

# Niemann-Pick Disease Group (UK) Annual Report 2014 -2015



[www.niemann-pick.org.uk](http://www.niemann-pick.org.uk)

# Contents

<b>3</b>	Reference and Administration	<b>15</b>	Information and Awareness
<b>4</b>	Trustees' Report	<b>16</b>	Think Again. Think NPC Campaign
<b>6</b>	Structure and Governance	<b>17</b>	Supporting and Influencing Research
<b>7</b>	Aims and Objectives	<b>18</b>	The International Niemann-Pick Disease Registry (INPDR)
<b>8</b>	What is Niemann-Pick Disease?	<b>19, 20</b>	National and International Collaboration
<b>9</b>	Research and Treatment	<b>21, 22</b>	Fundraising
<b>10</b>	Expert Care for Niemann-Pick Disease Patients	<b>23</b>	Our Objectives 2015-16
<b>11, 12, 13</b>	Our Activities	<b>24, 25, 26</b>	Financial Review
<b>14</b>	Annual Family Conference 2014		



# Reference & Administration

**Registered Charity Number:**  
**England and Wales: 1144406**  
**Scotland: SCO45407**

**NP-UK Registered Address:**

Suite 2, Vermont House  
Concord, Washington,  
Tyne and Wear  
NE37 2SQ

Telephone Helpline: 0191 415 0693

E-mail: [info@niemann-pick.org.uk](mailto:info@niemann-pick.org.uk)

Website: [www.niemann-pick.org.uk](http://www.niemann-pick.org.uk)

**Charity Trustees / Board of Directors**

David Roberts (Chairman)  
David Holton (Treasurer)  
William Owen  
Richard Rogerson  
Jim Green  
Prof. Frances Platt (Co-opted)

Miriam Evans (Co-opted)  
Janice Brooks  
Helen Carter  
William Evans  
Stefanie Rymza (Co-opted)

**Company Limited by Guarantee:**  
**Registered in England and Wales No: 07775835**  
**Company Secretary:** Mrs Toni Mathieson

**Auditors:**

James Anderson & Co,  
Chartered Accountants,  
Pentland Estate, Straiton,  
Edinburgh, EH20 9QH

**Bankers:**

HSBC, Grays Branch  
53, High Street, Grays, Essex  
RM17 6NH

**Medical Advisor:**

Dr Simon Jones  
Consultant in Paediatric Inherited Metabolic Disease  
St Mary's Hospital,  
Manchester Academic Health Sciences Centre  
Central Manchester University Hospitals NHS Foundation Trust



# Trustees' Report

## David Roberts, Chair, NP-UK



This year's Annual Report highlights the many ways in which NP-UK has made a positive difference to those affected by Niemann-Pick diseases.

Over the year we have fulfilled our key objectives and have made significant progress in bringing about the possibility of clinical trials in the UK for Niemann-Pick type C.

The work we do has a tremendous impact on the Niemann-Pick community and our achievements are described throughout this report. There are also many examples of how we fulfil our objective of providing much-needed advice and support to patients, families and the wider Niemann-Pick community. Our Clinical Nurse Specialist, Families Officer and dedicated Central Office team continue to provide a high quality service. Our many volunteers provide much needed assistance in delivering this service and the help and support that we receive, throughout the year, is very much appreciated.

I am pleased to report that our care and support service has now been expanded to include individual support for siblings and a project to help families improve emotional resilience is now embedded in the way we assist families in coping with the day to day challenges of looking after a loved one affected by this disease.

Next year we will build on our success and examine ways in which we can add capacity in order to meet the needs of the increasing number of adults who are affected by these diseases and those families who may not be aware of the services we can offer. In addition, this will help to improve the reach and flexibility of the services we provide.

Facilitating research continues to be at the heart of all we do and we continue to support the University of Oxford and Cardiff University by encouraging basic research into Niemann-Pick type C. During the year we were pleased to be able to extend our grant support to include University College London. Our collaboration with the International Niemann-Pick Disease Alliance enables us to have a voice in the international research community and this year we have been able to take part in discussions to bring about the possibility of clinical trials in the UK for Niemann-Pick type C. You can read more about these in the pages that follow along with the progress being made with the International Niemann-Pick Disease Registry project.

Increasingly, patients and their families have taken part in studies and questionnaires to provide information that may be useful in establishing the efficacy of new therapies. Some examples over the year include; wearable technology, the EyeSee Cam which measures eye movement relative to head movement and questionnaires that help gather valuable patient reported outcomes. We are extremely grateful to all those families who have taken part in these programmes, both over the year and in the future.

Our Executive Director has been at the forefront of our strategic commitment to collaborate with expert advisory groups, rare disease alliances and other lysosomal storage disease groups. Engaging with these groups, which include the LSD Patient Collaborative, Genetic Alliance UK, Rare Disease UK and the Specialised Healthcare Alliance, keeps us at the forefront of policy making and enables us to influence rare disease policy and the future of Highly Specialised Commissioning for rare diseases such as ours. This work is tremendously important and ensures we have a voice to improve the clinical standards of care for those affected by lysosomal storage diseases; it also provides a platform to campaign for equal access to drugs and therapies provided by the NHS.

As a small Charity, we could not achieve as much without the dedication and support of our Trustees. In addition to exercising overall control of the Charity they continue to create awareness of Niemann-Pick diseases by representing the charity at national and international events. Our Trustees are held in high regard by all those who seek their support and they frequently give up their time to take part in high level advisory group meetings and conferences, where they share their knowledge and skills via presentations, consultancy and through the promotion of awareness campaigns.

Looking ahead, we have defined our objectives for 2015/2016 over the following pages and these take into account the areas for improvement as well as strengthening our organisation to meet the continuing demands of the future. We rely solely on fundraising, donations and grants for our continuing work; the Trustees would like to thank all those who choose to support our Charity – including the many families, friends and volunteers - for their amazing fundraising achievements and donations. Your continuing support helps to provide our care and support service and contributes towards grants for research projects. We remain extremely grateful for your contribution to our success as a Charity.

We are also very grateful to the grant giving organisations and pharmaceutical companies who provide unrestricted educational grants for the benefit of families affected by Niemann-Pick diseases. Working together, as part of the wider Niemann-Pick community, ensures that we have the resources, skills and commitment to really make a positive difference to those affected by these devastating diseases.

