NPUK NEWS

30th Anniversary Special

SPECIAL MEMORIES

Head down memory lane with us as we celebrate thirty years of NPUK

Invisible Manners

An exciting look at our latest upcoming short film on ASMD NPB and other invisible conditions, featuring Billy Boyd, Weruche Opia, Isy Suttie and more...

RETURNING TO IN-PERSON MEETINGS FOR 2021/2022

Learn more about the return of our much loved in-person meetings...we've certainly missed you!

Don't be shy, get social with NPUK:

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

REDUCE · REUSE · RECYCLE

NIEMANN-PICK UK & LINGXI ZHANG ANIMATION PRESENT

INVISIBLE MANNERS

A SHORT FILM ON INVISIBLE CONDITIONS

NOT ALL THAT YOU CAN SEE, IS EVERYTHING THAT IS THERE.

FILM BY LINGXI ZHANG

WRITTEN & DIRECTED BY JOHN LEE TAGGART

STARRING: BILLY BOYD, WERUCHE OPIA, ISY SUTTIE

AADAM ASIF, GRAHAM KIRK, SANDY COWIE, NADIA SLATCH, DANIEL DE VICENTE, JUSTIN HOPKIN, JAMES KEEBLE, SALLY TUNGATE, CLÁUBIA VIEGAS BENDER, SARA STRÖER, CRAIG BURNS, ALEC KOUJAIAN, DAVID ROSE, JAMES BROOKS, PHILIP WILSDON, CAROL LEON & HOLLIE CARTER

EXECUTIVE PRODUCER: ELEANOR LILY TAGGART **MUSIC BY: LILLIPUT & BADGER**

POSTER ARTWORK: JAMES KEEBLE, LINGXI ZHANG & JOHN LEE TAGGART

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Registered as a company limited by guarantee in England and Wales

np uk

Inpda

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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 the amazing Aztec Colour Print for once again producing this vibrant magazine on behalf of our small charity.

Aztec has supported our printing needs for all other publications for the past few years, ranging from leaflets, to patient passports, medical alert 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 this special edition of NPUK News! As we begin to step out of the shadow of COVID-19 and the difficult months that accompanied it, I am confident we can look towards a brighter future together.

Earlier this year, we celebrated NPUK's 30th birthday, which served as the perfect opportunity to pause and consider how far our small charity has come. The NPUK of 1991 was certainly very different from the NPUK that we know today, the achievements and success of our organisation would never have been possible without the skill and commitment of the many people who have contributed over the years.

It all started with just ten affected families getting together at a satellite seminar focussing on Niemann-Pick diseases. It is difficult to imagine today, with our connected and close community, that this was the first time that each family had met another affected by this condition. It was decided in that moment that we are stronger together, whether this is raising awareness, supporting one another, or rallying for change... sentiments which have stood the test of time ever since, and are still reflected in our work today.

The strength and value of our community has been more than demonstrated in recent years, as we have faced the challenge of COVID-19 together. It has been hugely disappointing not to have been able to meet in person at our Annual Family Conference or regional get-togethers. Although the NPUK Staff Team were able to adapt new ways of virtual working and communication, it is great to see the slow and careful return to in-person meetings, clinic days, family fun days, and more. I particularly enjoyed catching up with old friends and making new acquaintances at our recent 'Reunion Lunch' and I will look forward to seeing everyone at our family Christmas Party, the details of which you can find on page 13 of this newsletter.

I hope you will find this special edition as interesting as I did! It was certainly helpful for me, in my role as Chairman, to reflect on the history of NPUK. I am always inspired by the dedication of those involved, by how much has been achieved and by the strong relationships and lasting friendships that have been formed as a result. We still have much to do as we write together the next chapter of NPUK's history.

Please do pass this newsletter on to a friend when you are finished reading. This edition especially, and the memories it contains, is meant to be shared and in so doing, you are broadening our readership and increasing potential support for our small but passionate charity!

I trust the remainder of your year is a happy and healthy one, I am very much looking forward to seeing you all, in person, once again.

Warmest wishes,

Will

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, the Pears Foundation, and The Hollie Foundation for their grant funding support

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

"...today you are still pioneering families..." - Susan Green, NPUK Founding Member

Get to know Lingxi Zhang, the artist & animator working on the upcoming *Invisible Manners* short film

Former NPUK Trustee and Fundraising Coordinator Tony Jellings reminisces on his many successes as part of NPUK's Executive Board

Pages 44-49 are dedicated to our many fantastic fundraisers...who during these difficult times mean more to us than ever.

It's difficult for us 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!

Big stars join Invisible Manners
Billy Boyd, Weruche Opia, and Isy Suttie join cast of latest film...

IN LOVING MEMORY

We remember all of those we have sadly lost in the NPUK Community in pages between 56 and 58

LAURA BELL
CLINICAL NURSE SPECIALIST

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

Laura is our Clinical Nurse Specialist on Niemann-Pick diseases. Laura is based in Manchester and works from Salford Royal Hospital, but is always kept very busy as she also offers home visits across the country and attends clinics and meetings with our community when necessary.

Hi Everyone,

I hope everyone is doing as well as can be, I know the last couple of years have definitely been a challenge for us all. At NPUK the past few months in particular have been very different, 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 lockdown 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 to hopefully 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!

Since my last NPUK News update I have continued to work closely with the metabolic centres and along with all of my colleagues from 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.

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 so we can overcome any challenges we face. I will continue to work with the medical professionals both within the hospitals and community settings to learn from these experiences.

I remember attending my very first conference 9 years ago to celebrate NPUK's 21st birthday and feeling so privileged to be a part of it. I have a lot of fantastic memories over the past years of working with some incredible families, Staff members (old and new) and of course all the Trustees. Long may it continue!

So as we move forward, I would just like to take this final opportunity to remind you that as Clinical Nurse Specialist for NPUK I continue to offer practical advice and support on all aspects of Niemann-Pick conditions from diagnosis and beyond. I can also help you navigate through your care and support teams and treatment as well as offering support in times of difficulty.

If you need any help, advice or just a chat I am always at the end of the phone and you are always most welcome to contact me at any time. As I hope you know already I am happy to hear from you, accompany you to any 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

NPUK CARE & SUPPORT "AFTER" COVID-19

As we slowly start to return to life as we once knew it, but with the understanding that we will likely be living with COVID-19 for the rest of our lives, additional precautions must be continued:

Laura and the rest of the Care & Support Team are thrilled to be able to resume in-person meetings with patients, family members, and friends within the NPUK Community, however as COVID-19 case rates still remain relatively high, we must consider our own risk and play our part in keeping ourselves and each other safe. Which is why to both limit the spread of the virus and to better ensure the safety of those we visit, we will be:

- We are fully vaccinated. Our Care and Support Team, and indeed all members of our Staff Team are fully vaccinated, which we hope adds some safety and reassurance for our community.

- In line with government guidance we will also be limiting the number people we mix with, as well as the frequency.

- Where possible, we will meet people outdoors or in well-ventilated places and will continue to wear face coverings in crowded, indoor settings when appropriate.

ANTHONY'S AMAZING ART

We were so impressed with Anthony (adult patient with NPC) and his fantastically intricate colouring efforts that we simply had to feature them in this edition of NPUK News. Well done Anthony, keep up the good work and keep sending these in!

LOUISE METCALFE
FAMILIES OFFICER

Hello Everyone,

I am thrilled and feel very privileged to be part of the NPUK team as the organisation turns 30. I have been with NPUK for five years now, previously in the role of Project Team Leader at NPUK, managing the grant received from the Big Lottery. As a result, I was able to get to know many patients, families, volunteers, trustees and organisations connected to NPUK. Now I am in the role of Families Officer at NPUK and I am really enjoying reconnecting with old friends and meeting new patients and families as the organisation continues to grow.

I cannot quite believe where the years have gone or indeed how much the charity has grown since I joined in 2016. I feel very fortunate to be part of such a wonderful team at NPUK and be involved in an organisation that gets bigger each year. People often ask me what I like about my job and I say that most of the time it really doesn't feel like work at all. I love meeting patients and their families and helping them with whatever it is they might need or just being there for a chat, either on a home visit, in a clinic or down the phone. If there is anything you think I can help you with, be it benefits, work, issues to do with schools or housing, equipment or just a listening ear, please do get in touch.

What is Louise's role here at NPUK?

Louise 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!

Meeting families, volunteers, trustees and all the amazing professionals treating, working on and researching Niemann-Pick diseases at our Annual Family Conference is always a highlight of the year and I am really looking forward to being able to do that again, when safe to do so. Our Christmas parties are always a lot of fun too and it's so nice to catch up with everyone at such a special time of year.

The last year has been hard for all of us when we haven't been able to get out and about as we would normally do. I know how difficult some families have found lockdown and it has been particularly hard for those that have been shielding and those that have not been able to work because of caring responsibilities. Being able to help families access our Emergency Response Fund for help with financial hardship

or to purchase things to help with their emotional wellbeing during this unprecedented time has been particularly rewarding. We have supported families with everything from exercise bikes to craft equipment to garden furniture and grocery bills. NPUK trustees have agreed to continue this funding for a little while longer so if you think you or your family could benefit from this, please do get in touch with me.

Having our Time to Talk sessions on Zoom every Friday has been a great way to connect with families and to stay in touch in a very informal, drop in basis. These have been so successful we have decided to keep these sessions running for the foreseeable future. Being able to meet up with some of the lovely Queen Bees (any lady with a connection to NPUK be it a patient, carer, friend or volunteer is most welcome) on Zoom and share a drink or too has been a lot of fun too.

However, with lockdown slowly lifting, we are cautiously starting to make plans to meet up with families in their homes and at small regional events. We held the first small event recently at a children's activity farm in Cheshire and it was lovely to see some of our children and their families again in such a relaxed environment. There was so much to see and do for the children and parents and NPUK staff members were able to catch up with one another too. The venue was recommended to us by a parent so if you know of anywhere local to you that might be suitable for one of our meet-ups be it for adults or children, that you or your family enjoy visiting, please do let us know.

If you would like to get in touch with me please call me on 07423106595 or email me at louise@npuk.org and I'll be delighted to hear from you.

Louise

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

Louise's Lemon Drizzle

Those who were able to attend our late summer Tatton Park event will already know how great Louise's lemon drizzle cake is! Well, as requested here's the recipe so you can create your own:

Ingredients: 225g unsalted butter, softened, 225g caster sugar, 4 eggs, 225g self-raising flour, 1 lemon, zested. 1½ lemons, juiced, 85g caster sugar (drizzle)

Step 1: Heat oven to 180C/160C fan/gas 4.

Step 2: Beat together butter and caster sugar until pale and creamy, then add the eggs, one at a time, slowly mixing through.

Step 3: Sift in the self-raising flour, then add the lemon zest and mix until well combined.

Step 4: Line a loaf tin (8 x 21cm) with greaseproof paper, then spoon in the mixture and level the top with a spoon.

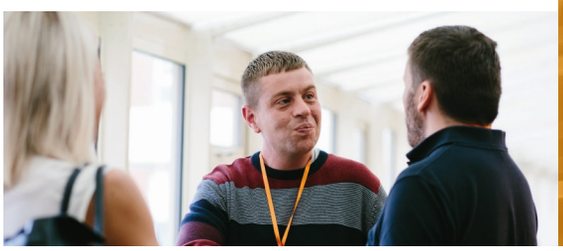
Step 5: Bake for 45-50 mins until a thin skewer inserted into the centre of the cake comes out clean.

Step 6: While the cake is cooling in its tin, mix together the lemon juice and caster sugar to make the drizzle.

Step 7: Prick the warm cake all over with a skewer or fork, then pour over the drizzle – the juice will sink in and the sugar will form a lovely, crisp topping.

Step 8: Leave in the tin until completely cool, then remove, serve...and enjoy! Will keep in an airtight container for 3-4 days, or freeze for up to 1 month.

STEVE NEAL
COUNSELLING & WELLBEING OFFICER



With the restrictions easing and people wanting to get out to live their "new normal" we have in turn been planning visits. We have certainly missed you and cannot wait to see your faces and to simply have quality time with our wonderful families. Our recent family meet up, where we were able to see some of you, reminded me just how much we have missed going on visits and also confirmed that it's what I love most about my job!

We have been deprived of so much the past 2 years, and have all felt the impact of the restrictions on some level, whether this be emotionally, physically, socially or mentally – no doubt this disruption has caused much uncertainty for so many. This has had a huge impact on our community's well-being, and consequently I have been focusing a lot of my work on promoting mental well-being and ensuring that we all remain connected. Just as we can develop problems with our physical health, mental health problems will be experienced by many of us over the course of our lives, so it really does need to be treated as a priority.

The support I have been offering, and continue to offer has taken place in many forms, one focus in particular is helping people within our Lads, Dads, and Carers group by having one to one phone calls or Zoom sessions to help work though any issues they might be experiencing - this has had a huge positive impact on a lot of our families. Another way we have been helping people is with the use emotional resilience tools, such as Cognitive Behavioural Therapy (CBT) which can help individuals better support themselves when going through difficult times.
We want our families to thrive, and these can really help change that cycle of negative thoughts and alter the way we react to destructive or unhealthy feelings.

Why are wellbeing and resilience important?
Resilience is important because it can help to protect against everything from the development of some of those everyday life problems we all experience, to more unique situations and problems that apply to each of us as individuals, along with other feelings like stress or anxiety. Resilience helps us to maintain our wellbeing in difficult circumstances and gives us the ability to cope with life's trials and to instead adapt to adversity. Working with families to develop different levels of resilience will change over the course of the journey we are all on.

To allow everyone to rise to this challenge we have developed a programme intentionally designed to help you embark on a more mindful journey that will help organise your week, with everything from; home life, working life and social life factored in. In short whilst working on your own well-being you can also help to bolster your mental health too.

If you would like to take part, please contact me!
I am always here for you.

Steve

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

Adjusting to life after lockdown

Moving from lockdown to the relaxation of COVID-19 rules: tips on taking care of your mental health

For many people, the easing of lockdown restrictions is a welcomed change, with the opportunity to finally see friends and family in person, but for some, even if we are happily anticipating these changes, the re-adjustments can take a toll on our mental health.

Fear and anxiety are possibly the most common emotional responses many of us will feel as lockdown eases. It took a lot of energy to get through the uncertainty of lockdown, and you may have worked out a system and routine that helps you, and perhaps your family, get through this turbulent time. So suddenly, this quiet routine no longer being necessary may appear alarming.

