 18. Initially it was a bit like being on a rollercoaster ride with an alien, his

behaviours were frustrating and they were frightening – I think we, as his family, didn't know how to deal with him at that time. It involved our whole world sort of falling apart; he lost his job, relationships broke down, and there was alienation from family and friends... so you can imagine it was a pretty traumatic time for all of us. We'd gone from everything being mapped out and life running along quite nicely to suddenly being thrown into, what can only be described as, a maelstrom of emotions, not knowing what was going on.

I suppose in retrospect the next 15 years were an undiagnosed phase in terms of Niemann-Pick disease. He was treated for bi-polar during this period, and this continues to be the case.

How did the diagnosis make you feel initially?

I suppose this comes more into the challenges really of that time, sort of accepting the diagnosis, or not. But then it wasn't my diagnosis to accept, it wasn't my illness and I think that is one of the challenges. Especially when your son is being diagnosed as an adult...I think I felt guilty – you start to think, was it our fault with it being genetic? Had we missed the earlier signs?

"...we had already lost our son in many ways to his mental illness..."

I felt like we were starting to suffer the loss of our son, again. The words that stuck with me from that time were that [Niemann-Pick disease type C is] "life limiting" and "life threatening". We had already lost our son in many ways to his mental illness, so I suppose we were now facing this almost surreal scenario, and also trying to understand it! What was actually being said to us? We were trying to take all of this on board, not that we were being overloaded, but we were asking a lot of questions and trying to understand the disease; how it was going to progress, what it was going to mean for Nigel in terms of his everyday life, and also perhaps a prognosis.

It became very frustrating I think. There were feelings of frustration surrounding the practicalities of trying to support Nigel as an adult in view of his age, and also that we weren't typical of many families that were receiving this diagnosis as Nigel lived independently, he didn't live with us but now had this dual-diagnosis [to

contend with]! We've always being a practical sort of family, not given to dramatics, we're usually like: "this is what it is, right. Now we've got to deal with it". But in a way, as I came to see the problems that other families had, I think I felt like a bit of an imposter as people had much more difficult lives and problems to deal with when compared with us. So I was comparing with other people's lives who had the same diagnosis...and yeah, they're all very different.

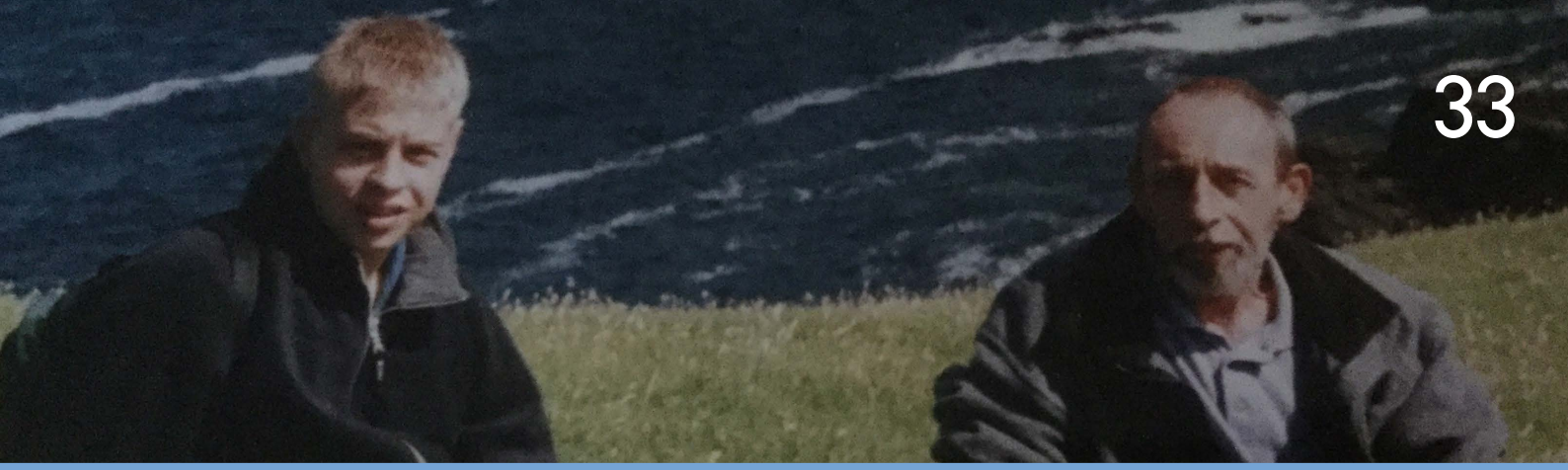
Is there anything you would like to share with the NPUK Community?

Well when I was talking about the concept of bereavement and loss, I'd also like to say we had to deal with another bereavement in the sense of losing John. So me losing my husband, and Nigel losing his Dad. So I suppose again that is something else that happened along our journey together...it's sometimes good to get these things out, you know? The things you try and push down on a daily basis when you're just getting on with the nitty gritty of life, otherwise you'd just feel like a wrung-out dish cloth wouldn't you? The way I would sum up [trying to contend with NP-C] would be, it's a little bit like trying to knit spaghetti.

How has the support of NPUK helped yourself and your family?

I didn't realise just how much support myself and Nigel have had from NPUK. We've had the clinical support from the outset which obviously is still ongoing; Laura [NPUK Clinical Nurse Specialist] coming to visit Nigel at his home and seeing how we are doing, checking if there are any questions that we needed to ask and telling us what support we would continue to have at Salford. We had all of the information, but knowing where we go from here is different, so I think this support has been invaluable really.

The emotional support has also been helpful, from all of the NPUK Team really!



I think looking at it now and analysing it, it is just like that metaphorical big hug, although admittedly other than Laura and Dr. Sharma we didn't really engage too much initially. But then subsequently when things sort of developed, and practical issues arose, Elizabeth [Former NPUK Senior Families Advocate] came along and now Karen [NPUK Families Officer] as well, Toni [NPUK Chief Executive] and Steve [NPUK Project Families Officer]... with the Conference events there has been further help and information, there's been social events with the Christmas Party, and other online things NPUK has put on such as seminars for individuals and families where you could go and find out a little bit about the genetic side and the research, all of the things that are going on there. I think for me I had to take a little step back from that, and just deal with the practicalities of the illness and the day to day living because that's how I operate. I will leave that stuff to the experts, and not overburden myself with things that are fascinating, interesting, and very important but that...I don't perhaps need, it's just something additional that I didn't need (at least there and then!)