*David Roberts*

*David Roberts , Chairman, NP-UK*





# Niemann-Pick Disease Group (UK)

## Structure and Governance

Niemann-Pick Disease Group (UK) (NP-UK) is a charitable organisation offering support and information to families affected by Niemann-Pick diseases and to associated professionals.

### ✓ History

The Charity was formed by a small group of parents in 1991 and in 1997 was granted charitable status as an independent charity, registered with the Charity Commission for England and Wales as an unincorporated association, registered charity number 1061881.

In January 2012, whilst remaining a registered charity, NP-UK became a charitable company limited by guarantee registered in England and Wales, company number 07775835, registered charity number 1144406.

### ✓ Organisational Structure

The Board of Trustees, all of whom are volunteers, primarily consist of people who have family members or friends directly affected by this group of diseases.

The Trustees meet at least three times a year and are responsible for all of the activities the Charity undertakes, with operational assistance from a number of Sub-Committees.

### ✓ Governance

NP-UK is governed by its Memorandum and Articles of Association, and is managed by a Board of Trustees, who for the purposes of company law, are also the Directors of the Organisation.

NP-UK conforms to the requirements of both Companies House and the Charity Commission for England and Wales and has adopted the provisions of the Statement of Recommended Practice (SORP) 'Accounting and Reporting by Charities' issued in March 2005. In all activities, the Trustees show due regard to the Charity Commission's guidance on public benefit.

### ✓ Risk Management

The Board of Trustees have considered and reviewed the possible risks to which the Charity may be exposed, and have established and implemented a risk management strategy, comprising a quarterly risk review and procedures to identify, mitigate and minimize the impact of risks to which the organization may be exposed.



# Niemann-Pick Disease Group (UK)

## Aims and Objectives

*The main aims and objectives of NP-UK are; to make a positive difference to the lives of those affected by Niemann-Pick diseases (NPD), relieve sickness and any distress which may arise there from, and to advance the education and awareness of families, professionals and the general public in all matters concerning the disease. NP-UK aims to meet these objectives through:*

- ✓ **The provision of a support and advice service for families and individuals and associated health and social care professionals**
- ✓ **The collection, collation and dissemination of relevant information**
- ✓ **The provision of support for national clinical centres of expertise for Niemann-Pick diseases**
- ✓ **The promotion of relevant research**
- ✓ **The promotion of national and international collaboration**

Through our objectives, we aim to increase awareness of Niemann-Pick disease amongst health professionals and the wider general public, to shorten time to diagnosis and enable research into effective therapies, ensuring that each affected individual has access to the best available treatment, support and information appropriate to their needs.

To ensure our objectives support the needs and wishes of those we support, we undertake regular consultation with our members, supporters and stakeholders. Our goal is to ensure the continuation, and the successful delivery, of the services we provide, whilst seeking sustainable expansion.

To achieve our objectives, we rely on:

- Funding from voluntary donations, corporate donors, charitable trusts and statutory bodies
- Our staff team, who provide our core support, information and advocacy services and raise much needed awareness of the social and economic challenges faced by those affected by Niemann-Pick diseases
- Most importantly, we would not exist without the fundraising support of our members, friends and families; their efforts are vital in assisting the achievement of our aims and objectives.

# What is Niemann-Pick Disease?

Niemann-Pick diseases are a group of rare, inherited, metabolic conditions that can affect children and adults. These conditions are caused by specific genetic mutations and are pan-ethnic. The commonly recognised forms of the disease are:

**Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick Disease or ASMD Niemann-Pick Types A & B**, rather than being two separate 'types' of the disease, actually represent the opposite ends of a spectrum of the same disease, both caused by a deficiency of the enzyme Acid Sphingomyelinase. Many variations exist within this spectrum, in terms of clinical symptoms and rate of progression.

## **ASMD Niemann-Pick Disease Type A (NP-A)**

ASMD NP-A is a rapidly progressive neurological disease that usually reveals itself within the first few months of a baby's life. Symptoms may include early feeding difficulties, failure to thrive and an abnormally large abdomen. Life expectancy rarely exceeds five years of age.

## **ASMD Niemann-Pick Disease Type B (NP-B)**

In ASMD NP-B there is generally little if any neurological involvement. Symptoms can include an enlarged liver and spleen, delayed puberty, susceptibility to respiratory infections and increased stress on the heart. Most patients will survive into their teens and adulthood.

**Niemann-Pick Disease Type C** is not caused by an enzyme deficiency, but the end result is the same; an accumulation of materials (lipids and other fatty acids) in the body's cells.

The presentation of NP-C is very variable and the onset of symptoms may occur at any time from early infancy to adulthood.

Symptoms of NP-C may include an enlarged spleen and liver and, in newborn babies, there may be prolonged jaundice. Speech can become slurred and swallowing problems may develop.

A symptom that is particularly suggestive of NP-C is difficulty with upward and downward eye movement. In those young adults, where onset is later, psychological problems and dementia can be major symptoms.

The disease is neurologically degenerative leading to progressive loss of motor skills and difficulty with walking. Patients may experience sudden loss of muscle tone, which can lead to falls, also epileptic seizures that are generally difficult to control.

## **Is there a Cure?**

At present, there is no cure for the Niemann-Pick diseases, although there is considerable research activity taking place around the world. Currently, those affected may benefit from supportive treatments - individual medication to treat the symptoms of the disease.

# Research and Treatment

There is no cure for the Niemann-Pick diseases. Over the years NP-UK has built a strong working knowledge in the field of NPD research. By developing and sustaining robust relationships with key figures and organisations across the world, we are able to keep up to date and to share information with our community in a timely way. In recent years, advances in characterisation and understanding of NPD has led to a number of possible therapeutic targets being identified:

## **Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick Disease Type B (ASMD NP-B)**

Genzyme, a Sanofi company, is currently undertaking a Phase 1b clinical trial in ASMD NP-B patients to evaluate the safety and tolerability of an investigational enzyme replacement therapy recombinant human acid sphingomyelinase (rhASM). Five adult patients are enrolled in the trial at two study centers, Mount Sinai in New York, NY, US, and St. Mary's Hospital in Manchester, UK.

## **Facilitating Progress**

Our extensive clinical experience, built up through the existence of a clinical centre and a clinical nurse specialist, assists us in encouraging research and trials here in the UK.