It is important to acknowledge that these feelings are reasonable, and even more so common. It is only gradually working towards a change in routine that we can move past such fears. Below we have outlined some tips on coping with fear and anxiety around lockdown easing:

Pace yourself

Recognising that you need to go at your own pace is important. It is easy to look on social media and think that everyone has got back to 'normal' in a matter of weeks, but this does not mean that you should be able to be pressured into doing things you do not want to. However, it is important to discuss your concerns and how you feel with those close to you, perhaps you could come up with a plan to meet others that suits you and allows you to move at your own pace.

Build up towards it

If you are struggling to shake off the lockdown way of life, but want to get back out there, try doing something that challenges you every day, or every few days. Don't worry if it does not go well at first, just consider how far you have come and what you hope to achieve.

Talk to your work

If you have started working from home during the lockdown, consider speaking to your work about the possibility of continuing to do so. If you are finding it hard to get to work or do particular shifts or activities because of anxiety or fear, speak to a manager or colleague about your concerns. If you have, or in the past have had, longer term mental health problems, you may be entitled to reasonable adjustments under the Equality Act, even if you have not disclosed it before.

Talk to people you trust

It is important to talk through how you are feeling with others. Don't dismiss your concerns, you may learn that others share similar concerns and fears. Remember that the NPUK team are always ready to listen to your concerns and talk through them with you, contact NPUK via phone: 0191 415 0693, or email: info@npuk.org

Although this article has offered some advice on getting back to 'normality', what is most important is to go at your own pace and evaluate what level of social interaction works for you. Get in contact with the NPUK team at any time, if you have any queries or concerns about what is included in this article.

RETURNING TO IN-PERSON MEETINGS

With our families, for our families!

We've certainly missed seeing our community as of late, which is why as soon as it was safe to do so we excitedly (but cautiously!) began to plan smaller meet-ups for families:

The first step back into in-person meetings took place on a beautiful Saturday at The Childrens' Adventure Farm Trust, Cheshire, which saw members of our Staff Team meeting together with a few NPUK families to enjoy a day together. Well thankfully the sun was smiling down on us, so we could enjoy a stroll around the farm, whilst re-uniting with old friends and catching up after such a long time...we really couldn't ask for better company and were very pleased with the day.

After the success of our first meet-up, we were eager to plan another, and so organised a Family Fun Day at Tatton Park, Cheshire. The beautiful grounds provided the ideal surroundings to reconnect with members of the NPUK community, support each other and, most of all, have fun. It seems we have two talented photographers in our midst too, as we got fantastic snaps of the day, courtesy of two of our NPC community members, Ben Gray & Shona Beveridge.

NPUK Reunion Lunch: October 16th 2021

On the 16th of October 2021 the NPUK community (patients, friends, family and NPUK staff/ trustees) finally had an opportunity to re-unite with one another after almost two years apart! Following our two small but successful events at the Children's Adventure Farm Trust and Tatton Park, we decided to go ahead with our plans for the "NPUK Reunion Lunch" which took place at the Best Western Pinewood Hotel, Handforth.

As you may know, we usually hold our Annual Family Conference and Interactive Workshop at this time, which spans a whole weekend. Although this year the full schedule of Conference was not feasible, the Reunion Lunch allowed us to catch up with other members of the NPUK Community and support one another - which is really the most integral part of all NPUK events! So although Zoom calls and social media chats are fun and have kept us going, it's safe to say there is nothing quite like meeting up with the NPUK community in real life and feeling that personal connection. Alongside the lunch, we had a slideshow of NPUK photographs, showing some of the key moments and memories from across the years, as well as activities related to the Invisible Manners short film...more on that later!

A huge thank you to everyone who attended and made it so special, we loved catching up with you all...roll on the Annual Family Conference 2022!

"...we hope that it will help to bridge the gap between the virtual world andthe face to face interactions we all prefer..."

- Toni Mathieson,
NPUK Chief Executive

NPUK Christmas Party 2021

It is with great excitement that we can invite you to our much-loved NPUK Christmas Party, a fun and festive celebration for our whole community! The event, as ever, is open to all members of our community including patients, families, friends and NPUK Staff / Trustees.

If you have not yet attended one of our NPUK Family Days or the NPUK Reunion Lunch, this is an especially perfect opportunity to reunite with the NPUK Family with some festive spirit! We've got a number of entertaining activities to get involved with, including Mirror Image Disco, a Magic Selfie Mirror and a visit from the big man himself, yes that's right...Santa!

The Christmas Party will take place on Saturday the 27th of November at the Holiday Inn, Kenilworth. For more information and to book your place email: info@npuk.org

Bill Owen, NPUK Trustee Research Report

I was reminded that 2021 is 30 years since we began our charity and Patient Advocacy activities. A great deal has transpired since then and much progress has been made against what is regarded as an intractable disease although much remains to be done. In the early days a group of affected families had got together under the leadership of Jim and Susan Green with the intent of identifying what could be done following the diagnoses of our children and how we could make a difference. At that time the NPC genes had not yet been identified, NP type A seemed a lost cause and Type B was just awaiting production of an enzyme replacement treatment and its availability to patients. We seem to be still waiting for this for complex and frustrating reasons.

Our committee members were recruited from the 'four corners' of the UK and we relied on the availability of someone's front room for our meeting and sometimes for overnight accommodation. The notes of meetings were in manuscript, no laptops, no internet, no mobile phones, no funds! Progressing every activity was a slow business and none more so than trying to identify research into the NP diseases. There were no active studies in the UK and not much better in the USA and the rest of the world.

The Niemann-Pick diseases, named after two physicians who described them, had been recognised since the beginning of the 20th Century and attempts had been made to classify the diseases based on clinical description and microscopy. Revisions to the classification have been made over the following decades and modern science and technology provide a much clearer picture of the molecular differences between the diseases and with other LSDs.

Somehow Susan had identified NPC research taking place at the National Institutes of Health (NIH) at Washington, USA where Dr Peter Pentchev had recently linked NPC in humans to the same disease in mice and had developed an assay – filipin staining, for diagnosis. In France Marie Vanier had also worked on the assay that detects unesterified cholesterol in cells with NPC. The cholesterol was found to be stored in lysosomes, the recycling centre of the cell and the NPC defect disrupted the metabolism of the cell leading to other pathologies and ultimately, cell death. Purkinje cells of the cerebellum were found to be particularly susceptible to the NPC defect and are mostly damaged beyond recovery by the time diagnosis is made.

The gene associated with NPC protein would not be identified until late 1997 with the protein sequence and topology showing the NPC1 protein to be integral with the lysosomal membrane. Although much has been learned about the role of NPC1 since then, its function and mechanism of action remain unclear. To provide a perspective, many lysosomal enzymes whose functions relate to degradation of complex molecules in the recycling process had been identified and linked to a range of 60 or so lysosomal storage diseases. Only much later did studies take place into the proteins of the lysosomal membrane, one of which is NPC1. It is reported that there are over 200 of these, the functions of which are largely unknown.

A great deal of progress has been made in studies of the NPC genes and the subsequent downstream in the

pathology that has enabled scientists to identify targets for intervention that may halt or slow disease progression. This research has resulted in the development of treatments currently undergoing clinical trials and others still in the development phase. In addition, new ideas arise from unexpected sources as it seems that NPC and its metabolic pathway is an area of academic study attractive to many scientists across the world.

In the world of rare genetic diseases funding is at a premium with studies requiring funds to support top professional scientists, PhD studentships, complex state of the art technology, animal models and the infrastructure in which they can carry out their work. This is expensive and because of the very low numbers of people affected, was not looked on favourably by those providing research grants. The situation may be improved now but in earlier days a great deal of funding was raised by patient groups to support small studies and promote international collaboration.

Of particular note is the Ara Parseghian Medical Research Foundation (now Michael, Marcia and Crista Foundation) based in Arizona. Their fundraising was exceptional and amounted to $50M from when they set up. This funding could be accessed by international scientists and has moved the pace of NPC research on by an immeasurable amount.

It is worth emphasising that NPC research is at the very fore front of cell biology with new mechanisms and classes of molecule emerging from the many studies being undertaken. Many other patient organisations have also contributed to the research knowledge discovery including NPUK and NPRF in the UK we have all done what we have been able and continue to do so.

MMC Foundation Conference 2021

Again, this year it was held as a virtual conference with presentation programme being run each Monday in June. Each session was scheduled over 3 hours and there were about 140-150 attendees.

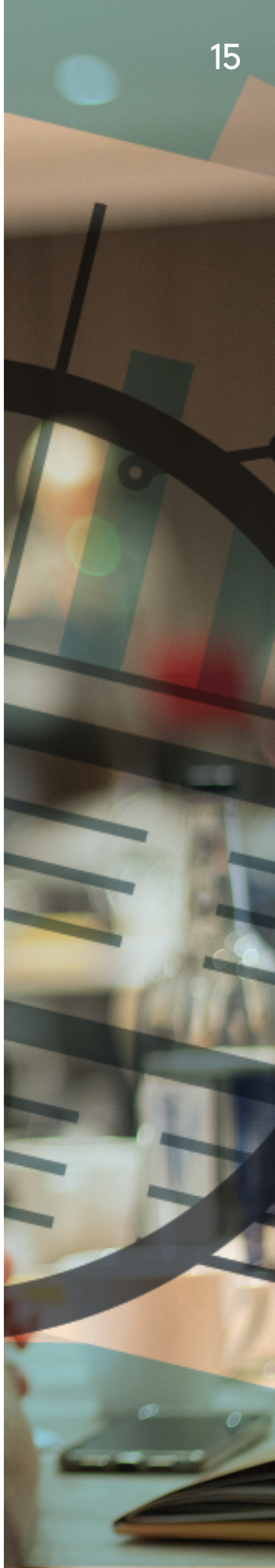
The sessions were as follows:

- Lipids and Organelles
- Cholesterol Transport and Signaling
- NPC Models and Approaches
- NPC Screening and Drug Development

The format is a mix of full presentations and short talks. The focus is mainly on the basic science of the cell and the impact of the NPC1 defect. Some of the presentations are very difficult to follow but some throw up new points previously unknown to the scientific world. The better this area is understood then the better the chance of developing accurate diagnosis and treatment.

Newborn Screening

It was encouraging to hear a presentation on NBS led by Dr Wasserstein of the Montefiore Children's Hospital, New York with support from the Firefly Fund. An update of their ScreenPlus Pilot scheme for 13 LSDs including all the Niemann-Pick diseases was given. A major advance in the NBS tests being employed is that two or three complementary tests combine to increase identification of true positives and to reduce the incidence of false positives. One of the tests is genetic and a library of pathogenic mutations has been built up over many years to provide a sound reference. This multiple assay approach will reduce the number of false positives, an undesirable feature in mass testing, in fact the approach is no longer just a screen but also a diagnosis. The major information missing being the clinical description which is not likely to be present at birth.



UK Archangel Initiative

Progress on the Archangel Initiative for UK NBS continues with a presentation of an analysis process undertaken though Archangel Trust using an algorithm based on the Wilson-Junger criteria (1968) to prioritise the various LSDs for NBS. I have queried the methodology and am awaiting further discussion on the outcome of the study. NP disease did not come out very well as one of the criteria used was diseases with later onset were not a priority.

It raises an important question as to 'when is a disease a disease?' We are very much aware that identifying disease presence – whether heart, cancer or other, is important if the disease is to be prevented from reaching the clinically symptomatic stage when the optimum chance for intervention is lost and in many instances, too late. Much to do I think.

Genetics England and NBS

Genetics England together with the National Screening Committee have undertaken a major public consultation on *The Implications of Whole Genome Sequencing for Newborn Screening – A Public Dialogue*. The report, 104 pages is available online and provides the results and public reaction to conducting WGS at the newborn stage. I have yet to read it but the public seem very much in favour given assurances on use and security of data. My initial reaction is that some of the Wilson-Junger criteria have been cut and pasted in the WGS approach thereby blocking a new look at what is really needed.

NBS In the EU - Eurordis

I have previously reported on activity by Eurordis moving forward with a Position Paper to influence EU/EC on improvements to the current NBS activities which vary between member states. A number of Key Principles are identified and include some major departures from existing criteria. Recognition is given to the central role of the family and, of major importance, recognition that diseases are 'actionable' even if effective treatment is not available. The actions that may be taken may also be described as benefits and it is the balance of benefits against harms which should be one of the major points in any assessment of an NBS proposal.There should be a lot more to come on this but communication with Eurordis is difficult.

General Observations on the Rare Diseases Landscape: clinical trials and early diagnosis.

Having reached the stage of having clinical trials in progress and other candidates being work in progress, we might feel some satisfaction that we are finally getting there – but caution is needed. Problems are being reported of failure to demonstrate efficacy, bankruptcy, company takeovers, high costs of medicines, method of administration, regulatory approval and more.

It seems strange to me that for many years the researchers work enthusiastically at resolving highly complex biological systems and yet rarely seem to question how a resulting intervention can be applied to an individual early enough to prevent disease onset and progression. The common response by researchers to this point is - 'earlier the better'. I suppose it's just one thing at a time? It's not widely reported but a likely reason for failure of some clinical trials is that the medicine is brought to the patient too late but how can patients be identified earlier? In general diagnosis, which may take years, mostly follows symptomatic presentation when damage has been sustained.

Disease identification is possible through NBS and pregnancy monitoring which are activities currently undertaken in UK clinics. As the baby grows there is also early stage monitoring of baby's development for the first 6 to 12 months but this is largely voluntary. NBS then, may be regarded as the focal point when a common

test – the heel prick test, is carried out and is the ideal opportunity to detect pre-symptomatic disease by use of genetic and metabolic markers.

It would appear from the recent Genomics England publication that the genetic element will be implemented over a number of years using WGS as a NBS test and find all genetic diseases over the lifetime of the UK population. This is remarkably ambitious, and it might have been less fraught with unknowns if the project had simply targeted the rare diseases complemented by metabolic biomarkers. Progress does not always follow the main unmet needs, but it is progress!

Rare diseases are out there – beware!

Those of us that experienced nature's rich genetic diversity first hand, eventually find out about topics such as disease incidence, carrier frequency, the one in four chance of two carriers having an affected child, the number of lysosomal storage diseases, the vast numbers of rare diseases with a genetic component, the number of potentially lethal genes we each harbour, are prompted to ask 'what is the risk probability of having a child with a rare condition?'

Population studies have been able to provide answers to most of these topics and figures are reviewed as studies reveal new, improved data. We know that NPC1 for example has a disease incidence of 1:100,000 and this relates to a carrier frequency of 1:170. This carrier frequency does not say anything about distribution and density of those carrying an NPC1 mutation and at the outset, none of us knows about our carrier status. We can meet a partner with the same affected gene anywhere it seems.

