



Programme

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

25th - 27th September 2020



Just a few reasons to join us...

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...

DIGITAL ANNUAL FAMILY CONFERENCE & INTERACTIVE WORKSHOP 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

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

**Saturday
26th September**
Niemann-Pick UK Annual Family Conference

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

**Sunday
27th September**
Niemann-Pick UK Annual Family Conference

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

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**

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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



Will Evans



David Holton



Jackie Imrie



Richard Rogerson



David Roberts



Bill Owen



Janice Brooks



Helen Carter



Joella Melville



Fran Platt
(Co-opted)



Stefanie Rymsza
(Co-opted)



Fiona Dunne
(Co-opted)



Jenny Charman
(Co-opted)

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



Toni Mathieson
Chief Executive



Laura Bell
Clinical Nurse Specialist



John Lee Taggart
Communications &
Campaigns Manager



Karen Thomas
Families Officer



Steve Neal
Project Families
Officer



Christine Jopling
Finance & Admin
Officer

6 OUR FIRST DIGITAL CONFERENCE

We are proud to introduce our milestone event, the NPUK Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases, like you have never seen it before... online and fully digital! Thanks to the WorkCast platform and the tireless efforts of the NPUK Staff Team, we are able to offer an event to the NPUK Community that encourages interaction, conversation, and fun social experiences that people have come to expect over the past 25+ years. Yes it's online, and no we won't be able to meet in-person, but trust us it's still going to be an incredibly special and informative few days.

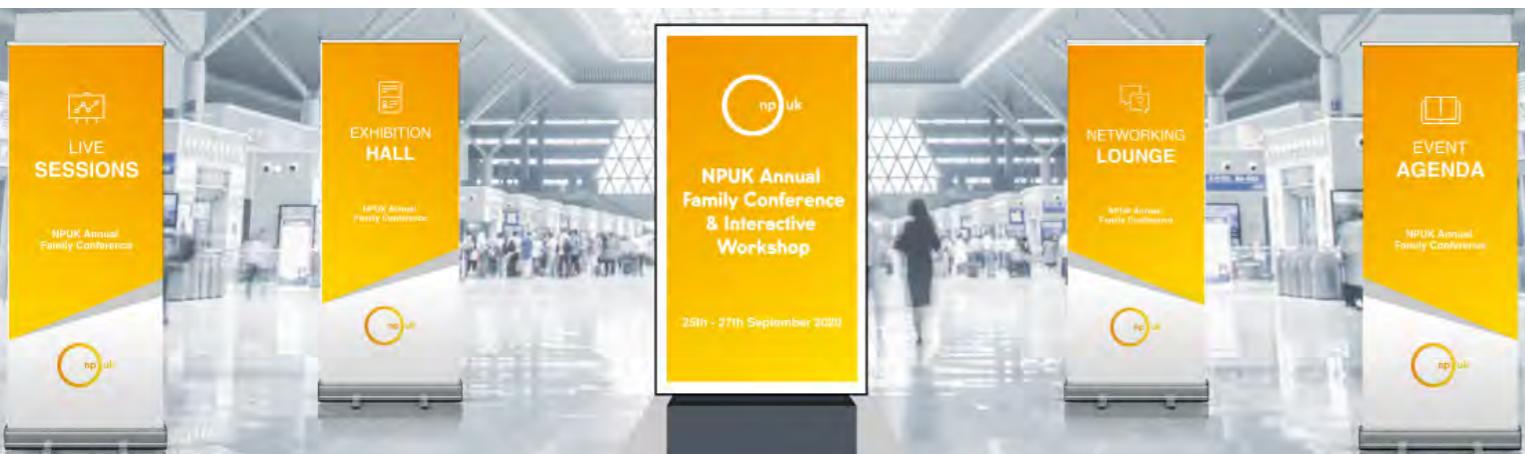
With that in mind we'd like to help you get acquainted with the fantastic WorkCast platform, which will be helping to provide this special version of our Annual Family Conference & Interactive Workshop. For those who have registered and are planning on joining us this year, this is what you will see in front of you upon arrival, the NPUK virtual lobby - the four banners in the lobby provide access to everything on offer over the course of the weekend:

The **Live Sessions** banner is where the presentations and activities on the main programme will largely be held – some videos/presentations will be shared on our social media and website, and others are restricted to the WorkCast platform “on demand”.

