NPUK NEWS

Spring 2019

IN THIS ISSUE

Annual Family Conference / Research and Clinical Trials / Fundraising News / Kid's Corner / Care & Support Team & much more!

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Chairman's Chat

Dr William Evans, NPUK Chair

Welcome to the first issue of NPUK News in 2019. I trust that you will find the newsletter full of information and updates, and I hope it provides a real flavour of what is happening across our community.

Since I last wrote, NPUK continues to offer support to our community, with our staff team working tirelessly to try to make living with these diseases a little easier.

One area that occupies a significant part of NPUK staff and trustees time is our involvement with other patient groups and umbrella organisations. As a collective voice through organisations such as Rare Disease UK, Genetic Alliance, the Specialised Health Care Alliance, Eurordis and the LSD collaborative, which Toni currently chairs, we can learn from each other and advocate for challenges that we all share. Three areas that have had recent attention are: a patient charter for newborn screening, driven by the Genetic Alliance; care coordination for those affected by rare diseases, a key part of the UK strategy for rare disease; and especially pertinent with several of our clinical trials looking to achieve regulatory approval soon, plus the NICE appraisal process for the reimbursement of rare disease therapies.

Recently NICE has advised the NHS not to fund Brineura (cerliponase alpha) for Batten disease, due to uncertainty of its long term cost effectiveness. It is important that we are aware of this and support others, such as the Batten disease community, facing these challenges.

Their challenge now, may well be ours in the coming years. Indeed it is certainly possible we may find ourselves with an approved drug for our diseases available across the world but not funded nor available in the UK. We will continue to work to ensure we are optimally positioned should we face such a challenge and as part of that we work with our fellow rare disease charities to lobby on these issues.

Sorry to paint a slightly negative picture but it is important that we are all aware of the challenges that we face and that occupy a significant part of the NPUK staff and trustees attention.

I hope you enjoy our newsletter. Once again John and the team have put together a publication that any organisation would be proud of. NPUK punches well above its weight!

Warmest wishes,

Will

Chair, NPUK

"...it is important that we are all aware of the challenges that we face and that occupy a significant part of the NPUK staff and trustees attention..."

Clinical Nurse Specialist: Laura Bell

Hi All,

I hope everyone is doing well and looking forward to some warmer and drier months now that Spring is upon us.

Since I last put pen to paper it has been a very busy few months with lots of travelling far and wide across the UK to visit families in clinics, at home and providing support at meetings in schools, local communities and with local health care services.

The team here at NPUK has been able to continue to support the designated clinic days that are held at the Specialist centres within the UK and these continue to be a success for both the patients, their families and for the members of the multi-disciplinary team. It can often be quite over whelming to meet so many professionals at the clinic days and our aim at NPUK is to support you, explain to you who everyone is, and explain how they can help support you and your loved ones. Holistic care is a very important aspect of your care and management and we understand that there is often a lot more than clinical advice needed to help you with your day to day living.

I often get asked by parents or carers what they can do to help their loved ones and I advocate that it is also very important to take good care of yourselves. Whether this be healthy eating, exercising and taking the time to do something for yourself.

Living and caring for someone with a complex condition is challenging and can have a significant impact on the quality of life. The care and support we can offer at NPUK can help you address any issues you have in aspect of your day to day living so if this is something we can help you with please do contact us and we are here to help you in any way we can.

If you need any help, advice or just a chat I am here at the end of the phone and you are always most welcome to contact me. As you know I am always happy to hear from you, to accompany you to appointments or meetings or visit you at home. I am very much looking forward to seeing some of you at our NPUK events happening throughout the rest of 2019!

Hope to speak to you soon,

Laura

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Senior Families Advocate:

Elizabeth Davenport

Hello Everyone,

As we are approaching summer it's the time to think about (hopefully) long summer days spent outdoors relaxing:

Well for some of us relaxing is not always easy even though we are physically still our minds can be thinking and planning different scenarios in our heads. Minds and their tricks are terrible and really love to invade precious family time and can disrupt sleep. As a result we can forget important things or even affect everyday tasks. I have started to try and train my mind and basically asked it to stop annoying me with scary or stressful thoughts and when they do creep into my mind I allow them a little time and then move them along. I cannot control what will happen but for now it will not take over my moment or my day.

By breathing slowly and trying to concentrate on each breath or sometimes an everyday object I can slowly stay in my moment till I feel less anxious. It has made me think more efficiently and I have noticed that I am sleeping a little better. As a result I have done some reading about mindfulness. At first I thought not another fad but it has really helped me so thought I would share this with you. If you want to know more then call me. It might also be a useful idea if you search online for mindfulness and see if any of their exercises help you!

As always I have been out and about meeting with the lovely individuals and family members of the NPUK Community by making home visits and also attending Clinic Days in Manchester and London. I also look forward to this year's Annual Family Conference & Interactive Workshop (20th-22nd September) so that I can see everyone together again; as ever it's a fantastic chance for families new and old to meet in one venue and share their experiences. For further details please see page 22 in this issue, or email the NPUK Central Office team at:info@npuk.org.

Have a great summer and even if it rains let's make the most out of it!

Elizabeth

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NPUK PROJECT FAMILIES OFFICER

STEVE NEAL

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

Throughout the past six months I have been supporting our community and working more closely with clinical staff, and outside agencies. I have been using coaching programmes and coaching skills in clinical practice, but a majority of my time has still been focusing on understanding and working with common life problems and obstacles to well-being that our families experience due to direct or indirect connection with Niemann-Pick disease.

This has allowed our families to develop an increase in confidence and engage more with the community. I am also supporting families who have a vast amount of knowledge and experience in regards to NPD, as they have stories to share that can help us all. This includes working with local media to help share a families story.

I have enjoyed meeting the families via home visits, but where I can and appropriate I try to meet outside the home in local parks, gyms or coffee shops, to help with emotional wellbeing. This has given me the ability to work with individuals, where we then can focus on issues or concerns with common obstacles they face with NPD. Working closely with Elizabeth and Laura, our roles and timings of how we support and make contact with families works extremely well and is a very powerful combination.

I am also making applications on behalf of families with grant giving organisations, providing advice on sources of funding for holidays, again with support of Elizabeth.

Over the next month I am working with John our Communications Officer, assisting a number of our parents create their own platform, via blogs, social media and YouTube to share their journey, raise awareness and help to combat isolation.

I have then been able to work with the team to sign post people to the NPUK Youth Council, and Lads, Dads & Carers groups. These groups now have passionate members of our community involved, who continue to make further connections whilst also giving each other consistent support - this has also helped to develop a culture where the support we offer is more accessible though communications and social media.

What I want to leave you with is this - we want NPUK family members to feel more empowered, but this starts with you reaching out to us in order to grow a supportive environment in which we can all benefit.

With best wishes,

Steve

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07787 818 885

Lads, Dads and Carers

The coming months will see us launch the well-being project for the lads and dads of our NPUK community.

Men of all ages are less likely than women to seek help for all types of well being problems, including our mental well being, physical well being and stressful life events.

The dads and lads Wellbeing Project aims are to:

• Champion the importance of men's health and overall well-being

• Help to reduce stress and anxiety levels among our group

• Help people to better understand issues that affect our mental health and wellbeing associated with NPD

• Work with health, social care and education professionals to ensure we offer the right support for our community

• Encourage us all to seek support for mental health and wellbeing issues

Road trip:

We're coming to YOU!

Steve Neal and Tony Somers will be hitting the road to come and talk to you! We look forward to meeting some of our wonderful Lads, Dads, & Carers, hearing their stories, and talking about the issues that affect them.

The aim is to help improve overall well-being and offer access to services for Lads and Das & Carers of all ages.

We will be using tools and strategies on mindfulness, health and fitness, stress management. and asking you all for your thoughts, ideas and experiences so we can all:

* Have more control over our health and the services that we would like

* Knowing how and when to seek support if you have a health concern

* Having more knowledge and information to be able to manage a health concern or conditions at home

* Knowing what groups and support are available near you!

Remember:

Being a man doesn't mean suffering in silence. Asking for help doesn't make you less of a man. If you would like us to visit you to help and advise you on a personal well-being programme, to share your ideas, or just a cuppa...then get in touch...

Contact Steve by email at: steve@npuk.org, or by phone on 07787 818 885

NPUK Project Team Leader:
Louise Metcalfe

Shaping our Future Together

In 2016, we received a grant of almost £450,000 from the Big Lottery's Reaching Communities fund to allow us to work with more patients and families affected by Niemann-Pick disease (NPD). Time has truly flown and we are now in the fourth year of this five year project.

Looking back at 2018, we have had another busy year with more patients being diagnosed with NPD, more patients and families experiencing symptoms and disease progression and more families needing support from us in a crisis.

Funding from the National Lottery allows us to continue and develop the work of Laura Bell, our Clinical Nurse Specialist, Elizabeth Davenport, our Senior Families Advocate and Steve Neal, our Project Families Officer. You can read more about what they do in their own features in this newsletter. As we cover the whole of the UK and Ireland, our staff have had another very busy year. Between them they have attended 47 Clinic Days or hospital appointments, made 69 home visits, attended 63 meetings in schools, hospitals and places of work. As ever, if you need any help or advice at any time from any of our staff, please do get in touch on the numbers shown.

One of our overall aims in this project is to increase people's knowledge about NPD both amongst our patients and their families, and the wider scientific and medical community. Our staff and Board Members have spoken about NPD at 22 conferences alone this year.

COMMUNITY FUND

We want patients and their families to have the skills and confidence to live with NPD and work with us and their healthcare teams to make sure they receive all the information they need to manage their symptoms, care and treatment. We know that by living with a rare disease such as NPD, patients, and in particular, their family members and carers, have to quickly become experts and advocates on NPD when dealing with professionals and support services as many have never heard of or worked with anyone with this condition previously.

Patients often tell us how frustrating it can be to have to repeat a lot of information about their condition every time they see a different professional and how useful it would be to have all the information about their condition in single document. Taking this on board, we developed patient passports and Medical Alert Cards for our NP-C and ASMD NP-B patients. If you would like a patient passport and/or Medical Alert Card, please contact us at the NPUK Central Office.

Our 25th Annual Family Conference (AFC) was our biggest to date and you can read more about this on page 22. This four day event allows our patients and their families, scientists,

pharmaceutical companies, doctors, nurses, therapists and other professionals from around the world to come together in one place to discuss the latest developments in NPD. It is also a valuable opportunity for families to get together, old and new, in a friendly and supportive atmosphere. As one parent commented, 'It was the only venue where we could meet other families dealing with this condition, which gave so much inspiration and comfort.'

Last year we had a further 11 patients diagnosed with a NPD. New patients and their families usually need a lot of intensive support around this time as they come to terms with a life changing diagnosis, learn more about the disease and the type of practical and emotional support they might need going forward from us at NPUK, their local healthcare teams and within their local community. Living with a rare disease such as NPD can be incredibly isolating and a lonely experience and we we try to bring people together, where possible locally, on social media, at family events, at our Conference or by helping them form their own support networks. One family member said, 'We have been dealt the most horrible card that no one will ever truly know or understand the pain and heartache of, but for such a hard, devastating, rare disease, we are sure lucky to have the support we do.'