I think some other ways in which NPUK have helped has been financially, helping with the benefits side of things and also the grant monies. Being able to access that, particularly this year during COVID-19 [with the NPUK COVID-19 Emergency Response Fund], it helped. When Nigel was shielding more we were able to decorate his living room, which was a project he had been talking about but not getting motivated to do – so that was an opportunity to do that. Steve has been actively involved in this and more since John and Nigel went to an NPUK Conference a few years ago, and more recently has helped with the Lads, Dads, and Carers group, trying to link Nigel with other people online. We're still working on that...he's decided it's not his forte for now, but who knows? Maybe one day, we live in hope. I'm personally coming out of my dinosaur phase now, starting to access more of the online support, so perhaps I might even start social networking!

If you have a story worth telling, we would love to feature you in the next issue - please email us at: info@npuk.org

Debbie Kaflowitz
a mother's story.

Debbie Kaflowitz originally shared her personal story with us at this year's NPUK Digital Annual Family Conference & Interactive Workshop: we thank her for allowing us to share it again in the following pages.

Our daughter Rachael had NPC, adult onset. From the time she was about 5 years old, I knew something was wrong. Her thinking was just not right. I clearly remember one day when Rachael was doing a magic trick with her friend Ali. They held up a cloth and Ali said, "See there is no hole." Rachael turned the cloth around and said, "Look, there is no hole on this side either." Most children learn that a hole goes all the way through just by living. Not Rachael. It gave me pause, but I wasn't really worried. She was only 5. Over the years, this continued on a larger scale. Even by 10 years old, she couldn't get the hang of the days of the week and months of the year. She knew her birthday was June 21st, but she had no idea the weather would probably be warm and there definitely wouldn't snow. She had no concept of time or numbers. If a teacher told her to turn to page 35, she had no strategy to find it. She would flip pages in every direction, hoping the right page would magically appear. She would say things like turn the heat on, I'm hot. I knew she had some kind of processing problem, but not one I had ever seen in all my years of teaching before. I certainly never considered it was a fatal disease.

In elementary school, I was constantly telling teachers I was concerned about her skills. They kept telling me how cute and well-behaved she was and that I was overreacting. I was a teacher myself and I knew something was wrong.

Her cognitive ability was getting worse and worse and finally they diagnosed her with a learning disability. By middle school, she was barely understanding anything in class. She would process pieces, but the rest of the information went right over her head. I'll tell you one funny story because certainly we must laugh. At about age 15, Rachael came home and said her class was going to be reading a book about tequila. Steve and I looked at each other and told Rachael there was no way the school would have them read a book about tequila. Rachael thought for awhile and said it's about a bird, too. We kept saying Tequila bird in our heads over and over again. I finally looked at her and said, "To Kill a Mockingbird?" Yes, that was it.

"...I certainly never considered it was a fatal disease..."

This lack of cognitive ability started to affect her relationship with friends. By 6th grade nobody really talked to her or invited her anywhere. By 9th grade she was pretty much invisible. In 11th grade when she left the public school because it didn't fit her needs, only 1 student noticed she was gone and called her.

However, Rachael did have things in her life that made her happy. She was wonderful with small children and babysat a lot. On the weekend, the little children in the neighborhood would knock on our door and ask if Rachael could come out and play. She took ballet lessons and performed. She was a counselor at a day camp during the summer and everyone loved her.

In junior year of high school, Rachael had her first big sign that something was very wrong. She had a complete mental breakdown. It was like her brain had disappeared. She couldn't find her way around the house. She would look at her toothbrush and say, "I know I should do something with that, but I don't know what." At first the doctor thought it was a brain tumor. Rachael started having manic episodes and diagnosed as bipolar. Next became psychotic, thinking people were after her. So Rachael started as bipolar, then added psychotic, then schizoaffective.

She couldn't go to school. One of us had to be with her at all times. I was teaching, so I would go to work first. All the most important subjects were moved to the morning. I would come home at noon and Steve would leave for work because he could move his meetings to the afternoon and stay late. It was an extremely difficult time. Rachael's psychiatrist had been trying all kinds of medications and combinations from the first mental breakdown, but nothing seemed to work.

Over the next 12 years, we were mostly focused on Rachael's mental illness. During that time, she was hospitalized in mental health units at 4 different hospitals, each one for about 8 weeks. However, at each one, the doctors would try lots of medicines and then throw up their hands, sending her home sometimes a little better but sometimes worse. She never lasted long on the meds from the hospital. Dr Behar had to continued juggling medications and dosages, maybe keeping one of the hospital's suggestions. At some points she was stable for a few months at a time, but it was never long lasting.

During one of those stable periods, Rachael was able to get a driver's license and hold down a part-time job bagging groceries. Eventually we had to take her license away and she lost her job.

When Rachael turned 21, she no longer qualified for any school services. At that point, we were at the mercy of state services, whose services were limited. At this time, our lives revolved around finding things for Rachael to do that made her happy. I searched high and low for a day program to fit Rachael's needs, but it was difficult to find the right program for her. First, she was over 18, so she needed an adult program. The majority of the people in those programs were mostly 50-60 year old men. She was also very socially aware, so putting her someplace where people sat in wheelchairs without interacting much wasn't good either. We finally found two very good programs that Rachael could attend.

This was an especially difficult time for Rachael. I have used the phrase one foot in the typical world and one foot in the disabled world many times. Rachael still knew the way her life was supposed to be. She hadnexpected to go to college, become a teacher like me, get married, and start a family. She knew her peers were moving forward while she was, at first standing still, and then going backwards. Even the children she babysat for were passing her. She would often cry, asking for her car keys and saying things like, "Who is going to marry me?"

Toward the end of those 12 years, other strange symptoms started to appear. She started falling for no reason. Her gait became awkward and she started walking on her toes. She would always look down when she walked because she said otherwise she couldn't see where she was going.

That's when Dr. Behar was convinced that either Rachael was never bipolar or schizoaffective in the first place and had an illness nobody could diagnose or she had two illnesses. His guess was that Rachael had a genetic illness that had mental health symptoms but also the symptoms that had just started to appear. Every year Dr. Behar went to a medical conference for psychiatrists. The next time he attended, he went to a session, called genetic illnesses that mimic bipolar, with Rachael in mind. He called me from the conference saying that the speaker had just described Rachael and he was going to find us a doctor there. After waiting months for an appointment, we finally saw a neurogeneticist who diagnosed Rachael in 5 minutes. He told us that after reading a letter from me and one from Dr. Behar, he was pretty sure he knew what it was. He just needed to check her eyes. NPC was then confirmed by a blood test. By this time Rachael was 26 years old.