The **Exhibition Hall** is a place in which you can find various booths hosted by a number of advocates and associated organisations. There is an opportunity for additional resources and interaction here – think of it as sponsor booth tables at in-person conferences!

The **Networking Lounge** is a space in which you can interact with other attendees, from health professionals and clinicians working in the field of NPD to NPUK families, they can all meet here to chat and catch up with one another. It works a lot like a standard live chat.

The **Event Agenda** is where you can access the programme for the entire Annual Family Conference & Interactive Workshop weekend – check which sessions, presentations, and activities are of interest so you can properly plan the days!



AGENDA ICONS EXPLAINED



If a session/presentation has this logo next to it on the programme it will be held on the main WorkCast platform/site



If a session/presentation has this logo next to it on the programme it will be held away from WorkCast platform on the NPUK Zoom



When you spot this Teddy on the programme it means it is a Children and Young Persons' activity or event

WorkCast EXPLAINED



Digital Annual Family Conference & Interactive Workshop 2020

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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



13:30 - 15:30

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)

15:30 - 15:45

BREAK

15:45 - 17:45

Session 4: Clinical Updates - Current status of active clinical programmes for ASMD and Niemann-Pick disease type C

Featuring NPUK Community Voice - Graham Kirk

Chair: Dr. William Evans

Presenters:

Update on Cyclo Therapeutics, Inc. Clinical Trials for NPC

N. Scott Fine, Chairman and CEO, Cyclo Therapeutics, Inc.
Sharon Hrynkow PhD, Chief Scientific Officer and Senior VP Medical Affairs

Acetyl-Leucine slows disease progression in NPC

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

Tatiana Bremova-Ertl, MD, PhD Department of Neurology University Hospital Bern, Switzerland

Taylor Fields Senior Vice-President, IntraBio Inc, Oxford

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

Professional Networking Events

NPUK: "Getting to Know You" Sessions

- Lads, Dads, and Carers / Adults Living with NP-C
- Mums / ASMD (8-9pm)

9:45

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

10:00 - 11:00

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 Munkaci, Ph.D. Senior Lecturer (Assistant Professor), Victoria University of Wellington, School of Biological Sciences, New Zealand

11:00 - 11:15

BREAK

11:15 - 12:45

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. Emrys 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 Biología Molecular Severo Ochoa, Universidad Autónoma de Madrid

Andrew Munkaci, Ph.D. Senior Lecturer (Assistant Professor), Victoria University of Wellington, School of Biological Sciences, New Zealand

12:45 - 13:30

BREAK

WorkCast

WorkCast

WorkCast

18:00 - 19:00

18:00 - 20:00

19:00 - 21:00

WorkCast

WorkCast

zoom

WorkCast

zoom

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 NP diseases and all those who share our goal.

NPUK Annual Family Conference 2020

26th September 2020



DIGITAL ANNUAL FAMILY CONFERENCE & INTERACTIVE WORKSHOP 2020

WorkCast

9:45 - 10:00

Welcome and Introduction

A very warm welcome to our first digital family conference! Dr William Evans, Chair NPUK and Toni Mathieson, Chief Executive, NPUK

WorkCast

10:00 - 11:00

Parallel Sessions - please select from session 1, 2 or 3

Session 1: COVID-19 and Niemann-Pick Disease

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 Munkaci, Ph.D. Senior Lecturer (Assistant Professor), Victoria University of Wellington, School of Biological Sciences, New Zealand

zoom

Session 2: Reflections

A dedicated space for those who have been bereaved. Join us for an informal chat and to share your memories, photos and stories of your loved one.

Hosts: Fiona Dunne & Steve Neal



WorkCast

Session 3: Children and Young Persons' Activity Programme

Join us for an exciting JoJo Fun Puppet Show!

