Programme

DIGITAL ANNUAL FAMILY CONFERENCE & INTERACTIVE WORKSHOP ON NIEMANN-PICK DISEASES 2020

25th - 27th September 2020
We would like to welcome you to our first Digital NPUK Annual Family Conference and Interactive Workshop on Niemann-Pick Diseases, a milestone digital event which seeks to celebrate our fantastic community and all those working to make a difference within the field of Niemann-Pick diseases.

This programme contains information on everything you need to know about this weekend including schedules for the Interactive Workshop, Annual Family Conference, and the varied exciting content you can expect to see throughout. If you have any questions or issues regarding the WorkCast platform at any time, don’t hesitate to get in touch with the NPUK Staff Team who will be happy to help. You can also email any queries to: info@npuk.org

Friday
25th September
Interactive Workshop on Niemann-Pick Diseases

Saturday
26th September
Niemann-Pick UK Annual Family Conference

Sunday
27th September
Niemann-Pick UK Annual Family Conference

Professional meeting providing research and clinical trial updates for scientists, clinicians, and professionals working in the field of Niemann-Pick disease

Day one of the Annual Family Conference, a meeting for patients and families. Great opportunity for interaction between the NPUK community and NPD researchers

Day two of the Annual Family Conference - chance for further interaction with scientists in the field of NPD, as well as opportunities to meet with the broader NPUK Staff Team

Need a few reasons to join in with this year’s Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases? We’ve got you covered below...

SUPPORT
“Support” has been integral to the planning of our Conference as our main intention is to provide a safe space for our community where individuals are aware that whatever they are facing, those around them have either experienced similar challenges or are ready to listen and support them in any way they can.

RESEARCH
Our Conference offers the opportunity for clinical, science and research professionals to share their knowledge and experience on behalf of the Niemann-Pick community. As we are unable to meet in person this year, our event is even more important to stimulate these conversations and to enable interaction between families and professionals.

CLINICAL UPDATES
Despite the COVID-19 situation our Digital Conference will aim to bring together clinicians, scientists, and pharmaceutical companies involved in the field of Niemann-Pick disease, providing an important opportunity to meet and discuss the latest clinical developments.

FAMILIES
We are aware how much our Annual Conference means to NPUK families and as 2020 has been particularly tough, our Conference is even more important to connect our community. Hence, although we have gone Digital, we have created a programme that provides multiple opportunities for families to interact with one another.

FUN
Nobody can say that our community don’t know how to have fun! Although we cannot hold our usual Conference activities online, we have created a schedule that provides the chance for everyone in our community to have fun in a relaxed environment. From puppet shows to Terry’s Quiz, there is an activity for everyone to enjoy!
Dear Families, Friends, and Supporters,

On behalf of the NPUK Board of Trustees, I have the pleasure of welcoming you to our very first Digital Conference weekend, which marks the 11th Interactive Workshop and 27th Annual Family Conference. It is of course very different to what we have grown accustomed to over the years, but we are confident that it will still be a meaningful and productive event.

In spite of the challenges COVID-19 has provided throughout 2020 NPUK has continued to work tirelessly on behalf of those affected by Niemann-Pick diseases to ensure access to optimum care, increase awareness and understanding of the condition and to facilitate research and treatment options where possible. This has been demonstrated by our digital output throughout the pandemic lockdown and once again by our busy, albeit digital, Conference programme, which includes updates from UK and International speakers covering the subjects of clinical trials, basic science research and practical care issues.

I hope you will find this new Conference experience informative and rewarding. If at any time you encounter issues within the digital platform please do get in touch with our Staff Team who will be happy to help.

I very much look forward to seeing you all in 2021, take care!

Will Evans
Dr. William Evans, NPUK Trustee & Chair

NPUK Board of Trustees

NPUK is governed by a Board of Trustees who are responsible for delivering the aims and objectives of the Charity. We encourage you to get to know them over the course of this weekend, you can also learn more at npuk.org/about-us

NPUK Staff Team

NPUK has a small but dedicated staff team who are responsible for delivering the services of NPUK and provide much-needed care and support.

Please don’t hesitate to get in touch with members of our Staff Team throughout the weekend, they will all be happy to help with any questions or issues you may experience on the WorkCast platform.