This is just one gene out of about 20,000 or 40,000 if we count the two genes we inherit. Studies have estimated that the number of potentially lethal genes we each harbour is over 200. Just a few years ago this was thought to be about 6. Even worse, the estimated number of proteins in humans is over 40 million, or is it more like 350,000 according to DeepMind the artificial intelligence computer?
What happens when any of these go wrong?
The accepted number of named rare diseases is between 6-7 thousand with new diseases being identified each month or so – see Orphranet Newsletters. We are probably all aware that some individuals suffer with a range of depression like illnesses but cannot be diagnosed. How many of these are there?

The figures I have given above may not be accurate, but I believe they provide an illustration of the potential dangers associated with human reproduction. Animals are no better. Nearly all of us are unaware of the dangers when we start out to create a family. Not only are the probabilities less than clear our education as teens fails to make us aware that there is even an issue to consider. This is a serious omission from our education. We are left to find out the hard way, it's not good enough when estimates show that 1:17 people in the UK will be affected by a rare disease at some point in their life.

A further concern relates to the oocytes or eggs carried by females for up to 40 years. With genetic mutations present in a recessive mode about 50% of the eggs, which are haploid – one set of chromosomes, will have a defective protein eg NPC1 but all similar recessive disease have the same issue. How is it that these eggs survive and remain viable of years? Or do they?

What can be done?

Prevention of many rare genetic conditions is within our grasp but there does not appear to be concerned action by Government, Heath Authorities, academic scientists, or patient advocates. The NBS/WGS initiative is a major breakthrough, but there is another, less painful and lower cost route. This is *pre-conception testing*, an approach that has been around for many years. In fact, the Human Genetics Commission favourably reported on a public consultation of using genetic testing as a disease prevention measure and chips for panels of rare diseases were available on university campuses for students to check their status.

If the intention is to introduce WGS into NBS, which it plainly is, then why not cut out the middlemen and go for route 1 targeting adults planning families? I have no idea how many couples this would involve but we could start tomorrow! As a postscript I recommend that affected families make all efforts to determine the carrier status of the wider families. Don't be put off by professionals telling you that it's very rare and low risk. What they are referring to is the risk probability. They usually forget to mention the risk impact which is as bad as it can be.

npuk: the story so far...

1991: Ten families affected by Niemann-Pick diseases met for the first time at a satellite seminar titled "Niemann-Pick Disease" and agreed to form a patient support group

1992: The first Niemann-Pick Support Group newsletter to families was sent out and led to the creation of a family directory in order to better meet their needs in the future

1994: NPUK produced and circulated the very first information leaflet to keep families up to date with the latest news

1996: NPD divided in two entities, based their metabolic defect: acid sphingomyelinase deficiencies, (types A, B & intermediate forms), and lipid trafficking defect, corresponding to NPC

2004: NPUK launched the (then new) website at www.niemann-pick.org.uk

Current site: www.npuk.org

2005: Toni Mathieson joined NPUK in a professional capacity in the role of National Development Manager, which developed into Chief Executive as NPUK grew

2006: The first 'Peter Carlton Jones Memorial Award' was presented to Emyr Lloyd-Evans from University of Oxford for his research on lysosomal calcium deficiency and NPC1

2007: Preliminary work began developing the International Niemann-Pick Disease Alliance, with the focus being to unite patient groups across the world

2013: Christine Jopling joined the NPUK Staff Team in the role of Finance and Administration Officer

2016: NPUK unveiled a huge re-brand, including a new website, updated logo, and an extensive social media strategy. John Lee Taggart joined the team to assist with communications

2016: Steve Neal joined the NPUK Staff Team in the position of Project Families Officer, having been a member of the community for many years prior

2016: Thanks to a five-year grant from the 'Big Lottery' NPUK welcomed Louise Metcalfe in the role of Project Team Leader Louise now holds the title of NPUK Families Officer

1997: The NPC 1 gene is successfully cloned

1999: Jackie Imrie joins in the newly developed role of Clinical Nurse Specialist on Niemann-Pick diseases to help support patients and families

2000: The NPC 2 gene is successfully cloned

2003: Toni Mathieson becomes involved with NPUK after her beautiful daughter Lucy, then just five weeks old, was diagnosed with Niemann-Pick Disease Type C (NPC)

2007: The Hollie Foundation (originally named the 'Hope for Hollie' Campaign) was founded

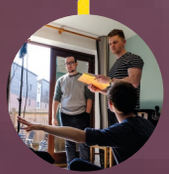
2009: Miglustat (Zavesca) approved for the treatment of progressive neurological manifestations of conditions, including NPC (although not a curative treatment)

2011: Jim Green stepped down after 20 years as Chair...guiding the group from humble beginnings to a well-known supportive small charity

2012: Laura Bell became part of the NPUK team in the role Clinical Nurse Specialist, having worked in the field of metabolic diseases since 2008

2019: Go Make Memories, directed by Carl Mason, is released to wide-spread acclaim

2020: In response to the COVID-19 pandemic, NPUK launched the "Coronavirus Emergency Response Fund" in collaboration with The Hollie Foundation



2021: Jenny Charman, founder of Harvey and the Brave Little Soldiers joined the NPUK team in the role of Fundraising Officer

2021: Proposed release date of short film, "Invisible Manners", featuring Billy Boyd, Weruche Opia, Isy Suttie and many more...

A massive milestone as NPUK turns 30!

Over the past thirty years Niemann-Pick UK has continued to go from strength to strength in our drive to provide care and support for all those affected by Niemann-Pick diseases. Below we have outlined just a few of the key moments we have shared...we have certainly come a long way since those early days!

In 1991, ten families affected by Niemann-Pick diseases (types A, B & C) attended a satellite seminar titled "Niemann Pick Disease" at the Research Trust for Metabolic Disease in Children (RTMDC) Conference, held at the University of Keele. This crucial occasion was the first time that each of the families had ever met another family affected by Niemann-Pick diseases. At the time, most of these families had been told the disease was so rare that they were likely to be the only family in the country affected. Susan Green (parent of two children with NPC) chaired the seminar, which led to the formation of a parent support group and evolved into NPUK as we know it today.

In 1994 we produced and circulated our very first information leaflet. This was an important step in our mission to provide practical and emotional support to our community. It is hard to imagine it now, with our community comms output, but at the time this was one of the few physical pieces of information on Niemann- Pick diseases. In 1997 thanks to a grant from Esso's Community Action Award

Scheme we were able to update the information leaflets to better inform our readers. Keeping our community in the know has been so key to fostering the supportive and connected community, which we continue to do today having created NPUK E-News, a monthly newsletter which provides a roundup of everything taking place within the NPUK community throughout the previous month, which serves alongside our bi-annual magazine NPUK NEWS.

In 2007 preliminary work began developing an International Niemann-Pick Disease Alliance, with the focus being to provide a way for patient groups across the world to co-operate together. In 2009 together with the NNPDF we jointly hosted an international meeting of NPD patient support groups, which resulted in the formation of The International Niemann-Pick Disease Alliance (INPDA) – a global collaboration of non-profit patient support organisations with a shared goal; to help those affected by Niemann-Pick diseases. The focus of the organisation was, and still is, to give Niemann-Pick

SO MANY PEOPLE HAVE

diseases a stronger global voice, and to ensure progress towards effective treatments for those conditions is accelerated. As you will know the INPDA has grown into an international network of non-profit groups and supporting families continues to be the INPDA's central focus.

In 2003 the first central office was set up in the home of our 'National Development Manager' (Washington, United Kingdom). In 2011 we moved to our NPUK office (HQ) also in Washington, Sunderland. The NPUK office is always a hub of activity and, during non-COVID times, a centre for our widespread community. We have welcomed many visitors such as former INPDA President/ NPUK Chair Jim Green, members of our Existing Board, the Care and Support team and many others, who we are able to update on the latest goings on in the community. As such NPUK HQ has become such an integral component to our small charity, acting as the centre point from which our widespread charity work can take place. More recently thanks to our lovely landlord we have had renovations carried out to our office space, making it a lot more modern and more importantly...on NPUK brand! The upgrade was complete with orange walls, white NPUK logos, and beautiful photography of our community throughout.

INCREDIBLE SUPPORTED US.

In 2012 Laura Bell joined the Team in her role as Clinical Nurse Specialist after previously working in metabolic diseases since 2008. Laura took over from Jackie Imrie, who still supports our charity in her capacity as an INPDR Consultant and NPUK Trustee.

In 2016 we unveiled a huge rebrand for our charity at the Annual Family Conference and Interactive Workshop, where we celebrated our 25th year together at Wyboston Lakes, Bedfordshire. This was an important moment to remember those no longer with us, who will forever remain in our hearts. We considered the positive steps NPUK has made during the past 25 years and our hopes for the future of the charity. NPUK's rebrand included a new website, an updated logo, a shortened paraphrased name in NPUK, and an extensive social media strategy focused upon raising awareness and supporting our community. We want to say a huge thank you to Emma Hesling and Bonafide Guests Only for their help in this area - it has certainly given a massive boost to our brand and presence in recent years!

2020 and the beginning of the COVID-19 pandemic was a huge challenge for us all and for those in the Niemann-Pick community in particular, feelings of isolation were only exacerbated by the pandemic. As such our support service changed, as we adapted to a digital way of doing things whilst continuing to assist those who require help. NPUK developed the #SocialNotDistant Campaign, which was a weekly schedule of activities, posts, and videos that our community could get involved with in order to keep everyone active, engaged and positive during a very difficult time. The success illustrated how powerful and supportive our community is!

We have only been able to reach such major milestones due to the ongoing support of you, our amazing community - here's to 30 more!

Susan's story.

As part of the NPUK 30th celebrations we wanted to share Susan Green's story, as alongside her husband Jim, she was integral to the founding of NPUK in 1991. In the following article she outlines what motivated this, and her hopes for the future of the charity.

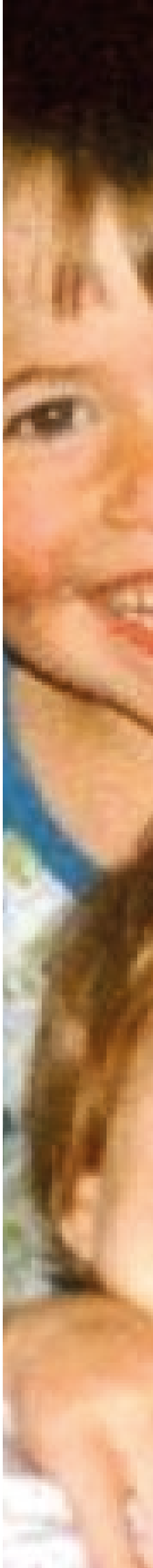
Jim and I were blessed with three happy healthy children, our sons Roy and Murray and our adopted daughter, Ailsa. Murray, our youngest son, had jaundice when he was born which took a while to clear up. Later, at the age of three, it was noticed that he had an enlarged spleen, various tests were done but all were negative. In spite of yearly monitoring no-one seemed to know why his spleen was enlarged. He was growing well and seemed healthy but, although he was bright and cheerful with a good general knowledge and a great love of books, he was not doing too well with his school work.

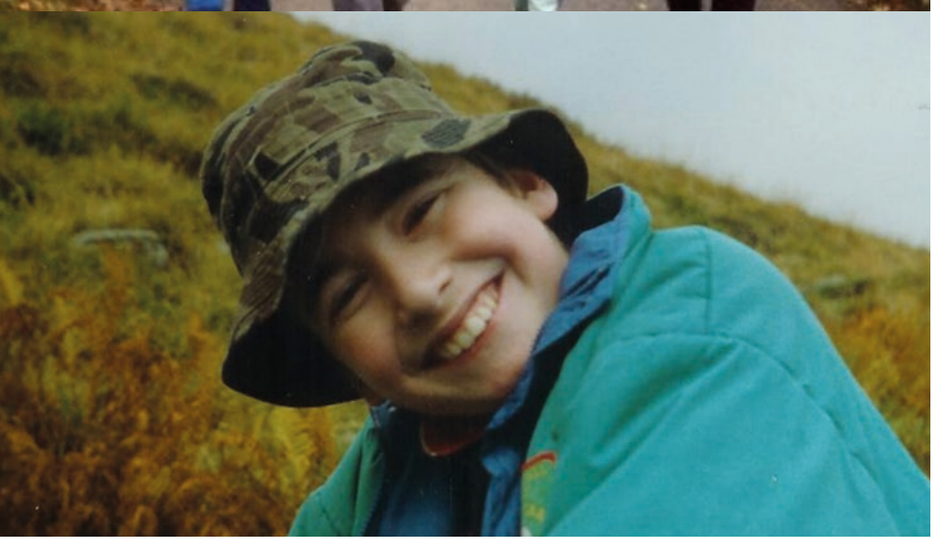
To the right you can see three healthy children...or so we thought at the time. Little did we know that we were shortly to begin our journey with Niemann-Pick Disease...

It was in August 1990 that Murray, then aged eleven, started having epileptic seizures and was rushed to hospital with a suspected brain tumour. We were very relieved when tests showed that it was not a brain tumour. It took another six weeks of tests before our neurologist said that he thought Murray might have a very rare metabolic disease. We had never heard of metabolic diseases. Another three months passed by before the results of his skin biopsy confirmed that Murray had Niemann-Pick disease type C.

We were told that NPC was so rare that we were probably the only family in the country with a child with this disease. We were told that it was degenerative and life limiting, and that there was no treatment and no cure. We were told that Murray would probably not live beyond his teens.

To say we were devastated would be a huge understatement. But there was more to come. We were told that Jim and I were carriers of this disease and that there was a one in four chance that Roy might also be affected. Six months later it was confirmed that Roy also had NPC. He was 14 years old and was showing no obvious signs of the disease. It was so difficult to understand that our sons were





going to die. They looked just the same as they always had.

Most people in the NPUK Community reading this will understand only too well how we felt. Since the moment of diagnosis we have been grieving for the loss of the healthy sons we should have had. Our dreams for their future, our future, have been shattered. We still live with the pain of this grief, and it doesn't go away.

We tried to find out as much as we could about Niemann-Pick disease but, in 1990 (before the World Wide Web and public access to the internet!) it was very difficult to get any information. What information we did manage to find was very difficult to accept. We didn't like what we heard. We had to learn a whole new language to even understand the symptoms! Hepatosplenomegaly, Vertical opthalmoplegia, ataxia, distonia, seizures, peripheral neuropathy, cataplexy, muscular spasms, spasticity and dementia.

We felt as if we had been parachuted into a foreign country where there were no signposts, no maps or guidebooks and we didn't understand a word of the language! We desperately needed to talk to someone who was familiar with this strange new country and could point us in the right direction.