While there was great sorrow with the diagnosis, there was also a little bit of relief. I had spent every free minute of the past 12 years, searching for the doctor who could fix Rachael and feeling guilty that I couldn't find someone.

Now we moved from what's wrong with Rachael to what do we do now. Actually not much changed for awhile. Rachael was already at her day programs Tuesday-Friday. She was still at the point where we could occupy her on the off days. Little by little Rachael started to decline. She went from walking on her own to using a walker outside the house. Next came holding on to one of us at home and using a wheelchair out of the house. She started becoming more childlike in her emotions and her cognitive thinking. Actually this was good. She now loved her day programs. They made her feel welcome and successful because painting, singing, other art projects were things she could do. Plus there was always someone helping her or simply paying attention to her and she loved that. That's what she liked most-people. Her friends were the instructors and aides. They loved her because she was one of the few people who could talk, laugh, and express her feelings.

"...while there was great sorrow with the diagnosis, there was also a little bit of relief..."

It was during this time when Rachael stopped asking for the keys to her car or the difficult questions of who is going to marry her. These upsetting thoughts could be triggered by running into a high school peer who was now out of college and holding a baby, but they was short lived. As long as she was out of the house, with people, who were NOT us, and occupied, she was happy.

Home was a different story. If someone asked me if I had any children, I would respond with yes, a 5 year old, a 13 year old, and a 25 year old, but they are all one person. The 13 and the 25 year old didn't want to be stuck in the house with her parents. Quite frankly, her parents didn't want to be captive with the 13 and 25 year old. Anything we did or said was wrong. For awhile we could take her to a movie, the zoo, a Broadway show, the beach in the summer, but those became more and more difficult as it became more difficult for her to focus her eyes ahead of her, as she became more easily tired, as she stopped being able to follow a story line.

We hired a companion around her age for Mondays, Saturdays, and eventually for Sunday for a few hours each day. They started by going out to lunch and then doing some errands in town and volunteering at the library. That world started to shrink, too, as it became too dangerous for her to get in and out of the car for errands, navigate herself anywhere, especially the bathroom. They list of foods she could eat safely was growing smaller and smaller. Finally Rachael and the aide had to stay at our house. At first she made bracelets, and when they became too difficult she made necklaces, which some of you have. Rachael would also do scrapbooking. She decided on backgrounds, where the pictures would go, and embellishments while the companion was her hands.

Little by little, Rachael's ability to do anything declined. She could no longer dress herself, bathe herself, or brush her teeth. She would wake up

many times during the night and try to get out of bed even though there were lots of bedguards. Steve and I started taking turns sleeping in bed with her. By this time, Steve and I were in our sixties and not sleeping every other night was taking a toll, so we hired night aides from midnight to 7 AM which really helped.

About a year before Rachael passed away, I noticed a decline again. Rachael was thin, but 5'7". I was 5" 4. Because Rachael couldn't help as much, I was having a harder and harder time getting her in and out of the car myself. Her food had to be cut into smaller and smaller pieces. When she got into bed, she didn't have the strength to move herself up so her head was on the pillow and neither did I. Although she loved to eat and could finish a big bowl of spaghetti or Mac and cheese, she was getting thinner and thinner.

On Sept 7th 2018, we brought Rachael to the emergency room with a very high fever that came out of nowhere. They admitted her with parainfluenza, which is characterized by cycling though high temperatures and then back to normal. 3 days into the hospital stay she developed pneumonia. After 2 months of our hopes being lifted and then dashed again, we knew there was nothing left to do and we moved her to hospice, where she passed away on October 25th.

Never Forgotten

Author Unknown

You are not forgotten, my love.
Nor will you ever be.
As long as life and memory last
your soul will live in me.

I'll miss you now...my heart is sore.
As time goes by - I'll miss you more.

Your loving smile, your gentle face,
No one can fill, your vacant place.

With this poem we remember Rachael Kaflowitz (21/6/1985 – 25/10/2018), Lorraine Carr (10/3/94-26/10/20), and Carla Wood (1/8/84-16/3/20).

Our love and memories of them will live forever, and are what inspires us to continue our work.

Helen Carter tells us the Hollie Foundation story...

The Hollie Foundation is a grant giving organisation that supports patients and families living with Niemann-Pick diseases - we sat down with their Chair and Founder Helen Carter to learn more:

What was the main inspiration behind setting up The Hollie Foundation?

My daughter Hollie who was diagnosed with NP-C at 2 years old and all the lovely families we had met since her diagnosis. We really wanted to help raise awareness of the condition and support others affected by it.

Did you have any previous experience of small charities and the charity sector at large?

Yes, at birth Hollie was classed as having an undiagnosed liver disease and during the first two years of her life we received invaluable support from the Children's Liver Disease Foundation. When we were subsequently informed two years later that Hollie may have NPC we reached out to NPUK and they made such a positive difference to our lives in the dark days of waiting for diagnosis, helping us to function and get through each day during and after diagnosis. This made us realise what a critical role small charities play in supporting vital information and comfort.

Are there any stand out projects or campaigns you feel have had a big impact?

Our annual charity balls have been very successful over the years and helped us to raise a fantastic sum of money and great awareness.

The funding from these events has enabled us to part-fund the essential post of NPUK Families Officer for many years.

What has been your proudest Hollie Foundation moment?

Seeing the community pull together for various fundraising events to support such a rare disease like Niemann-Pick, simply because they had been so inspired by hearing Hollie's story.

How much do your personal experiences inform the work you do?

They have a huge impact on the work we do. Our own personal experiences of living with the condition enables people to understand the disease better and raise awareness and funds. Hollie is now at an age where she wants to share her own story and experiences of living with a rare disease, and this alone has a big impact.

What impact has the current COVID-19 pandemic had on your organisation?

Our fundraising has been severely affected. All planned fundraising events had to be cancelled during the first lockdown and our grant programme has been significantly impacted. We are solely reliant on the wonderful people who have set up online fundraising events.

What is one message you would give to a potential Hollie Foundation fundraiser?

There is a real risk of our grant funding programme having to close over the coming 12 months without additional funds. We need your help more than ever now to enable us to continue supporting families and other charities with grant funding for enhancement of support services, equipment, adaptions around the home and other emergency funding.