You can also learn more about each individual and what their respective roles bring to NPUK, at the following web page: npuk.org/about-us/meet-our-team

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SPEAKERS’ PROFILES
We are calling on the NPUK Community and all of our other friends attending this year’s Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases, to simply make it your own! As the Conference is Digital, and is therefore taking place in your home, or any other venue of your choice, you set the rules and you choose what you watch and when! Please feel free to drop in and out of sessions, there is no obligation to attend every single presentation, and we do not expect everyone to watch all of the content all of the days! Many of our sessions/activities will be pre-recorded and available “on-demand” so access can be fitted around your schedule. If you need information, help, or advice, you can use the chat icon during sessions to ask questions or deliver feedback, it’s also a great way in which you can contact friends, family members, and professionals throughout the weekend - just like at our in-person event! If you received a goody bag in the post, we would love to see photos of the various NPUK Digital Conference swag - particularly the (temporary) tattoos!

The Event is free to families and friends - so be sure to invite others as you go so that they can join in with the fun!

We encourage you to “be social” throughout this weekend, please share what you get up to on Facebook, Instagram and Twitter with the relevant hashtags: e.g. #NPUKConf2020 #NPUKIW2020 #NPUKDigital

As this is our first ever Digital Conference, we are eager to hear all your thoughts and feelings on the sessions that you attend, please email feedback to: info@npuk.org
Interactive Workshop on Niemann-Pick Diseases 2020

25th September 2020

Session 1: Welcome and Introduction
A warm welcome to the 1st Digital Interactive Workshop on Niemann-Pick Diseases

Fran Platt, PhD, FMedSci, Prof. of Biochemistry & Pharmacology, Department of Pharmacology, University of Oxford

Dr. William Evans, Chair, Niemann-Pick UK

COVID-19 and Niemann-Pick Diseases
Expert discussion and debate regarding COVID-19 and its relationship with Niemann-Pick Diseases.

Chair: Dr. William Evans
Panel:
Professor Jonathan Ball, School of Life Sciences, University of Nottingham
Stephen L. Sturley, Ph.D. Department of Biology, Columbia University, New York, USA
Andrew Munkacsi, Ph.D. Senior Lecturer (Assistant Professor), Victoria University of Wellington, School of Biological Sciences, New Zealand

9:45 - 10:00

10:00 - 11:00

BREAK

11:00 - 11:15

Session 2: Research Updates
Key updates from science and clinical experts in the field of Niemann-Pick diseases

Chair: Fran Platt
Presenters: Beth Solomon, MS, CCC-SLP, Speech Language Pathology, National Institutes of Health, USA
Dr. Emre Lloyd Evans, Senior Lecturer, School of Biosciences, Cardiff University
Ahad A. Rahim, PhD, Professor of Translational Neuroscience, Associate Director of Research, University College London School of Pharmacy
Dr. Dolores Ledesma, Centro de Biologia Molecular Severo Ochoa, Universidad Autonoma de Madrid
Andrew Munkacsi, Ph.D. Senior Lecturer (Assistant Professor), Victoria University of Wellington, School of Biological Sciences, New Zealand

11:15 - 12:45

12:45 - 13:30

BREAK

13:30 - 15:00

Session 3: Improving Outcomes in Niemann-Pick Diseases
Global initiatives to further the understanding of patient experience, improve time to diagnosis and support therapeutic research

Chair: Marc C. Patterson, MD, Professor of Neurology, Pediatrics and Medical Genetics Mayo Clinic Children’s Center, Minnesota, USA

Presenters: Conan Donnelly, PhD, Registry Manager, International Niemann-Pick Disease Registry (INPDR)
Shaun Bolton, MSc, Operations Consultant, INPDR
Jackie Imrie, MSc, Specialist Recruitment Consultant, INPDR
Melissa Wasserstein, MD, Chief, Division of Pediatric Genetic Medicine Professor of Pediatrics and Genetics, The Children’s Hospital at Montefiore Bronx, NY, USA
Dr William Evans, Chair, Niemann-Pick UK
Claire Burbridge, MSc, CPsychol Senior Director, Clinical Outcome Assessments, Clinical Outcomes Solutions (UK Office)

15:00 - 15:15

15:15 - 17:45

BREAK

18:00 - 19:00

Round Table Discussion: Impacts and Burden of Niemann-Pick type C: Patient and Caregiver Perspective (Invitation Only)
A closed meeting hosted by NPUK, to discuss results of this Orphazyme sponsored patient and caregiver survey

19:00 - 21:00

NPUK: “Getting to Know You” Sessions
- Lads, Dads, and Carers / Adults Living with NP-C
- Mums / ASMD (8-9pm)

20:00 - 21:00

Professional Networking Events
**PRINCIPLES FOR INTERACTION**

Niemann-Pick UK (NPUK) works towards one clear goal: to improve the lives of people affected by Niemann-Pick Diseases (NPD). Our endeavours are directed into three key areas: offering care and support; sharing information; and the support of research and therapeutic development.