Eventually we made contact with some more families and started a support group - the beginning of the NPDG (UK) [now Niemann-Pick UK]. In listening to families it was obvious that they were all seeing different doctors and no one seemed to know much about Niemann-Pick disease! Nearly all of the families we made contact with had also been told the same thing – that this disease was so rare that they were probably the only family in the country with a child with Niemann-Pick Disease.

In listening to these families we realised that each one of the children were being seen by a different clinician, what we needed was one doctor who could see more than one family and learn more about this disease. Fortunately I was invited by Dr Sardarwaller to visit the Willink Biochemical Genetics Unit in Manchester "to see how they might be able to help". Dr Sardarwaller had recently retired as Director of the Willink and was keen to introduce me to the new Director, Dr Ed Wraith. Dr Wraith offered to hold a clinic for patients with NiemannPick Disease...and the rest is history!

Shortly after the boys were diagnosed, I had also written letters to everybody I could think of who might know about this disease (no internet or email in those days!). Eventually I received a letter from the National Institutes of health in Bethesda, USA. They said that they were conducting a clinical trial on the use of cholesterol lowering drugs for NPC and wondered if we would like to be involved...

Murray's seizures were proving difficult to control and he was too ill at the time, so Roy and I went by ourselves. We were made very welcome and met Dr Roscoe Brady who was in charge; Dr Peter

Pentchev, the biochemist, and a young Australian doctor who was working there called Dr Marc Patterson. We also met some parents from the US who had taken part in the first part of the trial, to see if the drugs could reduce the amount of cholesterol in the liver. The second part was to see if the drugs had any effect on the brain. Unfortunately there was a problem with funding for the next stage of the trial. Some of the families who had been on the trial had arranged a meeting with the doctors involved to see how they might raise funds to help with the research. Roy and I were invited to come along to what would turn out to be the first meeting of the National Niemann-Pick Disease Foundation. The folks at NIH said that they would be pleased to enrol both Roy and Murray on the trial when it got going again.

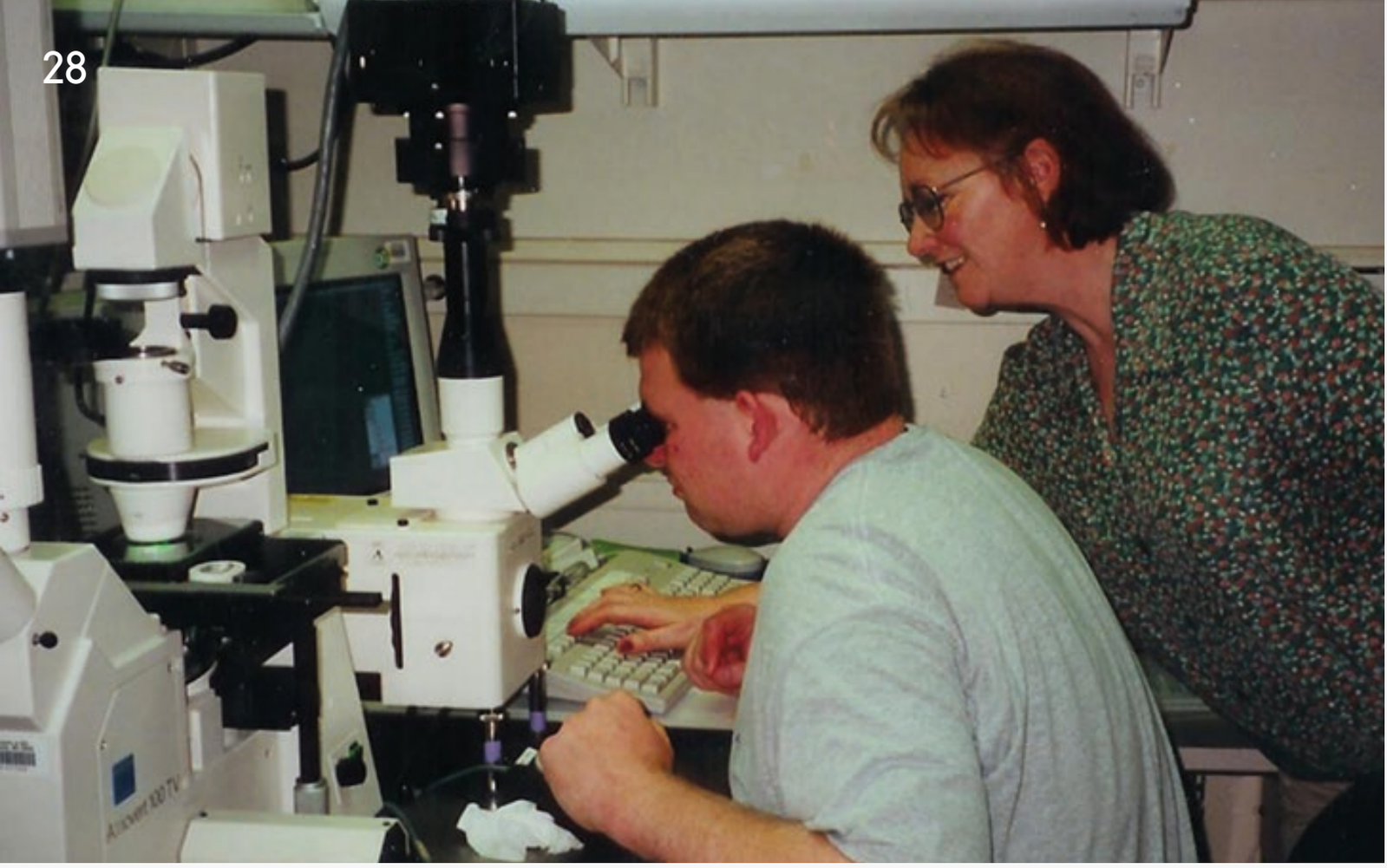
Jim and I knew that, if we wanted the boys to be part of the trial then we would need money to get us there on a regular basis. We went public with our story in the local newspaper. Not an easy thing to do! Our community in Hawick were fantastic and rallied round to help. In this photo I am clutching our tickets to Washington!

Unfortunately the second part of the trial did not take place. The clinical endpoint was to have been walking ability, which was the only thing they could measure. This meant that it would have needed to be a double-blind multi-centre trial with 100 patients, over two years. There was not enough funding. This was very disappointing. However they did want to see patients, including Roy and Murray on an annual basis to monitor the course of the disease. Later we heard that they were close to identifying the gene for NPC but needed blood samples from families with two or more children. We all gave our blood and we sent letters to all of the families we knew in the UK with two or more children, giving instructions on how the blood should be collected and shipped to the NIH. They all sent their blood!

The difficulties with not having a suitable marker that could use to measure whether a therapy was working, led to another small clinical study involving just three young adult patients. This was conducted by Dr Chuck Schwartz in Richmond Virginia. Roy agreed to take part. It involved having a tube inserted through his nose into the upper bowel to collect bile every two hours and two-hourly blood tests. The aim was to measure the size of the cholesterol pool in the body. The only difficult part for Roy was that he was not allowed anything to eat all day! At the end the nurses presented him with a chocolate cake! They had heard that it was his birthday next day. Roy was delighted!

It was good to be home again but we were realising that time was running out for Murray...

It was good to know that the research was progressing but unfortunately so was Murray's disease and time was running out. In October 1996, Murray, our wonderful, funny, amazing son, died. He was 18 years old. The doctors had been right in saying that he would not live beyond his teens. No amount of research could help Murray now. Our family's journey with Niemann-Pick disease continued and there were many joys and challenges to come...



In June 1997 Dr Gene Carstea came to our conference with the exciting news that the NPC1 gene had been found on chromosome 18! There was celebration all round; he discovery of the NPC1 gene, and later an NPC2 gene, enabled the development of genetic tests for pre-natal diagnosis and carrier testing. The genes also revealed a new way that cells handle cholesterol. They provided an understanding of a previously undefined pathway of cholesterol metabolism. This orphan disease might someday shed light on more common illnesses involving cholesterol, including heart disease, stroke and Alzheimer's disease. We were all so sure that a treatment or cure was just around the corner!

Roy continued to attend NIH every year and enjoyed meeting every one again while they collected samples, and information. His particular favourite was Jackie; she let him go to sleep when he was having his EEG! Roy also enjoyed the food!

Roy was always keen to be involved in research. He had watched the effect that this disease had on his brother and he was determined to help. He said, "Mum, if the doctors are working so hard trying to find some answers to this disease then we need to do our bit to help". So we continued to attend NIH every year and enjoyed meeting every one again while they did their tests and collected samples, and information. We were always made to feel welcome and the clinicians and researchers went out of their way to answer all of our questions. We were able to spend some time in the laboratories learning about the research and Roy was amazed to be able to look at his own cells through the microscope.

Roy and I usually managed to time our visits to NIH with attendance at the NNPDF family conferences. Roy would sit in on most of the presentations and was often found in conversation with researchers. Here he in the picture below with Dr Steve Sturley "the yeast man".

Meanwhile the clinical research was moving ahead here in the UK. We now had a large group of Niemann-Pick patients, being seen regularly by one doctor! The NPDG(UK) [now Niemann-Pick UK] contracted with the hospital Trust in Manchester to employ a Clinical Nurse Specialist to support patients and gather

clinical information. Although she was employed by the hospital Trust, her salary was paid for by the charity.

All this galivanting around can be tiring! But whilst this went on the clinical research was moving ahead in the UK. We had the largest group of Niemann- Pick patients being seen regularly by one doctor in the world! We had the first clinical trial starting for Miglustat, or OGT 918 as it was known then. It was exciting! It was Dr Fran Platt that I first heard talking about sugar A, at a Climb conference. Its real name nbutyldeoxynojirimycin was a bit too long! Then Dr Steve Walkley also gave a presentation on his work with N- BDNJ at an NNPDF conference. I asked him if he had met Fran and he said no, he had only read her research papers. So we invited them both to the next NPDG(UK) family conference. Theirs proved to be a very productive partnership.

We had the first ever clinical trial starting for a possible therapy for NPC. Miglustat, or Zavesca as it is now known, was a small molecule drug which it was hoped would slow down the progression of the disease. It was exciting! Roy was one of the first patients to be screened for enrolment in the trial in the UK. It was great to be involved in something for which we had waited so long.

On the second screening visit he met the speech therapists who did the swallowing assessment. He enjoyed that bit because he got to eat again!

Then there was the nerve conduction test – he definitely didn't like that! Next, the nutritionist, Roy always likes to talk about food! We then went to Sheffield by taxi to do the eye movement test. Disaster! Roy couldn't do it. He just kept falling asleep. It meant that he would not be able to be on the trial. Everyone was devastated! Except Roy who just wanted to sleep. There were even some tears and I found myself comforting them. I was horrified, if Roy couldn't do the test, how would the little ones manage? This could screw up all of the study! People kept apologising that Roy would not be able to take part and all I could say was, "that's OK, the important thing is that the trial goes ahead"...

The journey home was awful. I desperately wanted to talk to Jim but I couldn't because Roy was there. I didn't want to upset him although he seemed to have forgotten all about it – dementia can be a blessing at times! We were all bitterly disappointed but resigned to the fact that Roy would not participate.

We went to NIH as usual in the summer and Dr Fitzgibbons, the ophthalmologist who Roy saw every year, asked how he was managing with the trial. When I explained that Roy was not on the trial because he could not do the eye movement test Dr Fitzgibbons said, "that's nonsense, of course he can do it, that's what we do every year!" He made a phone call and it was agreed that Roy could do the test again with Dr Fitzgibbons who was the person doing the test for all the USA trial patients...Roy passed with flying colours!

Back to the Willink in November and Roy was finally enrolled on the trial. He was very pleased – I was stunned!

Helena and Jackie were delighted! The worst part was over, it was all uphill from there. That is except for the diarrhoea... but that's another story. I now know what the phrase "when the s**t hits the wall" actually means. Seriously, we were very well looked after at every stage of the trial.

We were delighted that some of Roy's symptoms actually improved on the trial, especially his speech and swallowing. That was an unexpected bonus. Taking Zavesca has helped to maintain Roy's quality of life for much longer than we expected. The fact that he was still able to eat heartily has helped a great deal to keep him in good health.

There are no miracles however, and the disease continued relentlessly. Roy was no longer able to walk and needed someone to push him around in his wheelchair. His dementia progressed, but he would understand what was being said to him and never lost his sense of humour.

As Roy used to say, "if it wasn't for the Niemann-Pick I would be fine!"


npuk
Thank you
RAREDISEASEDAY.ORG
hope
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Niemann - Pick UK


30

Roy was always keen to be involved in research. He felt it was important. He would say, "if the doctors are trying to find some answers to this disease then we need to do our bit to help". At age 33 Roy was no longer able to travel to NIH but we were asked if he would be willing to be part of an extension study for the N-acetyl cysteine trial at NIH. They needed more information on adults with NPC and would like to access Roy's medical records. I asked Roy and he replied as he always has in the past..."go for it!"

When the NPC1 gene was found Dr Peter Pentchev said, "the identification of the NPC1 gene is the fruit of a successful partnership between scientists and families and is a significant step in the research of Niemann-Pick disease type C. Scientists from many countries worldwide contributed to these findings by sharing data and cell lines from affected families. NPUK helped by locating families with children affected by the disorder and the investigators received major financial support for this work from both the APMRF and the NNPDF. He said that families with children affected by NPC have been important for much more. When he first began studying the causes of this disease, the lack of knowledge led to an 'incurably hopeless wasteland'. Even worse, the disease was one of the 'orphan diseases' that strike only a small number of people, leaving drug companies and most laboratories little incentive to understand them. The parents did not accept this hopelessness, he said, they took it on their own shoulders to enquire about what causes this disease. We had NOTHING but their courage. These pioneering families, their faith and perseverance, that's what led to the cloning of the gene". Well, we have come a long way since then. We now have approved therapies for NPC, and more clinical studies and trials taking place. Dr Ed Wraith and his team in Manchester were at the cutting edge of clinical research (sadly Ed died from a neurodegenerative disorder on the 10th April 2013). We have had a clinical research nurse as well as a clinical nurse specialist, publishing research papers based on clinical observation of a very large group of patients. We have a team of dedicated clinicians and scientists in the field of Niemann-Pick disease and new people being drawn into this field. Hundreds of research papers now appear on pub med. We have top quality pharmaceutical companies committed to developing therapies for NPD as well as International Registries of patients.

In 2009 there was a coming together of several Niemann-Pick support groups and clinicians from 12 different countries which led to the formation of the International Niemann-Pick Disease Alliance to provide a forum for patient groups and professionals working in the field of Niemann-Pick Disease. Today they are a global network of 23 non-profit groups (and counting!) supporting hundreds of families. By joining together through the INPDA, families affected by Niemann-Pick Diseases have a stronger global voice, and progress towards effective treatments is accelerated.

