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CONVERSATION!
#NPCONF22

24 JUNE
THE FLOREY INSTITUTE
OF NEUROSCIENCE AND MENTAL HEALTH

2022 NPC CONFERENCE

A conference for Niemann-Pick disease in Australia attended by patients, families, medical and scientific professionals.

Australian NPC Disease Foundation Inc
Research. Cure. Persevere.

cyclo therapeutics

Funded by the Australian Government Department of Social Services.

ABN: 87 767 010 514

c/- 3 Waller Street
Benalla VIC 3672
Australia

CONTENTS

ABOUT THE ANPDF

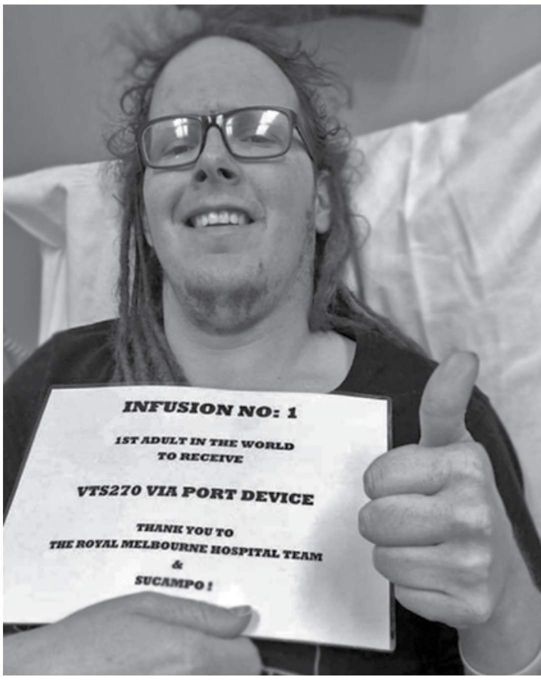
The ANPDF (Australian NPC Disease Foundation) is a not-for-profit organisation dedicated to improving the lives of Australians diagnosed with Niemann-Pick disease and their families.

Founded in 2009 by an Aussie mum with two recently diagnosed boys, this volunteer-powered charity is the culmination of parents trying to save their children from an incurable and fatal disease.

We may be small, but we strive to do it all.

Through activities that raise funds and awareness, the ANPDF supports families, funds scientific research and maintains key stakeholder relationships to ensure our Australian patients have access to as many treatment options as possible.

Each member of the ANPDF donates their time to every venture and has produced outstanding results. From our annual events (Charity Gala Dinner and Conference) to smaller one-off ventures (Glenarthon live Auction, Raffles, etc), we continue to keep our eyes peeled for every opportunity to achieve our goals to:



- **Provide an NPC-specific patient support group in Australia,** where none had previously existed. Our patient liaison officers make themselves available to all our families for support and guide the foundation towards providing relevant services where resources allow - including but not limited to the conference.
- **Advocate for Clinical Trials in Australia,** as majority of available trials don't make it to our shores. The ANPDF has been instrumental in gaining access to key treatments like Miglustat (Mar. 2010), Cyclodextrin (extended in 2017), Trappsol® Cyclo™ (current).
- **Fund scientific research for a treatment that will delay or halt Niemann-Pick disease.** We proudly support the Florey Institute of Neuroscience and Mental Health and have set clear parameters towards a potential treatment that, in its most hopeful outcome, will be deliverable to clinical trials within 8 years.
- **Raise awareness of Niemann-Pick disease to decrease diagnostic time**, thereby increasing time available for treatments. By hosting globally and nationally recognised events in Australia, we increase our platform to drive awareness within the medical community.

2022 ANPDF COMMITTEE

DEANNA CARPINO
President

Officially joining ANPDF in 2019, Deanna is inspired by her son Noah who was diagnosed with NP-C at 1yo. She launched the NPC Charity Gala in her first year and continues to drive the ANPDF forward wherever possible.

MANDY WHITECHURCH
Vice-President & Founder

Our fearless Founder continues to play a vital role as advocate and supporter of NP-C. When she isn't working and looking after her two boys with NP-C, she also represents the foundation as our Adult Patient Liaison.