As research advances and as an increasing number of clinical trials are initiated, it is even more critical that the information we provide to our community is up-to-date, accurate and balanced. This includes ensuring all information provided is, fair, not misleading in any way and capable of substantiation. In addition, we encourage transparency and accountability in collaborative working and the opportunity for open debate to inform the patient community.

Our annual events provide an ideal opportunity to be able to do this, as many of our families attend, alongside our healthcare professional colleagues and pharmaceutical company representatives. Throughout our activities and interactions, NPUK strives to maintain integrity, independence, impartiality and transparency in all we do.

Therefore, as we facilitate this exchange of clinical and scientific information, we would like to remind attending pharmaceutical and industry representatives that all communications and interactions at our conference must abide by the Association of the British Pharmaceutical Industry (ABPI) Code of Practice. Please visit www.abpi.org.uk for further information.

Another useful resource is the ‘Principles for interactions with biopharmaceutical companies: the development of guidelines for patient advocacy organisations in the field of rare diseases’. These Guidelines, which were developed by an Independent Expert Panel, recommend best practices and standards for interactions between patient advocacy organisations and industry. The Guidelines can be found on the NPUK website www.npuk.org.

NPUK also complies with this code (in particular Clause 27, Relationships with Patient Organisations) in our interactions with all pharmaceutical companies engaged in developing therapies for NPD. As such, we will not endorse or promote one particular trial above another.

However, we recognise the significant benefits that can arise from working collaboratively with all stakeholders, and will continue to offer unstinting support, encouragement and guidance to all who share in our goals; including pharmaceutical companies, researchers and academic institutions, health care agencies and other charities.

“...we encourage transparency and accountability in collaborative working and the opportunity for open debate to inform the patient community...”

NPUK believes these actions are in the best interests of the NPD community. We will continue to passionately support individuals and families affected by NPD diseases and all those who share our goal.
Treatment in Niemann-Pick disease type C patients with an HSP amplifier
Marc C. Patterson, MD, Professor of Neurology, Pediatrics and Medical Genetics Mayo Clinic Children’s Center Minnesota, USA

Clinical Development of Adrabetadex for NPC-1
Professor Paul Gissen
Consultant in Paediatric Metabolic Diseases, Great Ormond Street Hospital, London
Elizabeth Berry-Kravis MD PhD
Professor of Pediatrics, Neurological Sciences & Biochemistry Rush University Medical Centre, Chicago, USA

Session 12: ASMD Invisible Illnesses Campaign
Co-Chairs:
John Lee Taggart, NPUK Communications and Campaigns Manager
James Brooks, ASMD Community Member

Session 13: Children and Young Persons’ Activity Programme
Join us for some yoga for kids with Sarah Redshaw!

16:45 - 18:15
BREAK

18:15 - 19:00
Beth’s Baffling Brain Teaser & Raffle Draw
A quiz especially for the kids hosted by one of our dedicated Children & Young Persons’ Activity Programme Volunteer Team Leaders, Beth. Raffle draw with Melanie.

19:00 - 20:00
Double Sensation: Carl and Emma Burdon (Facebook Live)

20:00 - 21:30
The Colwell Quiz
Back by popular demand...The Colwell Quiz, hosted by the one and only Terry Colwell! (NPUK Community Member)

After the great feedback we had following Terry’s weekly quizzes during lockdown, as part of the NPUK: Social Not Distant Campaign, we are delighted to be able to offer a return to this popular format. Grab a drink of your choice, and join in with the fun via Zoom!

Don’t forget to share photos of yourself with your NPUK swag!
Please use the following hashtags when posting to your social media: #NPUKDigital2020 #NPUKFamily #NPUKAF2020 #NPUKWi2020
**Welcome and Introduction**
Dr. William Evans

10:00

10:15 - 11:30

**Parallel Sessions - please select from session 14, 15 or 16**

**Session 14: “Easy like Sunday Morning”**
Join the NPUK Team and community members for your morning cuppa and a bit of conversation

Hosts: Karen Thomas (NPUK Project Families Officer), Christine Jopling (NPUK Finance & Admin Officer), Janice Brooks (NPUK Trustee)

**Session 15: Dealing with Anxiety**
Join NPUK Project Families Officer Steve Neal & Counsellor Tony Somers to learn some helpful stress and anxiety management techniques

Hosts: Steve Neal (NPUK Families Officer)
Tony Somers (Counsellor and Author)

**Session 16: Focus on Gene Therapy**
Gene Therapy: what is it? Will it work for Niemann-Pick disease? Where are we now?

Chair: Dr. William Evans

Panel:
Charles H. Vite, B.S., DVM, PhD
Professor of Neurology and Neurosurgery, University of Pennsylvania School of Veterinary Medicine
Philadelphia, USA