NPUK Community Voice: Christine Willman

BREAK

WorkCast

11:30 - 12:30

Parallel Sessions - please select from session 4, 5, 6 or 7:**Session 4: Understanding Niemann-Pick Diseases****Chair:**

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

Dr. Helen Waller-Evans
Senior Lecturer, School of Biosciences
Cardiff University

Andrew Munkaci, Ph.D.
Senior Lecturer (Assistant Professor)
Victoria University of Wellington
School of Biological Sciences
New Zealand

Tatiana Bremova-Ertl, MD, PhD
Department of Neurology
University Hospital Bern

**Session 5: ASMD Niemann-Pick disease type A and B;****Research and Clinical Updates****Featuring "Global Community Voice": Sara Stroer**

Chair: Melissa Wasserstein, MD
Chief, Division of Pediatric Genetic Medicine, Professor of Pediatrics and Genetics, The Children's Hospital at Montefiore Bronx, USA

Clinical Development of Olipudase Alpha for ASMD Niemann-Pick disease type B

Dr. Robin Lachmann PhD, FRCP
Consultant in Metabolic Medicine
National Hospital for Neurology
London

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

Dr Dolores Ledesma
Centro de Biología Molecular Severo Ochoa, Universidad Autónoma de Madrid

**Session 6: The Mental Health Benefits of Exercise - "How the mind navigates the body"**

Paediatric Physiotherapist - Álvaro Berzal Gómez
Counsellor and Author - Tony Somers
Personal Trainer, Josh Bridgeman
NPUK Families Officer, Steve Neal

**Session 7: Children and Young Persons' Activity Programme**
Storytime with Cora

12:30 - 13:00

BREAK

13:00 - 15:00

Parallel Sessions - please select from session 8, 9 or 10**Session 8: Improving Outcomes in Niemann-Pick Diseases**

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

Presenters:

International Niemann-Pick Disease Registry
Conan Donnelly, PhD, Registry Manager
International Niemann-Pick Disease Registry

Shaun Bolton, Operations Consultant International Niemann-Pick Disease Registry

Jackie Imrie, MSc Specialist Recruitment Consultant International Niemann-Pick Disease Registry

**An Update on Newborn Screening**

Melissa Wasserstein, MD, Professor of Pediatrics and Genetics, The Children's Hospital at Montefiore Bronx, NY, USA
Pam Crowley Andrews, Co-Founder / Executive Director Firefly Fund

Archangel Newborn Screening Review Campaign

Georgina Morton, Chairperson ArchAngel MLD Trust
Pat Roberts, Director of Newborn Screening Project, ArchAngel MLD Trust

Session 9: ASMD, ERT and Me

Join us as community members share their experience and we learn what it is like to live with Enzyme Replacement Therapy

Co-Chairs: Sandra Cowie, Director Niemann-Pick Canada, President INPDA
James Brooks, ASMD Community Member

Presenters:

ERT and Me: My Story
Maddie Collin,
UK Gaucher Association

Our Story
Sara Stroer,
ASMD Community Member

NPUK – How can we help?
Karen Thomas,
NPUK Families Officer

15:00 - 15:15

BREAK

15:15 - 16:45

Parallel Sessions - please select from session 11, 12, or 13**Session 11: NP-C Clinical Updates - Current status of active clinical programmes for Niemann-Pick disease type C**

Chair: Dr. William Evans

Global Community Voice: Debbie Kafowitz

Presenters:

Beth Solomon, MS, CCC-SLP,
Speech Language Pathology,
National Institutes of Health, USA

**Update on Cyclo Therapeutics, Inc.
Clinical Trials for NPC**

Caroline Hastings MD (Phase I trial)
Director, Fellowship Program
Clinical Director, NeuroOncology Program
Children's Hospital and Research Center Oakland and Professor Pediatrics, University of California San Francisco School of Medicine

Reena Sharma MD (Phase I/II trial), Consultant Adult Metabolic Medicine, Honorary Senior Lecturer, The Mark Holland Metabolic Unit Salford Royal Foundation NHS Trust, Salford, UK

Sharon Hrynkow PhD, Chief Scientific Officer and Senior VP Medical Affairs

IB1001-201: Multinational clinical trial investigating N-acetyl-L-leucine (IB1001) for symptomatic and neuroprotective treatment of NPC

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

Tatiana Bremova-Ertl, MD, PhD
Department of Neurology
University Hospital Bern, Switzerland



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 #NPUKAFC2020 #NPUKIW2020

CHARLIE'S SELFIE TIPS

Never have light directly above or below yourself / the subject

Try and have the camera slightly above eyeline, as it makes your face look more defined and happier

Take multiple versions from multiple angles...remember you can always delete later but it's better to have different versions to choose from

np uk Digital Annual Family Conference & Interactive Workshop 2020



NIEMANN-PICK UK AND CINEBITES PRESENT

go make memories!