CHRIS MAKULSKI
Treasurer

Our newest executive committee member, Chris is a qualified accountant kindly dedicating his time and skills to managing the ANPDF's financial requirements and guide us into the future.

PIP JOHNSTON
Secretary

As both Vet and mum of three, Pip has brought her invaluable knowledge and compassion to her roles in the foundation as both Secretary and Young Patient Liaison. Thanks Doctor Pip!

ANDREW CARPINO
Public Officer

A constant support to his wife (Deanna), Andrew joined the executive committee in 2021. Leaving no stone unturned, he brings new ideas and strategies to the ANPDF's corporate partnerships as our Corporate Liaison.

GENERAL
Committee Members

☐ Marian Shoebridge
☐ Mary Calcagno
☐ Milva Carpino
☐ Ruth Gott
☐ Sarah Mercuri, Merch Manager
☐ Vince Demarte

Interested in joining the committee?
Email us at info@npcd.org.au.

SCIENTIFIC ADVISORY
Board Members

☐ Ya Hui Hung, Chair
☐ Pip Johnston, ANPDF rep.
☐ Ashley Bush
☐ Caroline Hastings
☐ Felicity Munro
☐ Joy Lee
☐ Mark Walterfang
☐ Tina Soulis

Thank you to all our committee members for their ongoing support.

WELCOME TO THE CONFERENCE

The 2022 NPC Conference in Australia is about sharing knowledge and experiences between patient families, clinicians and researchers.

Get the latest information on upcoming Clinical Trials both in Australia and overseas.

Gain new understanding, insights and tools from those who face the same challenges as you.

Share ideas and build new relationships within our Australian Niemann-Pick Community.

EVENT PARTNER

Cyclo Therapeutics, Inc. is a clinical-stage biotechnology company dedicated to developing life-changing medicines through science and innovation for patients and families living with challenging diseases including Niemann-Pick Type C1.

Hosted by

Australian NPC Disease Foundation Inc. (ANPDF) is a not-for-profit organisation dedicated to making a positive difference to the lives of those affected by Niemann-Pick Disease.

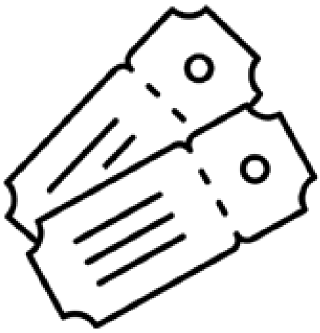
Through activities that raise funds and awareness, we are able to support patients and their families, as well as help fund scientific research towards better treatments.

Follow our journey at www.npcd.org.au

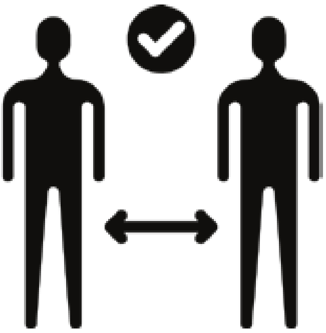
COVID-SAFE VENUE PLAN

All visitors can have the utmost confidence that our chosen venue, The Florey Institute, has developed an intelligence-led COVID-Safe Plan, in consultation with industry experts and government, that employs risk mitigation principles to ensure health and safety.

AN OVERVIEW ON HOW THE FLOREY INSTITUTE IS KEEPING YOU SAFE:

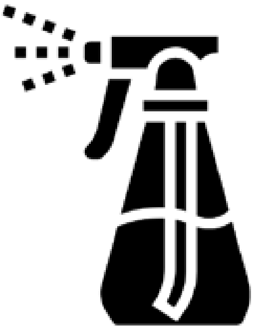


The conference is only accessible to pre-registered attendees.



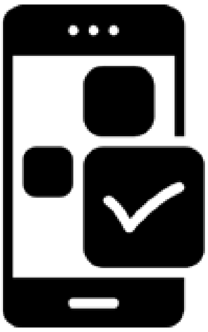
Physical distancing in conference room and break out areas to meet current distancing standards.

Sanitiser stations throughout the venue and dining areas.



Enhanced venue cleaning concentrated in high traffic areas.

Safe food and beverage service – All food is prepared under a HACCP accredited system.



Digital tickets to reduce shared contact.