The Alliance has also since launched the International Niemann-Pick Disease Registry, a collaboration between clinicians, scientists, researchers and patient associations across the world to collect clinical, diagnostic and outcome data from patients with Niemann-Pick disease. Data held in the registry will help aid progress in the understanding of Niemann-Pick diseases and potential treatments and therapies can be developed or refined in order to make a positive impact on the lives of people with Niemann-Pick disease across the globe.

All of this and more based on, "The commitment of pioneering families, their faith and perseverance". If you ever feel that you would like to take part in a clinical trial then, as Roy would say GO FOR IT! Today you are still pioneering families!

Remembering Peter Carlton Jones

NPUK Trustee Richard Rogerson

As part of our core objectives, which have guided our small charity for the last 30 years, we encourage scientific and medical research in the field of Niemann-Pick diseases, believing it vital to achieving positive progress for our community.

With this in mind we offer an annual award of up to £1,000, the Peter Carlton Jones Memorial Award, which is made available to an individual who is engaged in either research, teaching, treatment or care in the field of Niemann-Pick diseases, within the public or private sectors in the UK. As part of our NPUK: 30 celebrations we sat down with long-serving NPUK Trustee Richard Rogerson, to get a bit more background on this invaluable initiative:

"...he [Peter Carlton Jones] was introduced to NPUK through Tony Jellings [Former NPUK Trustee], who was a family friend. Peter became interested in the group and subsequently became a committed member of the Trustee Board. It was in his role as Trustee that Peter, along with his wife Lesley, attended the NPUK Annual Family Conference in Telford in 2004, during which Peter suddenly collapsed and sadly passed away. In honour of Peter and his valued work on behalf of the Niemann-Pick community, his fellow trustees developed an award scheme in his name..."

"...the first Award was presented in 2006 to Dr. Emyr Lloyd-Evans, then of the University of Oxford. Dr Lloyd-Evans has subsequently moved to Cardiff University, where he runs his own lab, continuing his work and interest in Niemann-Pick diseases and inspiring his colleagues and students to do the same...

...since its inception, the Award has been granted to 21 young researchers, generating worldwide interest and contributing to understanding and progress in the field of Niemann-Pick diseases..."

We are grateful to Peter and his family for their commitment and contribution to NPUK and for the ongoing and hugely valuable legacy of the Award that holds his name. We hope that it will support positive interest and potential progress in the field of NPD research for many years to come. We also thank Richard for his incredible support given over so many years.

THE MAKING OF INVISIBLE MANNERS

Invisible Manners is the title of our latest animated short film, which alongside a broader campaign on ASMD Niemann-Pick disease type B (NPB) and other rare, often overlooked conditions, hopes to raise awareness of the individuals and families affected by truly giving them a voice...

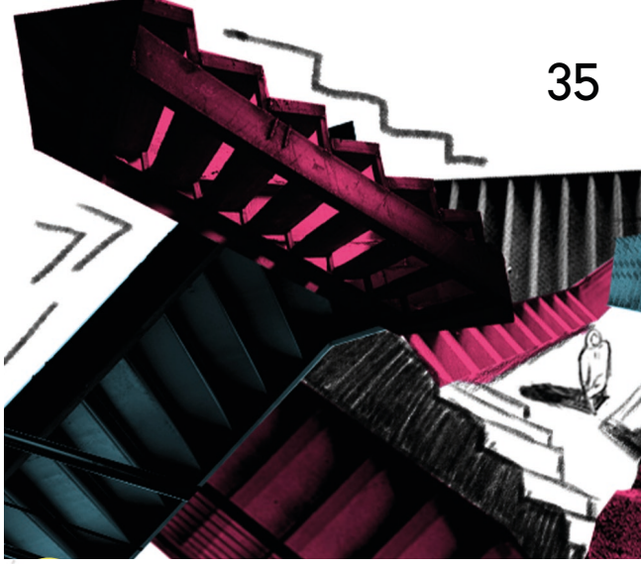
It was during 2019's Annual Family Conference & Interactive Workshop that we held what would become the first of many meetings with ASMD NPB families (both UK and international) to discuss how we could work in collaboration to raise further awareness of this often sidelined form of Niemann-Pick disease...and in doing so help to better represent the lives of those affected by it. One of the main points that came out of this initial discussion was how ASMD NPB can often times be considered an "invisible illness", and as such can all too often be misunderstood by the outside world. This can be similar for many conditions, including NPC and other related diseases...so perhaps our project should speak to a broader audience? Perhaps a film focusing on public perception of invisible conditions, and the need for kindness and empathy over judgment would hit the right note?

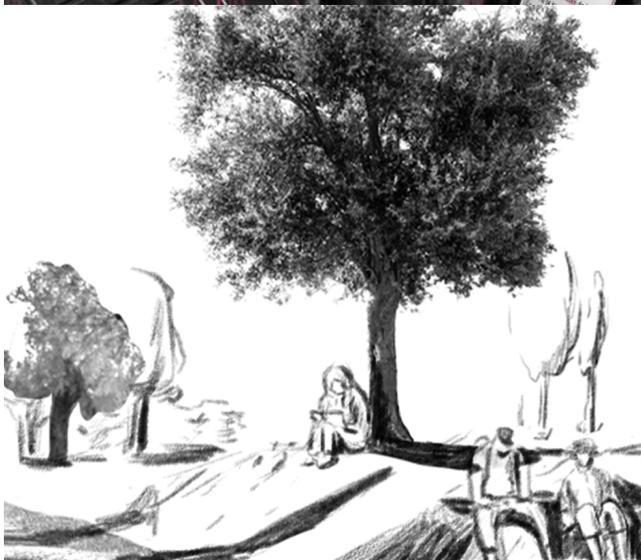
Well, development was very positive but then...a spanner in the works, yes COVID-19. It will likely come as no surprise that lockdowns and working from home don't really lend themselves very well to film making...and so to counteract this obstacle we found ourselves changing direction entirely whilst still playing to our strengths - namely our close relationships with the community and respected standing as a small charity. This is when we had an idea, we could write a narrative style poem based on the many shared experiences of our patients and families, which could then be read by our community (provided remotely by digital recording) and set to animation - in short, we could develop an impactful and truly collaborative film without ever having to jump through any COVID-19 related hoops!

This is where the incredibly talented artist and animator Lingxi Zhang comes in (read more on pages 36-39), who instantly wowed us with her portfolio and seemed to get the ethos of our charity and the project straight away. We recognised immediately that her artwork would provide the perfect scenescape for the poem, and were thrilled when she agreed to join the team. In one sense the work was just getting started...but we were excited with this new direction and the rejuvenated energy behind the project.

So began the process of storyboarding the scenes and ensuring each image presented reflected the experiences of those affected by an invisible condition. We wanted to make reference to the blue badge scheme, to invisible symptoms, to depression and the frustration of being misunderstood by so many...but also offer the positive chance for learning. At every stage of development it was necessary to continue to work closely with our community in order to best illustrate the many complex emotions and challenges an individual with an invisible condition can face on a daily basis. Lingxi's creative streak meant that we started to work to a more abstract profile, using a mixture of animation, photography, and narrated lines read by stars from both outside and inside of our community, to better tell this story. In doing so we could raise awareness of not only Niemann-Pick disease(s) but invisible conditions more broadly...as "not all that you can see, is everything that is there", and this is a feeling shared by many individuals and organisations, particularly in the world of rare diseases. Collaboration then, is key to progress.

Months later, we find ourselves very close to an official release date, and are on track for a late November/ early December 2021 premiere. We want to say a special thank you to Billy Boyd, Weruche Opia, Isy Suttie, Lilliput, Badger, and friends of the Niemann-Pick UK/INPDA International Niemann-Pick Disease Alliance community for their support on this project, which has meant that despite being a very small charity on a limited budget, we have been able to create something really special, which we can all be proud of. So seriously, thank you! We look forward to you all watching the film, and getting your feedback.





LINGXI

AN INTRODUCTION

Lingxi Zhang is an extraordinarily talented animator based in Paris, who has brought creativity and ingenuity to our latest short film project, "Invisible Manners", which hopes to bring to light both the complex challenges and public perception of invisible conditions, particularly ASMD NPB.

From the start Lingxi has really took this project and ran with it, inspired by the input from patients, families, and friends of the community - leading to her producing concepts that have gone above and beyond what we expected...

Back in July, Eleanor (NPUK Comms Assistant) sat down with Lingxi to chat about her involvement in Invisible Manners and to learn a little more about her inspiration:

Hi Lingxi, thank you again for taking the time to chat with us, I'm sure everyone will be eager to learn more about you! So, when did you start doing animation and how did you get into it?

I've always been interested in animation since my childhood, watching cartoons on my TV was the most important part of my day! I grew up


Étienne
&
Mathilde

with both Disney and Japanese anime...and so I was a typical student drawing in my math book during class. When it came to the day to choose what to study in university, I wrote down animation without an idea of what it'll be as a profession, but simply knowing that I like it.

So TV of course had an influence, but who is your favourite animator and what is it about their work which inspires you?

It'll be definitely Sylvain Chomet; a French animation director who made *The Illusionist* and *The Triplets of Belleville*. [The latter] was one of the first French films I watched, and I was totally shocked by both the storytelling and the story itself! For the first time an animation was not about princesses, superpowers, or cute animals going to adventure... it was about the life of an ordinary person [and was presented] with such a sensibility, empathy and depth. It opened my eyes to another possibility of animation, and I pursued it from China to France. So yes, it is a life-changing film for me

You have worked on many incredible and inventive projects, but what has been your favourite project to date (barring this one of course!)?

My favorite experience (apart form this one!) is a project called "En sortant de l'école" (*When Getting Out of School*), which is an animation series on French Television. The film is based on a poem (quite like this one!) and that's the only rule. They gave me maximum freedom to express myself

A selection of Lingxi's previous works, showcasing her adaptability

both in the narration and in the graphics. The whole project was accompanied by experts in every domain of animation production, and I had a wonderful experience in a script writing residence at a beautiful abbey called Fontevraud-l'Abbaye for one month.

Well unfortunately, we did not have any writer's residences to offer...but what drew you to this project in the first place?

After all of the professional experiences I have I hear myself saying that I want to do something which really matters...I guess this is natural and something which everyone experiences; in the beginning we look to ourselves and personal improvement, then we start to look outside so that we can go even further. So I really cherish this opportunity to work with a small charity, and to use my skills to tell the their [patients and family members] story.

That makes sense, so did you know much about rare diseases before being involved in Invisible Manners?

I have to admit that I didn't know much about rare diseases...but *Go Make Memories* gave me some of my first insights. The big difference with this project [compared to Lingxi's previous projects] is that I'm not imagining a life nor telling people how they should feel, but trying to express a voice of others - those who we rarely hear or we are not even aware of. If my work can help the community to be more seen and more heard, it will be my great pleasure.

Well, it is our pleasure to have you on the team! What was the inspiration behind the concepts and storyboards you have developed so far?

They're based on the poem *Invisible Manners* and conversations I have since had. I really wanted to visualise the "invisibility" through the art of animation. We developed a story with John that centred on our main character walking in a park, facing different obstacles and dealing with looks from others. It could be a threatening shadow, or a double reality that is only visible for people who suffer these disease...this will help to build a better understanding and awarenesses of these situations.

As you've mentioned, you've worked a lot with NPUK Communications & Campaigns Manager, John Lee Taggart, who is both Writer and Director on this project. But what has this process been like?

It is my great pleasure to meet and work with John, as he is so passionate about the work, and I get so much positive feedback on the way which really helps to push the film forward! The process of the film is not always linear, we had several rounds of character design, concept art and storyboards. With each of these we had discussions, and became clearer on what we really want to tell and the best way to do it.

Perfect...so lastly then, what are your hopes for this film?

I hope the film is first of all appreciated by the community, the film is for them and their opinion means a lot to me. In the long term I hope it's seen and talked about by as many people as possible, whether in festivals, on the internet or in our friendship circles. The objective is to make the "invisible" visible to the public, so we can make a change for the better.

From left to right: Former INPDA President Jim Green, INPDA Executive Communications Officer Lisa Chavez, Professor Marc C. Patterson, M.D, NNPDF Executive Director Joslyn Crowe, and NPUK Communications & Campaigns Manager John Lee Taggart

MARKING GLOBAL NIEMANN-PICK AWARENESS MONTH

On the 19th October the International Niemann-Pick Disease Alliance (INPDA) and Niemann-Pick diseases groups across the world, marked Global Niemann-Pick Diseases Awareness Day, a day for pushing for further awareness of Niemann-Pick diseases, whilst looking back at how far we have come over the years and planning for the future.

To mark this day, the INPDA released a short film with the intention to raise further awareness of Niemann-Pick diseases and enable more research towards potential treatments and cures. Although at times pushing for awareness can be overwhelming in a world where so many important campaigns, charities and awareness groups battle for recognition, the INPDA hoped that the video would encourage viewers to play a small role in raising awareness, with an action as simple as a like, comment or share of the short video. We did not quite expect the reaction it received, with 88 shares from the INPDA Facebook page! This was an absolutely incredible result for the INPDA and just illustrates that people are ready to listen and lend their support to the cause. If you have not yet watched the Awareness video, you can check it out on the INPDA Facebook page, perhaps even give it a share and help us to reach triple figures!

October is Global Niemann-Pick Diseases Awareness month. Although any day is a chance to raise awareness of Niemann-Pick diseases, during October we aim to really move beyond the bounds of our own community and reach those who may never even have heard of Niemann-Pick diseases, encouraging them too to take action in the fight for further research into Niemann-Pick. Let's have a look at what a few of the global Niemann-Pick groups got up to throughout the month...

To commemorate Global Niemann-Pick Awareness month the Australian Niemann-Pick Type C Disease

Foundation launched their #NPCDiaries campaign. Via this campaign, the Australian NPC Foundation shared individual's stories of living with Niemann-Pick disease Type C through, short, moving, and informative videos, which were shared across their social media, with links to their website for more information.

Associação Niemann-Pick Brasil held a live talk with Dr Rita Dos Santos Mendes, Neuropediatric Doctor of Pequeno Prince Children's Hospital on the 22nd of October, via Instagram, which was a really informative and useful chat. On Global Niemann-Pick Awareness Day, the group also shared some key moments in the history of Associação Niemann-Pick Brasil, in a short video which compiled a collection of photographs of community members over the years.

During October Asso Vaincre Maladies Lysosomales shared a powerful video on their social media, in which Tristan spoke about the affects of a diagnosis of Niemann-Pick Type B on his life. The video illustrates the importance of moving research forward and producing hope for patients affected.