Many local services are struggling with dwindling public resources to provide the care and support our families need in their home or at school. This has been reflected in the number of families we have worked with in a crisis situation in the last year. Patients may need support with agencies when they give up work, become carers, are bereaved, move to Universal Credit or apply for DLA or PIP for the first time. The gap between moving to Universal Credit and actual payments has led, in some cases, to families going into debt and arrears.

Please do contact Elizabeth Davenport if you are struggling with any of these issues - we know how easy it can be to be overwhelmed by mountains of paperwork and the negativity of the information you often have to provide.

We are here to help you and want to give you the skills and confidence to navigate the benefits system but appreciate that the land-scape is changing, particularly with PIP and Universal Credit, and people may need more support for some time yet.

If you would like to know more about our Big Lottery project, please do get in touch as we would love to hear from you. Please email Louise at louise@npuk.org or call the Central Office on 0191 415 0693.

Louise

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0191 4150693

BREAKING DOWN BARRIERS

Research Report:

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

Introduction

This Spring Newsletter article provides a brief overview of progress relating to Niemann-Pick therapies and diagnostics. We are not alone in development of the new technologies and examples from other research are very encouraging.

Gene therapy is bringing the possibility of effective treatment to genetic diseases of the eye and the ear although it must be recognised that relatively small numbers of cells are involved in these diseases unlike, NPC1.

Similarly, in the field of diagnostic research the press have recently reported that a woman has the ability to smell Parkinson's disease with which her husband was subsequently diagnosed. It seems that molecules given off by cells undergoing stress can be detected on the skin, presumably in perspiration. This is quite remarkable given that Parkinson's is a disease of the brain.

I report on progress being made in the detection of molecules specific to NPC disease that can be detected in blood and, importantly, dried blood spots taken from all babies at birth. A test to detect these molecules offers an opportunity to administer therapy at the pre-symptomatic stage and to prevent the birth of other children at risk of the disease.

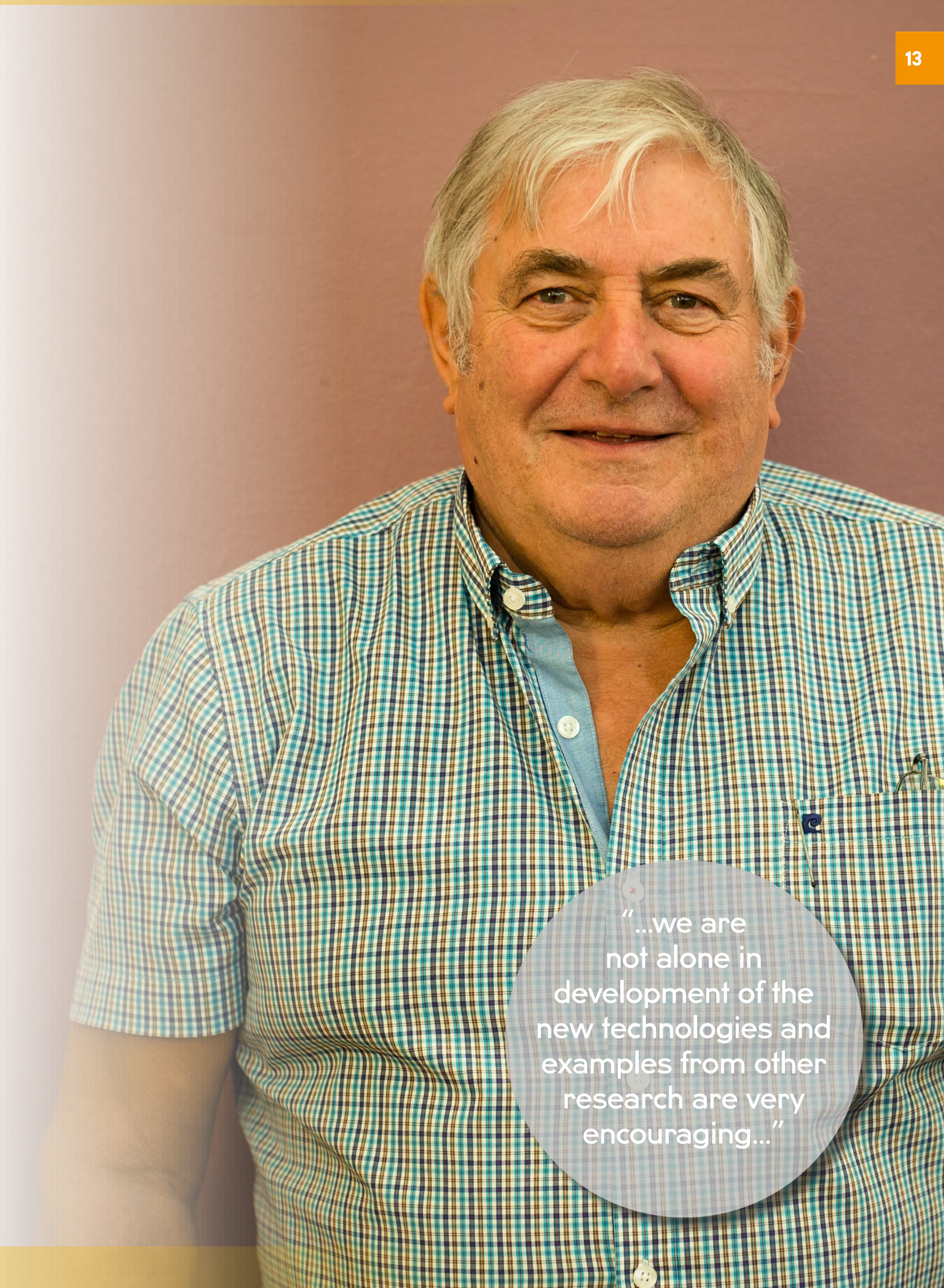
Clinical Trials Progress

The history of clinical trials for Niemann-Pick diseases (NPD) has been fraught with difficulties most of which remain. Although Zavesca (miglustat) has been available for nearly 20 years and approved for use in many countries its use has, at best, resulted in delaying disease progression. The current clinical trials using cyclodextrins and arimocromol are hoped to improve the longer term outcome and realise some degree of symptomatic improvement. Recent announcements from the companies conducting the trials have described mixed results and there is a great deal more analysis to be done before these results are fully understood.

Perhaps more encouragingly, a recent conference sponsored by US charities in conjunction with the FDA entitled "Patient Focussed Drug Development" gave patients and parents an opportunity to comment on their recent experience in participating in the clinical trials. The comments were generally supportive of the trials and described improvements in the various severity domains such as mobility, speech and cognition. How this family perception will correlate with the formal trial results remains to be seen.
It has been clear for some time that early intervention in the treatment of neurological diseases is essential to the achievement of halting the onset of the disease and, the avoidance of loss of normal human functions such as cognition and swallowing.

Currently diagnosis cannot be made until after the onset of neurological symptoms by which time irreversible damage has been done; even then there is often a delay of many years due to the rarity of the disease and variability of presenting symptoms.

It remains important that existing experimental clinical trials continue and that new interventions are developed and it is encouraging to see that some of the


"...we are not alone in development of the new technologies and examples from other research are very encouraging..."

new treatment approaches are aimed at symptomatic improvement which will make the day to day lives of those affected and their families more bearable. There are however, some fundamental obstacles to achieving real success in beating the disease with the main areas being early, pre-symptomatic diagnosis and a better understanding of the cellular and genetic biology in human populations.

Basic/diagnostic science

As a patient organisation we continue in our efforts to support the basic and diagnostic research at universities and hospitals in order that we can help those currently affected by NPD and to identify ways of preventing the diseases such that future families, who may be genetically at risk of having children with NPD, can avoid having affected children. Although we attempt to set research priorities, the situation is not under our control and new information often requires re-evaluation of how we should proceed.

However we have established a number of priorities that appear to stand the test of time. These are:-

• Development of a therapy that is highly effective, safe, tolerable, can be simply administered and is affordable to the NHS.

• Identify how early diagnosis may be accomplished. Ideally prior to the onset of neurological symptoms.

• Improve understanding of the cell biology, the function of the NPC1 protein, the trafficking of each mutant protein, the toxic contribution of each storage metabolite to cell death. Also the point at which cells deteriorate beyond rescue.

The first of these, therapeutic development, is making progress on a wide front with, perhaps, the greatest promise being demonstrated by gene therapy studies. The task should not be underestimated as both central nervous system and all peripheral organs and tissues will need to be transfected with a corrective gene. Total cell numbers are in the billions!

(Left to right) David Priestman shares a moment with Harley-John Mason (NP-C) at the NPUK Annual Family Conference & Interactive Workshop on Niemann-Pick disease(s) 2018

The current situation is that significant progress has been achieved in the UK and other countries on developing this technology but the challenge is now to move from animal models to humans – clinical trials. It may take some time before this happens and we will continue to monitor progress.

It has become clear that the success of any therapy, however promising in laboratory studies, requires administration at the earliest possible stage if it is to be of real benefit. Administration following symptomatic diagnosis will not achieve the best results. New born screening (NBS) is currently a way in which babies with the molecular genetic and metabolic signature of diseases such as NPC can be identified. The problem is that a test is required which must satisfy many stringent measures and be capable of integration into the NHS systems. Conducting tests on a few tens or even hundreds of babies is one thing, but NBS needs to be applied to hundreds of thousands of babies – it is a large scale production issue. NBS cannot take place without a test.

About two years ago clinical researchers in the USA and the UK identified previously undiscovered bile acid products. After undertaking studies using dried blood spots (DBS) from children subsequently diagnosed with NPC and making comparative measurements with NPC carriers and children with normal NPC genes determined that one of the bile acid derivatives would meet the technical criteria for NBS. Unfortunately, progress on the development of this test is stalled due to the unavailability of the study leaders and a technical difference between the USA and UK test results.

In addition to the development of the test, patient groups in the USA working with professional colleagues are planning a NBS pilot scheme for the test in a number of States over the coming two years. This will yield valuable information on the quality of the test and on genetic variation of NPC1 gene in the population. It is unlikely that anything this ambitious will be undertaken in the UK.

It may seem at times that research into the basic workings of the cell is a long way off the provision of a treatment but this is where it all begins. Better understanding of how the many metabolic pathways in the cell function guides investigators to the identification of therapeutic targets and proof of principle studies. Despite considerable funds being brought to bear on NPC1 research, the function of the protein remains unknown. Although the cholesterol metabolism is implicated by many of the NPC1 mutations, this is not the whole story as is demonstrated by the variant class of mutations where cholesterol storage is barely detectable.

We often hope that a new therapy will be a panacea and correct all the problems we see. Unfortunately this is not always the case and although some benefits may be apparent, other problems do not go away. Basic research can assist by identifying symptomatic therapies that may be capable of treating and improving individual symptoms such as walking, swallowing or cognition. We are seeing some of these therapies undergoing clinical trials and if successful, can be used in combination providing a better quality of life.

A further contribution arising from basic science research is the identification of how the NPC defect gives rise to cellular toxicity and what pathways are activated which result in inflammation – activation of the immune system, loss of cellular functions and ultimately, cell death. Intervention in some of these areas can assist in delaying disease progression and prolonging life. The main point is to identify the problem as early as possible. This allows therapies a good chance of working.

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

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

National Genomic Healthcare Strategy

Health Minister Nicola Blackwood's speech at the Rare Disease UK Parliamentary reception to mark Rare Disease Day and launch the National Genomic Healthcare Strategy.