We will continue to play a key role in supporting and facilitating research projects at Cardiff University, the University of Oxford and at University College London and will collaborate with laboratories, clinicians and pharmaceutical companies on all aspects of current and future research for the benefit of our community.

## **Niemann-Pick Disease Type C (NP-C)**

A Phase I Study of Cyclodextrin (HP- $\beta$ -CD) for NP-C continues at the National Institutes of Health in Maryland, USA. Alongside this, a Phase 1 study of Histone Deacetylase Inhibitors (HDACi) has commenced at the same venue.

In addition, Orphazyme, a Danish Biotech Company, is progressing plans for the clinical development of Arimoclomol, a small molecule inducer of heat shock proteins, for NP-C.

## **The Peter Carlton Jones Memorial Award**

We nurture the interest of young researchers through our annual "Peter Carlton Jones Memorial Award", which is presented to an individual engaged in either: research, teaching, treatment or care within the public or private sectors in the United Kingdom.

The Award, of up to £1000 is granted in response to the submission of a research project which provides an original contribution to the scientific or public understanding of the Niemann-Pick diseases and/or their treatment or cure.



# Expert Care for Niemann-Pick Disease Patients

NP-UK continues to support and work collaboratively with the designated highly specialised centres caring for those affected by Niemann-Pick Disease and other Lysosomal Storage Disorders (LSDs).

In England, LSD patients have had access to a nationally managed specialised service since 2004, providing equitable access to expert clinical care and treatment.

## **Paediatric Highly Specialised LSD Centres:**

- Great Ormond Street Hospital, London
- Birmingham Children's Hospital
- Manchester Children's Hospital

## **Adult Highly Specialised LSD Centres:**

- National Hospital for Neurology and Neurosurgery, London
- Royal Free Hospital London
- Queen Elizabeth Hospital, Birmingham
- Addenbrooke's Hospital, Cambridge
- Salford Royal Foundation Hospital, Manchester

Patients in **Wales, Scotland** and **Northern Ireland** also have designated specialist hospitals providing care for LSD patients:

- Wales - University Hospital of Wales, Cardiff
- Scotland - Yorkhill Children's Hospital, Glasgow
- Northern Ireland - City Hospital, Belfast

We provide a **unique care and support service** to those affected by Niemann-Pick diseases and their families.

We are the only UK charity offering care, support information and advice to the whole Niemann-Pick community including patients, parents, carers, extended family, health and social care professionals, science and research academia.

Our service is closely monitored to ensure that each activity is of value and benefit to the community we support.

**Elizabeth Davenport, NP-UK Families Officer**, provides an individual non-clinical advice and information service for affected individuals and their families, plus counselling and support following bereavement.

**Laura Bell, our Clinical Nurse Specialist**, provides those affected and their families with expert care and practical advice within the clinic setting or at home.

Our **NP-UK Central Office Team**, Executive Director, Toni Mathieson, Finance and Administration Officer Christine Jopling and Information Officer, Sue Woodhouse, support the day to day management of our Central Office, operating our 24 hour helpline and ensuring the smooth running of all our activities.

# Niemann-Pick Disease Group (UK)

## Our Activities



We are the only charity in the UK that provides specialist information plus practical and emotional support that aims to assist families living with Niemann-Pick diseases, whilst supporting progress towards therapies and clinical trials.

Trustees have a broad range of skills including clinical expertise and the direct experience of being a parent or carer. This deepens our understanding of the disease and enables us to influence not only our work but that of the social care, scientific and clinical communities.

Our staff team are central to achieving our objectives; providing support, information and advocacy services and raising much needed awareness of Niemann-Pick diseases plus the social and economic challenges faced by those affected.

### **This year, we have:**

- Provided support and information to families and associated professionals via our staff team, 24-hour help line, comprehensive website, newsletters and bulletins.
- Continued to fund the post of a dedicated Clinical Nurse Specialist for Niemann-Pick Diseases, with support from family fund raising and BBC Children in Need.

### **Public Benefit**

The Trustees confirm that they have complied with the duty in section 4 of the Charities Act 2006 and, when planning our activities for the year, Trustees carefully consider the Charity Commission's guidelines on Public Benefit.

In working towards our charitable aims, we undertake a wide range of activities in the three key areas of Care and Support, Information and Research, each of which aims to ensure that those affected by, or connected to Niemann-Pick disease, receive care appropriate to their health and emotional needs.

- Offered UK wide networking and mutual support opportunities for families through our Annual Family Conference, Clinic Days and social networking sites.
- Expanded our care and support service to include individual support for siblings and a project to help families improve emotional resilience.

# Niemann-Pick Disease Group (UK)

## Our Activities

### Care and Support Service

Through our team of skilled staff, we provide an individual care, support and advocacy service for those affected by Niemann-Pick diseases, their families and associated health and social care professionals. The service is flexible and offers a wide range of support on a needs led basis.



Our **Clinical Nurse Specialist** provides expert support, advice and information regarding all aspects of the Niemann-Pick diseases:

- An individualised advice service via phone/email/internet
- Clinical care and help in navigating the NHS
- Home visits
- Genetic counselling
- Bereavement Counselling
- Teaching sessions for Schools
- Kindly supported by grant funding from BBC Children in Need



*“Alone we can do so little,  
together we can do so much”  
Helen Keller*

### NP-UK Clinical Nurse Specialist and Families Officer

Our Clinical Nurse Specialist and Families Officer provide a unique support service to those affected, with the aim of ensuring that each family or individual receives the level of support they require to meet their needs.

Due to the rarity of these conditions, many health and social care professionals have little or no knowledge or experience to assist them in caring for effected individuals, therefore our expert support service is much-needed and appreciated at both a local and national level.

The flexibility and responsiveness of this service takes into account the differing needs of our community, which is pan-ethnic and spans all age groups.

Our **Families Officer** provides non-clinical advice, information and support on an individual basis:

- Home visits and assistance at clinic appointments
- Advice, reassurance or just a friendly voice at the end of the phone
- Benefits advice, housing, aids and adaptations
- 
- Special Educational Needs and help with Statements
- Kindly supported by grant funding from the Hope for Hollie Foundation





# Niemann-Pick Disease Group (UK)

## Our Activities



### Interactive Family Care and Support Service

Our Interactive Family Care and Support service first launched in 2010, thanks to a grant from the Big Lottery Fund Reaching Communities Programme. It has now become an integral part of our activities. The service uses interactive technology to improve community access to advice and information from the NP-UK Clinical Nurse Specialist (CNS), and their own health care team.