Ahad A. Rahim, PhD, Professor of Translational Neuroscience, Associate Director of Research, University College London School of Pharmacy

Cristin Davison, Project Manager, Support of Accelerated Research for Niemann-Pick C (SOAR-NPC)

William Pavan, Ph.D. Senior Investigator Genetic Disease Research Branch National Institutes of Health

11:30 - 11:45

11:45 - 12:45

**BREAK**

**Parallel Sessions - please select from session 17 or 18**

**Session 17: NPUK: Support. Community. Progress**
Check in with the NPUK Care & Support Team who are here for you, our community! We will also hear from another inspirational member of our community before hearing from John regarding the success and legacy of our short, Go Make Memories

Chair: Joella Melville, NPUK Trustee

Hosts: NPUK Staff Team

**Session 18: Children and Young Persons’ Activity Programme (Magic Show!)**

We hope you enjoyed this year’s Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Disease. Please send any and all feedback to: info@npuk.org - surveys will be available shortly.

Your feedback is important to us.

Informative? 
Supportive? 
Entertaining?
KIDS' CORNER

Our Digital Annual Family Conference is not just for adults, it's for everyone! With this in mind this section is dedicated to the younger ones in the NPUK community...

...but Big Kids are certainly welcome to have a try of the games and puzzles too!

Can you spot the differences between these two pictures?

(Pssst...there are six to find in total. Good luck!)

Children and Young Persons' Activity Programme:
Saturday 26th September 2020
10:15 - 11:15 JoJo Puppet Show
11:30 - 12:30 Storytime with Cora
13:00 - 14:00 Singing and Stories
15:15 - 16:15 Kid's Yoga
18:00 - 19:00 Beth’s Baffling Brain Teaser

Sunday 27th September 2020
11:45 - 12:45 Magic Show

ANIMALS WORDSEARCH

Children & Young Persons’ Volunteer Team Leaders:
Katie Reynolds
Beth Scowcroft
Jan Patterson

These are the amazing people who will be helping to support this year’s Digital Children & Young Persons’ Activity Programme - you can access the programme on the WorkCast event site (Kid’s Quiz will be via. Zoom)
We are incredibly thankful to each and every one of our speakers over the course of both the Annual Family Conference and the Interactive Workshop on Niemann-Pick Diseases, your contribution to this weekend and your ongoing support as an advocate to those in the NPUK community is much appreciated.

For further details on our speakers please visit the NPUK WorkCast event page.
Dr. Conan Donnelly joined the INPDR from the National Cancer Registry Ireland, where he spent nearly three years as Senior Epidemiologist before taking on the role of Research Manager. Conan has a strong background in research and management, bringing key skills and experience to the role of Registry Manager and helping the INPDR to further develop its ground-breaking global patient disease registry.

Fiona Dunne joined the NPUK Board of Trustees last year, after being a member of the NPUK Community for many years. She brings both passion and personal empathy to the Board. Her son Harry (NP-C) passed away in 2010 and sadly his sister Grace also passed in 2018. We are proud to have Fiona as part of the Trustees team.

William Evans is a GP and joined the Board of Trustees of NPUK in 2010 and was elected Chair in 2016. Will is married to Miriam and has 3 children, their eldest son Sam was diagnosed with NPC when he was only a few months old in early 2008. Will represents NPUK and our community at scientific conferences, meetings with pharmaceutical companies, drug regulators and other rare disease charities. He also writes articles and gives presentations to the rare disease community, families affected by NP-C, doctors, nurses and scientists.

Taylor Fields is the Senior Vice-President of IntraBio Inc. At IntraBio, she leads all efforts related to the development and commercialization of IntraBio's novel treatment and therapies, and is principally responsible for the company's clinical and regulatory development programs. Ms. Fields obtained a B.A. in International Studies and English from the University of Michigan, and received a Masters in systems design from the University of Oxford.

N Scott Fine is the Chief Executive Officer and Chairman of Cyclo Therapeutics, Inc., a biotechnology company focused on developing novel therapeutics based on cyclodextrin technologies. Additionally, he is a member of the Boards of Directors of Kenon Holdings Ltd. (NYSE: KEN) and International Mobile Telecom Investment Stichting. Mr. Fine devotes time to several non-profit organisations, including through his service on the Board of Trustees for the IWM American Air Museum in Britain. Mr. Fine has been a guest lecturer at Ohio State University's Montz School of Law and at Forham University Law School.