NPUK Communications Officer John Taggart was in attendance to represent our charity alongside other Genetic Alliance UK member groups. For those who missed our original post we share Nicola Blackwood's speech on the next few pages:

"...it is a such a pleasure to mark Rare Disease Day with you all. I want to start by saying thank you to Genetic Alliance, Rare Disease UK and all of you whether you are patients, carers, researchers, clinicians or campaigners – it is down to you that we have come so far in recent years with rare diseases research and care. we all know that.

For too long too many parents had to cope with a sick child with a rare disease but with little information as to what the disease is, let alone where they might find treatment.

"...for too long the treatment of those with a rare disease was been seen as a public policy afterthought rather than as a priority..."

Health Minister Nicola Blackwood giving a speech at the Rare Disease UK Parliamentary reception to launch the National Genomic Healthcare Strategy.

For too long GPs were frustrated by being unsure where the best centre of expertise is to treat a particular rare disease

For too long the treatment of those with a rare disease was been seen as a public policy afterthought rather than as a priority.

And this is why the establishment of the National Rare Disease Policy Board and Forum was a genuine turning point I think. It sent out a strong message to the whole system that information, diagnosis and treatment of those with a rare disease is not only now at the heart of the policy machine but it was also much more strategic and being developed with patients as its guide.

My own experience as an Ehlers Danlos patient has been entirely typical I think and has led me to my own conclusions about rare diseases policy.

I was undiagnosed for 30 years and went through all the usual experiences of the diagnostic odyssey – getting very sick from childhood and being referred to many doctors who each tried their best, ordering more and more complicated and invasive tests but ended up suggesting a psychiatrist or prescribing me something that just made me sicker.

Finally, a wonderful neurologist with experience of EDS realised what had been going on and referred me to a specialist who diagnosed me in 20 minutes. Twenty minutes – after all those years.

Over the next 18 months I acquired a fleet of specialists. Initially it was a disaster. As they started trying to find the right medical regime for me I got much, much sicker and I found trying to co-ordinate all the tests and appointments and new medications – while still working – impossible.

I do not think there is a flat surface in this building I have not collapsed on – including on this very podium where I tried to give speech a couple of years ago.

Then the NHS stepped in and saved me. The occupational therapist here in Parliament, my GP and the whole team at UCL Autonomic Unit literally picked me up and held my hand – helping co-ordinate my care and getting me the support I needed at work. Gradually, the pieces fell into place and I have clawed my way back to stable health. I will not pretend to any of you it was easy. It was not. And there were many setbacks along the way. But I do know I am incredibly lucky.

Firstly, my battles are nothing compared to many of you here today and for that you have my unending admiration.

Secondly, without my family I simply would not have made it. They have sacrificed beyond measure to care for me and I can never repay them.

Thirdly, I owe so many NHS workers – nurses, doctors, my GP, pharmacist, paramedics and more – my stable health today. I am not sure I will ever be able to communicate to them quite how dramatically they have changed my life.

But this process has also taught me indelible lessons about how urgent it is to improve care for rare diseases for everyone – not just the lucky ones like me.

This is why we must never relent in our campaign to bring an end to the diagnostic odyssey – it is pernicious and even after diagnosis the damage it does to mental health of patients and their families must not be forgotten.

RARE DISEASE UK

We must press even harder on clinical awareness and groundbreaking research so more patients can be diagnosed and treated earlier.

Finally, co-ordinating your care can feel almost impossible when you are ill and the complexity of services for rare disease must not act as a barrier to access for care.

That is why the publication of the UK Strategy for Rare Diseases in 2013 represented such a significant achievement for everyone here today.

It put the emphasis firmly on raising awareness, improving diagnosis, and enhancing research and patient care and we have come a long way.

Today DHSC and NHS England have made good on the promise to publish annual updates to the implementation plan.

One year on we can celebrate some incredible milestones – let me just highlight a few.

Care co-ordination:

Firstly, on the issue of care co-ordination I have asked NHS England to implement a rare disease 'insert' from April 2019. This refers to a set of provider criteria to sit alongside NHS England specifications for services treating patients with rare diseases and allows NHS England to hold providers to account for the way in which they treat patients with rare diseases.

There will be up to 3 criteria (depending on the nature of the service): ensure there is a person responsible for co-ordinating the care of any patients with rare diseases give every patient with a rare disease an 'alert card', including information about their condition, treatment regime and contact details for the individual expert involved in their care ensure that every paediatric patient has an active transition to an appropriate adult service, even if that adult service is not the commissioning responsibility of NHS England I hope that this will make a real difference for patients on the ground. I will be keeping a close eye on whether it does.

Research:

Many of you will know that the National Institute for Health Research (NIHR) has established the new BioResource for Translational Research for Common and Rare Diseases.

By March 2018, 37 individual rare diseases had been adopted and by 2022 we expect this number to have increased to 100.

The in-depth phenotyping for rare diseases, linking that to genomic data promises to provide an invaluable research environment for rare diseases discovery.

At Public Health England the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) has made brilliant progress expanding their registries this year.

They have achieved 100% population coverage for conditions diagnosed either antenatally or postnatally, up from 49% in 2017 to 2018.

I truly believe this will improve our understanding of the causes

diagnosis and treatment of rare diseases.

I would like to thank everyone in this room who has played a part in developing these vital research projects.

EU Exit and European Reference Networks:

I know though that many here are concerned about the potential impact of Brexit on our research capabilities. I want to stress the government is a strong supporter of the European Reference Networks (ERNs).

We are keen to agree a future relationship with the EU that includes continued participation in the ERNs. We have made this position clear to the European Commission. In return, the European Commission have shown that they recognise the significant expertise of UK clinicians. Both sides appreciate the importance of maintaining UK participation.

Refresh of the UK Strategy for Rare Diseases:

As we work hard on what our future relationship with the EU will look like, I am also aware that we need to work hard on what the future of the UK Strategy for Rare Diseases post 2020 will look like.

I can reassure you that, post 2020, there will be an overarching framework to improve the lives of all those living with rare conditions. I am sure you will appreciate that it is too early to know exactly what this would look like, but I am personally committed to ensure that the rare disease community are closely involved as we move forward on this.

Genomics:

The really big news, of course, is that in December 2018, the 100,000 Genomes Project completed its sequencing phase – a fantastic achievement by NHS England, Genomics England and other partners including Health Education England.

I know that this project has delivered life-changing results for patients – 1 in 4 participants with rare diseases are receiving a diagnosis for the first time – but it wouldn't have happened at all without the support and participation of many of you here today so I want to personally thank you for the historic role you have played in helping us transform life chances for so many others with rare diseases. Not only that, but this project has paved the way for using genomics in 'everyday' healthcare.

Last year, NHS England launched the Genomics Medicines Service (GMS), making the UK the first in the world to integrate genomic technologies, including whole genome sequencing, into routine clinical care.

As the NHS Long Term Plan in January, seriously ill children who are likely to have a rare genetic disorder, children with cancer, and adults suffering from certain rare conditions or specific cancers, will all be offered whole genome sequencing from 2019 under the new Genomics Medicines Service.

This all speaks to a wider ambition. We want to lead the world in the use of data and technology to prevent illness, not just treat it. We want to diagnose conditions before symptoms occur and we want to deliver personalised treatment, informed not just by our general understanding of disease but by our own personal, de-identified medical data – including our genetic make-up.

Now you will know that the NHS Long Term Plan outlines our vision for the NHS over the next 10 years: the plan focuses on prevention and early detection and has been developed

with frontline staff, patients and their families. The plan also sets out the ambition to focus targeted investment in areas of innovation, particularly genomics. This will enable more comprehensive and precise diagnosis, and allow patients to access more targeted treatments to reduce the use of harmful medications and interventions.

In order to make this a reality, I am delighted to announce that we will be working with the National Genomics Board, people in this room and the broader genomics community to develop a National Genomic Healthcare Strategy. This will tackle not just rare diseases, but it will be built on the foundations that the rare diseases community helped build with the 100,000 Genomes Project.

The National Genomic Healthcare Strategy will set out how the whole genomics community can work together to make the UK the global leader in genomic healthcare.

It is vitally important that this is not just a government exercise: we will be leading a national conversation and I want to encourage everyone with an interest – patient, carer or professional – to share their views and contribute to a coherent, national vision.

Conclusion:

There is so much more I could say but I just want to close with this. I know living with a rare disease or caring for someone for a rare disease can feel relentless. Unseen.

Please do not lose faith. In the National Rare Diseases Policy Board, the Forum and in me as your minister, you have people advocating for you right at the heart of the system.

But we know we cannot do it without you. The mountains we have climbed were only conquered when we worked in true partnership – and there is still so much more to do. That is why Rare Disease Day is all about you..."

Shared with thanks to Rare Disease UK:
gov.uk/government/speeches/we-must-bring-an-end-to-the-diagnostic-odyssey-of-rare-diseases

NATIONAL GENOMIC HEALTHCARE STRATEGY

KEY POINTS:

The strategy sets out how the genomics community can work together to make the UK the global leader in genomic healthcare. It also breaks down how the government will improve services by:

- Ensuring every person with a rare disease will have a dedicated person responsible for co-ordinating their care

- Ensuring that every patient with a rare disease will be given an 'alert card', including information about their condition, treatment regime and contact details for the individual expert involved in their care

- Ensuring every child with a rare condition will be transferred to appropriate adult services when they reach the age of 18, even if that adult service is not the commissioning responsibility of NHS England

HM Government

CLINICAL TRIAL UPDATES:

When Niemann-Pick UK (NPUK) was founded in 1991 we could have only dreamed of having so many different Niemann-Pick disease clinical trials ongoing. We have put together a small breakdown of each trial to keep you up to speed on the latest updates:

CTD Holdings Inc:

CTD Holdings Announces Plan to Launch Clinical Trial of Trappsol® Cyclo™ in Alzheimer's Disease: "Alzheimer's Disease is a devastating illness for patients and their families. We expect to launch a Phase I trial using Trappsol® Cyclo™ as a first step on our pathway to market approval of Trappsol® Cyclo™ for an Alzheimer's Disease indication," said N. Scott Fine, CTD's Chairman and CEO. "We are planning to meet with FDA in the near term to discuss our plans."

Full press release available, here: https://www.npuk.org/ctd-holdings-announces-plan-to-launch-clinical-trial-of-trappsol-cyclo-in-alzheimers-disease/

Mallinckrodt:

Following Mallinckrodt's announcement regarding early phase data from their VTS-270 study on 6th November, a comprehensive list of questions posed by the global NP-C community, was submitted to the company. You will find Mallinckrodt's response to these questions at the following link: https://www.npuk.org/mallinckrodt-responds-to-global-community-questions-on-vts-270-trial/

Orphazyme:

Orphazyme reports positive results from full data set of Phase II/III arimoclomol trial in Niemann-Pick disease Type C (NPC): "Treatment with arimoclomol adjunct to routine clinical care resulted in a 74% reduction in disease progression (p-value =0.0506) as measured by the primary endpoint, 5-domain NPC Clinical Severity Scale (NPC-CSS). In the predefined subgroup of patients of 4 years and older (44 out of 50 randomized patients in the trial), the treatment difference was statistically significant with a minimal disease progression at month 12 in the arimoclomol-treated group (p-value =0.0219)."