### Mortimer the Monkey

Mortimer the toy Monkey was introduced at our Annual Family Conference 2014. Mortimer was named by a group of siblings whose family members are affected by Niemann-Pick diseases. Mortimer now works alongside Families Officer Elizabeth Davenport to support children and their siblings by helping them to talk about their thoughts and feelings regarding Niemann-Pick disease.



### Rare Disease Day 2014

NP-UK is pleased to be a friend of the Rare Disease Day Campaign, now in its 4th year and helping to raise much needed awareness of the impact and challenges faced on a day to day basis by those affected. The 2014 them focused on care; encouraging everyone in the rare disease community to **Join Together for Better Care.**

- A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population
- There are between 6,000 and 8,000 known rare diseases
- 1 in 17 people will be affected by a rare disease at some point in their lives.
- 75% of rare diseases affect children.



### Family Fun Days

This year, NP-UK Families Officer, Elizabeth Davenport hosted three Family Fun Days for our community, with support and funding from The Pride in Leyton Appeal, The Campbell Burns Metabolic Trust, Daytrippers, Merlin's Magic Wand and the Hope for Hollie Foundation. Families were treated to an Easter celebration at Gullivers Land, a visit to Legoland and an amazing Christmas Party with Santa Claus as special guest!



The Campbell Burns  
Metabolic Trust  
Walking with you



# Niemann-Pick Disease Group (UK) Annual Family Conference 2014

Our 21st Annual Family Conference, held September 2014, at Wyboston Lakes, Bedfordshire, saw over 200 UK and International delegates in attendance. The Conference theme, "Today's Vision: Tomorrow's Reality" recognised the upsurge of activity in this field, particularly in regard to clinical development.

Expert speakers provided delegates with the latest news in research and potential therapies for Niemann-Pick disease, plus related care issues, breakout sessions and practical workshops. As well as hearing the latest research and clinical updates, this weekend event is about sharing information, meeting new people and making new friends.

Our Children and Young Persons' Activity Programme, staffed by a team of dedicated volunteers, enjoyed a Fun Packed Day at Woburn Safari Park on Saturday, followed by arts, crafts, music and science and workshops on Sunday.

**"The children really enjoyed their day out and the activities on Sunday"**



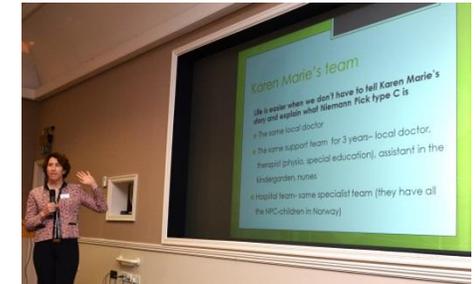
**"Thank you so much for an excellent weekend"**



**"Fantastic and inspiring conference!"**

**"Subject and timing of sessions were great"**

**"High standard of organisation"**



On Friday 26<sup>th</sup> of September 2014, the **5<sup>th</sup> Interactive Workshop on Niemann-Pick Diseases** provided the opportunity for professional delegates to share current information regarding all aspects of Niemann-Pick diseases, including clinical management, research and potential therapies, and to debate the future of treatment and care for those affected. As in previous years, the Workshop was well attended, with delegates from the UK, USA, Denmark, France, Spain, Germany and Italy.

Many thanks go to our volunteers and our Conference Sponsors, **Actelion Global, Actelion Pharmaceuticals UK, Genzyme, A Sanofi Company and Dr Heti Davies**, without whom this event would not be possible.

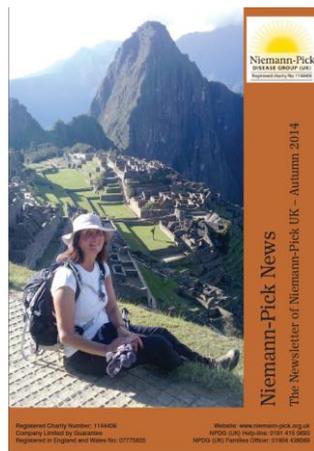


# Niemann-Pick Disease Group (UK)

## Information and Awareness

NP-UK recognises the need to provide current, relevant, information and news regarding all aspects of the Niemann-Pick diseases in a timely manner.

We produce a wide range of informative materials and educational resources to assist families and professionals in dealing with all aspects of the Niemann-Pick diseases. These are regularly updated to reflect the changing needs of those we support.



During the year we have:

- ✓ Distributed our Newsletter "Niemann-Pick News" plus e-bulletins to our members and supporters free of charge
- ✓ Continued to develop and distribute educational information on Niemann-Pick diseases to support families and professionals
- ✓ Encouraged close working relationships with other patient groups to share information, stimulate interest in, and further the knowledge and understanding of NPD
- ✓ Organised an Annual Family Conference to encourage the sharing of knowledge and experience between families and professionals
- ✓ Organised a Workshop specifically for health professionals, providing the opportunity to share the latest information regarding clinical care and scientific advances
- ✓ Given talks and presentations at conferences and fundraising events across the UK, including educational talks to schools, hospitals and hospices

We also have a strong on-line presence through our website and social media channels such as Facebook and Twitter.



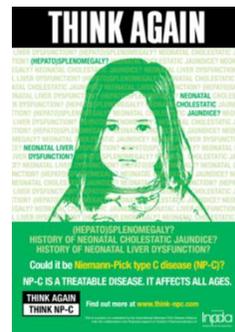
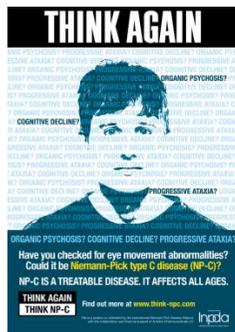
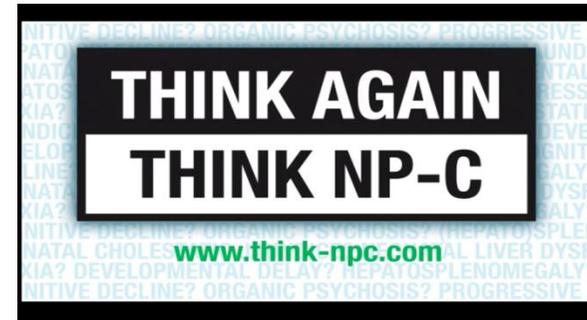


# Niemann-Pick Disease Group (UK) Think Again. Think NPC Campaign

The **Think Again. Think NP-C Campaign** was launched in September 2014.