Stay Safe! And enjoy the 2022 NPC Conference in Australia.

VENUE INFORMATION

The Florey Institute of Neuroscience and Mental Health is the largest brain research group in the Southern Hemisphere. With more than 600 research and support staff and educating 90 post-graduates. The conference is located in the Kenneth Myer Building, Cnr Genetics Lane & Royal Parade.

THE FLOREY INSTITUTE

Kenneth Myer Building
30 Royal Parade
Parkville, VIC 3052

t +61 3 9035 3000
e info@florey.edu.au
w florey.edu.au

PARKING

Enter via Royal Parade between Genetics Lane & Grattan Street.
Hours: 6:00am to 10:00pm.
Fees: $12.00 up to 4 hours, or $25.00 all day.

FROM THE AIRPORT

Distance	21 km / 20 min (car)
Est. Taxi fare	$50
Sky Bus fee	$19.75

PUBLIC TRANSPORT

Stations	Melbourne Central and Flagstaff Stations
Trams	Tram 19, Stop 11-University of Melbourne/Royal Pde
Taxis Silver Top	1300 976 404
Black Cabs	13 22 27
Transport info	www.ptv.vic.gov.au

NEARBY HOTELS

Naughtons Hotel	43 Royal Parade	(03) 9347 2255
Mercure	Cnr Flemington & Harker St	(03) 9329 1788
The Larwill Studio	48 Flemington Rd	(03) 9032 9111
Jasper Hotel	489 Elizabeth St	(03) 8327 2777

GENERAL INFORMATION

CATERING

All day meals will be provided including lunch and tea breaks. Coffee and tea will be available all day.

If you have advised any special dietary requirements on your registration form, these will catered to on the day. Please ask the catering or support staff to find the correct meal option.

MOBILE PHONES

Please respect the presenters and other members of the audience by ensuring your phone is switched off or set to silent whilst in session.

DISABILITY ACCESS

Access is enabled and available at all entrances and elevators.

Disabled toilets will also be located on the same floor as the conference.

SPEAKER CONTACT DETAILS

We understand there may not be time to answer every question or query within each presentation's timeslot. If your question was unanswered during the conference, we invite you to follow up with our speakers directly at the below email addresses.

CAROLINE HASTINGS	Caroline.Hastings@ucsf.edu
CONAN DONNELLY	Conan.Donnelly@inpdr.org
FIONA HELLIER	dsa@dementia.com.au
FRANK PFRIEGER	fw-pfrieger@gmx.de
GAIL HILTON	gail@childhooddementia.org
JACKIE IMRIE	Jackie.Imrie@inpdr.org
JOY LEE	Joy.Lee@rch.org.au
LOUISE HEALY	education@rarevoices.org.au
MARC PATTERSON	Patterson.Marc@mayo.edu
MARK WALTERFANG	Mark.Walterfang@mh.org.au
SEAN HOSKING	sean.hosking@mh.org.au
YA HUI HUNG	yahui.hung@florey.edu.au

Alternatively, you may contact the conference organisers via email: info@npcd.org.au

SPEAKERS

Discover more about Niemann-Pick Disease type C (NPC) and the people it impacts with our speakers.

FRANK PFRIEGER
University of Strasbourg

MARC PATTERSON
Mayo Clinic

CAROLINE HASTINGS
UCSF Benioff Children's Hosp.