Full press release available, here: https://www.npuk.org/orphazyme-reports-positive-results-from-full-data-set-of-phase-ii-iii-arimoclomol-trial-in-niemann-pick-disease-type-c-npc/

Sanofi Genzyme:

Sanofi Genzyme: ASCEND and ASCEND-Peds Studies Complete Enrollment: "Sanofi Genzyme's Global Project Team would like to announce that our pivotal registration study (ASCEND Study) for adult patients with chronic, systemic acid sphingomyelinase deficiency (ASMD) Types B and A/B (or intermediate type) and our pediatric study (ASCEND-Peds) also treating Type B and A/B ASMD pediatric patients have successfully completed enrollment for the targeted number of patients for each of these trials."

Full press release available, here: https://www.npuk.org/sanofi-genzyme-ascend-and-ascend-peds-studies-complete-enrollment/

These clinical trial updates were the most recent at the time of printing, for the latest articles please visit our website at www.npuk.org.

Annual Family Conference & Interactive Workshop on Niemann-Pick disease 2019

This year's Annual Family Conference will yet again be held in the familiar surroundings of Wyboston Lakes, we are so excited to be together again:

This year we will return to Wyboston Lakes, Bedfordshire, over the weekend of 20th-22nd September 2019, to celebrate our 26th Annual Family Conference & 10th Interactive Workshop on Niemann-Pick Disease(s) (more information on this milestone event on page 24). Wyboston Lakes is one of our favourite locations, with more special memories made over the years than we can likely count! It has proven itself time and time again the perfect place for our community to get together and share experiences, updates, and everything in between in a safe welcoming environment.

One of the main things that the NPUK community say they value most about our charity is simply getting the opportunity to meet up with and be with one another, which is why our Annual Family Conference is something that we all look forward to every single year! It's a compact yet emotionally charged couple of days which always leaves us wishing we could have longer. Yes there is Facebook, emails, and telephone calls should we need it - but nothing beats having NPUK Community members together physically under one roof.

Toni Mathieson, NPUK Chief Executive, has been working hard as always to ensure that this year's programme will once again offer you the opportunity to hear the latest developments regarding therapies and clinical trials for ASMD Niemann-Pick disease types A and B, and Niemann-Pick type C, plus related care issues, breakout sessions, and workshops. Expert professionals and NPD family speakers from around the world will provide interactive presentations focusing on the practical management of Niemann-Pick diseases, clinical trials, and potential therapies, plus much more.
More specifically, we are happy to announce that individual workshops will also be on offer to the NPUK Community, such as smaller group meetings for Lads, Dads and Carers, Reflections, and the NPUK Youth Council - to name but a few!

There will also be an extensive Children and Young Person's Activity Programme, including day-trips, and fun activities

so that parents and carers can watch professional presentations and take part in workshops safe in the knowledge that their children are being taken care of by our dedicated NPUK volunteer team. After all, our Conference is all about inclusivity - it's not just for the professionals, parents, or children...it's for everyone!

There's lot more in store but we wouldn't want to spoil the suprise - safe to say that this will be the event of the year and you certainly won't want to miss out!
If you wish to attend this year's Annual Family Conference & Interactive Workshop on Niemann-Pick Disease(s) then we would love to have you!
Please get in touch as soon as possible as places are limited and tend to fill up quickly.

Pre-register by contacting info@npuk.org. Booking forms will be available soon. For furtherinformation please visit the NPUK website at www.npuk.org or contact at the NPUK Central Office by phone on: 0191 415 06 93.

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

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

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

10th Interactive Workshop

This year's Interactive Workshop marks the 10th year of this important event, which brings together health and research professionals working in the field of Niemann-Pick disease

The Interactive Workshop, which this year tales place on Friday 20th September, provides an opportunity for both UK and International professionals to share current information regarding all aspects of Niemann-Pick diseases, including clinical management, research and potential therapies. Hosted by Professor Fran Platt, University of Oxford, the meeting will feature clinical and scientific presentations and workshops on relevant topics, including the latest research and clinical updates for ASMD Niemann-Pick disease and Niemann-Pick disease type C.

This year's programme will once again offer the opportunity to hear the latest developments from around the world in the areas of basic science, clinical trials and therapeutic options for this group of diseases, plus much more, with time for discussion and questions with each of our expert speakers both during and after their presentations. The event will be followed by the NPUK Annual Family Conference, which is the largest gathering of families affected by Niemann-Pick disease in the UK.

As well as highlighting the most recent advances in research and clinical care, these events provide an ideal opportunity to strengthen our network by encouraging families and professionals to meet and to share their thoughts and experiences.

Professionals and families alike tell us how much they value these events and the opportunities they provide, which often lead to new friendships, research collaboartions and much more. Dr. Emyr Lloyd-Evans (Senior Lecturer at Cardiff University) explained to us last year how important these events are when it comes to shaping research and working collaboratively towards effective therapies for NPD:

"...without the interactions with the families and patients at these meetings, you don't perhaps see these little aspects of the disease because they're not in papers that we read - you have to be able to interact with the families and the patients to pick up on these things...and that's impacted directly on our research..."

We look forward to welcoming delegates to Wyboston Lakes once again and to celebrating ten years of interaction and collaboration with yet another informative and successful event. To register your interest, please contact info@npuk.org.

YOUTH Council

The NPUK Youth Council (NPYC) provides an opportunity for young people to feed into the work of NPUK, so that they can have a direct impact on the future of our charity:

At our Annual Family Conference last year we held a workshop to discuss the Youth Council and enable young people to tell us what they would like this initiative to look like. Their feedback was instrumental in the creation of the group and will also be vital going forward.

We recognise the value of the experiences, views, and ideas of the children and young people in our community - we know that their opinions are very important and can really make a difference! With this in mind we have decided to offer the chance to Youth Council members to meet with the NPUK Board of Trustees at least once a year to talk about their activities, and issues, as well as to provide updates on the progress of the NPYC. We trust that this will help to cultivate increased self-confidence, purpose, and skills going forward.

After a successful meeting between NPUK Project Families Officer Steve Neal and volunteers assisting with the NPYC we have produced an 'Introduction Leaflet' which will be shared with all of those with interest in joining the NPUK Youth Council. For those already part of the group keep your eyes peeled for your own copy in the post soon, for those considering joining in please get in touch with Steve at: steve@npuk.org and we can organise delivery.

The NPYC will:

- ensure that the voices of our young people are heard and that their views, concerns, and ideas are listened to

- provide an opportunity to raise issues that are important to the children and young people within the NPUK community

- share the views of young people with professionals (e.g. scientists, doctors etc.) to highlight the needs and experiences of those affected by Niemann-Pick diseases

- support the young people of our community to increase their skills, knowledge and confidence

Shona Beveridge:

"my job as a youth trainee"

In the following article, NPUK Volunteer and Community Member Shona Beveridge shares the story of her new job and how it's going so far!

I have been doing my SVQ3 (The Scottish Vocational Qualification 3 in Health and Social Care - Children and Young People), in my spare time and also during my hours at the office.

Kerry is my Assessor for my SVQ, I am doing this qualification so I can apply to be a "Childhood Practitioner" which is a role my boss thinks is achievable if I can get some of my units. I am a Youth Trainee for 12 months , so hopefully I should be able to achieve some units.

However on Mondays and Thursdays I am doing a placement as a youth trainee under Drew McNeil's Youth Trainee Scheme.

This is in a Local Primary school, (Cradlehall Primary) where I will help in the Nursery and Primary 1, otherwise known as "reception", as you English people might know!

In the meantime I am on a trial for my physical condition Niemann-Pick disease type C, I currently go to Birmingham every fortnight as the trial I'm on is only run at two centres in the UK: Great Ormond Street Hospital in London and Birmingham Children's Hospital.

It's a long way to go for treatment, but now I have moved to the adult services, my adult neurologist who runs lots of other trials, is interested in running the trial I'm on at my local hospital, but that could take a while to set up.

Thanks Shona we look forward to your next update, keep up the fantastic work!

Shona (middle) shares a fun moment with her sister Kelsie, father Callum and NPUK Finance & Administration Officer Christine Jopling at last year's Conference

Picnic in the Park

Pack a picnic, bring a rug and come and join NPUK staff, families and friends at Craigtoun Country Park near St Andrew's in Fife, Scotland on Saturday 22nd of June 2019.

There's plenty of things to do in this 47 acre park for visitors of all ages including an adventure play area, trampolines, a boating lake and crazy golf to mention just a few. We've hired the Picnic Hut to use as a base to meet, explore the park from and to eat our lunch in.

It's a great opportunity to make some new friends and to meet other families. For more information and to let us know if you are coming, please ring us at the NPUK office on 0191 415 0693 or email us at info@npuk.org.

See you in Scotland!

We have limited spaces available for the National Rotary KidsOut Day on Wednesday 12th June, 10am-4pm at Drayton Manor Theme Park. The day promises to be a fun get-together of the NPUK Community, volunteers, and members of the NPUK Staff Team, so if you are interested please get in touch with us as soon as possible by email at: info@npuk.org or by phone at: 0191 415 06 93.

Fundraising FOCUS

In the following pages we shout out to all of our fantastic fundraisers who have set out to raise awareness of Niemann-Pick disease and rally donations for NPUK in the process. We rely entirely on grants and voluntary donations to continue our support for those affected by Niemann-Pick disease...so these people are truly heroes in our eyes!

Rebekah Cayzer - Wigan 5K: Rebekah has taken part in sponsored walks in support of NPUK in the past, and stepped up her game (no pun intended!) to challenge herself further...this time with the Wigan 5K.

Before the event Rebekah wrote: "...I am doing the Wigan 5K for Niemann-Pick UK. They help to support my cousin Matthew, his family and other children with this condition and their families!..."

Georgina's Gang - Hint of Pink 10th Anniversary Ball: Looks like it was a fun night at Georginas Gang's Hint of Pink 10th Anniversary Ball! Not only did it get people together for a night of fun for a fantastic cause, there was also a cheque presentation to NPUK for a staggering £5,000. Jackie Imrie, NPUK Trustee was in attendance and by the looks of the photo to the left got right into the spirit of the evening!

Georgina's Gang was set up by family and friends of Georgina Pearce (NP-C, 6/11/96 - 31/10/16) with the original aim of supporting Georgina and her family, who live in the Bailiwick of Guernsey, and also other families in their situation. They have supported NPUK for many years and we are very appreciative of all of their amazing efforts.

Paula Perkins serving another satisfied customer at the crafts and tombola tables at one of our Annual Family Conferences

Rachel Geere - Winchester 10K Road Race: Back in February Rachel Geere, as part of the fundraising super team Harvey and the Brave Little Soldiers, took part in the Winchester 10K Road Race in support of NPUK. When speaking of her motivation Rachel wrote:

"...my main motivation is [to run] in aid of a fantastic cause in support of Harvey and the Brave Little Soldiers who raise funds for Niemann-Pick UK, a charity very close to my family's heart as my sister-in-law's nephew was diagnosed with NP-C a couple of years ago.

To help fund the research required to cure this horrible disease, and to support the children and families, any donation no matter how big or small is greatly appreciated..."

Jade McCann - Trbute to Tj: This year in a beautiful tribute to her brother Tj (11/8/2015-17/1/2018, NP-C) who sadly passed away last year, Jade decided to ask for donations to NPUK for her birthday in lieu of gifts.

Jade commented: "...for my birthday this year, I'm asking for donations to Niemann-Pick UK. I've chosen this nonprofit because not only does it mean the world to me but I'm doing it for my brother: RIP Tj Love you..."