On Global Niemann Pick Awareness Day, Associazione Italiana Niemann Pick Onlus released a short film via their social media in which families shared their own stories about how Niemann-Pick diseases have affected their lives from diagnosis to the day-to-day necessary care. The moving video was then shared across numerous platforms such as the news website, La Nuova Calabria, as well as a write up in the Italian newspaper, Il Risveglio del Canavese!

Throughout October, Niemann-Pick B-RS, led a campaign encouraging followers to share images of themselves wearing the Niemann-Pick B-RS t-shirt in a hope to raise awareness of Niemann-Pick Type B. This is such a great and simple way to raise awareness of the disease, and it so fantastic to see people representing our charity.

Fundacion Niemann-Pick de Espana utilised social media during Global Niemann-Pick Diseases Awareness month, by encouraging followers to learn more about the diseases and donate to their small charity, via a simple donation tool they had set up on their website.

The National Niemann-Pick Disease Foundation (NNPDF) were super busy during Global Niemann-Pick Awareness Month. On their social media the NNPDF created a profile picture frame to encourage individuals to show their support of the month. Via their Facebook page the NNPDF also shared personal stories of individuals affected by ASMD and NPC, encouraging individuals to support and donate where they could. They even had the backing of actress, writer, and producer Erika Alexander, who encouraged individuals to donate to the NNPDF and find a cure for this "relentless disease".

Niemann-Pick UK took over Rare Revolution Magazine's Socials on Global Niemann Pick Awareness Day (19th of October) ... don't worry it was all above board as part of their Tuesday Takeover series, in which they encourage a different rare disease charity to use their platform as a space to promote further awareness for their given rare disease. Throughout the day we posted quotes from NPUK and INPDA members via Twitter, provided answers to some burning questions about Niemann-Pick UK via Instagram and shared some updates about what our charity has been up to recently via Facebook. We were very pleased with the takeover, as it reached a lot more people than expected and helped to give NPUK a voice and raise awareness for Niemann-Pick diseases. Thank you to everyone who got involved in the takeover!

If you have any suggestions or ideas about how we can raise awareness or perhaps you even have some thoughts on Global Niemann-Pick Awareness Month 2022, we'd love to hear from you! Simply contact: info@inpda.org

INPDR UPDATES

With INPDR CEO, Conan Donnelly

The International Niemann-Pick Disease Registry (INPDR) was created by a collaboration of patients, health professionals and medical researchers, to document the Niemann-Pick patient experience; to improve diagnosis, treatment and care, to advance research and ultimately, to improve health outcomes for all those affected. In the following article, INPDR Chief Executive Officer, Conan Donnelly gives us an update on the latest progress.

Since our last update, there has been a lot going on at the INDPR, and much progress has been made. We have almost completed a review of the Patient Reported Database (PRD), which includes making a number of significant and valuable changes. When you next visit the PRD, you will see new questionnaires on quality of life and unmet need, plus additional updates that will improve your user experience.

If there is anything that exemplifies the purpose of the INPDR it is the PRD – it truly has the potential to contribute to better patient outcomes and is only made possible through the collection of comprehensive data in the INPDR alongside the strong Niemann-Pick community leadership that ensures the patient perspective is at the heart of the research.

The PRD is an opportunity to share your experience of Niemann-Pick disease. When you join the 400+ others that are currently contributing, you are helping to tell the real story of patients and families affected. Your data will help to support natural history studies, clinical research, health technology assessments and to determine the efficacy of therapeutics. In this way your data can affect real and positive change; to learn more or join the INPDR Patient Reported Database (PRD), visit: https://registry.inpdr.org/insight/prd/ Earlier this year, we were pleased to announce a collaboration with the University of Aston, Birmingham to support a PhD study which will further develop and strengthen quality of life questionnaires that were developed specifically for use in Niemann-Pick disease type C. Many of you were involved and contributed to the initial phase of this work, which has the potential to create and validate new outcome measures for use in clinical trials and other research. This will support the collection of information that truly reflects patient and family experience and helps to ensure that clinical research and therapies have a meaningful impact.

We were pleased to receive continuing grant support from the Ara

Parseghian Medical Research Fund (APMRF), helping us to make further progress in developing the Registry, particularly in the USA. In line with this grant, we are pleased to welcome our new United States Clinical Research Associate, Solomon Mbua, who starts work in early November. Please check our website and social media for the full official introduction - we look forward to seeing the positive impact he will make on behalf of the Niemann-Pick community.

We now have four US clinical sites, and therefore more US patients contributing their data to the Registry. This includes Rush Medical Center in Chicago and Benioff Children's Hospital, University of California & San Francisco, with more to follow soon.

An important step forward in September was a Letter of Support from the European Medicines Agency (EMA) for the use of the INPDR in clinical research studies. This support prepares the INPDR for supporting the essential studies that monitor safety and efficacy of new therapies.

A highlight for me in October was having the opportunity to join the NPUK 'Reunion Lunch', in Wilmslow. After starting work just prior to the lockdown in 2020, this was a long-awaited opportunity for me to meet and speak to patients and families. I appreciate that the work of the INPDR and the research that will come from it will not happen without the support and involvement of patients and families and it was my pleasure to spend time getting to know and understand more about the community we support.

To keep up to date with news and information about the INPDR, please 'Like' and follow us on Facebook @INPDR to ensure you don't miss out! For questions or further information regarding the INPDR, please contact info@inpdr.org

INPDR GETS SOCIAL @INPDR

A small collection of the recent posts from INPDR, for more follow @INPDR on Facebook, Twitter, and LinkedIn

PROGRESS TOGETHER

As ever, over the past ten months our fantastic fundraisers have gone above and beyond for Niemann-Pick UK, ensuring that we can continue to provide our vital support services- we want to give a shout out to a few of our latest fundraising superstars!

In March, **Emil and Jen Pruden** took on the gruelling **4x4x48 challenge**, running four miles every four hours for forty-eight hours! The previous year Emil had taken part in the challenge and this year his partner Jen joined him, all in the name of raising funds for NPUK. This was an incredible effort, the pair smashed their fundraising target and totalled £1,520.21. We are still so honoured and empowered by this tremendous fundraiser.

Long-time NPUK volunteer **Katie Reynolds** took on an impressive challenge of **walking 100 miles** in March! Katie explained a little bit about this fundraiser: "...those of you that know me, will know that NPUK are a charity especially close to my heart as our gorgeous Lucy gained her wings aged 4 in 2007. Seeing how much the charity means to so many people is the most amazing thing ever..." This was such a powerful fundraiser - thank you Katie for fundraising and for the support, time, and care that she has given to the NPUK community over the years!

It seems March was a busy month as **Cleo Walker** also set herself a challenge and pledged to run and **walk 50 miles** throughout the month to fundraise for NPUK. As Cleo ran, she added updates on her progress to her social media. Cleo almost quadrupled her fundraising goal, and her determination and support means so much to our NPUK community!

This year **Prosale staff members** nominated Niemann-Pick UK as their

DRAISING

Bruntsfield Primary School's P6S class held a pirate themed party to celebrate the end of term, and Alexander's (pictured bottom right with his brother William, both in costume) final week before leaving mainstream school. Alexander was lavished with gifts, and the school also raised £265 plus Gift Aid to NPUK...thank you ship mates!

charity to raise funds for and came up with various inventive ways to fundraise. As such, in April, Prosale Installation Manager, **Lauren Slender**, **ran 100km** throughout the month. In February, to mark the Polish celebration**Tłusty Czwartek**, known as Fat Thursday, the team offered **donations in return for traditional Polish donuts** to celebrate the day. Also, in August the Prosale team took part in the **Tough Mudder** challenge (pictured on the next page) with funds raised going to NPUK. Overall, the tremendous team raised a whopping £2,746.10 for NPUK! Clearly, this is a fantastic effort by the whole team, and it means so much that Prosale have been thinking of our small charity throughout this year.

A huge thank you to **Sofia Jameson** who, earlier this year, as part of her Duke of Edinburgh Award, **walked 80 miles** to raise funds for NPUK, alongside raising awareness by sharing information about NPUK and her fundraiser via her social media. This was such an inspiring fundraiser, and we want to say a huge congratulations to Sofia on her Duke of Edinburgh Award success!

In 2020 brothers **Dan and Jason Carter** decided to enter an ultra-marathon, but, like many things, their plans were put on hold. But, in June, the pair **ran the length of Hadrian's Wall**, a mere 70 miles within 26 hours. They raised an outstanding £5,088.93 for NPUK!

Mimi & NPD, this year printed **NPUK car stickers** to help raise awareness of NPC and raise money for NPUK. This was such a fantastic idea, and it really makes us smile when we see a car representing NPUK.

In June, **Sally Cowen and her daughter, Beth**, ran the **Town & Gown 10km**, in memory of Julia Corbett. Sally writes: "...Juliet was my mother's god daughter and the youngest of four children. She was lovely, loving, and much loved. Born in 1970,

married at the age of 21 and four years later pregnant with her son Max. She had begun to tremble and was mis-diagnosed as having MS. Juliet has been an inspiration all of my life, an infectious smile and an amazing singing voice. [---] Throughout Juliet's illness she never stopped smiling. Her memory lives on through her husband Martin and son Max..." This was an incredible effort by the pair who raised an amazing £695.89, we really appreciate such a show of support, particularly during a tough two years for fundraising.

NPUK fundraising powerhouse and NPC Mum **Helen Beveridge** in July took on an incredible fundraiser to raise cash for our small charity. Helen **swam between 25-29 kms in the sea between Jersey and France**, dodging the jellyfish! This was a superb effort, especially after having little time to train due to numerous lockdowns. As soon as we announced this fundraiser, Helen had the backing from the whole community, and she absolutely knocked her fundraising target of £400 out of the park... and then some, raising £3,116.26!

Tim Bamber this year has really shown his dedication knows no bounds as he undertook **numerous fundraisers** in the name of NPUK. Tim wrote: "...another year and another fight! Unfortunately, we find ourselves watching my beautiful cousin Mel suffer in very much the same way we saw my cousin Nat suffer... this disease is horrible and it breaks my heart! This is why I'm doing what I'm doing!" Tim's fundraisers have included, among many others, the Mad March Hare 67-mile bike ride in March, the 112 Fred Whitton sportive bike ride in May, an Iron Man triathlon in June and the Outlaw triathlon in September- let's hope this month he's putting his feet up! In total Tim raised a stunning £1,406.25 for NPUK!

A massive well done to **Millie**, Gloria Neal's (NPC: 1/7/54-8/11/19) granddaughter, who made the most of the heatwave this summer and set up a **jumble sale** during July with profits going to NPUK. By the end of the day Mille had raised an amazing £58.50, terrific work!

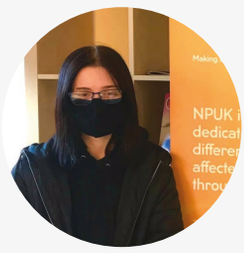
Jodie Ryan took on a challenge which would send a shudder down most people's spines...**skydiving for NPUK**! Jodie was inspired to take on this momentous challenge by Joshua (NPC,) and his family, and wanted to support NPUK. We are always amazed by the dedication of our fundraisers, but Jodie literally went above and beyond this time, incredible effort!

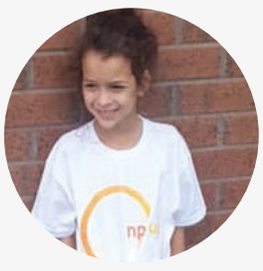
The NPUK Golf Day returned in September, which celebrated its fifteenth year. **Craig Mathieson**, NPUK Chief Executive, Toni Mathieson's, brother-in-law, originally created this event in memory of his niece Lucy who lost her battle to the disease in 2007. A huge thank you to all the golfers, sponsors, and attendees for making the day truly special. Craig had hoped to reach the £100k mark as a combined total for all NPUK Golf Days following the event, and we are pleased to say this target was reached! The event even made headlines in NPUK HQ's local newspaper, the Sunderland Echo, and it was fantastic to see our small charity recognised for the work we do.

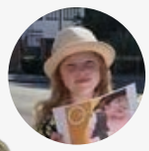
September also saw the return of the **Great North Run**, which celebrated its 40th year on the 12th September. Amongst the participants, were a few fantastic supporters of NPUK, as well as many more cheering them on. Sister super team, **Chrissy Mason** and **Katherine Turner**, dressed in matching tutus (in NPUK branding colours, of course!), took on the challenge, made local headlines, and raised a combined total of £2,326.79!

NPUK Chair, **Will Evans**, and his wife **Miriam Evans** also took part in the run. Miriam and Will's eldest son, Sam, aged thirteen was diagnosed with NPC when he was a baby, and the couple have been heavily involved in the NPUK community since that time. The pair even featured on BBC News, proving an insight into their motivation for running as well as speaking about NPUK and Niemann-Pick disease more broadly. Miriam and Will raised a phenomenal £7,031. 29 - it was incredible to see our small charity feature on such a large platform as the BBC News, and even more so to see the dedication of our fantastic runners on the day!








The Prosale Team were still all-smiles after their Tough Mudder fundraising challenge!

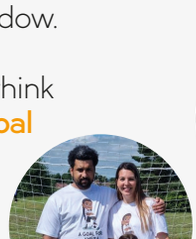
In October, **Karen Bowman** took part in the **London Marathon**, raising funds in the name of NPUK. Karen bravely signed up the marathon when a last-minute dropout looked like we may have lost our place for 2021. But Karen came through, and with limited time to train, absolutely smashed the London Marathon in just 3 hours 42 minutes! We think you will agree this is a tremendous effort from Karen, and we are so grateful for such an incredible show of support for our small charity.

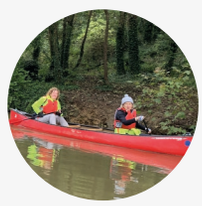
Jordan Hamilton held a fundraiser back in March 2020 which featured optional fancy dress, a disco, bouncy castle, face painting, glitter tattoos, and a whole lot more...in short it was a **party** well worth attending, and for a good cause too! The main aim was to raise as many funds as possible for NPUK whilst providing amazing prizes and a day of fun in memory of Jordan's son Freddie who sadly passed away in November 2018, aged 20 months, from Niemann-Pick disease type A. We hope all who attended had a fantastic time, many thanks to all involved in making it happen.

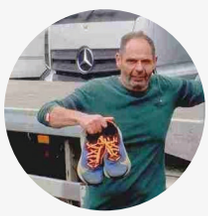
In September, **Phillip Winetroube**, friend of NPUK Chief Executive Toni Mathieson, took part in the **Berlin Marathon** and kindly donated the money he raised to Niemann-Pick UK. It was the first Berlin Marathon to take place since the Covid-19 pandemic began, and it has been so wonderful to see the gradual return of such events once again.
A massive thank you to Phillip, who raised an incredible £2,158.75 for Niemann-Pick UK!