Many thanks to Helen for sharing her insight into this fantastic small charity! If you have been motivated to support the work they do, you can find out more and donate at: www.theholliefoundation.com, or check out their social media accounts at the handle below.

@theholliefoundation

inpda

NPUK
is part of the International
Niemann-Pick Disease Alliance (INPDA)
a global network of Niemann-Pick advocacy
groups working together to facilitate progress in the field.
Find out what some of the inspiring groups have been up to, here:

Niemann-Pick Argentina have been working with their community via Zoom throughout the COVID-19 situation. During Niemann-Pick Disease Awareness month (October), they celebrated the 15th anniversary of the founding of the Association!

The Australian NPC Disease Foundation has been preparing for a face-to-face 2021 Conference and Gala if possible. They also have been trying to work with the Australian government for access and approval of much needed drugs for patients with early onset NPC.

Niemann-Pick & Batten Brazil Association during October have been posting various videos of families, partners and collaborators within their community, on their social media.

Niemann-Pick Canada has recently partnered with the medical data science company AllStripes and other leading Niemann-Pick Foundations in recruitment for a sibling study research project focused on early diagnosis and intervention resulting in better outcomes and quality of life for patients with NPC.

In France, **Vaincre Maladies Lysosomal (VML)** have been planning a web conference for NPC and NPB families taking place in November. The web event will be an opportunity to share the latest on NP disease, clinical trial updates and the impact of COVID-19 for the community.

Niemann-Pick Selbthilfegruppe have been involved in the newly founded organisation of lysosomal storage diseases (LysoNET) as a representative in collaboration with other parent groups. The focus of this organisation is to find ways to support public and clinical awareness for rare issues, and therefore recognise that working together will make progress more attainable.

The Italian Niemann-Pick Association started a series of initiatives aimed at raising further awareness of Niemann-Pick disease following the impact of Global Niemann Pick Awareness Day. This initiative has included a communications campaign through social networks and the publication of articles dedicated to the topic in prestigious magazines such as Focus.

Niemann-Pick Association of Fuenlabrada, in response to the COVID-19 situation, have begun to do online events such as selling tickets for their Christmas lottery and a virtual charity race which was a huge success. It's been fantastic to see the positive energy of their community throughout this year!

The Niemann-Pick Foundation of Spain has supported the recruitment of patients with NP-C to be involved in a unprecedented trial beginning in October 2020 following the success of basic research carried out at the Severo Ochoa Institute showing the effect of Efavirenz, antiretroviral, in improving the synaptic function of mice.

NP Suisse, following the INPDA virtual meeting on July 26th and the European virtual meeting on August 27th, decided to support the Berne Hospital in its efforts to take over the Abrabetadex treatment under compassionate use. The first delivery of Abrabetadex to a former Birmingham Swiss patient took place on September 16th.

The Hollie Foundation has been working hard to maintain its grant funding program despite the impact of COVID-19 on their fundraising income. The Hollie Foundation partnered with Niemann-Pick UK and contributed towards the Coronavirus Emergency Response Fund, in recognition of the negative impact that the Coronavirus pandemic may be having on individuals and families within the community.

To support families in their community, the **National Niemann-Pick Disease Foundation (NNPDF)** held their annual Family Support & Medical Conference in July, but rather than a weekend together in San Antonio, Texas, as planned, they held a virtual event that spanned over 3 days and in which over 300 families and speakers took part!

The Wylder Nation Foundation continues to make strides on their FAAH inhibitor program aimed at addressing the neurological manifestations of ASMD, and potentially other Lysosomal Storage Diseases including NPC.

The following article has been adapted from the International Niemann-Pick Disease Alliance e-newsletter. For full versions, including previous issues, please visit the official INPDA website at: www.inpda.org/newsletter

for further information please visit: inpda.org

Celebrat

achievements so far and planning for the future

The International Niemann-Pick Disease Registry (INPDR) aims to improve standards of diagnosis, care and treatment for the global Niemann-Pick patient community. In the following article, INPDR Communications Consultant, Angela Wilson gives us an update of work so far.

It's impossible not to mention how challenging the last year has been for so many of us in the Niemann-Pick disease community and beyond. We hope all of the NPUK family are managing to keep safe and well. The team at the INPDR, many of whom you already know, are first and foremost human so we offer solidarity to everyone navigating the twists and turns of COVID-19 and various restrictions.

For the INPDR, the recent situation has offered us the chance to make some developmental improvements in the background – some of which won't be immediately obvious to everyone but they will support and enable the collection of data that reflects the true patient experience and impact of Niemann-Pick diseases.

The INPDR has recently received a generous grant from the US based Ara Parseghian Medical Research Fund. This is a significant achievement and will support a two-year project to expedite growth of the Registry and recruitment in the US, including the appointment of a Clinical Research Associate. Ultimately, this grant and the work it enables will benefit the global Niemann-Pick community, as it will help to make sure the Registry offers rich and powerful data that is reflective of the global patient experience.

The INPDR continues to recruit patients from the UK and across the globe, via both our Clinician Reported Database and our Patient Reported Database. We've also been working on making sure the Registry is as accessible as possible. For example, we will be offering postal consent, translated information documents and we have added some tutorials to our website to help those wishing to sign up to the Patient Reported Database.

ing

Visit www.inpdr.org and check out the 'How to join' page in the 'Patients' section. To ensure the data we collect is of the highest quality, we have recently undertaken a review of data collected so far. We have collaborated with the expert clinicians and researchers who form part of our Scientific Advisory Committee to agree a set of amendments that will improve data collection and quality and enable the greatest impact on research, care and treatment for Niemann-Pick patients.

During Niemann-Pick disease Awareness Month we ran a social media campaign highlighting our achievements so far. It was pleasing to see how far we've come since the Registry was established in 2013 and we cannot emphasise enough how our progress is your progress. We have some ambitious plans for the future but we can't achieve them without you.

If you want to find out more about our plans we have just published our Strategic Business Plan 2020 -2022, which sets out our aims for the next phase of our development and explains ways in which the INPDR can positively impact outcomes for Niemann-Pick patients, their families and the wider rare disease community.

Finally, a massive thanks to NPUK, their families and friends for welcoming us to their first ever digital family conference. It was such a loss to not have the opportunity to see people face to face but we really enjoyed taking part and hope we were able to share useful information and answer any questions you may have.

We were so grateful to still be able to connect with the heart and soul of our work – patients and their families – thank you to NPUK for making it happen.