Prof. Paul Gissen got his PhD in Human Molecular Genetics from the University of Birmingham investigating genetic causes of rare paediatric diseases when he developed specific research interest in intracellular trafficking disorders. Paul is currently a Consultant in Paediatric Inherited Metabolic Diseases at Great Ormond Street Hospital, London and a group leader at the Laboratory for Molecular Cell Biology, University College London.

Álvaro Berzal Gómez began to use his experience in child neurology to help those affected by debilitating physical conditions - he now has a number of trusted clients and is dedicated to improving their quality of life.

Christina Guldborg is Director Clinical Outcomes at Orphazyme A/S and is responsible for the validation and standardized implementation of outcomes measures in clinical trials. Christina has more than 30 years of experience in the pharmaceutical industry.

Caroline Hastings is a Professor of Pediatrics at UCSF Benioff Children's Hospital Oakland, and serves as the director of NeuroOncology. She is on the clinical faculty at UCSF Benioff Children's Hospital Oakland, and serves as the Director of the Pediatric Hematology/Oncology Fellowship training program. Her academic interests include clinical trial development in high risk and relapsed leukemia, pediatric brain tumors, and in particular Niemann-Pick Type C disease, a progressive fatal metabolic storage disease.

Dr. Sharon Hrynkow is Chief Scientific Officer and Senior Vice President for Medical Affairs at Cyclo Therapeutics, Inc., a Florida-based clinical stage pharmaceutical company working to develop cyclodextrin-based drugs for the treatment of Niemann-Pick Disease type C and Alzheimer's Disease. Dr. Hrynkow leads the company's clinical, medical and scientific programs. Prior to joining Cyclo Therapeutics in 2015, Dr. Hrynkow devoted 20 years to public service, including in leadership roles in global health at the National Institutes of Health and the Department of State.

Jackie Imrie is a Trustee of NPUK and obtained a degree in Biochemistry, Genetics and an MSc in Genetic Counselling after being a paediatric nurse. She worked at the William Harvey Medical Unit, Manchester seeing patients with many inborn errors of metabolism. In 1999, funded by NPUK, Jackie became the first support nurse for families with Niemann-Pick disease. She currently serves as a consultant for the International Niemann-Pick Disease Registry (INPDR).

Debbie Kaflowitz, 13-month-old baby, with NPC, has a chance to receive the INPDR’s novel treatment and therapies. Debbie’s daughter, Rachael Kaflowitz, died from Niemann-Pick Type C, surrounded by her loving family. Debbie and the rest of her family members remain committed to both increasing the awareness of this rare condition and raising money to find a cure for Niemann-Pick, so that someday others may not have to go through the same challenges and heartache. Debbie has dreams of being with the NPUK Community in person at some point...but for now, digital will have to do!

Sean Kassen In this role as Director of the Ara Parseghian Medical Research Fund at The University of Notre Dame Sean is responsible for administering all of the functions of the Fund which includes advancing Niemann-Pick Type C (NPC) disease research, fundraising, managing the grants program, and raising awareness of NPC disease. Shaun received his Ph.D. in Cell and Molecular Biology from the University of Notre Dame in 2008.

Ciara & Graham Kirk have been members of the NPUK Community for many years, offering their unique perspective to positively further our charity - most recently with our upcoming campaign on “Invisible Illnesses” as well as joining in with the fun of the “Social NotDistant Campaign” during the COVID-19 lockdown!

We are proud to have them both on board for our NPUK Community Voice feature this year!
Robin Lachmann
Dr. Lachmann is one of two consultants in the Charles Dent Metabolic Unit at the National Hospital for Neurology - one of the National LSD Centres for adults. He is Chairman of the Metabolic Disorders Clinical Reference Group which advises NHS England. Dr. Lachmann has a special interest in LSDs which involve the brain. This stems from his PhD which was on developing viral vectors for gene delivery to the brain. With the help of a Big Lottery grant from the NPUK he spent some years working on the NP-C mouse. Although he no longer works with mice, he does still look after patients with NP-C.

Dr. Dolores (Lola) Ledesma
Dr. Ledesma’s scientific career in neuroscience started during her PhD at Autonoma University of Madrid (1992-1995), studying the post-translational alterations of Tau protein in Alzheimer’s Disease. Dr. Ledesma currently leads a team focusing on the contribution of cholesterol and sphingolipids to neuronal physiology and on the pathological consequences of their alterations in lipid storage disorders such as Niemann-Pick disease.