Also in September **Matt Lawrence** ran the **Robin Hood Half Marathon**, donating the funds raised to Niemann-Pick UK. Matt is a headteacher and had been inspired to get involved in Niemann-Pick UK after learning of one of his pupils, Amera Adeyemo, diagnosis with Niemann-Pick Type C, as he wanted to find a way to support Amera and her family. A massive round applause for such a dedicated fundraiser!

Amera's school as a whole were also inspired to support her in whatever way they could. **Breadsill Hill Top Primary School** hosted a **penalty shoot** out at the school to raise money for NPUK, calling the fundraiser "A Goal for Amera", in which Amera's classmates were encouraged to score as many goals as possible in a 15-minute window. Amera chose to hold a penalty shootout as she is a football fan and a supporter of Derby County, and we think it was a pretty brilliant idea! The school also sold **"A Goal for Amera T-Shirts"** on the day, with funds raised also going to Niemann-Pick UK. In total, Breadsill Hill Top Primary School raised a fantastic £2,405.96 for NPUK, and we want to thank each and every person who attended the fundraiser for getting involved and donating to NPUK.



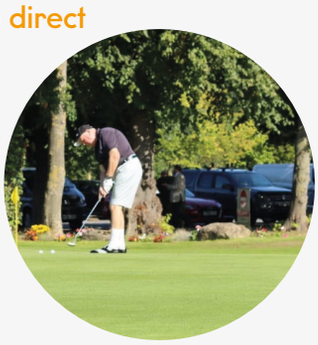




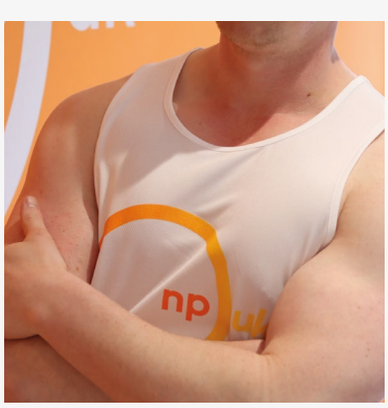
Our fantastic team of dedicated (and sporty) **NPD Super Scientists** were once again giving their all to raise funds for NPUK this September. The scientists decided to do approximately **70 miles around Oxfordshire and Buckingham** by bike and canoe! On the first day they cycled 54 miles, battling three punctures and various diversions, and on the second day they took to the water, paddling 12 miles and then walking the final distance. Although this feat was surely gruelling at times, it was well worth it as the fantastic team raised an incredible £1,951.56 for NPUK! Of course, as ever, we are overwhelmed by the Super Scientists support, who always live up to their name.

So far this year we have had a record number of individuals from our community setting up **Facebook Birthday Fundraisers** for NPUK. Since January 2021 Facebook fundraisers have raised over ... for NPUK. It is always such a privilege to see members of our community thinking of us, and particularly during recent years, we really have appreciated every penny donated towards our small charity so that we continue doing our vital work. So, a huge thank you to everyone who has set up, donated to, or shared a fundraiser, you are all very much appreciated!

We are so thankful for all the fantastic people who have offered **donations to NPUK every month via direct debit** - this income is incredibly useful and very much appreciated.
For more information on setting up a direct debit, please email: info@npuk.org



NPUK SHOP



Have you had a chance to check out our new NPUK Shop yet?

After a few delays to the launch of the shop, we are now well into the swing of things, with our online shop having been up and running over the last few months!

See, fundraising does not have to be a mammoth task like a sponsored skydive (although this was fantastic work Jodie!), it can be as simple as donating a few quid for an NPUK mask, and repping our charity while you join the morning commute!

Our NPUK merchandise items are simple but stylish in design, and through simply wearing our logo you can encourage more people to take notice of NPUK and the work we do. It's a bonus that funds raised will, as ever, go towards our support service... meaning whether you buy a bag, sweatshirt, wristband, or whatever else...you are directly helping patients, families, and friends affected by Niemann-Pick diseases.

The shop stocks all things NPUK; from keyrings to beakers, tote bags to t-shirts, with new stock being added regularly. As you can see from the images to the right, the NPUK Staff Team (Steve, Laura, John and Louise) couldn't resist trying on the gear themselves! So if you have not checked out our online shop yet, what are you waiting for?

Have a browse and see if anything takes your fancy. Remember Christmas is just around the corner and most of the items would make ideal stocking fillers!

The shop can be found via our website (npuk.org). Happy shopping! However if you have any issues with the shop you can contact our Comms Assistant Eleanor at: eleanor@npuk.org, who will be happy to help, ensuring that you get your NPUK swag as soon as possible!

INTRODUCING OUR NEW FUNDRAISING OFFICER!

Jenny Charman has joined the NPUK Staff Team in the newly established role of Fundraising Officer. We feel Jenny is a great fit due to her vast experience with the fundraising group, Harvey & the Brave Little Soldiers, and her personal family connection through her nephew, Harvey (NPC).

Tony Jellings
a fundraising powerhouse.

As part of our NPUK 30th birthday celebrations, former NPUK Trustee & Fundraising Coordinator, Tony Jellings, kindly shared an article with us, exploring his family's connection to Niemann-Pick Disease, and the fantastic work that he has been an integral part of over the years...

Juliet was the youngest of our four children. She was lovely, loving and much loved. She was born in 1970. Married at the age of 21 and four years later pregnant with her son Max. At that time she had begun to tremble. She was diagnosed incorrectly as having MS. Max was born in April 1995. One year later Juliet was diagnosed at the National Hospital for Neurology and Neuro-Surgery as having Nuclear Vertical Gaze Palsy.

While on a visit to our home in Norway, our very good friend Richard Rogerson (of whom more later) undertook some research on our behalf to ascertain more about this condition and to find out whether a support group existed. This led us on our journey home to Sussex making a detour to Scotland where we had arranged to meet with Jim and Susan Green to learn more about Niemann-Pick.

We were presented with a great deal of highly impressive medical detail and terminology and left Hawick feeling despondent and confused.

By the end of the year, having become a member of the support group's management board and having learned that there was no formal provision for fundraising, I offered to take on the role of fundraiser which was accepted, perhaps with some suspicion and reluctance.

At this time also the group operated under the aegis of the Research Trust for Metabolic Diseases in Children. I was not happy with the levy of 10% being imposed by the RTMDC on all funds raised by the group and consequently I began to agitate for the group to become officially registered with the Charity Commissioners. Registration subsequently took place in April 1997.

In the meantime, being concerned to strengthen the management committee I persuaded my friends Richard Rogerson and Sue MacPherson, both of whom had no family ties with the group, to bring their respective skills and knowledge to support the group. They both agreed and respectively became Vice Chairman and Treasurer. Several years later, again at my instigation, my friend Peter Carlton Jones, formerly Head of Public Affairs for Anglo American, was appointed to the Board of Trustees with responsibility for PR matters.

So far as the credibility of the group in the eyes of the general public was concerned, I felt it was important that we should have a team of distinguished patrons. So with the agreement of the board, I contacted a number of different people- mostly friends- many of whom accepted the invitation to become a patron. Those who did being the Earl Cairns, then the Chairman of BAT Industries, the company for whom I worked as Head of Personnel before my retirement and Professor Martin Rosser, Juliet's consultant at Queen Square. Others who accepted were Sir Robin Catford, formerly on the staff of Margaret Thatcher at 10 Downing Street, Dora Bryan, actress and comedian, Dominic Walker,- Bishop of Monmouth, Lord Bassam of Brighton, then a Whip in the House of Lords, Nick Mathias, director of IMG Artists and Guy Johnston, award winning international cellist- all friends of my wife and myself.

All these patrons have in their own way not only added to the credibility of the group but have helped in many different ways with fundraising.

Over the years, since 1996, our fundraising activities have been many and varied. From being very much a one man band, I was gradually able to persuade more active members of the group to become involved and this was an extremely welcome development.

Apart from appeals to Charitable Trusts, Institutions, businesses and Government, fundraising activities have included the following:-

Donations including in memoriam donations, birthday and anniversary gifts (ranging from £1 to over £5, 000), sponsored events (e.g. walks, triathlons, marathon, cycle rides, parachute jumps, swimmathons, etc etc), jumble sales, Christmas sales, fashion shows, garden parties, charity lunches, whist-drives, coffee mornings, auctions, raffles, sale of donated gifts (e.g. paintings from reputable artists), lectures, talks to schools and rotary clubs, sale of our own charity Xmas cards, collecting boxes, appeals to golf clubs and, very significantly not only from a fundraising point of view but also from a raising of our profile point of view, series of concerts.

Early in 1999 I arranged for an explanatory video, introduced by Dora Bryan, to be made professionally and to be used for educational as well as fundraising purposes. I expect this has become very dated now but it proved very helpful at the time.

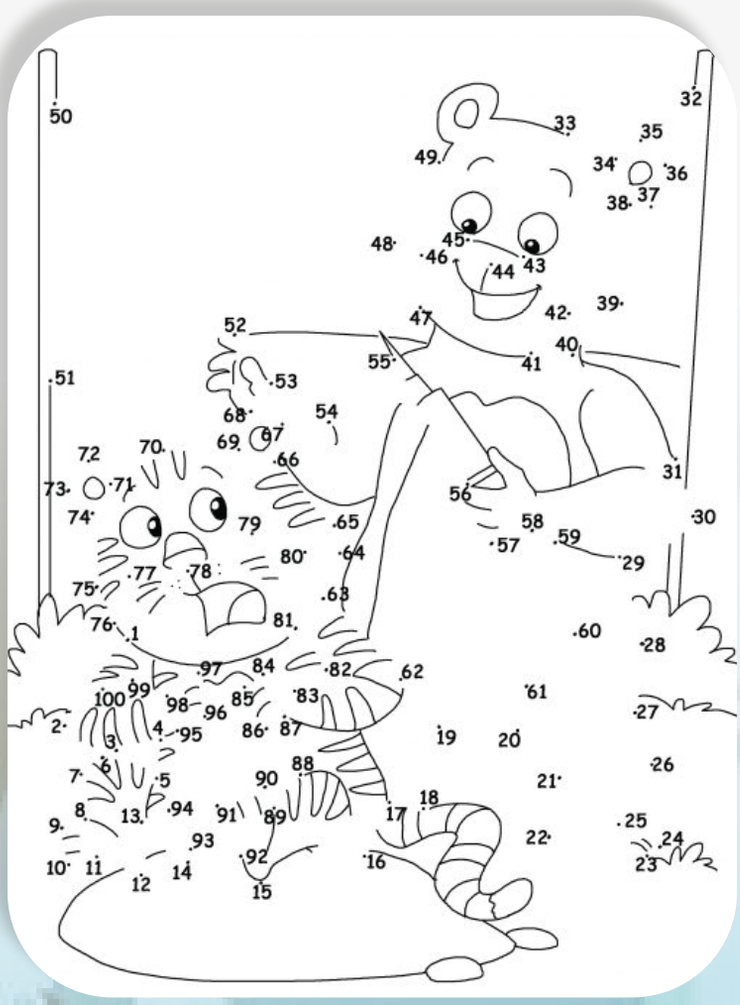
Fundraising has enabled the Group to meet its core expenses, produce a regular Newsletter, maintain a telephone helpline, subsidise the annual family conference and regional conferences and most importantly to fund the research project at Addenbrookes Hospital, to employ a Specialist Support Nurse and a National Development Manager (now the role of Chief Executive, held by Toni Mathieson).

After spending the last 18 years of her life in a care home Juliet died on the 26th August 2017. A total of over £1,000,000 was raised during my time as Fundraising Coordinator.

KIDS' CORNER:

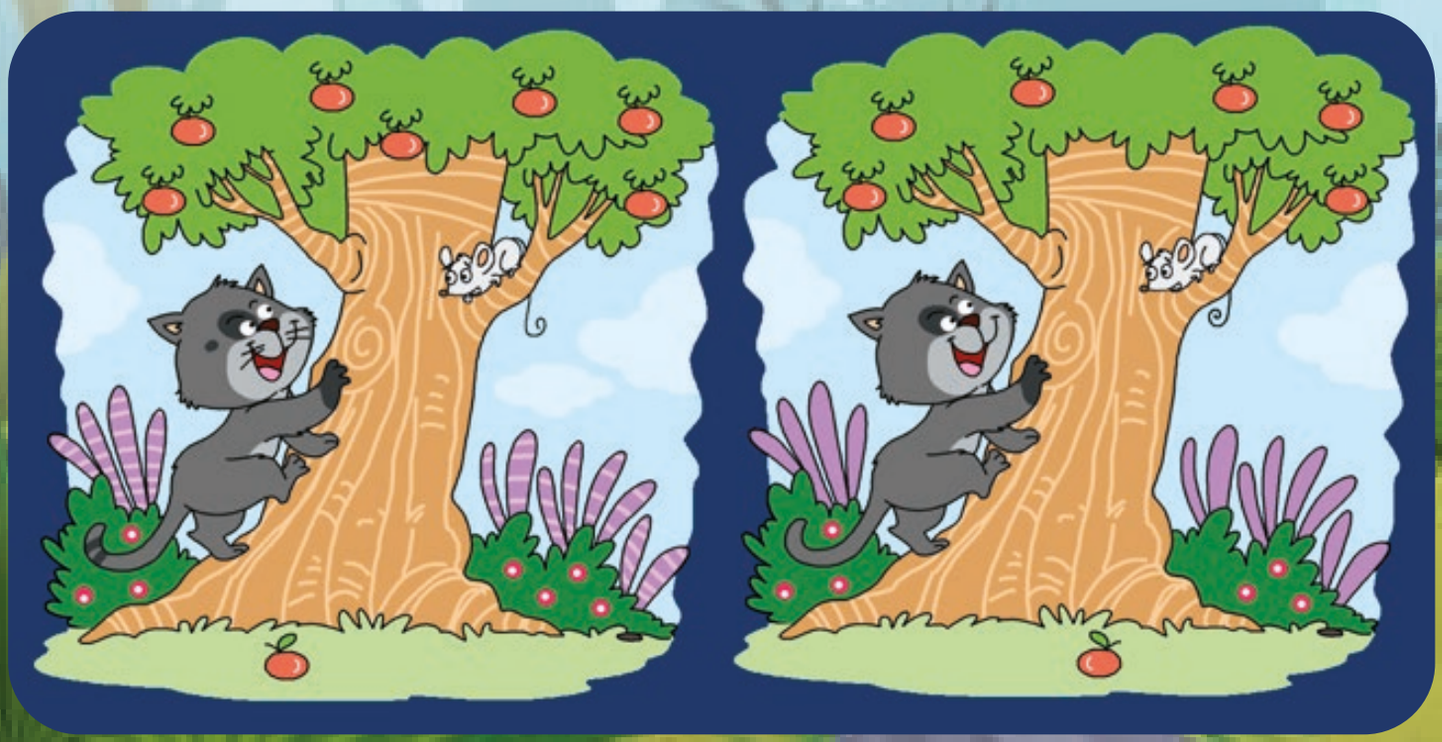
We are back with more games and acitivities for the younger members of the community (big kids permitted too!)