SEAN HOSKING
Royal Melbourne Hospital

MARK WALTERFANG
Royal Melbourne Hospital

YA HUI HUNG
The Florey Institute

FIONA HELLIER
Dementia Support Australia

JACKIE IMRIE
INPDR

CONAN DONNELLY
INPDR

GAIL HILTON
Childhood Dementia Initiative

LOUISE HEALY
Rare Voices Australia

JOY LEE
Royal Children's Hospital

Mistress of Ceremonies
FELICITY MUNRO

SCHEDULE OF TALKS

8:30 AM	***CONFERENCE CHECK-IN, TEA & COFFEE***
9:00 AM	**Conference opening and Welcome to country** **Felicity Munro**, ANPDF Committee Member, Aust.
9:10 AM	**Niemann-Pick Type C Disease: Past, present and future** **Frank Pfrieger** PhD – Tenured Scientist, Institute of Cellular and Integrative Neurosciences, France
9:50 AM	**Updates on clinical trials in Niemann-Pick disease, type C** **Marc Patterson** MD FRACP FAAN FANA – Professor of Neurology, Pediatrics and Medical Genetics, Mayo Clinic, USA
10:20 AM	***MORNING TEA BREAK***
10:35 AM	**Cyclo Therapeutics' Development Program** **Caroline Hastings** MD – Director of fellowship program in pediatric hematology and oncology, UCSF Benioff Children's Hospital, USA
11:20 AM	**Behind the Scenes: A look at the preparation and set up of clinical trials** **Sean Hosking** – Clinical Trials Coordinator – Royal Melbourne Hospital, Aust.
11:45 AM	**Royal Melbourne Hospital Clinical Research** **Mark Walterfang** MBBS (Hons) PhD FRANZCP – Professor of Neuropsychiatry, The University of Melbourne; Consultant Neuropsychiatrist, The Royal Melbourne Hospital, Aust.
12:15 PM	**The Florey Institute Research** **Ya Hui Hung** BMus BSc (Hons) PhD – Senior Research Officer, The Florey Institute of Neuroscience and Mental Health, Aust.
12:45 PM	***LUNCH + NETWORKING***
1:45 PM	**Dementia Support Australia** **Fiona Hellier** – Team Leader – Dementia Support Australia, Aust.
2:15 PM	**International Niemann-Pick Disease Registry** **Jackie Imrie** MSc, Clinical Research Manager and **Conan Donnelly** PhD, CEO, International Niemann-Pick Disease Registry, UK
2:45 PM	**Childhood Dementia Initiative** **Gail Hilton** – Project Manager, Childhood Dementia Initiative, Aust.
3:15 PM	**Rare Voices Australia** **Louise Healy** – Education and Advocacy Manager, Rare Voices Australia
3:45 PM	***AFTERNOON TEA***
4:00 PM	**Live Q&A with Frank Pfrieger**
4:10 PM	**Current Australian Clinical Experience** **Mark Walterfang** and **Joy Lee** MD FRACP – Metabolic Consultant, Royal Children's Hosp., Aust.
4:40 PM	**National Framework for NP-C care and management workshop**
5:40PM	**Conference Closing** **Deanna Carpino** – ANPDF President, Aust.

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Soroptimist International of Melbourne, Inc.

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National Disability Conference Initiative Grant funded by the Australian Government Department of Social Services.

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The Australian NPC Disease Foundation thanks our Community Partners for their ongoing support - without you all, this work would not be possible.

JOIN THE CONVERSATION

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Australian NPC Disease Foundation Inc

Research. Cure. Persevere.

STAY CONNECTED

with Australian NPC Disease Foundation Inc

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- @NiemannPickOz
- /ANPDF

Find us online at
www.npcd.org.au

KEYNOTE LECTURE

NIEMANN-PICK TYPE C DISEASE: PAST, PRESENT AND FUTURE

My presentation will provide background information about the disease. To this end, I will outline the somewhat tortuous history of its discovery, summarize selected aspects of current research and provide an outlook on the development of new diagnostic and therapeutic approaches.

Frank W. Pfrieger, PhD
Institute of Cellular and Integrative Neurosciences.
Centre National de la Recherche Scientifique,
University of Strasbourg.
67000 Strasbourg, France.

Frank is working as tenured scientist at the Institute of Cellular and Integrative Neurosciences affiliated with the French Centre National de la Recherche Scientifique and the University of Strasbourg (France).

He studied biology at the University of Konstanz (Germany) and obtained his PhD in 1994 from the same university after experimental work with H.D. Lux at the Max-Planck Institute for Psychiatry in Martinsried (Germany).

From 1994 to 1997, he worked as postdoc with B.A. Barres at Stanford University (CA, USA). In 1997, he was recruited as independent group leader to the Max-Delbrück Center for Molecular Medicine in Berlin-Buch.

In 2001, he moved to Strasbourg as group leader of the German Max-Planck Society in a bilateral cooperation with the CNRS. Since 2005, he holds his current position.

His research aims to understand the mechanisms of neurodegeneration in NPC disease and to develop new diagnostic and therapeutic approaches.