Emyr Lloyd-Evans
Dr. Lloyd-Evans is a senior lecturer at the School of Biosciences, Cardiff University. He runs a research lab with a focussed interest on lysosomal diseases and diseases of ageing and lectures to undergraduate medical and biomedical students on lysosomes, Ca2+ signalling and lysosomal diseases. In recent years, the lab has an emerging interest in the development of new drug screening strategies for lysosomal disease and the use of zebrafish models of lysosomal disease for high throughput in vivo drug testing. The lab collaborates with numerous academic, industry and biotech partners to develop new therapies for these diseases.

Eric Low
Eric has worked in the fields of medical research, market access and patient organisations for over 25 years. He currently runs a consultancy business specialising primarily in strategic market access, life sciences and healthcare policy, patient centricity and patient organisation development.

Toni Mathieson
Toni became involved with NPUK back in 2003 after receiving the news that her beautiful daughter Lucy, then just five weeks old, had Niemann-Pick Disease Type C (NP-C). After receiving amazing support from the charity during those difficult months, and in the following years when Lucy’s brother and sister, Hannah and Samuel, passed away from the same condition, she decided to lend her support to the charity. In her role as Chief Executive, she uses the empathy and compassion from her personal experiences to provide hope for other families affected by Niemann-Pick disease.

Allison May Rosen
Allison May Rosen is an award-winning communicator who specializes in providing strategic communications, content development, and training services for healthcare organizations. Allison has more than two decades of experience working in the private and public sectors, including a dozen years at a global healthcare agency where she advised major health companies, trade associations, universities, non-profits and foundations.

Joella Melville
Joella has worked with NPUK in a professional capacity since 2009 and joined as a Trustee in March 2016. Her background in healthcare communications has meant that her central focus has been on helping to raise the profile of both Niemann-Pick disease and our small charity, in the UK and at an international level.

Georgina Morton
Georgina Morton is the Chair of ArchAngel Trust and mother to Ava, who is living with metachromatic leukodystrophy (MLD). The ArchAngel MLD Trust was established to support medical teams around the world who are working to help people with the rare genetic illness Metachromatic Leukodystrophy (MLD). ArchAngel also awards grants to help UK families with MLD affected children and is spearheading a campaign to have all UK babies screened for MLD (and other rare diseases) at birth.

Andrew Munkacsi
Dr. Andrew Munkacsi is Senior Lecturer in the School of Biological Sciences and Director of the Chemical Genetics Laboratory at Victoria University of Wellington. A major focus of his research is Niemann-Pick type C disease, a neurodegenerative disease caused by lysosomal accumulation of cholesterol and sphingolipids. His team uses a suite of genomic, proteomic and lipidomic analyses to identify modifiers of cholesterol and sphingolipid accumulation that are thus candidate targets to modify the onset and progression of Niemann-Pick type C disease.

Steve Neal
Steve joined NPUK in early 2017 to support our Big Lottery funded project “Shaping the Future Together”. He has brought a range of skills and experience to his role as Project Families Officer, as well as personal insight into NP-C, which helps him in his work to support families. Steve is well known to the NPUK community (his mum, Gloria was affected by NP-C) and has previously been a volunteer with the Activity Programme at NPUK Family Conferences.

Prof. Marc Patterson
Professor Patterson’s research has focused on neurometabolic disorders, with special interests in Niemann-Pick disease, Type C, Gaucher disease and Congenital Disorders of Glycosylation, areas in which he has published and spoken widely. His awards include a Fulbright Award, The Lewis P. Rowland Teaching Award at Columbia University, and several names professorships. Professor Patterson is strongly committed to care and advocacy for children and families with neurological disabilities.

Dr. William Pavan
Dr. William Pavan received his B.S. in animal science from the University of Massachusetts, Amherst and his Ph.D. in physiology from the Johns Hopkins School of Medicine, Baltimore. He completed his post-doctoral fellowship in the laboratory of Shirley Tilghman, Ph.D., at Princeton University in which he studied the developmental genetics of mouse coat color pigmentation. His primary areas of interest include the development and diseases of melanocytes, the cells responsible for pigmentation of skin and hair, as well as the lysosomal storage disorder Niemann-Pick disease, Type C.

Prof. Fran Platt
Fran works in the Pharmacology Department at the University of Oxford. Her laboratory is interested in understanding and treating lysosomal storage diseases. A major current focus of the lab is in NPC disease with ongoing studies on the pathogenic cascade, mechanisms of immune activation, biomarker identification and trialling therapies in a mouse model of this disease.
**Dr. Ahad Rahim**

Ahad has worked on gene therapy and the development of gene transfer vectors for 10 years while working at the Institute of Cancer Research and more recently University College London in the laboratories of Professor Adrian Thrasher and Dr. Simon Waddilington. Ahad’s laboratory at UCL focuses on the development of gene therapy strategies for a range of intractable and ultimately lethal neurological disorders.