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

Registered charity England and Wales (114408) and Scotland (SC045407)
Registered as a company limited by guarantee in England and Wales: 07775835

STARRING LLEYTON COOMBS EMILY STRIDE CHRISTOPHER HUGHES
BODHI RAE BREATHNACH ANDREA HALL
MUSIC BY CHRIS KÖBKE CINEMATOGRAPHER JAMES WESTLAKE
PRODUCTION DESIGNER ISA SHAW-ABULAFIA POSTER DESIGNER ROB HOTCHKISS
ANIMATION BY JAMES RICHARDSON WRITTEN BY ALEX SUTTON
EXECUTIVE PRODUCER JOHN LEE TAGGART DIRECTED & PRODUCED BY CARL MASON



Supporting those affected by Niemann-Pick

NPUK Annual Family Conference 2020

27th September 2020



10:00

Welcome and Introduction

Dr. William Evans

WorkCast

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

BREAK

11:45 - 12:45

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!)



KIDS' CORNER

1
2
3
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8
9
10



Can you spot the differences between these two pictures?

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



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!

CHILDREN & YOUNG PERSONS' VOLUNTEER TEAM LEADERS:



Jan
PATTERSON



Katie
REYNOLDS



Beth
SCOWCROFT

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)

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

alligator / ant / anteater / badger / bat / bear / bee / bird / bunny / butterfly / camel / cat / cheetah / chipmunk / cow / coyote / crab / crocodile / deer / dinosaur / dog / dolphin / donkey / duck / eagle / eel / emu / falcon / ferret / fish / flamingo / fly / fox / frog / goat / hamster / horse / kangaroo / koala / lemur / leopard / lion / lizard / lobster / mole / moose / mouse / newt / octopus / owl / panda / panther / parrot / peacock / penguin / pig / possum / puma / quail / raccoon / rat / seal / shark / shrimp / squid / swan / tiger / toad / turkey / turtle / walrus / wasp / whale / wolf / yak / zebra /

ANIMALS WORDSEARCH

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



Can you help Terry the Turtle find his shell?

Our Digital Speakers

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



Pam Andrews

Pam Andrews is Co-Founder of The Firefly Fund, a non-profit organisation she developed with her husband, Chris. Most importantly she is the proud mother of Belle & Abby Andrews (NP-C) who serve as a constant inspiration to all of us in the wider Niemann-Pick community.



Prof. Jonathan Ball

Jonathan Ball is professor of molecular virology at the University of Nottingham. His research relates to emerging viruses, viral vaccines and treatments, and blood-borne infections. He is also Director of the Centre for Global Virus Research at the University of Nottingham.



Laura Bell

As Clinical Nurse Specialist for Niemann-Pick diseases, Laura is always available to offer clinical support and advice to individuals and families in the UK and Ireland regarding any aspect of Niemann-Pick disease, plus home visits whenever necessary. She brings a wealth of experience to her role, and an unrivaled dedication to better quality of life for those she assists.



Elizabeth Berry-Kravis MD PhD

Elizabeth Berry-Kravis MD, PhD is a Professor of Pediatrics, Neurological Sciences, and Biochemistry at Rush University Medical Center in Chicago. She has run an NPC program since 2013, been Co-PI of the Vtesse phase 2/3 registration trial of VTS-270 (hydroxypropyl-beta cyclodextrin) for NPC and has been PI of an investigator-initiated expanded access program for VTS-270 through which 42 patients with NPC have been treated at 20+ sites in the USA.



Shaun Bolton

Shaun Bolton is a Clinical Research Coordinator based at the Queen Elizabeth Hospital Birmingham who is working with the International Niemann-Pick Disease Registry team. Shaun has worked in the NHS since 2013, working heavily with NPUK during that time. Shaun helps to support the INPDR as it moves into the future.



Tatiana Bremova-Ertl

Tatiana focuses on neuro-ophthalmology and neuro-otology of neurodegenerative diseases, especially inborn errors of metabolism. Through her work she aims to address the relevance of heterozygosity in these disorders by establishing novel, robust biomarkers which will potentially lead to earlier diagnosing and recognition of a rare conditions by practicing clinicians.



Josh Bridgeman

Josh is a world class strength and conditioning trainer, who was first introduced to the Niemann-Pick UK community during the Social Not Distant Campaign (during the COVID-19 lockdown). Josh volunteered his time for functional fitness sessions and has kindly agreed to join us again during our first digital Annual Family Conference!