NOTES

Marc C. Patterson MD FRACP FAAN FANA
Mayo Clinic Children's Center, USA

Marc Patterson was born and educated in Australia, and trained in neurology, child neurology and neurometabolic disease at the University of Queensland in Australia, at Mayo Clinic, and at NINDS/NIH. He is currently Professor of Neurology, Pediatrics and Medical Genetics at Mayo Clinic.

He was Director of the Child Neurology Training program at Mayo 2008-2016), and Chair of the Division of Child and Adolescent Neurology (2008-2017). Dr Patterson had previously served as Professor and Director of Pediatric Neurology at Columbia University in New York (2001-2007).

Dr Patterson is currently an Editor for the Journal of Inherited Metabolic Disease, and JIMD Reports, Editor-in Chief of the Journal of Child Neurology and Advisory Editor of Child Neurology Open.

His research and practice have focused on rare diseases in children, including multiple sclerosis and neurometabolic disorders in general, with special interests in Niemann-Pick disease, type C (NPC), other lysosomal diseases, and congenital disorders of glycosylation, areas in which he has published more than 300 peer-reviewed papers and book chapters.

He serves on the scientific advisory boards of several rare disease foundations. Dr Patterson has received funding support from NIH, industry, and private foundations.

UPDATE ON CLINICAL TRIALS IN NIEMANN-PICK DISEASE, TYPE C

There are currently several clinical trials, or expanded access programs, available to patients with Niemann-Pick disease, type C. The most recent addition to these, is a double-blind, randomized, placebo controlled, multi center International trial of intravenous hydroxypropyl-beta-cyclodextrin, for which there will be a separate presentation [NCT04860960].

This trial, and all but one of the remainder, are designed to ameliorate the clinical course of Niemann-Pick disease, type C. The exception is the planned trial of N-acetyl-L-leucine, a randomized, double-blind, crossover, placebo controlled trial, whose primary outcome measure will be ataxia [NCT05163288].

NOTES

CLINICAL TRIALS AND RESEARCH

INTRAVENOUS TRAPPSOL® CYCLO™ IN PATIENTS WITH NIEMANN PICK DISEASE TYPE C1: UPDATES ON THE RESULTS FROM PHASE 1 AND PHASE 1-2 STUDIES AND LAUNCH OF THE INTERNATIONAL PHASE 3 PIVOTAL TRANSPORT NPC TRIAL

CURRENTLY ENROLLING

Niemann-Pick Disease Type C1 (NPC1) is a disorder of intracellular cholesterol and lipid trafficking that leads to the accumulation of cholesterol and lipids in the late endosomal/lysosomal compartment, resulting in systemic manifestations (including hepatosplenomegaly and lung infiltration) and neurodegeneration.Preclinical studies have demonstrated that systemically administered 2-hydroxypropyl-β-cyclodextrin (HPβCD; Trappsol® Cyclo™) restores cholesterol metabolism and homeostasis in peripheral organs and tissues and in the central nervous system (CNS).

A Phase I clinical trial assessed the safety, pharmacokinetics, and pharmacodynamics of 2 doses of HPβCD in peripheral tissues and the CNS in adult subjects with NPC1. A Phase 1-2 trial continued to assess these parameters using 3 dose levels in children and adults, and as well evaluated early clinical effects over a 48-week period. Detailed results from these trials will be presented which demonstrate an excellent safety profile and support the mechanism of action of systemically administered HPβCD in mobilizing intracellular cholesterol stores in subjects with NPC1 as demonstrated in previous preclinical studies.

These data support the need to test these encouraging results in long term studies and with precise clinical assessment tools. The background, eligibility, and methodology of the multi-center, double-blind, randomized, placebo-controlled Transport NPC Phase 3 international trial, currently enrolling, will be presented.

NOTES

Caroline Hastings MD

Director, Pediatric Hematology Oncology Fellowship Program Children's Hospital & Research Center Oakland; Professor of Pediatrics, University of California, San Francisco, School of Medicine, USA

Caroline Hastings received her M.D. from the University of California, Davis in 1986 and subsequently completed her Pediatric residency and fellowship in Pediatric Hematology and Oncology at Children's Hospital & Research Center Oakland, California. She is on the faculty at UCSF School of Medicine and Benioff Children's Hospital Oakland, and serves as the Director of the Pediatric Hematology/Oncology Fellowship Program and clinical site Director of the Pediatric NeuroOncology program.