**Pat Roberts**

Pat Roberts is the Director of the Newborn Screening Programme and the Secretariat to the UK LSD Patient Collaborative Group, of which Niemann-Pick UK (NPUK) is a member. Her work with Save Babies Through Screening Foundation UK (now Krabbe UK) has helped to increase awareness of this condition and other lysosomal storage diseases through collaboration and targeted campaigns. Pat is also the Global Liaison Director at KrabbeConnect USA.

**Dr. Reena Sharma**

Dr. Reena Sharma is a consultant in Adult Metabolic Medicine at The Mark Metabolic Medicine Unit, Salford Royal Hospital NHS foundation trust since 2011 and is honorary senior lecturer at the University of Manchester. She is European coordinator, Chief investigator and Principal investigator for various industry sponsored as well as academic clinical trials.

**Beth Solomon**

Beth Solomon is the Lead Senior Speech Language Pathologist of the Speech Language Pathology Section of the Rehabilitation Medicine Department at the NIH Clinical Center. She is currently involved in a host of research protocols investigating phenotypic expressions of Niemann-Pick Disease, Kennedy’s Disease, and many more.

**Tony Somers**

Tony Somers is a professional counsellor, life coach, trainer and self defence expert. In addition, he is a trained fire fighter and a 6th dan martial artist. Tony set up anthony somers.com in 2007 having spent the previous 17 years as a trained fire fighter working for the West Midlands fire service. Tony is respected by his clients and has built an enviable track record as an accomplished counsellor/coach and self defence professional.

**Sara Stroer**

Sara Stroer is a tireless advocate for all Niemann-Pick families and has collaborated on a number of International Niemann-Pick Disease Alliance (INPDA) campaigns, offering a new perspective for the German patient support group, Niemann-Pick Selbsthilfegruppe, and a stronger voice for other ASMD NP-B family members such as herself. Sara joins us this year to tell her family’s story, the reason for her dedication and drive!

**Stephen Sturley**

Stephen trained as a biologist (B.Sc.) and geneticist (M.Sc. and Ph.D) at Birmingham University (UK) and developed a research program that has applied genetic and biochemical approaches to understanding the metabolism of fats and sterols in humans. Consequently, Stephen and his colleagues established a gene discovery strategy that has identified several novel pathways of eukaryotic lipid homeostasis. Particular success has been attained with regard to sterol and lipid transport in human diseases such as obesity, diabetes and neurodegeneration (mainly through our studies on Niemann-Pick type C (NP-C) disease).

**John Lee Taggart**

John is the Communications & Campaigns Manager for both NPUK, the INPDA, and the INPDR - his role centres on the development of engaging content for, and about, the Niemann-Pick community. His role has increasingly led to collaboration with other rare disease organisations and advocates, working to strengthen the shared rare voice. In 2019 he was Executive Producer on the short film, Go Make Memories, which was developed to raise awareness of Niemann-Pick disease - the film has since won on to win a number of awards and featured at a number of film festivals to wide acclaim.

**Paula Jane Taggart**

Paula is an Iyengar yoga teacher who has been practicing for 35 years, and teaching since 2002. She places emphasis in her classes on accessibility for all ages and levels, believing that everyone can take part in yoga and reap positive benefits as a result. Paula first volunteered for us earlier this year during our Self Care Campaign, and will be supplying short video presentations throughout the Conference weekend.

**Karen Thomas**

Karen joined the NPUK team as our new Families Officer in October 2019 and quickly hit the ground running by developing close relationships with the NPUK Community. All of her previous work experience has involved providing support, information and guidance to individuals. As a Families Officer her role is to offer practical and emotional support to those affected by Niemann-Pick disease and their families.

**Charles Vite**

Charles is an American College of Veterinary Internal Medicine (ACVIM) board-certified veterinary neurologist with expertise in clinical neurological, neuroanatomy, neuropathology, neuroradiology, and electrophysiological techniques. Charles currently manages the National Referral Center for Animal Models of Human Genetic Disease (RCAM, NIH OD P40-10939; PI: Vite). The RCAM consists of breeding colonies of more than 40 models of human genetic diseases, including many with CNS diseases such as dogs with globoid cell leukodystrophy and mucopolysaccharidoses, and cats with Niemann-Pick disease type C1 and alpha-mannosidosis.

**Helen Waller-Evans**

Helen Waller-Evans is a lecturer at the Medicines Discovery Institute at Cardiff University, where she runs the lysosomal disease section. As a dedicated cell biologist, Helen has been researching lysosomal storage disorders, including Niemann-Pick type C for the last eight years. Six of these years were spent with Emyr Lloyd-Evans, a world-leading lysosomal disease expert, where she gained expertise in lysosomal cell biology.