Every single piece of data in the Registry makes a difference – we are greater than the sum of our parts - so if you have any questions or suggestions about our work, the difference it can make and how you

Visit www.inpdr.org for further information.

np uk FUND

Now more than ever our fundraisers are essential to NPUK ensuring it can continue to provide our vital support services - here are just a few of our fundraising heroes!

In February, before pubs became a distant memory, **Emma Floisand**, **Pete Clarke** and friends arranged a meat raffle fundraising event at the Angel Vaults, Evesham. This event managed to raise a total of £1015! Thank you to everyone who arranged the event and took part.

The Big Yellow Friday Comedy Night took place on the 28th February and NPUK were one of the three charities that the proceeds from this fundraiser went towards. Organised by Keith and Julie Algie the night raised an amazing £7,168.75 inc. gift aid! We are so pleased to have been involved and grateful to the community for raising funds for us.

During lockdown **Terry Colwell** decided to have a bit of a re-style and shaved his head to fundraise for Niemann-Pick UK. This bold move managed to raise £600 and we are incredibly grateful for his commitment to NPUK and we hope, as the days grow colder, he has a good selection of woolly hats.

Our **super scientists** working in the field of Niemann Pick Disease completed a biking challenge as part of the GNR fundraising campaign in September. If you have attended our Annual Family Conference & Interactive Workshop in the past you may be aware that our super scientists take part in a fundraising challenge for NPUK in the lead up to the Conference. Although we did not have the physical Conference this year, our scientists were still enthusiastic regarding taking on a challenge to fundraise for NPUK. The team cycled from the Department of Pharmacology at the University of Oxford, through Oxfordshire, raking up a total of 60 miles! They raised an amazing £2,716.92 which as ever, will be divided between ourselves and the Niemann-Pick Research Foundation. We are incredibly grateful for their ongoing support.

Often those in the wider community who know/ learn about Niemann-Pick UK become very proactive in their fundraising. This often means lending, adapting, and using their skills/abilities to raise money. For example, this year, **Chris and Shell, the Gemini Stitchers,** made some beautiful small bags which they have been selling and £5 from every bag sold was donated to NPUK.

RAISING

We are grateful to **Deb Callaghan** who made face masks in support of NPUK, raising funds and also providing some for use by our community. It is always very encouraging to see people be so inventive in their fundraising for Niemann-Pick UK!

This year so many wonderful individuals in our community have set up Facebook Birthday Fundraisers for NPUK. Since January Facebook Fundraisers have raised over £4,760 for Niemann Pick UK! In particular, during this time when fundraising is more difficult and larger scale events have been put on hold, the value of these online fundraisers and the support of our wider community means so much. We want to say thank you to everyone who set up and/or donated to a fundraiser...there have been 30+ of you lovely folks, and it means so much!

We have never been more thankful for all of those amazing people who provide donations to NPUK every month via. **direct debit** - this source of income is absolutely essential, particularly at a time when grants and other forms of funds will likely be harder to come by. For further information on setting one up, please email info@npuk.org.

Our fundraising superhero, **Dan Carter,** ran 70 miles in 15 hours and 30 minutes, 35 miles on 5th June and another 35 miles on the 6th June! Dan raised an amazing £1,210 for NPUK! **Shona Beveridge** also set herself an impressive lockdown challenge and walked 26 miles in her garden in 6 weeks beginning on the 26th of April as part of the 26-challenge fundraising campaign. Shona raised an astounding £1,500, tripling her original target of £500 and continues to be an inspiration to the community. Well done Shona!

Sarah McGauley, Ollie McGauley and Rachel Cooke pledged to walk a mile a day during September to raise money for NPUK and keep fit. Sarah wrote about the group's inspiration to take on such a momentous challenge: "Taylor Burrough aged 8 is a brave young lady who has been diagnosed with the rare genetic conditions known as NPC although this condition can be life limiting there is so much that is not known about the condition. Taylor will have lots of challenges in her life so the support of NPC team will be invaluable to her and her family." The group raised an incredible £725; we thank them for their enthusiasm and commitment!

KIDS' CORNER:

fun and games galore!

We are back with more games and acitivities for the younger members of the community (big kids permitted too!)

We hope you enjoy these fun games! If there are any activities andchallenges that you enjoy which were not included, please get in touch with us at: info@npuk.org and we will be sure to include them in the next edition of NPUK News!

SPOT THE DIFFERENCE (FIVE TO FIND!)

WorkCast

We are pleased to announce that all of the content which was provided for the Children & Young Person's Programme during September's Conference is now available on-demand on Workcast:

workcast.com/?cpak=8781578378966284&pak=6908238042624992

2020 WORDSEARCH (FOURTEEN TO FIND!)

W Q Z N O H Q X R Y N M P I Z A K U Q S
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S Y T G P L Q C W Z Y B R Z H Z T M U V
J I G D B U B J N U R S E S O O S B I W

ANSWERS: quarantine, toilet paper, wash your hands, virus, cleaning, gloves, social distancing, mask, homeschool, doctors, hand sanitiser, nurses, disinfecting wipes, stay at home

CREATE YOUR OWN NPUK MASCOT

NPUK Families Officer Karen Thomas wants you to use this space to draw your own NPUK character...best fit will become our official charity mascot! Please send complete artwork to karen@npuk.org. Good luck!

DIGITAL THINGS EVERYONE SHOULD TRY!

With 2020 proving to be an incredibly challenging year for everyone, we must now adapt to digital devices and services to ensure that we keep ourselves supported, informed and entertained!

In this article we have put together a fun selection of the best digital activities you, and your family, can try to keep your spirits up over these next few months:

1) Turn your home into a laboratory!

The Glasgow Science Centre has been bringing a bit of science into everyone's homes to help keep families inspired during these challenging times.

Check out their GSC at Home videos archived on the Glasgow Science Centre website which include some fun activities like making your own wind turbine as well as some fascinating science experiments and lessons.

glasgowsciencecentre.org/learn/gsc-home

2) Settle down with a story

If you have any avid readers in the family, why not check out the Roald Dahl website? The website has several pre-recorded readings from Roald Dahl books by a few famous faces.