Her academic interests include clinical trial development in high risk and relapsed extramedullary leukemia, brain and spinal cord tumors in infants and children, and rare metabolic diseases including Niemann-Pick Type C disease.

She was the first physician in the US to develop and use hydroxypropyl-β-cyclodextrin in children with NPC and developed the first FDA approved compassionate use protocol. Building on her experience with compassionate use of cyclodextrins, Dr. Hastings served as the Principal Investigator for a US phase I clinical trial supported by CTDH to test intravenous cyclodextrins formally in NPC. She also served as a Senior Clinical Advisor to CTD's EU/Israel phase I/II clinical trial, Co-Chair of CTD's Family and Physicians Listening Circle, and member of CTD's Scientific Advisory Board.

Dr. Hastings is currently conducting clinical trials (inclusive of gene therapy) in patients with Gangliosidosis Type I and Metachromatic Leukodystrophy, as well as natural history and registry studies in rare disease.

Dr. Hastings is involved with advocacy on a national and regional level and serves on the Executive Board of Trustees for the American Society of Pediatric Hematology Oncology and is the President elect of the society. She leads awareness and philanthropic events and has personally raised several million dollars to support local programs in research and clinical care as well as pediatric clinical trials in rare disease.

Dr. Hastings enjoys spending time with her husband and three daughters, traveling, all animals, reading about sociology and economics and creating the perfect garden.

CLINICAL TRIALS AND RESEARCH

BEHIND THE SCENES: A LOOK AT THE PREPARATION AND SET UP OF CLINICAL TRIALS

The running of a sponsored clinical trial is heavily dependent on the painstaking preparations made from everyone involved from the sponsor to the patient and all those in between. My presentation will focus on the process completed by the research team at the institute level. Prior to institute involvement – I will give a brief overview of what is involved in the preparation of the Protocol and Investigator Brochure.

- Site Selection – Choosing a site requires a clear and calculated feasibility process. I will explain how an institute is chosen and how an Investigator determines the capability of their team.
- Ethics – The most important part of all research is operating in the realm of Human Ethics.
- Governance – Once main site is identified and the HREC has been submitted we are still faced with the great task of submitting Research Governance for each of the institutes.
- Patient Selection and recruitment – Here we will look at the process in which a patient is chosen and the efforts taken in navigating recruitment criteria.
- Running the trial – It is only after all these processes that we actually get to the running of the research project.
- Coming to an end – Through hard work and years of effort we come to the end of a trial. I will show the processes necessary to close the trial out.

In conclusion, using examples of the patients I will reflect upon the relationship we have had with those effected by NPC and give a brief insight in to the journey we all took together.

Sean Hosking
Royal Melbourne Hospital, Australia

I started my medical career in the Australian Defence force in 1999. Trained as a field medic then transferred to nursing. Left the army and completed my Nursing in 2004. Worked in Orthopaedic surgery and urology for 6 years.

After being trained in managing clinical trials in a ward setting I developed a love of research. I applied for the position of Epilepsy research coordinator in June of 2010 and was interviewed by the head of the department Professor Terence O'Brien.

I began working on clinical trials in Epilepsy and other neurological conditions that year. In 2016, I met Professor Mark Walterfang when he approached me about a new and exciting trial for NPC. We began working together closely on this very long trial and have been managing NPC research ever since.

NOTES

Mark Walterfang MBBS (Hons) PhD FRANZCP
Royal Melbourne Hospital, Australia

Professor Mark Walterfang has worked as a consultant neuropsychiatrist at the Royal Melbourne Hospital for more than 20 years, having completed specialist training at Neuropsychiatry, RMH, and his PhD at the Melbourne Neuropsychiatry Centre, focusing on neuroimaging.

He is completing a second research doctorate in Niemann-Pick type C disease, having managed adolescent and adult patients with this illness at Neuropsychiatry since 2000.