Donations on Facebook have been a transformative feature for us here at NPUK, in less than a year YOUR help has raised over £8,000.

We would like to say a huge thank you to Deb Callaghan, owner of Essential Beauty, for choosing Niemann-Pick UK as their chosen charity 2019!

Our Chief Executive, Toni Mathieson is a regular client of Essential Beauty and through her conversations with Deb has shared her story of living with NP-C. Deb has generously chosen to support NPUK throughout 2019, with members of her esteemed 'Essential Beauty Club' providing a donation of £2.50 made direct to NPUK. Over the course of the year this will undoubtedly build up, helping to raise funds and also further awareness of Niemann-Pick disease to the wider community! Huge thanks go to Deb for her friendship and understanding, and to her lovely clients for their amazing support! For further information on both Essential Beauty, please visit: www.essentialbeautysalon.net

Gillian Hancox and Morag Hughes - Rebel Swimmers: The entire NPUK Community were taken aback by the inspirational efforts of the two "Rebel Swimmers", Gillian Hancox and Morag Hughes who took on the epic Polar Bear Winter Challenge in support of Niemann-Pick UK. Before the event, Gillian wrote:

"...we love to swim in open water so we decided to join The Polar Bear Winter Challenge 2018/19. People from all over the World are involved including Canada, Norway and the Shetland Isles. We must only wear swimsuits, goggles and thin silicone swimming caps. NO wet suits, gloves or shoes. So far, so good, we are both exceeding the challenge limits, especially Morag! We are raising money for research into Niemann-Pick disease which is an inherited disease affecting all ages. We have a friend who has three daughters, two of whom have the disease and the other is a carrier..."

The Rebel Swimmers smashed the event, and raised hundreds of pounds for NPUK - this just goes to show that our fundraisers are some of the best (and bravest) out there! Well done ladies!

Erith Contractors: NPUK Trustee David Roberts joined Erith Contractors' Managing Director David Darsey for a cheque presentation earlier this year, and was pleased to be able to thank them in person for their continued support:

"...I would like to express my sincere gratitude for arranging the presentation of this generous donation on behalf of the Institute of Demolition Engineers. The money raised will help us to provide the much needed care and support service for our families and to facilitate research in to this devastating disease..."

Terry Colwell - Harwoods Jaguar Land Rover Basingstoke: Late last year NPUK Community fave Terry Colwell and his fun-loving co-workers at Harwoods Jaguar Rover Basingstoke helped to provide some Christmas cheer in support of Niemann-Pick UK!

Terry, who is a Sales Executive at the company decided to nominate NPUK for their annual Christmas jumper day - a promotion in which employees are invited to wear a fun Chrimbo jumper in place of their usual attire and are invited to make a donation on that day.

We're pleased to announce that every single department got involved, as they know how much of a positive impact donations can make to individuals and families affected by Niemann-Pick disease.

Great effort everyone - thanks so much for thinking of us!

donr

After the 31st of March our current facility (provided by Vodafone) will unfortunately be unavailable, but don't worry our new provider Donr has us covered!

Supporters of NPUK can become regular givers using just their mobile number in less than 15 seconds - no more obstacles!

Our tech allows supporters to become regular donors in just 15 seconds, leading to increased conversion rates. Donr is integrated into the major four UK mobile network operators (EE, Three, Vodafone, and O2) which allows NPUK to utilise innovative carrier billing technology.

As a supporter of our charity you can donate to NPUK directly by texting 'NPUK' plus your text amount (e.g. for a donation of £10 text 'NPUK10') to 70085.

Remember that we rely on voluntary grants and donations, such as those received through text giving services, to continue to support the individuals and families of our community affected by Niemann-Pick disease.

For more details on Donr and for further information on making a donation to NPUK please contact John, our Communications Officer by email at: john@npuk.org or by phone on: 07984 366 334.

David Key - London Marathon 2019:

David Key took on the massive challenge of the London Marathon this year, inspired to take part by his son Alexander (age 8, NP-C) in a bid to raise funds for both Genetic Disorders UK and Niemann-Pick UK, and also to raise the broader awareness of Niemann-Pick disease.

No doubt the training has been hard, especially during the frosty winter months, but thankfully David found some inner motivation as he mentiones here: "...I always said that I would never run a marathon, not least because I knew I probably wouldn't be able to! But then I saw some of the Invictus Games this year, got a bucket load of inspiration from those incredible athletes, and applied. Training is going well so far [---] it will all be worth it if I can raise awareness of these devastating diseases and help raise some money too..."

Amazing effort David, we're always in awe of marathon runners!

Andrew Hall - Harvey and the Brave Little Soldiers: Ho ho ho! Well look who it is, Harvey and the Brave Little Soldiers have done it again - this time by getting Santa Claus on board to do a bit of fundraising on behalfof NPUK! Harvey's grandad Andrew Hall didn't think twice about slipping into the Santa suit last year for a good cause - drumming up support on the streets of Birmingham. We hope that thisfeature is an annual occurance...you certainly suit the beard Andrew!

Team Roman - Ramble: After seeing Josh Cullip (NP-C) on Countryfile with Matt Baker, Team Roman decided to take part in a ramble around a local lake (College Lake in Tring) to support him. They also took on the challenge in memory of Roman - who is the guiding force behind all of Team Roman's fundraisers and awareness campaigns. The group raised an amazing £200 in the process - prompting Roman's mum Scarlet to comment:

"...after the ramble on Sunday, I dont know about you guys but my legs ache a lot today... but my heart is so full. I just want to take a minute to say a massive THANK YOU to Linda Janes for everything she does for NPUK and Keech Hospice! We are very lucky to have you in our lives and love your continued involvement in something that means so much to us! Roman is looking down with such pride at all you do! Also, thank you to the members of Team Roman for your continued support - it means the world! Without you all, Team Roman would not be possible! We value you more than you know..."

In Memory of Calum Burdon

Masquerade Black Tie Ball

Saturday 5th October 2019

Ribby Hall, Wrea Green

Woodlands Suite

Live Entertainment - Auction/Raffle

4 Course Meal £50.00 per person

Table of 10 £500.00

Arrive 18:15/Carriages 01:00

Brian House Children's Hospice

Charity Fundraiser for two great Charities

np uk

For Booking Enquiries Contact:

Carl: 07966309865 Lianne: 07954651205 rcbcharityball@yahoo.com

Pete Clarke & family - Meat Raffle:

Thank you to Pete Clarke and family, as well as the fantastic people at the Angel Vaults pub in Evesham for their brilliant fundraising efforts. As a result of your Christmas meat raffle you raised a staggering £1,019.

We love to see local communities such as this getting together to make a positive difference - much appreciated!

Thornden School: Many thanks to the year 9 pupils of Thornden School in Chandlers Ford, Eastleigh who clubbed together to raise funds for NPUK, inspired by the efforts of the television appearances of those within our community such as Joshua Cullip and Shona Beveridge.

Cheswick Green School: The thoughtful students of Cheswick Green School decided to do all they could in support of NPUK by putting on a Christmas fundraiser motivated by fundraising super-team Harvey and the Brave Little Soldiers.

The children raised a grand total of £375 - excellent work!

ONLINE DONATION PLATFORMS:

Unfortunately the BT MyDonate website will close on 30 June, its owner BT has announced, which will mean NPUK fundraisers who currently use the platform will have to look for an alternative.

When it was launched by BT in 2011, MyDonate was the first commission-free fund-raising service. However, fundraising technology has moved on significantly and many other fundraising platforms were now available for people and charities to use, several of which were also commission free.

Don't worry, you can still use Virgin Money Giving or Just Giving, which are incredibly user-friendly and offer a great service. For further informationplease get in touch with NPUK Communications Officer John, at: john@npuk.org.

Virgin money

JustGiving™

HOW TO PLAN YOUR NEXT NPUK FUND-RAISER:

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

1 **Choose something fun:** The only limit to fundraisers is your imagination. There are so many events to choose from...check out our fundraising pages (pages 28-34 for inspiration!)

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

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

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

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

Shona Beveridge and Matt Baker share a smile together during the BBC Children in Need Rickshaw Challenge 2017.

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

NPUK Awareness Films

This year we have launched a new campaign featuring the inspirational stories of the amazing people that make up the NPUK Community.

Released primarily on Facebook, but often shared on other social media platforms, these short awareness films seek to inform those outside of our community of Niemann-Pick disease and the effect that it can have on the lives of those individuals and families who deal with it on a daily basis. With this in mind we chose to strip back the large majority of the scientific language which can often confuse or perhaps even alienate those without a close connection to the condition, and instead focused on the personal story which surrounds a Niemann-Pick diagnosis.

We are pleased to say that we have had an incredibly positive response to these videos so far, enjoying a far greater reach than any previous posts. We believe that this shows the power that we have in each and every one of our stories - and also that the NPUK community, as it grows in number, is a force to be reckoned with!

We want to say a massive thank you to everyone who has taken part in our campaign so far - the stories have resonated not just with our community but to so many outside of it across the globe.

If you would like to take part in our NPUK Awareness Films campaign, then please get in touch with John our Communications Officer by email at: john@npuk.org.

Our first video featured **Joshua Cullip** (age 12, NP-C) and detailed his amazing journey on Countryfile alongside The One Show's Matt Baker.

Alfie Burns' (age 10, ASMD NP-B) story clearly touched both the NPUK community and those outside of it - it currently has over 17k views and counting!

We were excited to share **Shona Beveridge**'s (age 18, NP-C) story with those outside of our community - she continues to inspire with all that she does!

Our latest video featured Amarii (age 4, NP-C) who despite his Niemann-Pick diagnosis has a love for life and a smile on his face! His story has already reached over 10k people.

Alfie Burns
MK Dons

Our very own NPUK superstar Alfie Burns took centre stage at MK Dons' Stadium MK in March:

As mentioned on the previous page of this issue of NPUK News, Alfie Burns (ASMD NP-B) was one of the first of our community to take part in our new social media awareness videos - accruing over 17,000 views in just a couple of months, an unprecedented figure making his film our most successful on Facebook to date!

Clearly Alfie's story, one of defiance and bravery in the face of such a diagnosis is one that resonates with both the NPUK community and those outside of it alike. But this wasn't to be the end of Alfie's impact this year - as he was treated to a birthday to remember for a lifetime!

Pictured to the right alongside MK Dons captain and club hero Dean Lewington, Alfie was first out on the pitch as a mascot for the Crawley Town home match in which MK Dons won 1-0! It likely goes without saying that every one in the NPUK Community was a fan for the day!

The good people at MK Dons were also kind enough to use the power of social media to spread the word of Alfie's journey with ASMD NP-B and the effect that it has on both himself and his family. By sharing our awareness video on their Facebook page (which currently has well over 40,000 followers) they were able to achieve over 3,000 independent views to date (and counting!)

Massive thank you to everyone at MK Dons for setting up such a fantastic day for Alfie and his family, also we want to show special appreciation to Antoni Fruncillo, Media Manager, for his help every step of the way - which includes providing this legendary image below capturing the proud moment in all of its glory!

Go Make Memories

We are currently working on a new short-film which we hope will raise further awareness of Niemann-Pick disease and shine the light on the daily lives of the NPUK Community.

At NPUK one of our main focuses is to continue to increase the awareness of Niemann-Pick disease and the effect that it has on the individuals and families affected by the condition. Such a diagnosis can often lead to feelings of isolation, which can have a negative ripple effect for the whole family - not just the individual directly affected. It is our hope that through this upcoming short film, we can help showcase this to those within the community and outside of it.