If your little one is not such a big reader, then don't worry - there are also various other fun activity ideas to do indoors, such as quizzes, puzzles, and crafts. Something for everyone to enjoy!

roalddahl.com/things-to-do-indoors

3) Take a trip to the zoo

With physical trips to the zoo impossible, Edinburgh Zoo have set up live camera links to animal enclosures, such as Yang Guang, their giant panda or visit Penguin Rock, home to gentoo, rockhopper and king penguins! The website also provides a downloadable animal study and story worksheet and information on animal behaviours, so you will know just what to look for when watching the live stream videos.

edinburghzoo.org.uk/webcams

@NiemannPickUK

@niemannpickuk

@NiemannPickUK

6) Get moving

If you have not already, why not try some of Joe Wicks' workouts? Wicks' workouts took the UK by storm during the lockdown beginning in March.

The workout videos, especially the "P.E. with Joe" series, are perfect to get kids off the sofa and moving! Joe also has many videos for older exercise enthusiasts too.

5) Immerse yourself in incredible natural history

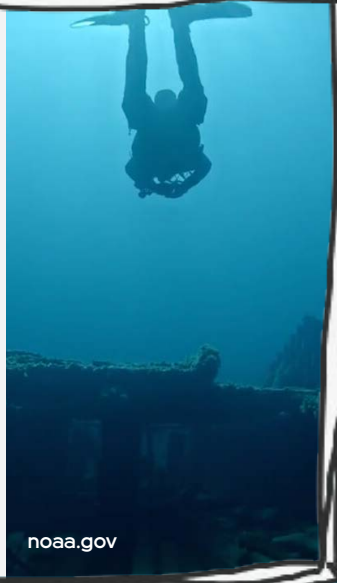
The National History Museum has not experienced so much disruption to their reliable stream of visitors since the Second World War, but they have adapted and are committed to continuing to educate avid enthusiasts about the world around us.

From exploring the deep oceans with their virtual venture into the life of a blue whale to virtually wandering the museum itself via their virtual tour from Google Arts & Culture... there is bound to be something for everyone on their website!

6) Dive into the ocean

If your little ones love the ocean and sea life, then why not take a virtual tour of the marine sanctuaries and ocean habitats?

The National Oceanic and Atmospheric Administration (NOAA) has a collection of virtual dives providing an immersive 360 view of national marine sanctuaries. From a shipwreck to a sea lion encounter, these virtual dive galleries are sure to keep the family entertained.



A more extensive A-Z list of ideas for fun but accessible family days out is available on a recently posted blog article on the NPUK website at: www.npuk.org - of course these will need to be followed in line with the latest goverment advice

We would love to see all of you getting involved and having fun! Make sure if you're posting photos you add the hashtag #NPUKDigital #GoMake Memories

DA

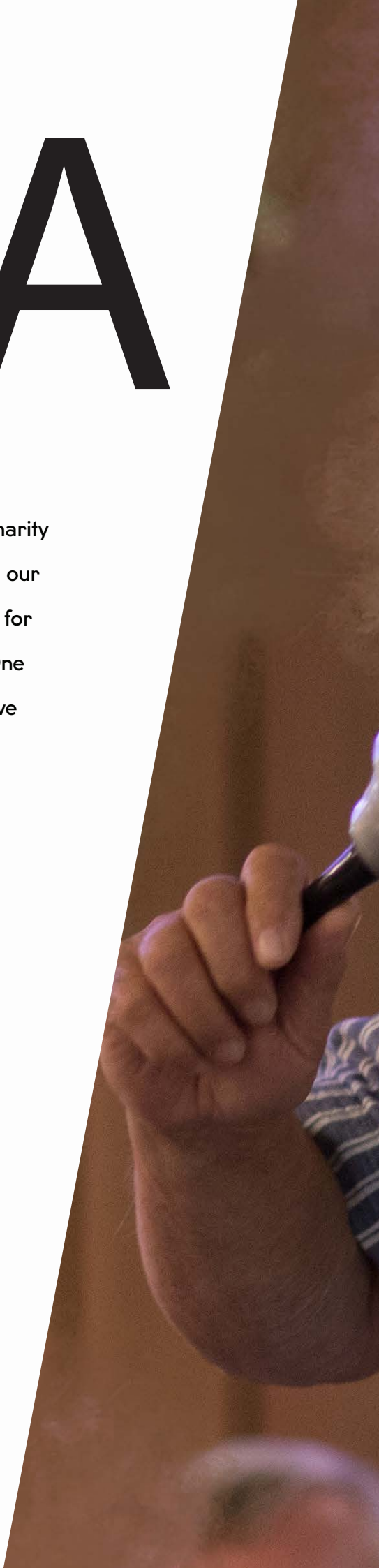
It is our Board of Trustees who push our small charity forward and keep us on track - ensuring we reach our objectives and aiming to secure a positive future for our organisation, and of course, our Community. One such Trustee is the legendary Dave Roberts, who we interview in the next few pages:

What made you want to become a trustee?

I wanted to become a Trustee after attending a board meeting as an observer and talking to my brother-in-law, Bill Owen.

Bill was already a trustee and he would often talk about the incredible work the charity was doing to support families and the need to create awareness of the disease and encourage basic research which could lead to potential treatments.

That first meeting was an eye opener. I had no idea how such a small group of trustees could possibly complete all the outstanding actions before the next board meeting, and I came away thinking I would help in any way possible.



VE

Dave gets everyone's attention at our Annual Family Conference 2019 - must have been time for the next session!

Did you know much about the charity sector before you became a trustee?

I didn't know anything about the charity sector really. Like most people, I would put money into collection boxes, make donations or support local fund raising events but I never thought beyond that immediate action of doing a little bit to help someone.

What do you think is the biggest challenge currently facing charities?

Without doubt, every charity is facing the impact of COVID-19. All charities have had to cope with falling income at a time of increasing demand for their services and many charities have had to furlough their employees and reduce the services they provide.

The biggest challenge is therefore how to raise income at a time of increased uncertainty and increasing unemployment. I think more charities will have to diversify their fundraising activities to include virtual streaming and digital fundraising events alongside traditional fundraising, online donations and grants.

The other great challenge will be how charities imagine and transition themselves to a new and longer term way of working that enables them to recover and survive the challenges ahead.

"That first meeting was an eye opener"

How will COVID-19 change the way NPUK operates moving forward?

COVID-19 and the restrictions placed upon us, forced most charities to change the way they worked. Thanks to a tremendous team effort by all our staff we became very successful at adapting the services we provide by remote working and "going digital" This enabled us to meet government guidelines and safeguard the health and safety of our staff and families.

A personal view about moving forward is that we will build on the amazing work our whole team did to support families and keep them connected during the lockdown. I think we will get back to some face to face contact when necessary but retain the best examples of remote working and video connections where and when we can.