AN UPDATE ON CLINICAL RESEARCH INTO NPC IN AUSTRALIA

Over the last 15 years, patients with NPC have been increasingly involved in clinical research and clinical trials in Australia. In this presentation we will highlight some past, recent and ongoing research into NPC in Australia, particularly with patients of the Royal Melbourne Hospital and its collaborators.

We will highlight research examining brain changes in NPC and the effect of medication on these changes; additionally, we will also describe a range of other clinical studies across a range of health disciplines that looked to further elucidate changes in organ systems in NPC and how these changes could be used to track illness and treatment.

NOTES

CLINICAL TRIALS AND RESEARCH

MESSAGE IN A FAT BUBBLE: AN MRNA GENE THERAPY FOR NIEMANN-PICK DISEASE TYPE C1

Pathological mutations in the NPC1 gene result in the fatal neurodegenerative disorder Niemann-Pick Disease Type C1 (NP-C1).

Messenger RNA (mRNA) gene therapy is an emerging approach to restore functional genes. This presentation will provide an update on the NPC1 mRNA gene therapy being developed at the Florey.

NOTES

Ya Hui Hung PhD
The Florey Institute of Neuroscience and Mental Health, Australia

Ya Hui Hung is a molecular biologist/ biochemist at the Florey Institute of Neuroscience and Mental Health.

Her interest in rare genetic diseases began with her PhD studies on Menkes disease and copper metabolism at the University of Melbourne (Australia). Her research interest in Niemann-Pick Disease Type C (NP-C) evolved from her early postdoctoral studies investigating the relationship between copper and cholesterol metabolism in the development of neurodegeneration and Alzheimer's disease.

Her NP-C research has contributed to mapping of the metallobiology landscape in NP-C, and identified early neurological changes in the heterozygous Npc1 mice that may have clinical implications for human NPC1 carriers.

She is currently exploring new therapeutic options to treat NP-C, with a particular interest in mRNA-based gene therapy.

NIEMANN-PICK DISEASE TYPE C

#NPCONF2022 #NC2022

A rare, fatal and progressive genetic disorder affecting...
people.

DEMENTIA SUPPORT AUSTRALIA; PARTNERING TO SUPPORT YOUNG PEOPLE LIVING WITH A DEMENTIA DIAGNOSIS

Dementia Support Australia (DSA) are a national service that is locally based and provide tailored personal advice to carers of people with dementia where behaviours may be impacting negatively on the carer and/or the person with dementia.

This presentation provides a brief overview of the work the dementia consultants have done with families with a child with dementia, how to access the service and what support is available. A case study will be discussed.

NOTES

Fiona Hellier
Dementia Support Australia

I am an experienced Registered Nurse having worked in the hospital & health care industry for many years. I have worked across the acute healthcare sector, community nursing and aged care in clinical and management roles in regional and metropolitan areas.

I am currently completing my Masters of Gerontology through Flinders University with a focus on dementia care. I have been working with Dementia Support Australia for over four years and manage a team of carers who work alongside staff and community carers modeling and coaching approaches and engagement to assist in supporting with behaviour management.

I am part of the DSA Childhood Dementia Team and have completed the training put together with the assistance of BDSRA and other professionals. I have supported several families with children living with dementia to date.

ADVANCES IN THE NIEMANN-PICK PATIENT-LED INTERNATIONAL REGISTRY

The International Niemann-Pick Disease Registry is a web-based, patient-led, independent registry for the collection of clinical and patient-reported from Niemann-Pick Disease patients. The INPDR is a community resource to illuminate the natural history of NPD and improve outcomes for patients and their families.

Over the last year the INPDR has expanded into new countries, developed enhanced quality systems, extended data collection across both the clinical and patient-reported database and supported and published research. we will present this progress and give examples of how the registry can have an impact on the families affected by Niemann-Pick disease.

We will be inviting families to participate in the registry and show you how you can join and contribute to this global movement.

Jackie Imrie MSc

International Niemann-Pick Disease Registry, UK

Having qualified as a paediatric nurse I obtained a Degree in Biochemistry and Genetics and MSc Genetic Counselling. I worked at the Willink Metabolic unit Manchester for 20 years seeing patients with many inborn errors of metabolism.

In 1999 I started as the first Niemann-Pick nurse, funded by NPUK, setting up the national database and supporting families and professionals.