Janice Brooks

Janice has been a supporter of NPUK for many years and a trustee for most of these. She first became involved with NPUK when her grandson was diagnosed with Niemann-Pick type B (NP-B) at three years of age. Ever since she has been working hard to further the work of NPUK in providing care and support for those affected by NPD.



James Brooks

James Brooks was diagnosed with ASMD Niemann-Pick disease type B at three years old and has been a familiar face in the Niemann-Pick UK (NPUK) community for many years alongside his dedicated family. Despite his condition James lives a full life; he recently graduated from the University of Roehampton, works a part-time job and regularly contributes to ASMD discussions for NPUK campaigns. We are thankful to have such a positive role model here at NPUK!



Claire Burbridge

Claire is a chartered psychologist specialising in health research, working as Strategic Lead at Clinical Outcomes Solutions (COS) - a health research consultancy. Claire is based in the UK and specialises Clinical Outcomes Assessments (COAs) - ways of assessing outcomes in clinical trials that are important to how a patient feels or functions. Claire has worked in this area for almost 20 years at COS and within the pharmaceutical industry, and has conducted research in a number of different disease areas. Claire led a team of researchers at COS in conducting surveys to explore the experience of living with NPC from the patient and caregiver perspective - these insights are been used to identify what is important to patients and their families.



Helen Carter

Helen's daughter, Hollie, was diagnosed with Niemann-Pick Disease Type C (NP-C) in July 2007 at two years of age. As a result of this Helen and her husband set up the Hope for Hollie (now referred to as The Hollie Foundation) campaign in 2008 to raise awareness of NP-C in our local community. Helen now serves on the Board of Trustees for NPUK and is a tirelessly dedicated advocate for our charity and indeed all families affected by Niemann-Pick disease globally.



Maddie Collin

Maddie Collin was diagnosed with Type 3 Gaucher disease in 1996 at the age of 17 months. She joined the UK Gaucher Association Board in 2015, in order to learn more about current issues facing the Gaucher community, and to represent Type 3 patients at meetings. Last year she joined the UK Gaucher Association as the charities' Project Coordinator and has recently joined the team at Aparito.



Sandra Cowie

Sandra is an adult with ASMD Niemann-Pick Disease Type B who serves as a Director of Niemann-Pick Canada and President of the International Niemann-Pick Disease Alliance Sandra currently lives and works in Toronto, Ontario, Canada. Sandy uses both her personal experiences with Niemann-Pick disease and her passion for the community to drive for an improvement to awareness and support for individuals and families across the world.



Dr. Cristin Davidson

Cristin Davidson has been part of the NPC research community for the past 15 years. Her work on NPC disease began at Colorado State University with care and study of the NPC1 cat colony. Her post-doctoral and subsequent work continues in the vein of therapeutic interventions, with efforts now directed at gene therapy for NPC1 disease. In addition to bench science, Cristin is the project manager for SOAR-NPC, a collaborative group of NPC researchers and families supporting therapy-driven projects.



Dr. Conan Donnelly

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

Fiona 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

Will 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

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

Mr. 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 Moritz School of Law and at Fordham University Law School.



Prof. Paul Gissen

Prof. 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

After a number of prestigious internships in the area of neurological physiotherapy, Álvaro 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 Guldberg

Christina Guldberg 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

Dr. 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

Sharon H. Hrynkow, Ph.D. 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

Jackie 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 Willink Metabolic 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 Kafowitz

Debbie's daughter, Rachael Kafowitz, sadly passed away on Oct. 25, 2018, after a long battle with 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. Sean received his Ph.D. in Cell and Molecular Biology from the University of Notre Dame in 2008.



Ciara & Graham Kirk

Ciara and 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 ArchAngelTrust 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 Munkaci

Dr. Andrew Munkaci 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 NPD, 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 disorder.



**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 Waddington. 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 gone 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 neurology, neuroanatomy, neuropathology, neuroradiology, and electrodiagnostic 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 Emrys 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.



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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

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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

This, our first Digital Annual Family Conference & Interactive Workshop on Niemann-Pick Diseases, is just one way in which we have attempted to change with the times by harnessing new and innovative digital technology to better support those we advocate for and represent.

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.

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

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.



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.

Till next
year...

Making a difference for the
Niemann-Pick community

npuk.org

#NPUKConf20 #NPUKFamily

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