This awareness campaign aims to reduce the time to diagnosis by supporting healthcare professionals who are unfamiliar with NP-C to recognise the key signs and symptoms of the disease.

Activities are taking place across the world, including here in the UK, to reach the right people with the right messages, so that patients can be diagnosed sooner.



In the UK, relevant clinicians have been targeted at the National Hospital for Neurology and Neuroscience and Evelina Hospital in London; Salford Royal and Manchester Children's hospitals, with more to follow as we continue to identify the best way to reach our target audience.

We aim to get information on NP-C, and how to diagnose it, in front of as many healthcare professionals who may have undiagnosed (or indeed misdiagnosed) patients as possible.

The powerful messages and visuals of this campaign aim to capture the attention of our target audience and encourage them to take action – to THINK NP-C.

As it currently takes, on average, 5 years to diagnose NP-C, the value of an early diagnosis cannot be underestimated.

Although there is not yet a disease-modifying treatment for the disease, the symptoms can be managed more effectively with an accurate diagnosis. Patients and carers are then also able to access appropriate support – a lifeline for so many families.



# Niemann-Pick Disease Group (UK)

## Supporting and Influencing Research

Facilitating progress towards therapeutic interventions for these diseases is central to everything we do and has been for the past 23 years.

With increased research activity and clinical trials on the horizon for ASMD Niemann-Pick type B and Niemann-Pick disease type C, this year, our efforts have been focused on providing clear and accurate information to our community. In addition, there has been a greater need for collaborative working to ensure the patient voice is heard and can influence progress.

Our Board of Trustees, the majority of whom have family members or friends directly affected by Niemann-Pick disease, understand the urgency to find effective therapies and the fact that progress never seems to be fast enough.

We actively encourage and facilitate progress in research through the relationships we foster and the networks we contribute to and aim to maximise the benefits of funding, by working in partnership with like-minded organisations in pursuit of a better understanding of Niemann-Pick disease. By sharing knowledge and expertise and by working together, we believe progress will be even faster.



This year we have continued to:

- ✓ Maintain links with clinicians, scientists and the pharmaceutical industry in the development of potential therapies for Niemann-Pick diseases, both in the UK and in the USA.
- ✓ Actively support and, where appropriate, fund scientific research into Niemann-Pick disease. This year, we have supported studies at Cardiff University, the University of Oxford and the University College London.
- ✓ Nurture the interest of young researchers through 'The Peter Carlton Jones Memorial Award', granted in response to the submission of an original contribution to the scientific or public understanding of the Niemann-Pick diseases and/or their treatment or cure.
- ✓ Monitor scientific advances and effectively communicate news in a number of different formats – through our website, newsletter, social media and e-bulletins and face to face at our Annual Family Conference.
- ✓ Attend relevant scientific and research events around the world, including but not limited to, the Ara Parseghian Medical Research and Scientific Conference, Brains for Brain, WORLD, the British Paediatric Neurology Association Annual Conference and the Society for the Study of Inborn Errors of the Metabolism (SSIEM) Annual event.
- ✓ Support the development of the International Niemann-Pick Disease Registry, leading the Work Package for 'Project Dissemination' and raising awareness of this important tool at NP patient events worldwide.



# The International Niemann-Pick Disease Registry (INPDR)

The International Niemann-Pick Disease Registry (INPDR) is an EU funded project to establish a global registry for Niemann-Pick diseases.

Why is the INPDR needed?

A registry is an important tool for collecting and recording information about a disease, such as the effect it has on a patient and how it progresses. At the moment registries are typically held by pharmaceutical companies to investigate a single drug. This can lead to several registries for the same disease, and limited access to the data they hold.

The INPDR is a disease registry owned by patient groups and clinicians involved in the care of NPD patients.

It will enable progress by allowing authorised access to anonymised clinical data, helping to identify and recruit patients to clinical trials, coordinate research efforts and improve patient outcomes globally.



The INPDR is owned by the International Niemann-Pick Disease Alliance and is being developed and coordinated by University Hospital's Birmingham NHS Foundation Trust (UHB) in the UK, with 11 Associate Partners across 7 EU countries (including NP-UK) and 17 International Collaborating Partners in the USA, Canada, Italy, France, Estonia, Netherlands, France, Australia, Brazil and Argentina.

This €2m Project commenced in April 2013 and is co-funded by the EU Health Programme and the partner organisations. Niemann-Pick UK is leading Work Package 2, delivering the dissemination of the project.

This much-needed project is truly a collaboration between clinicians, scientists, and patient associations across the world, with the collective aim of improving care and treatment for Niemann-Pick patients worldwide.

***"The INPDR will generate a highly-needed public resource that unites diverse international attempts to better understand the biology and clinical picture of Niemann-Pick diseases. The research community may learn how to better diagnose the disease and why some patients are doing better than others. For the individual patient, the information therein might speed up inclusion into clinical trials."***

Dr Heiko Runz, MD



# Niemann-Pick Disease Group (UK) National and International Collaboration



## The International Niemann-Pick Disease Alliance (INPDA)

The International Niemann-Pick Disease Alliance (INPDA) was formed in 2009 and provides a forum for the exchange of information, experience and knowledge, with the aim of accelerating progress.

The INPDA brings together non-profit Niemann-Pick support organisations from around the world. Members include: Argentina, Australia, Brazil, Canada, France, Germany, Italy, the Netherlands, Spain, USA and the United Kingdom.

The Alliance provides a network that enhances the transfer of information, maximizes the use of resources and demonstrates that collectively, Niemann-Pick diseases are not as rare as was once thought. Further information can be found at [www.inpda.org](http://www.inpda.org)

The INPDA is currently engaged in delivering three key projects that aim to make a difference in the areas of scientific, clinical and therapeutic research, patient care and access to treatments and improving time to diagnosis:

- The International Niemann-Pick Disease Registry,
- The Loire Valley meeting,
- The Think Again. Think NP-C Campaign

## The UK LSD Patient Collaborative

Lysosomal storage disorders (LSDs) are a group of approximately 40 rare inherited metabolic disorders that result from defects in lysosomal function, including the Niemann-Pick diseases.