Having retired from full time work for NPUK, I did Consultancy work in the field of NPDs for over 10 years, which included raising awareness to aid quicker diagnosis and contributing to scientific and clinical papers. I am currently leading recruitment as Clinical Research Manager with INPDR. I am also a Trustee for NPUK and MLD support association UK.

Conan Donnelly

International Niemann-Pick Disease Registry, UK

Conan Donnelly is an epidemiologist and has been Chief Executive of the International Niemann-Pick Disease Registry since 2020.

He has worked in disease registration for over ten years now and has seen firsthand the importance of patient-reported and clinical data in improving patient outcomes.

Conan is currently leading the registry through some exciting changes including the rollout of an expanded patient-reported database, global expansion of the clinical registry and facilitating several research studies to improve the care and outcomes of patients and families affected by Niemann-Pick Disease.

COMMUNITY CONNECTIONS

CHILDHOOD DEMENTIA - THE PROGRESS SO FAR

The Childhood Dementia Initiative started in November 2020 in Sydney. It is the first organisation in the world representing more than 70 rare genetic conditions that cause dementia in children. The organisation was set up by Megan Donnell, the former Founder and CEO of Sanfilippo Children's Foundation with three major goals:

- to increase awareness and advocacy,
- to accelerate therapeutic development
- and to improve care and quality of life.

Gail will provide an overview of childhood dementia, including the definition and statistics. The presentation will then explore the goals and objectives of the Childhood Dementia Initiative and will look at the progress of the last 18 months since the organisation began. Gail will also provide a look forward and discuss ways in which families and researchers can be involved with projects now and in the future.

NOTES

Gail Hilton
Childhood Dementia Initiative, Australia

With a masters degree in Development Studies and undergraduate degree in Biology, Gail has more than 15 years of experience in managing projects and delivering services within the paediatric, adolescent and young adult health sector.

After many years in the oncology space, Gail has been involved with childhood dementia as a health issue for three years – first at Sanfilippo Children's Foundation and since its inception as Head of Programs – Care & Quality of Life at the Childhood Dementia Initiative.

Gail is responsible for all connections with families and health and social care professionals and is leading a number of projects to build the evidence base around the needs of families impacted by childhood dementia across Australia in order to drive improvements in care.

Louise Healy
Rare Voices Australia, Australia

Louise has post-graduate qualifications in psychology and has managed a consulting business for the past seven years. Her connection with rare conditions began when her first child was born with a rare metabolic disorder.

Louise has been involved in rare disease support and advocacy for over 10 years and she is the current Vice President of the Metabolic Dietary Disorders Association (MDDA). She has led successful advocacy campaigns for access to medicines and support programs for people with rare disorders of protein metabolism.

Louise is a previous RVA Board member, current member of the Queensland Genomics Community Advisory Board and a founding board member of the Global Association for PKU.

NEWBORN BLOODSPOT SCREENING

Newborn Bloodspot Screening is a successful public health program that has been offered to all babies in Australia for over 50 years. Most conditions screened for are rare and many people will never have heard of them until they receive a positive newborn screening result.

Over recent years policy attention has turned to how to best ensure that this important program, currently implemented by state health systems remains current, equitable, safe and consistent.

This presentation will cover the role of newborn screening, the importance of embedding screening into essential care and support systems for families, population screening principles and how researchers and newborn screening specialists are investigating what role genomics may play in future screening.

NOTES

WORKSHOP

#NPCONF2022
#NC2022

NATIONAL FRAMEWORK FOR NP-C CARE AND MANAGEMENT

led by Prof. Mark Walterfang and Dr. Joy Lee

Beginning with an overview of what's currently available in Australia and access to treatment, this session will primarily aim to create an opportunity for feedback from key stakeholders, patients and families. This will be step one and result in setting the agenda, timeline and key parameters of the project moving forward.

FRAMEWORK WORKSHOP

NOTES

Australian NPC Disease Foundation Inc

Research. Cure. Persevere.

AUSTRALIAN NPC
DISEASE FOUNDATION
c/o 3 Waller Street
Benalla, VIC Australia

Email
info@npcd.org.au
Website
www.npcd.org.au