Helen has taken the skills she gained during this time to the Medicines Discovery Institute, where she is now working on developing cell based screens to identify potential new therapies for lysosomal storage disorders.

**Melissa Wasserstein**

Dr. Melissa Wasserstein is the Chief of the Division of Pediatric Genetic Medicine at the Children’s Hospital at Montefiore and an Associate Professor of Pediatric and Genetics at the Albert Einstein College of Medicine (AECOM). Dr. Wasserstein has published more than 70 articles and chapters. Melissa is a clinical investigator whose research focuses on evaluating the natural history of ASMD Niemann-Pick disease A/B, and has served as Principal Investigator on many clinical trials evaluating novel treatments for rare disease.
SAVE THE DATE

14th - 17th October 2021

Niemann-Pick UK (NPUK) would like to cordially invite you to next year’s event, which will be the 28th Annual Family Conference & 12th Interactive Workshop on Niemann-Pick Diseases - which we hope will be a return to an in-person meeting.

The provisional date for this meeting is the 14th - 17th October 2021, and we are pleased to announce that we will be holding the Conference at our home away from home for many years, Wyboston Lakes Bedfordshire.

We hope you enjoyed this event and found it to be a positive and informative experience - please send any feedback or further queries by email to: info@npuk.org, or you can call us at: 0191 415 06 93

www.inpdr.org
Connecting
During COVID-19

This year has been very challenging for everyone, which has certainly included Niemann-Pick UK and our close-knit small community.

Our communication strategy during these unprecedented times was centred upon continuing the engagement of our community and the meaningful interactions that take place between our families. Hence, we wanted NPUK individuals and families to have a sense that they were still involved in the NPUK community. Our online polls provided those who follow NPUK on social media to choose what activities and posts would be shared that week. From amusing videos to self-care advice, there was something for everyone!

Like many organisations we have been working on our video calling expertise to move the engagement with our community online. This has included the NPUK team holding weekly video calls and our fantastic community getting together, including the Queen Bees and Lads, Dads and Carers Sessions taking place over Zoom. Our “Time to Talk” sessions have also been a roaring success. This series of video calls enabled members of our community to have a catch up from home with one another and our Care & Support team over their morning beverage.

In response to the COVID-19 crisis and in recognition that many individuals and families within our community had been impacted by the lockdown, the Coronavirus Emergency Response Fund was created. NPUK trustees designated £10,000 to create a new grant scheme for patients and families. The Hollie Foundation, in acknowledgement of our shared goals, contributed a further £2,500.

WAYS WE STAYED CONNECTED:

As an immediate response to the COVID-19 situation we launched our Social Not Distant campaign which spanned over ten weeks and scheduled, via social media, various activities, interactive sessions and informative posts to engage with.

One of the highlights of our new Digital approach was the NPUK After video series. Over ten weeks we heard from individuals in our community via video recordings sharing what they have been missing the most and what they were looking forward to doing in the future. This weekly series provided a bit of positivity and levity, with many people appreciating hearing from friends and family members who were currently in shielding/lockdown. None of this would have been possible without the enthusiasm of many individuals in our community who got involved!

Huge thanks to our Sponsors

The Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases would not be possible without the generous support of our sponsors - many thanks to each and every sponsor, it is truly appreciated.

The Big Lottery Community Fund has enabled us to expand our community reach thanks to their grant support with the “Shaping Our Future Together” initiative.

BBC Children in Need continue to provide essential grant support to NPUK, enabling access to the vital care and support offered by NPUK Clinical Nurse Specialist, Laura Bell.

Cyclo Therapeutics, Inc. provided financial support for this event via an unrestricted educational grant without control or influence of the content of the programme or the selection of speakers.

The Hollie Foundation has once again provided a grant in support of this year’s Children & Young Persons’ Activity Programme, and continue to part-fund the essential role of our Families Officer, Karen Thomas.

Mallinckrodt provided financial support for this event via an unrestricted educational grant without control or influence of the content of the programme or the selection of speakers.

Orphazyme provided financial support for this event via an unrestricted educational grant without control or influence of the content of the programme or the selection of speakers.

Sanofi Genzyme provided financial support for this event via an unrestricted educational grant without control or influence of the content of the programme or the selection of speakers.
11th Interactive Workshop on Niemann-Pick Diseases & 27th Annual Family Conference

Till next year...

#NPUKConf20 #NPUKFamily

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Making a difference for the Niemann-Pick community

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