The UK LSD Collaborative consists of representatives from the Association of Glycogen Storage Diseases, the Batten's Disease Family Association, the Gauchers Association, the MPS Society (Mucopolysaccharide Diseases), Save Babies Through Screening Foundation (representing Krabbe Disease), The Cure and Action for Tay Sachs (CATS) Foundation and Niemann-Pick UK.

The Collaborative has undertaken to jointly promote and share understanding of LSDs, to advance standards of care and to enhance the wellbeing of all those affected.

It is now a recognised forum and has provided expert patient perspective to NHS England in respect of highly specialized services for LSDs, access to treatment and therapies, home care, transition and the approvals process for new therapies.



# Niemann-Pick Disease Group (UK)

## National and International Collaboration

### Students 4 Rare Diseases (S4RD)

Students 4 Rare Diseases was founded by a group of 3rd year medical students at Barts and the London Hospital to create a learning module for students on rare diseases. The initiative was supported by the UK LSD Collaborative, of which NP-UK is a founding member.

The LSD Collaborative provided support to S4RD that assisted them in successfully applying for a Genzyme PALS Award grant in late 2013.

The Second Unusual Suspects Symposium organised by the S4RD team, which includes representatives from four London Medical Schools, was held at the Royal Society of Medicine in April 2014 with the aim of bringing future doctors and rare disease patients together to emphasise the importance of diagnosis and appropriate management.



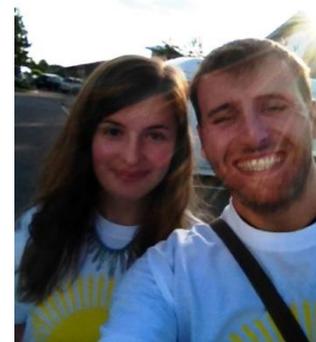
In addition, in September 2014, S4RD won the best poster prize at the International Symposium on Newborn Screening Regional European Meeting in Birmingham.

### Engaging with Volunteers

We organise a programme of community events and activities throughout the year.

These include Family Fun days, Clinic Days, expert meetings and conferences for the whole family to participate in.

Our event programme is important and we rely on our childcare volunteers to keep our children and vulnerable adults safe whilst ensuring they have a happy and memorable time.





# Niemann-Pick Disease Group (UK) Fundraising

We would like to thank all of our members, families and friends, corporate donors and charitable trusts for their invaluable support this year.

We are constantly amazed by the fantastic support shown to us by our army of volunteer fundraisers, including family members, friends, corporate and on-line donors who go to often extreme lengths to fundraise on our behalf.

This year's events have included Golf Days, Charity Balls a Zumbathon, and a 70's Themed Disco Night!

Although it is impossible for us to mention all of them here, we are more than grateful for each and every one of their efforts, which support all areas of our work, including research, family support and advocacy.

Here are some of the highlights...





# Niemann-Pick Disease Group (UK)

## Fundraising

### Corporate Giving

We are grateful to our corporate supporters, namely the pharmaceutical companies working in the field of Niemann-Pick diseases to progress treatment, research and care for this group of diseases. Their contributions, in the form of unrestricted educational grants, are greatly valued and assist us in our work to inform and educate our community and the wider general public about Niemann-Pick diseases.

We are at all times careful to maintain our independent position and therefore the acceptance of a grant does not imply endorsement of a company's products.

We also grateful to the many businesses and companies who tirelessly support our work in so many ways, such as hosting a collection box or holding a dress down day. In many cases this support is due to a member of staff having a personal connection to someone affected by NPD. These efforts are greatly appreciated.

We are always keen to engage with businesses interested in our work and in ways in which they might be supportive.

Opportunities include payroll giving, matched funding of staff fundraising efforts, event sponsorship, 'in kind' donations of goods or services, and the provision of volunteers

**THANK YOU!**

### Trusts, Grants and Family Campaigns

As we continue to expand and improve our services to meet the changing needs of our community, seeking support from grant-giving bodies has become essential to building the capacity of our organization and to providing an increasingly important source of income.

We make regular applications to trusts and foundations whose aims and objectives are in line with our own or those that support specific themes relevant to our areas of our work. When successful in the grant making process, we carefully monitor and evaluate the progress of these projects to understand their impact and the difference they make to our community and also to enable regular reports to those who have so generously supported them.

We are also grateful for the support of Family Campaigns, whose tireless fundraising efforts provide support for all areas of our work. This year, we were grateful for the support of:

- ❖ Roald Dahl's Marvellous Children's Charity
- ❖ BBC Children in Need
- ❖ The Hope for Hollie Foundation
- ❖ Brian Murtagh Trust
- ❖ Grace's Wish
- ❖ Georgina's Gang
- ❖ Joan Ainslie Trust
- ❖ Big Lottery Fund Awards for All
- ❖ Campaign for Calum
- ❖ The Katie Elliott Fund



# Niemann-Pick Disease Group (UK)

## Our Objectives for 2015-2016

### Care and Support

- We will further develop our services to include a sibling support service
- We will research and develop our services to better support an increasing number of newly diagnosed adults
- We will continue to meet the needs of our community through the provision of our care and support service
- We will engage with our community to ensure our services are meeting their needs
- We will research and develop our services to better support our BME families

### Collaboration

- We will continue to promote the Think Again Think NPC campaign and further develop ways to reach our target audience
- We will build on our strong relationship with the INPDA and LSD Collaborative to exchange information, advance standards of care and enhance the wellbeing of all those affected.
- We will support National Rare Disease alliance groups for the benefit of our all those affected by Niemann-Pick diseases
- We will remain active in providing an expert patient voice and perspective to NHS England in respect of highly specialized services for LSDs, access to treatment and therapies and the approvals process for new therapies

### Providing Information

- We will continue to develop our on-line presence.
- We will research ways in which we might improve the information we provide for BME families
- We will develop and distribute information in support of an increasing number of clinical trials
- We will continue providing up to date and accurate information on all aspects of Niemann-Pick Disease; by means of our Central Office and Clinical Nurse Specialist, educational and on-line resources, newsletters/e-bulletins, the website and telephone help line