We hope you enjoy these fun games! If there are any activities and challenges 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

(There are 10 to find!)



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Whenever you complete these games (well done!) make sure you pass them on to a friend so they can challenge themselves too... why not see who can solve the puzzles the fastest? Good luck!

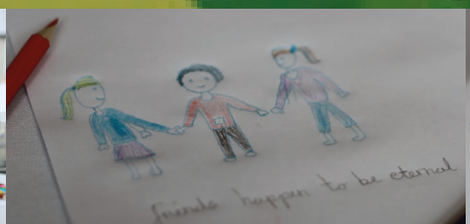
WORD SEARCH: how many can you find?

BATMAN / BETTY BOOP / BUGS BUNNY / CHARLIE BROWN / DAFFY DUCK / DONALD DUCK / FAT ALBERT / FRED FLINSTONE / GEORGE JETSON / GUMBY / HOMER SIMPSON / MICKEY MOUSE / MR. MAGOO / PINK PANTHER / POPEYE / PORKY PIG / ROCKY AND THE BULLWINKLE / SCOOBY DOO / SPONGEBOB / SUPERMAN / THE GRINCH / TOM AND JERRY / TWEETY BIRD / UNDERDOG / WILE E. COYOTE / YOGI BEAR

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

NPUK HAS TALENT: We invited our community to flex their creative muscles at this year's NPUK Reunion Lunch in support of Invisible Manners, our latest short film...and we were not disappointed! Below are just a few of the artistic offerings we received, which we will certainly be incorporating into the campaign as the project continues to progress...




TIME TO MAKE SOME PLANS!

Although in 2020 we had to find a new way of doing things, with many of us relying on technology for school, work and even family activities, we are sure you will agree, that nothing beats being there in-person.

With that in mind we have found a few family fun days out that, although following government guidelines, are a great way to get back out there and start making memories!

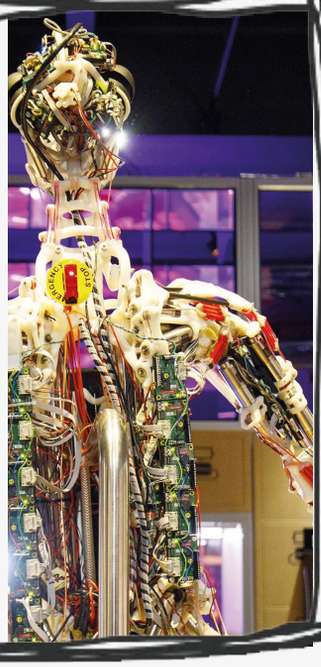
@NiemannPickUK

@niemannpickuk

@NiemannPickUK

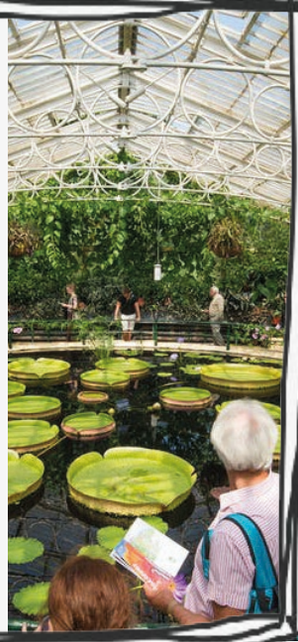
1) Become a Culture Vulture

The museum is always a great day out for the family, and there will no doubt be something to occupy everyone's attention. Although for over a year we have, for the most part, been unable to access the wonders of the museum, many have re-opened in 2021.
For example, the Science Museum in London is open as usual, and visits are available upon booking. They also ask that visitors continue to wear face coverings, unless exempt.



2) Get into Nature

Get back out into nature and visit the botanical gardens. As many botanical gardens and centres are largely outdoors this makes social distancing a little easier and ensures that the space is well ventilated. Kew Gardens in Richmond, for example, have worked hard to ensure that visitors feel safe. A one-way system is employed for some attractions, and they ask visitors to maintain social distancing and wear face coverings (unless exempt).



3) Have a Roarsome Day Out

Embrace your wild side and go on a safari. Knowsley Safari, Preston, is one of many parks across the UK that have embraced a COVID safe way of doing things. If you travel by car then this is an easy way to have a socially distanced family day out, as the 5-mile safari drive offers incredible sights across the park. However, they also have a foot-safari, rides, café, and restaurant, which all follow COVID-19 safety protocol.

4) Go on a Trip to Hollywood... Sort of!

Although a lot of us have spent the last year lounging on the sofa watching telly, cinema has still been a great miss. There is nothing quite like watching something for the first time on the big screen. So why not check out what is on at your local cinema and find out about their latest COVID-19 restrictions? Most cinemas continue to implement social distancing measures across all areas.

5) Take a Turn around the Town

Walking tours are a great way to learn more about a new place or get acquainted with the history of your hometown. Of course, not every Walking Tour available will be designed to keep little ones interested, but there are many specifically for families that are great fun. For example, in Bristol the Blackbeard to Banksy Walking Tour offers a fun and fact filled way to explore the city. The tour is wheelchair friendly, and they are constantly reviewing the news and information from government to keep both tour guide and audiences safe, find out more information about their COVID procedures by contacting them via email: aftbristol@gmail.com.

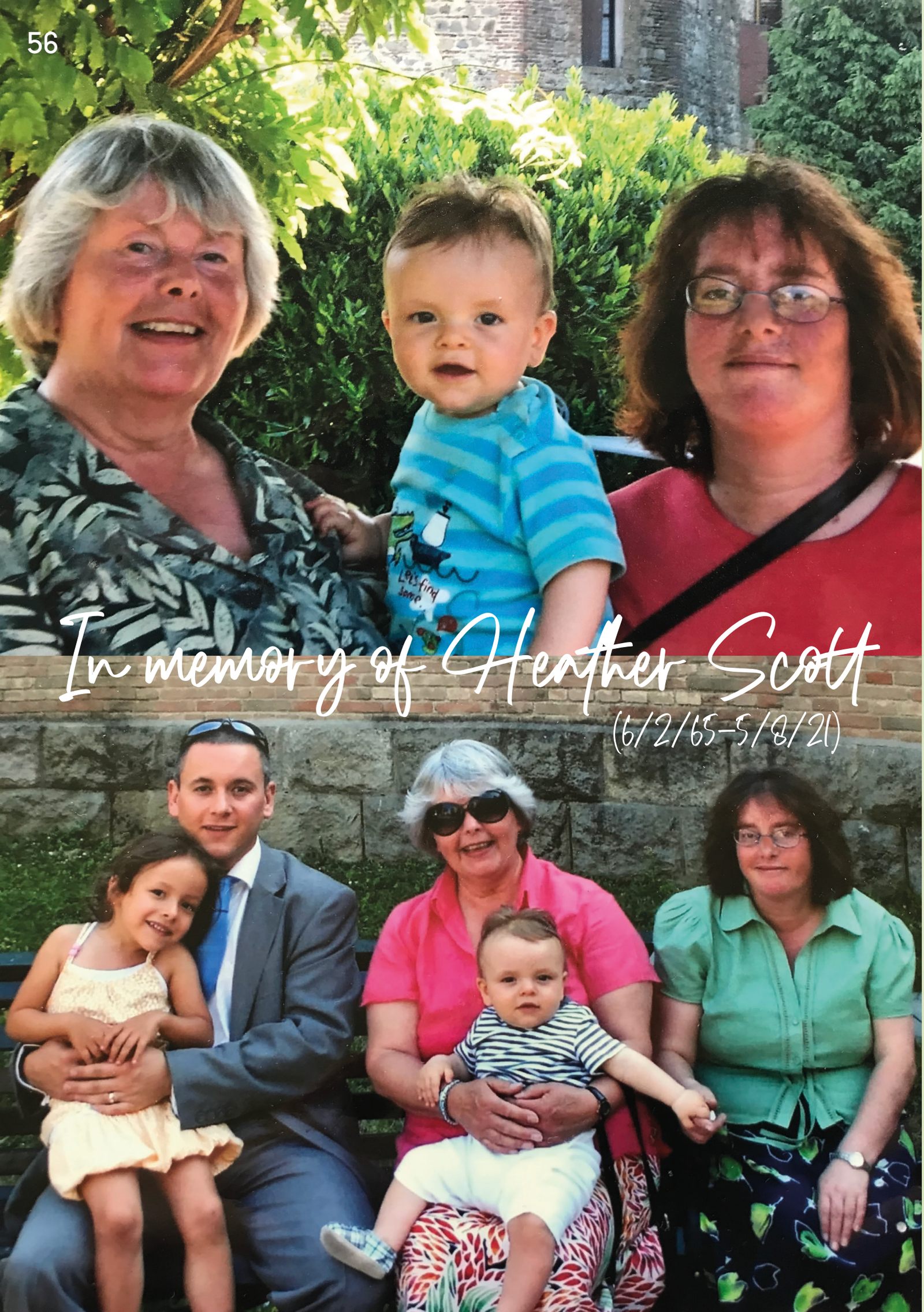
6) Bring your own Lunch

If all else fails, what is better than a picnic with the family?

Whether by the beach, at the park, or in your garden, it is an easy way to get outside and into nature, plus it is easier to regulate your own social distancing. The National Trust have compiled a comprehensive list of the most picturesque spots in the UK, nationaltrust.org.uk/lists/perfect-pic-nic-spots., definitely worth a look to make sure you make the most of good weather!

We would love to see all your snaps of family and friends getting out there and having fun, so if you are posting to social media make sure to use the hashtag #GoMake Memories

Although COVID restrictions have eased for many of us, what is most important when getting back to 'normal' is that you and your family feels safe doing so. If you want to speak with someone about your upcoming plans or receive further guidance on COVID safety and how to plan for this, get in contact with the NPUK team either by phone: 0191 415 0693 or email: info@npuk.org



In memory of Heather Scott

(6/2/65-5/8/21)

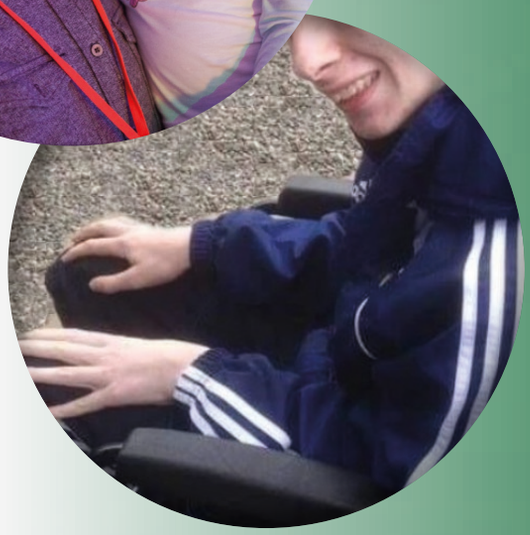
They say good things come in little packages, and as we think back on Heather that was most certainly true; born to John and Syl prematurely at just 4lb 60z, always the smallest in classes at school, and later a beautiful woman at 4'11"...this never seemed to matter, as her character simply radiated from her, filling any room with life, energy, and joy. It's no surprise then, that she was an incredibly popular individual and had many friends in the surrounding areas of Durham, Leeds, and beyond - this made for a life well lived, with many special memories to hold dear.

Family life was incredibly important to Heather, who was always surrounded with love; as a little girl she loved helping Grandad Wilson in his corner shop in Leeds, was close to both her Grandmother's, Mary and Irene, as well as her Auntie Pat. In 1973 her little brother Richard was born, and the family was complete...they later moved from Leeds to Durham City where she lived and worked for many happy years. Heather was always known as hardworking and dedicated, and experienced a number of different jobs, her favourite being when she held the role of Audio Typist at Durham Constabulary. It spoke to her especially as aside from the work there was a warm and playful atmosphere which Heather particularly enjoyed - the days would fly by with laughter as the Policemen, who had clearly taken a shine to her, would constantly joke and tease. This made for many fond memories.

As the years passed Heather experienced some travel adventures too, in particular in 2003 when visiting Italy with Mum and Grandma to attend her (now fully grown up) little brother's wedding, staying at Abano Terme near Padover, and visiting Venice with her new sister-in-law Marzia. She later returned in 2009, this time as a proud new Aunt, for her nephew's baptism.

Heather was a person you would likely find difficult to put in a box; yes she loved wildlife, in particular mallard ducks, and spent many a weekend in her youth hiking the wilds of the Northumberland and North Yorkshire Moors with a friend to see what she could spot, and in later life would enjoy feeding the ducks at the river in Warkworth with her Mum...but she also liked nothing more than cosying up on the sofa with her Grandma Irene to watch Countdown as well as many other TV quiz programmes! And when she wasn't doing either of these things, you may have been able to catch her listening to heavy metal music...a love affair she had begun in her teens and early twenties alongside friends with motorbikes, and carried on throughout her life! Every tune brought back those special memories, likely some of the brightest and best times she had enjoyed.

Heather's Niemann-Pick diagnosis occurred later in life, providing answers to many issues she had encountered over the years, but of course not stopping the gradual decline, which meant it became harder and harder for her to take care of herself. However with South Quay care home in Blyth, she found a haven for the last five years of her life... where she was embraced for the special character she was by both staff and fellow residents alike...whether it was when moving and shaking to heavy metal music, or simply when sat comfortably with a wordsearch puzzle in hand, she was content, she was happy...she was Heather.



In loving memory...

Don't think of them as gone away...
their journey's just begun,
life holds so many facets
this earth is only one.

Just think of them as resting
from their sorrows and their tears
in a place of warmth and comfort
where there are no days and years.

Think how they must be wishing
that we could know today...
how nothing but our sadness
can really pass away.

So just think of them as living
in the hearts of all those they touched...
for nothing loved is ever lost -
and oh, how they were loved so much.

Lorainne Carr (10/3/94–26/10/20)

Amana Johura (26/5/17–4/10/21)

Daniel Cross (3/6/98–25/8/21)

Stephen White (1/6/61–18/10/21)

Zubaida Malik (14/11/2017–26/10/21)

With grateful thanks to our grant providers:

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Pears Foundation

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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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