Recently we met with a group of NPUK families to discuss their experiences with NPD, and since then our Communications Officer John and Director/Producer Carl Mason (IMAGINE, 2015) have been working closely with a screenwriter to develop a screenplay which we feel reflects the shared experience many of our NPUK community members have been through.

One of the main things that we discovered through the aforementioned focus group and conversations with other members of the community were that the feelings surrounding initial diagnosis can range from that of shock and disbelief to anger and doubt. Many families shared stories which ended with the doctor or health professional telling them that they should "go make memories" with their child, a possibly well-meaning, but nonetheless harsh and brutal way to deliver such news. It wasn't until these individuals found out that they weren't in fact alone, thanks to support networks such as NPUK and more importantly the wide community of people also faced with a Niemann-Pick diagnosis, that they

began to feel more able to cope with the unknown road ahead.

The film itself will be different to other awareness shorts you may have watched in the past as rather than using simply live action to tell a linear narrative, we have decided to instead use a mixture of both live action and animation so that we can better display the progression of the condition as it deteriorates, and the subsequent effect this has on the family.

Carl Mason, commented: "...the animation art style will be 'hand-drawn', rough, kinetic and textural; focusing on 'anonymous' character design with no lip sync, to allow for the animation, music and sound design to all be in production simultaneously.

It is our hope that Go Make Memories will impress the judges on the film festival circuit both this year and the next so that it can reach a wider international audience, leading to further awareness of NPUK, our community and the impact of Niemann-Pick disease.

We will be showcasing the film in full at our upcoming Annual Family Conference & Interactive Workshop on Niemann-Pick Disease(s) on the 20th-22nd September 2019.

We are excited to share more about thisproject as and when it develops further - you can keep up to date with the latest newssurrounding Go Make Memories at the Niemann-Pick UK Facebook page. Many thanks to The Hollie Foundation and all families who have helped us with this project to date - you have been invaluable!

INTERESTED IN BEING INVOLVED WITH FUTURE NPUK VIDEO PROJECTS?

If you would like to take part in future video projects, whether that be short video interviews, vlogs, short films or anything in between, then we would love to hear from you! After all it is the stories of the NPUK Community - those individuals and families affected by Niemann-Pick disease, that helps us to push for further awareness amongst the general public.

We are planning to capture more video content in the coming months, in particular at the upcoming Annual Family Conference & Interactive Workshop. So if you have a story worth sharing and want us to help you tell it, then please get in touch with John, our Communications Officer, by email at: john@npuk.org or by phone on: 07984 366 334, and we can make that a reality!

INPDA

GLOBAL COLLABORATION

The International Niemann-Pick Disease Alliance (INPDA), is our network of NPD support groups across the globe, of which NPUK is a founding member.

We are looking forward to the biggest INPDA event, the **Biennial INPDA "Face to Face" meeting**, which will be hosted by the Vaincre Les Maladies Lysosomales (VML) in beautiful Lyon, France on the 14th-16th June.

The meeting is a very special moment in every INPDA member group's calendar as it is a unique opportunity to come together alongside expert health professionals and industry representatives working in the Niemann-Pick disease field.

For further information on the event please follow the INPDA Facebook page or visit the INPDA website: inpda.org.

International Niemann-Pick Disease Registry (INPDR):

The International Niemann-Pick Disease Registry (INPDR), is a single, disease-specific registry created for the global Niemann-Pick community. A registry is essentially a collection of data that can greatly help to increase knowledge and under-standing about a condition – now widely seen as essential, especially in the rare disease field.

The INPDR is a not-for-profit registered company based in the UK, with an independent Board of Trus-tees and Scientific Advisory Committee. It was created by NPD patient groups and experts from across the world, with the aim of improving standards of diagnosis, care and treatment for patients every-where. It includes both clinical reported data and patient reported data for Acid Sphingomyelinase de-ficient (ASMD) Niemann-Pick disease, and Niemann-Pick disease type C.

Currently six European countries are entering clinical data and global roll-out is underway. Patient re-ported data has been entered in 5 countries; this allows

patients to self-enrol in the INPDR and con-tribute to progress, independent of clinician input – an important option in countries where clinical da-ta entry is yet to go live.

Continued development will provide a sustainable patient-led database of at least 1000 patients. A brand new front-facing website is in development and will be launched soon. We would like to en-courage patients, families and clinicians to register their interest and get involved with this valuable initiative, which will do much to support the collection of natural history data and facilitate research and therapy development.

For further information about the INPDR please visit: inpdr.org, you can also keep up to date with the latest news via the INPDR Facebook page.

The Ara Parseghian Medical Research Fund: Niemann-Pick Type C (NPC) externally-led Patient-Focused Drug Development (PFDD) meeting

The PFDD meeting is a groundbreaking initiative driven by those affected by Niemann-Pick disease and their advocates that culminated in a structured meeting to provide the FDA, drug developers, and NPC stakeholders with perspectives from around the globe. The meeting took place on March 18th in Washington DC, USA, with NPUK Chief Executive Toni Mathieson in attendance representing our charity and the community we support.

With permission from Hope for Marian, we decided to share the following piece written by Kayla Miller which we will feel perfectly sums up the impact of NP-C but also the unity that was felt at the meeting.

"NPC shows no mercy. It will turn speech into muffled noises. It will steal joy from laughter as you fall from cataplexy. It will take your hearing and make the sound of birds become a distant memory. It will take a one year old who is potty trained and cause her incontence by the age of five. By seven you'll need assistance...braces, walker, or a wheelchair.

NPC will make your speech understandable at times and at others you'll stammer and point in frustration or rub your belly and say "yum" as you try to express your needs. It will turn your favorite gummies and juice into a choking hazard.

It will bring more pneumonias than most see in a lifetime and intensify the flu into a terrifying stay in the pulmonary intensive care unit. You will enjoy art by drawing lines, using colors and making dot dot smileys with your aid, Miss Amanda.

It will cause you to ask others if they remember things, because you're unsure yourself and have been prompted to remember too many times. And though you once knew your shapes, colors, numbers, and letters, you'll eventually hold on to spelling your name - K-A-M.

NPC will take away your ability to remember your transfer school bus friends, who you've come to know for three years. On your best days, you won't be able to remember most of your friends names, and on your worst days, NPC will keep you from remembering the names of family and close friends. It will take more than just quantity of life, eventually it will take the quality, too.

As a parent, it will cause you to view time differently and it will make you face your biggest fears of losing your child's personality and quality of life along the way."

You can hear Kamryn's story from her mom Kayla in her own powerful words at 4:33:41 and our own at 3:18: https://youtu.be/iHcDkdrUk3E

Thank you to every NPC family who sat shoulder to shoulder and shared our shared hope, pain, and voice with the FDA. We have no approved treatments, no cure, but we have a united voice for a path forward together.

Thank you to Kayla Miller (mother to Kamryn, 7, NP-C) for these powerful words.

What is the Ara Parseghian Medical Fund?

The Ara Parseghian Medical Research Fund at Notre Dame is a non-profit organization dedicated to funding medical research projects to find a treatment for Niemann-Pick Type C (NP-C).

The Parseghian Fund is named in honor of Ara Parseghian, the much beloved and well-known Notre Dame Football Coach, whose three youngest grandchildren were diagnosed with NP-C in 1994.

ARA PARSEGHIAN
MEDICAL RESEARCH
FOUNDATION
A GOAL FOR LIFE

study opportunity

HELP US UNDERSTAND NIEMANN-PICK

A market research study is being conducted to better understand the patient and caregiver experiences living with Acid Sphingomyelinase Deficiency (ASMD) / Niemann-Pick disease.

If you have been diagnosed with ASMD / Niemann-Pick Type B or Intermediate Type A/B, you may qualify to participate in an interview and online survey about your experiences.

Interested?

Contact Jon Carroll for more information and to see if you qualify.

Email: jon.carroll@kqhealth.com
Phone: +44 (0)7816 983 812

This Rare Disease Day the International Niemann-Pick Disease Alliance (INPDA) of which NPUK is a founding member, decided to do something a little different...

Every year on Rare Disease Day the spotlight is placed firmly on all rare diseases, giving us the opportunity to come together to raise collective awareness and look forward with renewed hope for effective therapies and treatments. The INPDA (International Niemann-Pick Disease Alliance), a network of 18 NPD patient groups from 15 countries across the world, is committed to raising awareness every day - not just on Rare Disease Day.

We are not just faceless organisations - our members support and represent hundreds of inspirational people! So we wanted to use this annual opportunity to share with the world the 'Global Faces of Niemann-Pick', a collaborative project which proudly showcases the individuals who give us

hope in spite of the harsh realities of this devastating group of diseases.

"We are stronger together" - a phrase we have heard over and over again, but that's because it's true, and it is the spirit of these words which brought the International Niemann-Pick Disease Alliance (INPDA) to life in the first place! We are here to support one another and it is in fact our shared experiences which bring us together, and makes for such a welcoming atmosphere, one which transcends countries and cultures.

It's fantastic to have such a visual representation of this through the project, we look forward to more inclusive and collaborative efforts in the future...watch this space!

Thank you to everyone who got involved and shared your pictures with us, you can view the graphic on the INPDA Facebook page - for your own digital copy please get in touch with INPDA Communications Officer John Taggart at john.taggart@inpda.org.

Help & INFO

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

Stress is the feeling of being under too much mental or emotional pressure. It's when we think a task is too big for our resources. Sometimes we are so stressed we forget what not being stressed feels like.

Short-term stress is not a bad thing, but long-term it can be harmful. It's important to work out how stress affects you. The first step of trying to reduce stress is recognising when you're getting stressed.

TIPS TO REDUCE STRESS

Plan your time; trying to cram in too much will end up making you feel stressed when you do not achieve everything.

Reduce your to-do list - try delegating tasks or, if they are really not essential, cross them off your list altogether!

Just say no - know your limits and stick to them.

Avoid people who stress you out. If you know a particular person touches all your hot buttons, limit the amount of time you spend with them.

Take control of your environment. If trips to the shops are really difficult, can you shop online instead?

Be more assertive. If someone calls and you really do only have five minutes, tell them and be firm when the time is up.

Be positive. If something goes wrong, try to find a more positive way to view it. Try to focus on the five things that worked today, not the one that didn't.

Take notice of the world around you and what you are feeling. Reflecting on the experiences of the day will help you appreciate what matters to you.

Learn how to let go of anger and resentment and forgive others.

To read the rest of this article on stress and tips on stress management please visit: www.contact.org.uk/advice-and-support/your-child-your-family/family-life/coping-with-stress/

Universal Credit - *explained*

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

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

Managing Debt:

Living with a Disability

In the following article we explain how to deal with debt if you have a disability and show you where to get help if you need it.

It can be easy to push your debt concerns to one side, but as with anyone, ignoring the issue will only worsen the situation.

Although having a disability can make it particularly hard to tackle uncontrolled debts, especially if you have a low income, there are steps you can take to help address the problem. If you are finding it hard to pay back your debts, the following may help:

Get in touch with your lenders:

It is important that you contact your lender/s as soon as you get into difficulty.

They often help by renegotiating your borrowing and could reduce the money you need to pay back each month to a more achievable level. This may mean paying more in interest overall, but it can help make debts more manageable.