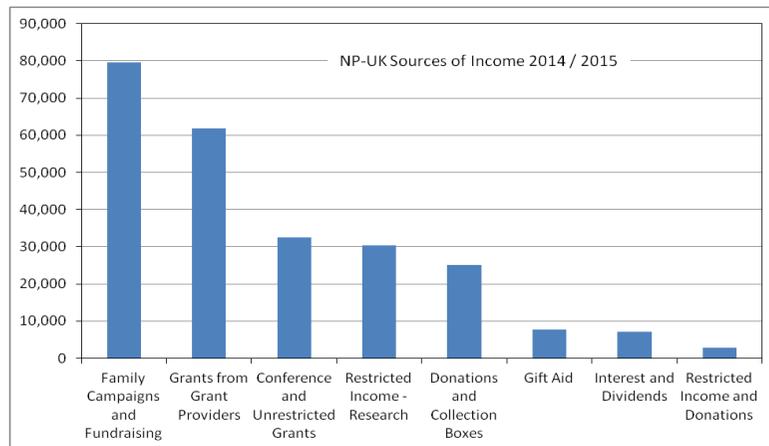
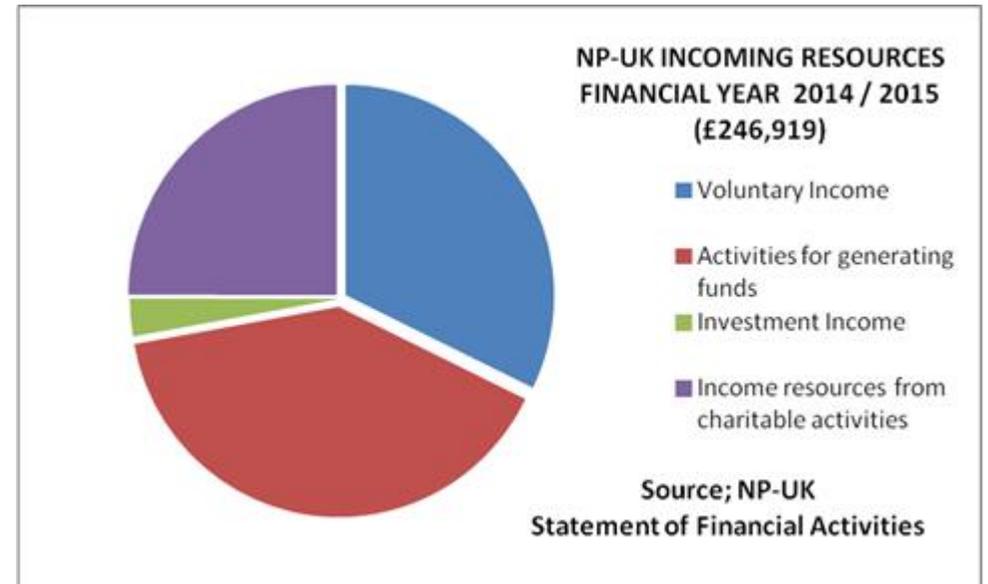
### Research

- We will fully support the International Niemann-Pick Disease Registry project
- We will continue our support for Niemann-Pick research by making grants available to laboratories working in this field
- We will encourage and facilitate clinical trials when the clinical conditions and funding becomes available
- We will promote Niemann-Pick disease research through the relationships we foster and the networks we attend
- We will continue to provide the annual "Peter Carlton Jones Memorial Award to encourage the interest of young scientists/researchers.

# Niemann-Pick Disease Group (UK) Financial Review

The total income for 2014 / 2015 was £246,919 of which £177,875 was raised by donations and fundraising activities through the generosity of families, friends and supporters, plus unrestricted grants made by pharmaceutical companies. (£15,100).

Without this support, we would not have been able to provide our core and support services that so many families have come to rely on.

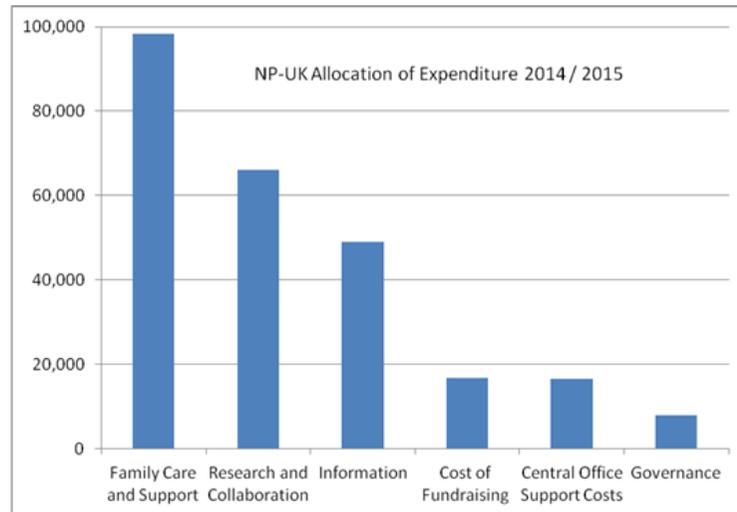


In addition, £61,864 was raised through grant giving organisations and we received an investment income of £7,168 which was equivalent to a return on investment of 3.17%

The total income received from all sources was £5,326 better than 2013/2014 but fell £14,346 short of expectations due to the lead time in preparing and submitting grant application forms.

# Niemann-Pick Disease Group (UK) Financial Review

Total expenditure for the year was £254,584 and £7,665 was taken from reserves to balance the shortfall in income; the distribution of funds is shown below:



## Net Assets

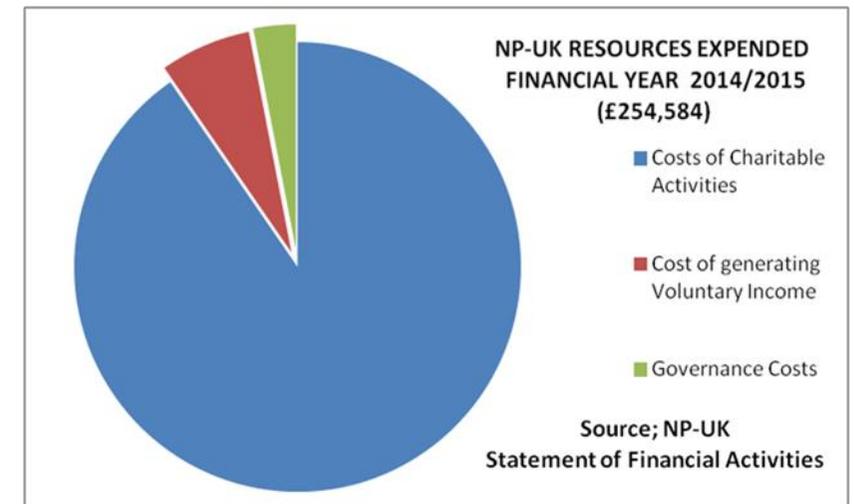
Net assets at the end of the year amounted to £267,847 of which £226,356 was held as an endowment fund to be used, when required, to assist in financing the family care and support service and £41,491 was held in bank accounts of which £19,033 was restricted for use as described in the financial statement.