We have shown this can be a practical alternative and there are many amazing examples of how Zoom made it possible for our care and support team to interact and support families and at a social level, enabled different groups to meet up and share their thoughts and experiences or just have a good catch up between themselves and, of course, there was Terry's famous quiz and Carl and Emma's sing along. There have also been

some excellent examples of digital fundraising events.

More recently, our digital family conference has shown how we can attend virtual meetings and connect with families and children by doing things differently.

These examples show what is possible by embracing a digital way of working but in a world of increasing uncertainty, we will work with our community to find creative ways to solve the challenges we face. We all miss socialising and meeting up with each other, but the day will come when we are able to have the best of both worlds.

What would you say to someone considering becoming an NPUK trustee?

I would say come along to one of our board meetings as an observer to see the work we do.

Trustees will have many different reasons for wanting to becoming a trustee but the one thing they have in common is a passion for supporting the Niemann-Pick community and a commitment to making a difference to their daily lives and hopes for the future. It can therefore be very rewarding to know that families, healthcare professionals and scientists both appreciate and benefit from the work we do.

It is important that the Board of Trustees have a broad range of skills when making decisions that affect how the charity is run and actions we take. In making these decisions, trustees bring with them knowledge from the job they do. Some examples are, project management, governance, finance, company law, marketing and healthcare. However and most important is the need to have trustees who are connected to families affected by Niemann-Pick disease. This brings a unique insight to the problems they face and helps to shape the service we provide.

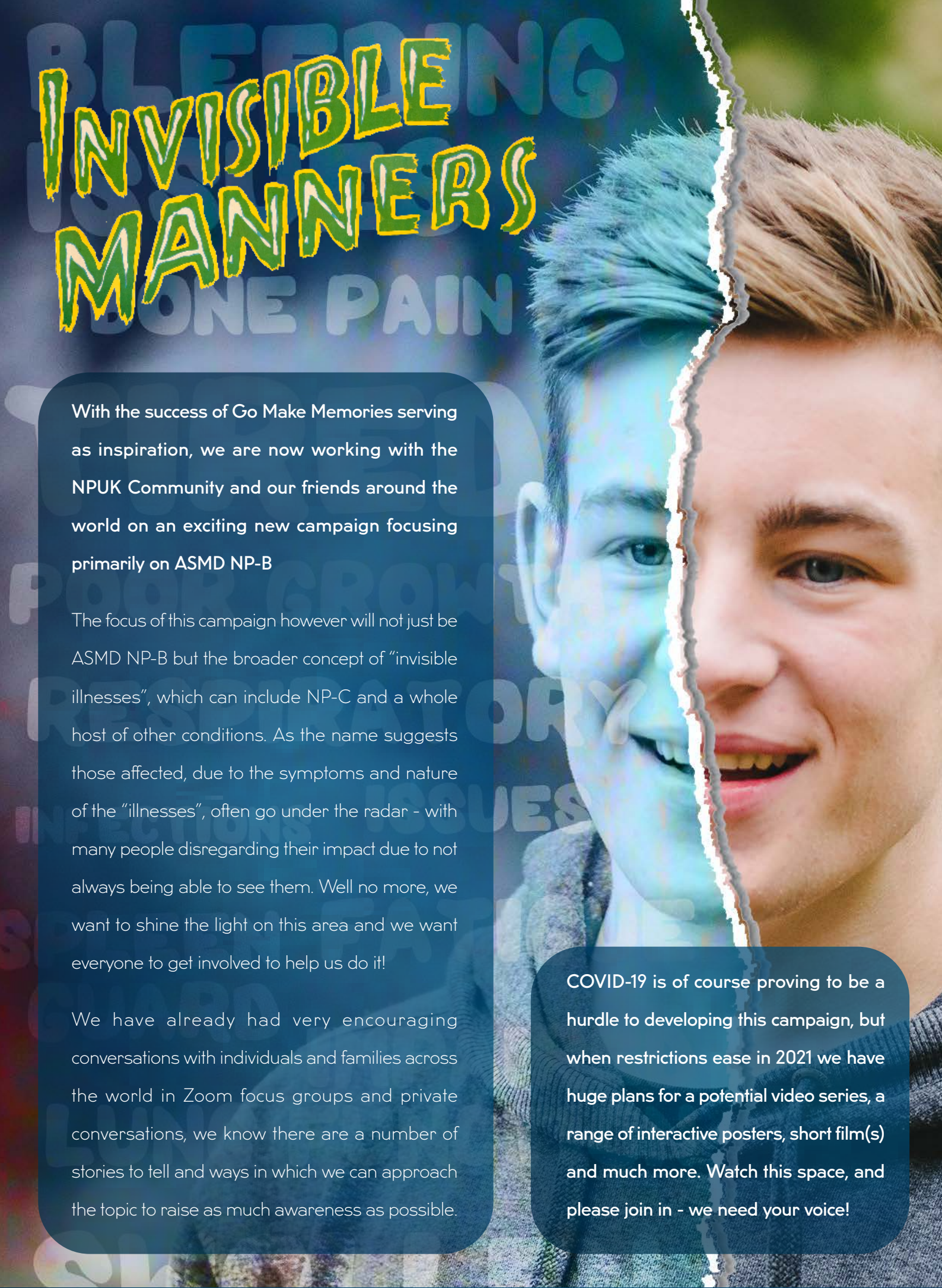
If you are thinking of becoming a trustee, you may not really know how you can contribute or what difference you will make. My advice is, do not worry. There is so much to do and think about that it won't be long before you are joining in conversations and making decisions that will help shape the charity, fulfil our aims and objectives and make a positive difference to lives of all those affected by Niemann-Pick disease.

One final note is that anyone from any background can become a trustee and the benefits are likely to be a mixture of feeling good about putting your skills to good use, personal development, meeting new people and joining professional networks, gaining new knowledge and skills and making a contribution to the success of the charity.

Got questions for Dave?

You can get in touch with him by email at: davidr@npuk.org

Invisible Manners



With the success of Go Make Memories serving as inspiration, we are now working with the NPUK Community and our friends around the world on an exciting new campaign focusing primarily on ASMD NP-B

The focus of this campaign however will not just be ASMD NP-B but the broader concept of "invisible illnesses", which can include NP-C and a whole host of other conditions. As the name suggests those affected, due to the symptoms and nature of the "illnesses", often go under the radar - with many people disregarding their impact due to not always being able to see them. Well no more, we want to shine the light on this area and we want everyone to get involved to help us do it!