Determine your budget:

Total up your weekly or monthly earnings, as well as any benefits you receive, along with everything you spend your money on during this period. If the money coming in is less than your outgoings, identify non-essential areas where you can cut back your spending.

Determine how much you spend on your accommodation, utility bills, food and anything that relates to your disability; pay for these first and then see how much money remains for other items.

Avoid further debt:

While further borrowing may seem attractive when you are not making ends meet, avoid taking out more money as it will only perpetuate your problems with debt.

The only exception is if you can consolidate your debts to make them cheaper (via a balance transfer for instance).

Ask for help:

Here at NPUK our Care & Support team can support you in speaking with an impartial debt adviser, who will help to create a plan to clear your debts which suits your individual needs. Contact us by email at: info@npuk.org for further details.

Article shared with thanks to money.co.uk

NEED ADDITIONAL SUPPORT?

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

For a list of organisations which may be able to help, please visit: www.cashfloat.co.uk/blog/money-borrowing/top-debt-charities-uk/

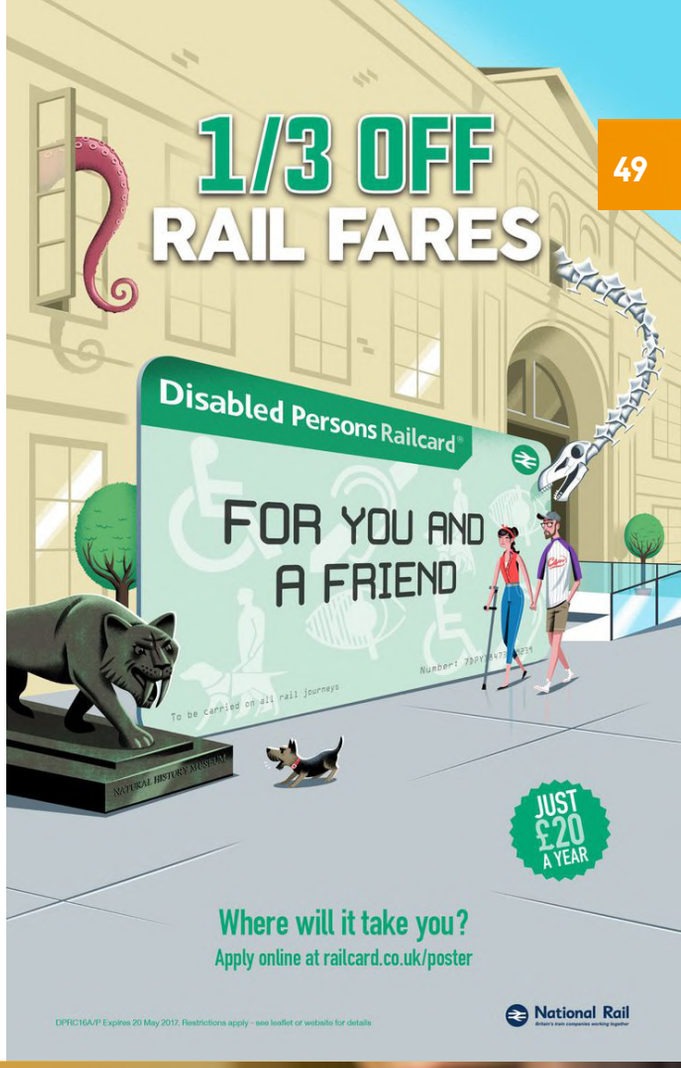
Disabled Persons Railcard

Get 1/3 off adult rail fares for travel on the National Rail network in Great Britain - time to get planning some extra-special summer day-trips:

If you're travelling with another adult they will also get 1/3 off their rail fare, so you can save money for a friend or family member too! There are no time restrictions on the Disabled Persons Railcard, so you can use it to get a discount on tickets at any time of the day.

- One year Disabled Persons Railcard: £20
- Three year Disabled Persons Railcard: £54

For further information and to find out if you qualify please visit: disabledpersons-railcard.co.uk



Did you know that NPUK have some iPads available for adult patients and their families?

COMMUNITY FUND

As part of our National Lottery project "Shaping Our Future Together" NPUK has a limited amount of iPads for us by adult NPD patients and their families. We are interested in seeing how people with a rare disease like Niemann-Pick use interactive technology and how it helps them manage their disease. People already taking part in the project have told us about the many different ways in which they use their iPad.

Some use it to play games, make lists or research NPD treatments or trials. Others are using their iPads to relax with their families and friends by listening to music or watching films together. Many of you are making memories by taking lots of photos and videos on your iPads. You can also use your iPad to contact any of us at NPUK or connect with the professionals involved in your care or to link in with other NPUK families.

From time to time we will contact you to see how you are using your iPad. How you use your iPad is completely up to you but please do tell us if you have found a particularly useful App or feature that we can pass on to other users.

If you would like to know more about our iPad project please ring Louise at the NPUK Central Office on 0191 415 0693 or email louise@npuk.org

SELF CARE TIPS:

Inspired by the mental health charity Mind, we have put together these graphics which we hope will help those struggling with certain thoughts and feelings.

Make sure to note the page number so you can go back to these tips any time the world seems difficult, and also please feel free to pass these tips on to any friends or family members who they may assist:

FEELING ANGRY, OR FRUSTRATED?

- RIP UP SOME PAPER INTO SMALL PIECES
- HIT A PILLOW / CUSHION
- THROW ICE CUBES IN THE BATH SO THEY SMASH

ANXIOUS, PANICKY, TENSE?

- SLOW DOWN - TAKE TEN DEEP BREATHS, COUNTING EACH ONE
- USE A CALMING VISUALISATION AND POSITIVE SELF-TALK
- FOCUS ON RIGHT NOW

DISSOCIATIVE, SPACED OUT?

- FIND A SAFE, QUIET SPACE AWAY FROM CROWDS
- CREATE AND REPEAT POSITIVE AFFIRMATIONS
- DRINK A GLASS OF ICE COLD WATER SLOWLY

DEPRESSED, SAD, LONELY?

- BUILD A SOFA FORT AND WATCH YOUR FAVOURITE TV SHOW
- WRITE ALL OF YOUR NEGATIVE FEELINGS DOWN ON A PIECE OF PAPER, THEN RIP IT UP
- TAKE IT SLOW, REACH OUT TO A FRIEND WHEN YOU'RE READY

If you are struggling to cope with any of these feelings please remember that we are here for you, get in touch with us at any time by phone on: 0191 416 06 93 or by email at: info@npuk.org.

HANNAH'S BEARS

Hannah L. Blunt creates fantastic personalised memory bears which we are sure many of the NPUK Community would love!

Suitable for everyone from those hoping to create a unique keep-sake for a friend or family member to bereaved parents who wish to create an item imbued with love and a personal connection, Hannah can deliver. Below she explores the experiences which led her to offer this fantastic service:

"...the idea for making memory bears out of loved ones' clothes came about after a traumatic time in my life, where for a substantial amount of time, I had to rely on others.

When I had recovered, I began sewing as a hobby. I wanted to support and bring comfort to others, like how I had been helped and so the memory bear idea was born.

To date, I have made so many different memory bears out of beautiful babygrows, football shirts, beaver and scout jumpers and even paramedic shirts! They can be made from almost any fabric, though I do tend to find that the less stretchy the material, the better.

Every bear is lovingly made, sewing a lifetime of memories into each individual one. Memory bears give comfort to so many people. Me knowing this, is the best part about creating them..."

Hannah has created items ranging from a bear for Rememberance Sunday, the Cub-Scouts, specific football teams and much more.

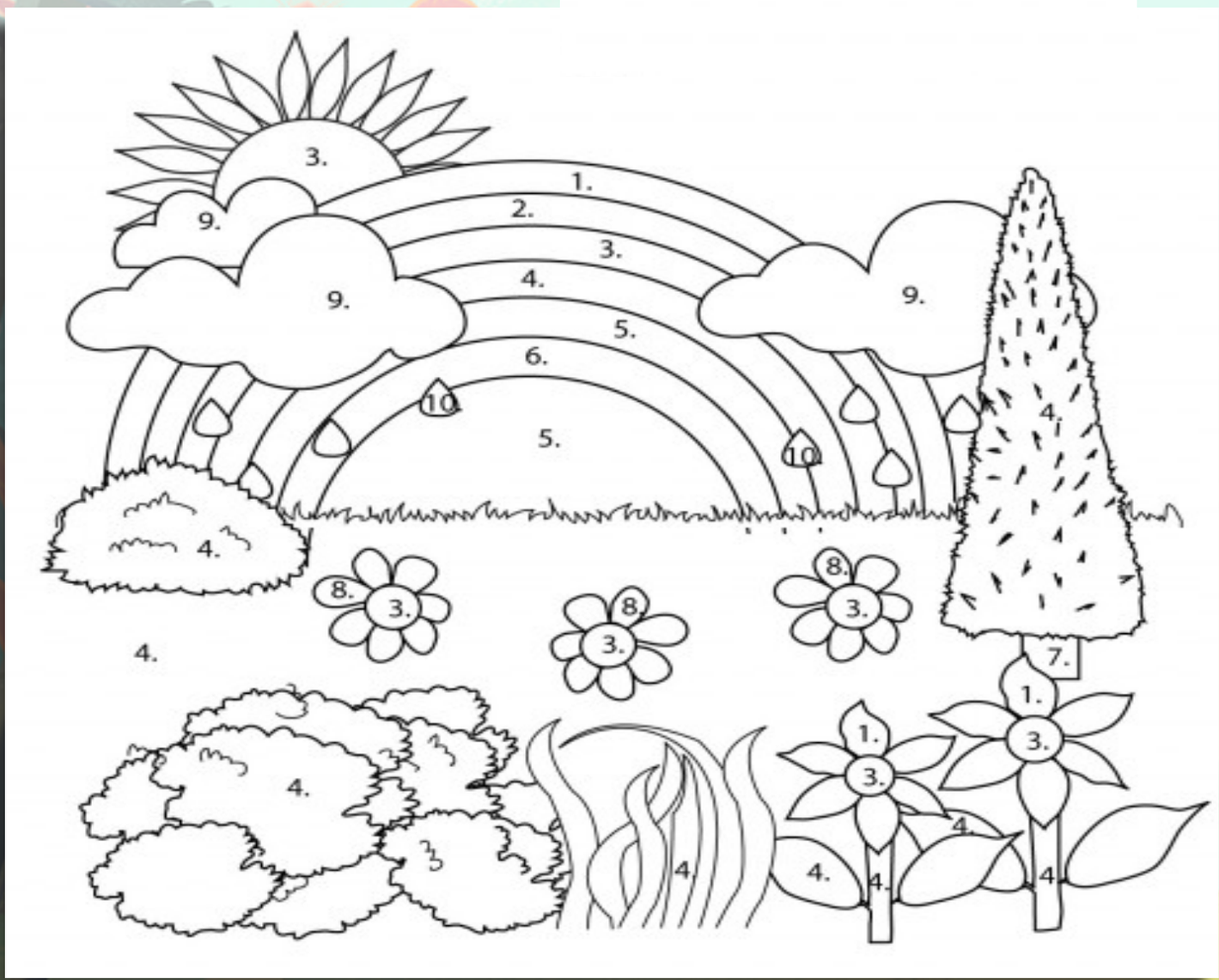
For further information please get in touch with Hannah directly on Facebook or by email at: h.blunt@hotmail.co.uk

KIDS' CORNER

In this new feature we wanted to give a fun place for the younger members of the NPUK community.