The balance of £22,458 was used to finance the day to day operating expenses of the organisation.

This year, the cost of generating voluntary income included fundraising costs plus an additional cost in support of a fundraising strategy which placed greater emphasis on obtaining grant funding; the benefits of this investment will be realised in 2015/2016.

Fundraising costs therefore increased from £7,919 (3.28% of income) in 2014 to £16,579 (6.79%) in 2014/2015.

Total governance costs were £7,805 and included £1,248 fees for the independent examiner.





# Niemann-Pick Disease Group (UK)

## Financial Review

### Reserves

Our current Finance Strategy is based on incoming funds matching the expenditure required to deliver our objectives and the services that we provide to families and beneficiaries.

Our Reserves Policy is subject to an annual review and for 2015/2016 takes into account the balance between the sources of income and the risks associated with periods of reduced income.

Notwithstanding cash flow forecasts, the required level of reserves is currently established as £125,000. This represents seven months when incoming funds are, on average, £10,564 below monthly expenditure plus £25,000 to fund the day to day operation of the Charity and £25,000 to provide urgent and unplanned, grants to UK laboratories to fund research in line with our Grant Application Process.

The free reserves at 30 April 2015, excluding the endowment fund, amounted to £22,458 and the Trustees are committed to building up the level of reserves in line with the reserves policy.

During this period, the financial security of the Charity is partly provided for by the endowment fund.

### Risk Management

The Board of Trustees have considered and reviewed the possible risks to which the Group may be exposed, and have established the following risk management process:

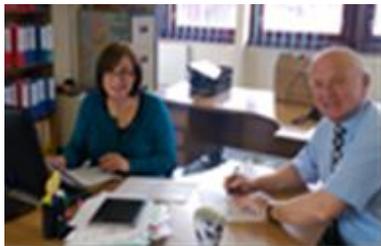
1. The development and implementation of a risk register
2. Quarterly risk reviews
3. Procedures to identify and mitigate risk

The major risks identified this financial year include:

- A reduction in Grant Funding impacting upon our reserves
- The competitive fundraising environment and reducing opportunities affecting our potential to secure funds.
- NHS and Welfare Reforms will create additional demand for our care and support services
- A rise in newly diagnosed adults will create additional demand for the services we provide

### Outlook

The Trustees are satisfied that the necessary actions are in place to address the shortfall in grant funding and that net assets are sufficient to ensure the day to day running of the Charity and the services we provide.





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

VERY RARE PROGRESSIVE IRREVERSIBLE CHRONICLY DEBILITATING LYSOSOMAL STORAGE DISEASE<sup>1,2</sup>

**NP-C affects all ages<sup>1</sup>**

**Incidence of NP-C is 1 in 120,000 live births<sup>2</sup>**

Likely an underestimate due to lack of clinical awareness<sup>1</sup>

**NP-C takes on average 5 YEARS to diagnose<sup>3</sup>**

**That's...**

1,826 DAYS

260 WEEKS

43,824 HOURS

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

**THINK AGAIN THINK NP-C**

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

**Individual symptoms are non-specific to the disease<sup>1,2</sup>**

**If you are a:**

- Paediatrician**  
LOOK FOR ATAXIA, DEVELOPMENTAL DELAY, HEPATOSPLENOMEGALY
- Paediatric hepatologist/neonatologist**  
LOOK FOR HEPATO/SPLENOMEGALY, NEONATAL CHOLESTATIC JAUNDICE, NEONATAL LIVER DYSFUNCTION
- Adult neurologist/psychiatrist**  
LOOK FOR COGNITIVE DECLINE, ORGANIC PSYCHOSIS, PROGRESSIVE ATAXIA

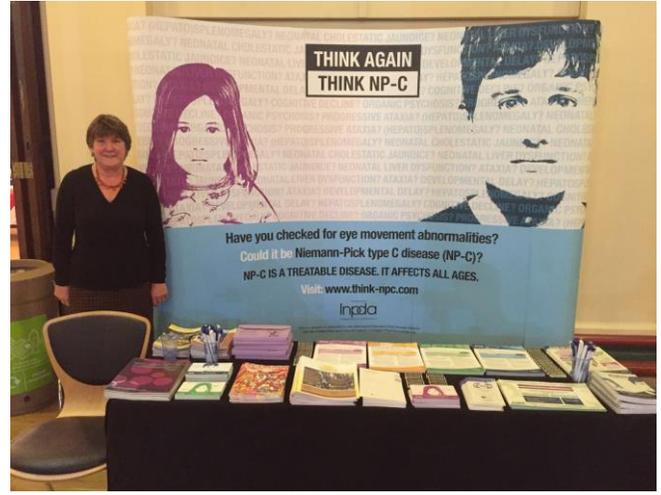
- References**
1. Patterson M, Hendrickz, Wetherling M, et al. on behalf of the NP-C Guidelines Working Group. Recommendations for the diagnosis and management of Niemann-Pick disease type C: an update. *BMJ Open* 2012; 16(02): 330-344.
  2. Walter, M. Niemann-Pick disease type C. *Orphanet J Rare Dis* 2010; 5: 16.
  3. Wilburg FA, Smith J, Prineas M, et al. Development of a suspicion index to aid diagnosis of Niemann-Pick disease type C. *Neurology* 2012; 78(23): 1560-1567.
  4. Mengel S, Niemann M, Lueggen C, et al. Niemann-Pick disease type C: hepatosplenomegaly: an expert-based clinical description. *Orphanet J Rare Dis* 2013; 8: 166.

To help reduce the time to diagnosis visit [www.think-npc.com](http://www.think-npc.com) today

**THINK AGAIN THINK NP-C**

**Niemann - Pick UK**  
Registered Charity No. 1146848

**Inpoda**  
FOUNDED 2009  
International Niemann-Pick Disease Alliance



[www.niemann-pick.org.uk](http://www.niemann-pick.org.uk)