We have already had very encouraging conversations with individuals and families across the world in Zoom focus groups and private conversations, we know there are a number of stories to tell and ways in which we can approach the topic to raise as much awareness as possible.

COVID-19 is of course proving to be a hurdle to developing this campaign, but when restrictions ease in 2021 we have huge plans for a potential video series, a range of interactive posters, short film(s) and much more. Watch this space, and please join in - we need your voice!

np uk pop-up shop

helping to raise funds and a-wear-ness

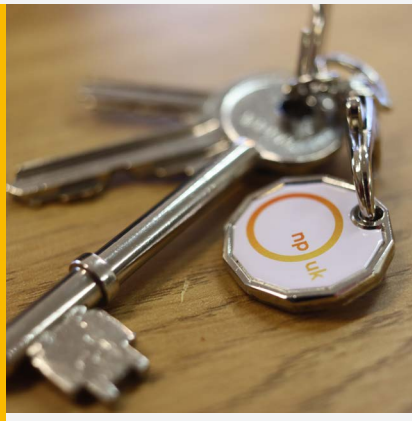
Following your positive response to the merchandise developed for our 1st Digital Conference and with a small amount of stock remaining, we decided to make an exclusive selection of NPUK merch available to our community, in return for a suggested donation

Items include t-shirts, hoodies, face masks and key rings, with all donations going to support our work with patients and families. To the right are sneak previews of our face masks, and other items which are already proving very popular in the NPUK office!

We trialled some of the products in the Goody Bags for our Digital Annual Family Conference & Interactive Workshop in September, with very positive results. We have now increased our range to include quality items that you can enjoy using and wearing in your daily lives, whilst raising much-needed funds and a-wear- ness for our small charity.

Watch this space for launch details and make sure you are following us on Facebook @NiemannPickUK to hear the latest!

If you have any queries, please get in touch with us by either email at: info@npuk.org or by phone on: 0191 415 06 93.



The NPUK trolley coin
John's house keys not included, I'm afraid

The NPUK hook keyring
Helpful for doors, entering pin codes, and more

THINK AGAIN
THINK NP-C

THINK AGAIN. THINK NP-C is a campaign which aims to support healthcare professionals unfamiliar with NP-C to recognise the key signs and symptoms of the condition and reduce the time to diagnosis. It has been translated to a number of different languages, and as a result has had an impact in many countries. For further information please visit: think-npc.com

NG RARE PROGRESSIVE IRREVERSIBLE CH
SIBLE CHRONICALLY DEBILITATING LYSOS
TING LYSOSOMAL STORAGE DISEASE[1–3] RAR

ARCHANGEL
MLD TRUST

Newborn Screening
Together for Change!

Owing to the rarity of serious genetic disorders like Niemann-Pick, many children are misdiagnosed or diagnosed at a point where the disease has already progressed too far for treatments and therapies to be of benefit. With a number of potentially effective and life changing therapies in development for Niemann-Pick and other ultra-rare conditions, now is the time to lobby for changes to the UK Newborn Screening policy.

Earlier diagnosis could provide the opportunity for children to receive life changing innovative therapies as they become available and to access expert clinical care and advocacy support.

Currently, UK babies are screened via the heel-prick test (also known as the blood spot or Guthrie test) at 5 or 6 days old. This sample is sent to a laboratory, where it is screened for just 9 rare disorders. Babies born in other high-income countries are tested for up to 59 rare diseases.

ArchAngel MLD Trust has joined forces with Nickie Aiken MP, with the support of other rare disease patient organisations including NPUK, to create a campaign calling for urgent expansion of the UK Newborn Screening Programme. To support and learn more about the campaign visit: archangel.org.uk/newborn-screening-campaign.

The current system for accepting new disorders onto the UK programme remains a complex and lengthy process. The criteria and standards for screening are set and monitored by the National Screening Committee and Public Health England, and many patient organisations and clinicians face huge difficulties trying to get conditions added to the programme.

Despite tireless work by a number of dedicated patient organisations and clinical experts representing other serious conditions, few changes have been made to the UK Newborn screening programme since its inception in 1965. UK NBS policy now requires urgent review.

ARA PARSEGHIAN MEDICAL RESEARCH FUND
UNIVERSITY OF NOTRE DAME

Niemann-Pick Type C Caregiver Preference Survey on Benefit Risk for Potential Treatments

Dear Friends in the NPC Community,

We invite families or caregivers of persons with Niemann-pick Type C disease to take a new survey that is designed to collect information about your priorities and preferences for treating Niemann-Pick Type C.

It is organised by the NPC Therapy Accelerator, a collaboration for the entire NPC community guided by the patient perspective. In the survey, you will read about hypothetical treatments for Niemann-Pick Type C. Some parts of the potential treatments described in this survey are based on real and investigational treatments and some are hypothetical. The information collected here is important to help the U.S. Food and Drug Administration (FDA) understand your thoughts and feelings about potential treatments for NPC.

This survey is intended for parents and current and past caregivers of those with Niemann-Pick Type C. Those who have multiple children with Niemann-Pick Type C should complete one survey for each child and only one survey should be submitted for each child (e.g., two parents should not complete the survey separately on behalf of the same child). For parents/caregivers of older children or adults with Niemann-Pick Type C who are involved in their treatment decisions, you may consult with your child when responding.

Your participation in this survey is optional and if you start the survey, you can stop at any time. The survey takes about 30 minutes

You can access the survey, at the following link: www.surveymonkey.com/r/NPC_B-R_Survey

"...there will be no cure without research and no research without funding..."

ARA PARSEGHIAN

NPUK is a member of the UK LSD Patient Collaborative Group. The collaborative is comprised of patient organisations representing those affected by lysosomal storage disorders (LSDs) who have joined forces to create a strong lobbying and action group for LSD patients and their families in the UK. This provides a stronger voice, particularly at this challenging time - for further information visit: lsdcollaborative.org.uk

LSD COLLABORATIVE

WE ARE THE 300 MILLION

SHOW YOUR SUPPORT THIS RARE DISEASE DAY

FEBRUARY 28TH 2021

RARE DISEASE DAY®

There are over 300 million people living with a rare disease worldwide

With grateful thanks to our grant providers:

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NPUK

WHO WE ARE...

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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 & Campaigns Manager, 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.

PASS THIS COPY ON TO A FRIEND!

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