We hope you enjoy these 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!

1. red
2. orange
3. yellow
4. green
5. blue
6. purple
7. brown
8. pink
9. white
10. light blue



LAB EQUIPMENT WORDSEARCH

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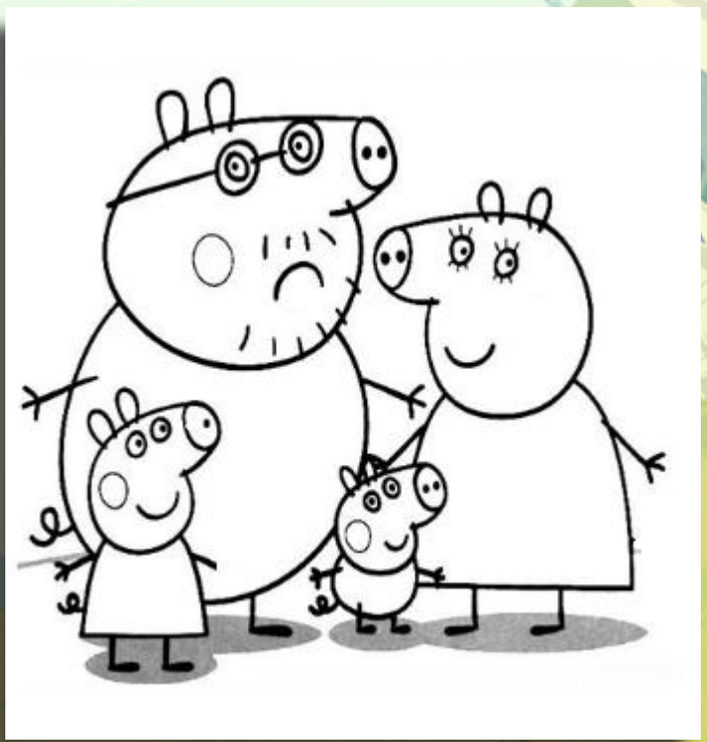
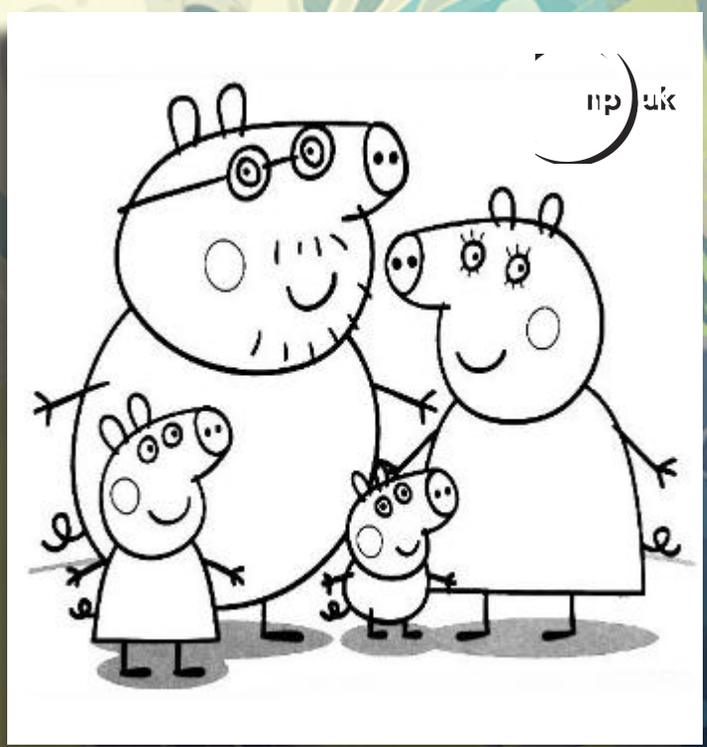
TRIPOD STAND
PIPET
DROPPER
REAGENT BOTTLE
WOOD SPLINTS
THISTLE TUBE
MICROSCOPE
FORCEPS
STOPPER
TRIANGLE
METER STICK
BEAKER
TEST TUBE
FUNNEL
BURET
PROBE

SPOT THE DIFFERENCE (SIX TO FIND!)





In loving memory

of those always in our hearts

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

Freddie Quinn 15/03/17 - 23/11/18

On the 23rd of November 2018, we sadly lost our precious baby to niemann pick disease- type A, aged only 19 months.

Freddie was a kind, beautiful, fun loving little boy who never ever lost the precious smile from his face no matter what he was going through.

Freddie was diagnosed on the 27th of March 2018 soon after his first birthday. In that time from diagnosis to Freddie's passing, we as a family realised just how important it is to make as many special memories as possible. We threw Freddie a Christmas on the 4th of August, visited Zoos and safari parks and almost every weekend made a trip to the aquarium which was Freddie's favourite place.

Freddie's life, albeit short, was filled with so much love and comfort whether it be from family, nurses, consultants, community nurses etc and I couldn't have asked for anything else for our beautiful boy than the love he received.

We are so so grateful to have had our son and the best brother in the world, and one day Freddie, we will meet you in Neverland. Forever in our hearts and thoughts, love mummy, daddy and your little sister Frankie.

Ruby Williams 24/10/13 - 2/4/17

Written lovingly by Ruby's mum, Kelly Williams:

It's been two years since you went away,

And I'll always miss you more than words can say,

You're my own beautiful angel in the sky,

Playing amongst the fluffy clouds up high,

I'm always your mummy, you're always my Boo,

Nothing will break the loving bond between me and you,

I love you my little lady and always will

There's a space in my heart only you could fill

I'm the proudest mummy there could ever be,

And though your angel wings have set you free,

You live forever deep inside my heart,

And we will never truly be apart.

The following poem is in loving memory of Arunjit Singh (9/10/16 - 16/12/18):

No one know how much we miss you,

No one knows the bitter pain

We have suffered since we lost you

Life has never been the same.

In our hearts your memory lingers,

Sweetly tender, fond and true.

There is not a day, my dear,

That we do not think of you.

On the 8th April 2019 the world lost a beautiful soul, Bonnie Strachan-McMillan (10/3/17 - 8/4/19, ASMD NP-A). The following poems on this page were originally shared at Bonnie's funeral service, by her mum Kayla and dad Jack:

My Mum, she tells a lot of lies, she never did before, but from now until she dies she'll tell a whole lot more. Ask my Mum how she is, and because she can't explain, she will tell a little lie, because she can't describe the pain.

Ask my Mum how she is, she'll say "I'm alright", if that's the truth then tell me, why does she cry each night? Ask my Mum how she is, she seems to cope so well. She didn't have the choice you see you see nor the strength to yell. Ask my Mum how she is, "I'm fine, I'm well, I'm coping".

For God's sake Mum, just tell the truth just say that your heart is broken. She'll love me all her life, I loved her all of mine. But if you ask her how she is, she'll lie and say she's fine. I am here in heaven. I cannot hug from here. If she lies to you do not listen, hug her and hold her near. On the day we meet again, we'll smile and I'll be bold. I'll say:

"You're lucky to get in here Mum with all the lies you've told!"

Love you always my Bonnie Lass, loads of love from Mum
(Written by Kayla Strachan)

You are the sky, I am the earth.

We align, it's perfectness.

Right in front and for miles,
like waters we know like The Clydes and the Niles.

We will run and laugh, jump and smile.

We shine like stars, forever baby.

Not once since I have known you,
did I not see you not ever wee lady.

Before you were born I knew you would be mine forever.

You are my dreams since always and forever -
nobody could keep us apart. Nobody could stop us from being together.

I love you, I love you, and I love you.

(Written by Jack McMillan)

It is with a very heavy heart that we share, on behalf of parents Jim and Susan Green, the sad news of their beloved son Roy's passing.

Roy, age 42 NP-C, passed away peacefully in November 2018, with Jim and Susan by his side. Roy's loss has been so deeply felt, by them and by the many people around the world to whom he provided such friendship, hope and inspiration.

Jim and Susan filled Roy's life with love and we can think of no better parents. Inspired by their children, their work has been instrumental in the development of NPUK, INPDA and the INPDR, work which continues to make an impact today.

Our hearts break for Susan, Jim and their family, and all who knew and loved Roy. Words do not seem enough at such times; we continue to hold them all in our hearts and to send our sincere sympathy, love and prayers to all.

Nadia Slatch remembers her beautfiul son, Zayn (28/10/12 - 10/04/17, NP-C) with these touching words:

2 years ago was the worst day of my life. It was the day I held you tight as you took your last breath. It was the day you gained your Angel wings & left an empty hole in our hearts. Life hasn't been the same since you left & don't think it ever will be the same again.

Sometimes I still can't believe you have gone, I can't express the pain I feel & how much I miss you, a part of my heart is missing & I feel empty inside. To lose a child is pain beyond belief, no words can describe the stabbing pain feeling that hits you when you are reminded that your child has gone forever. I always think about your special ways & I know even though you have left this world you will remain forever in my as my guardian angel.

Even after 2 years I still find myself missing you every single minute of each day, sometimes i may seem fine but then the reality hits me that you have gone & I can't control my sadness knowing you will never return. Your siblings miss you immensly & i love how they talk about you every day. I always question why you were taken so soon & never got to experience normal things & have a normal life...but i know deep down after all the suffering you went through you are now happy running free in paradise.

Zayn's legacy will carry me through & light the way for me, I have memories to last a lifetime & I promise I will forever make you proud. Keep running free my darling Angel, until we reunite again.

Love & miss you always Forever loved & remembered.
Always in my heart - my angel.

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

NG RARE PROGRESSIVE IRREVERSIBLE CHR SIBLE CHRONICALLY DEBILITATING LYSOSO TING LYSOSOMAL STORAGE DISEASE[1–3] RARE

NP-C affects all ages[1]

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

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

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

That's...

43,824 HOURS

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

THINK AGAIN THINK NP-C

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

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

If you are a:

Paediatrician

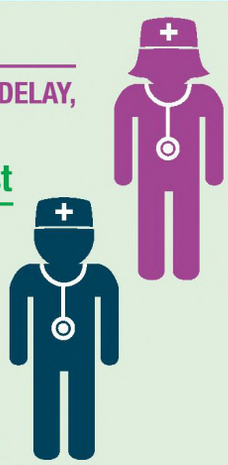
LOOK FOR ATAXIA, DEVELOPMENTAL DELAY, HEPATOSPLENOMEGALY

Paediatric hepatologist/neonatologist

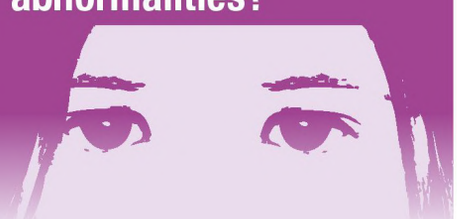
LOOK FOR HEPATO/SPLENOMEGALY, NEONATAL CHOLESTATIC JAUNDICE, NEONATAL LIVER DYSFUNCTION

Adult neurologist/psychiatrist

LOOK FOR COGNITIVE DECLINE, ORGANIC PSYCHOSIS, PROGRESSIVE ATAXIA



Have you checked for eye movement abnormalities?



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

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With grateful thanks to our grant providers:

COMMUNITY FUND

BBC Children in Need

THE hollie FOUNDATION

NPUK

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Note from the Editor...

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

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

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

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