



# Niemann-Pick Disease Group (UK)

Registered Charity No. 1061881

Providing effective support and making a positive difference to families affected by Niemann-Pick diseases through the provision of

***Care and Support : Information : Research***



## Annual Report

2010-2011

## *A Year of Development and Progress*

**W**ithin the pages of this Annual Report you will find an overview of our activities, plans and challenges. This year, thanks to the efforts of many supporters it has been a year of development and progress. It has also been a time of change that has seen us beginning the move to the next stage in our journey towards solving the challenge presented by the Niemann-Pick Diseases.

Such a journey is never fast enough for those on it and is fraught with difficulties. However our aim in the NPDG(UK) has always been to help speed up progress whilst trying to ease the load and clarify the direction.

Every now and then it is important to reflect and plan for the journey ahead. This ensures that maximum use is made of the resources available in supporting families, stimulating and facilitating research and providing information. The need for such an assessment led us to undertake a Strategic Review exploring external factors, potential risks, current constraints and the availability of resources – all of which could affect the future direction of the Group.

The Review took place in April 2011; a summary of the results can be viewed on our website.

Research has taken us to the point where, with fair weather the diseases will be embarking on trials of potential new therapies in the year ahead. We are involved in actively supporting these trials and are helping in every way we can to ensure their speedy completion. It is important to ensure their progress is as quick as possible so that the results can inform our next steps and will benefit all those affected by the diseases. That is where our research sub-committee has concentrated its efforts and, thanks to the fund raising efforts of many, we have been able to fund new research projects and to facilitate collaborations designed to assist progress in the trials.

Supporting affected individuals and families has always been a core part of our objectives. You will read in this report about the employment of a new Families Officer, new information booklets and the development of a new Big Lottery project. All designed to support and ease the immense load carried by families on this journey.

I hope in reading the report you feel the progress and that it encourages and supports all of those who travel this journey together.

This will be my last annual report as I step down as Chairman this year. I would like to thank everyone who has joined us on this journey, for their support, energy and friendship. Together you have made a huge difference.

Best wishes



Chairman NPDG (UK)

The main aims and objectives of the NPDG (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.

The NPDG (UK) aims to meet these objectives by:

- 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.

To achieve our objectives, we rely on funding from voluntary donations, corporate donors, charitable trusts and statutory bodies, plus the fundraising support of our members, friends and families. Our goal is to ensure the continuation, and the successful delivery, of the services we provide, whilst seeking sustainable expansion.

### Structure of the NPDG (UK)

The Niemann-Pick Disease Group (UK) is a charitable organisation offering support and information to families affected by any type of Niemann-Pick disease and to associated professionals.

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

During this year, 2010/11, the NPDG (UK) was governed by a Board of 10 Trustees within the context of an approved Constitution. All trustees and office bearers were elected at the AGM. The Board, made up of people who have family members or friends directly affected by this group of diseases, has met on five occasions this year, with a number of additional teleconferences. It is responsible for all the

activities the Group undertakes. All of the Trustees are volunteers and each takes on responsibilities within the Group to co-ordinate/support an aspect of our functions, with a number of Sub-Committees providing operational support. The Group's Trustees, honorary officers, and professional advisors are listed on page 12 of this report.

Our four employees 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.

- Since 1999, the Group has funded the salary of a full time Clinical Nurse Specialist, Jacqueline Imrie, providing expert care and practical advice, plus home visits whenever necessary. Genetic counselling and advocacy services are also provided.
- In 2011, the NPDG (UK) strengthened its support service with the addition of a Families Officer, Elizabeth Davenport, providing non-clinical advice, information and support.
- The Group's Executive Director, Toni Mathieson, and Information Officer, Sue Lowe, manage the NPDG (UK) Central Office, operating a 24 hour helpline and ensuring the smooth running of the Group's day to day activities.

Most importantly, the Group would not exist without the support of our members, friends and families; their efforts are vital in assisting the achievement of our aims and objectives.

### Risk Management

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

- The development and implementation of a risk register
- Quarterly risk reviews
- Procedures to identify and mitigate risk

The major risks identified this financial year include:

- The retirement of the Clinical Nurse Specialist and the need to plan for succession
- Succession planning for Trustees and the development of skills

■ Economic and social pressures which will adversely affect incoming resources and coincide with the ending of grant funded projects in 2012 / 2013

Actions are being developed to address these risks and will be communicated when formalised through newsletters, bulletins and letters as appropriate.



When planning our activities for the year, the NPDG (UK) Trustees carefully consider the Charity Commission's guidelines on Public Benefit. Through our work, we aim to ensure that each individual affected by, or connected to, this disease, is able to access the best possible care, support and information appropriate to their needs. In working towards our charitable aims, the Group undertakes a wide range of activities in the three key areas of Care and Support, Information and Research:

## Care and Support Services

The NPDG(UK) provides a unique care and support service to families affected by the Niemann-Pick diseases. The service has been developed over a number of years and is closely monitored to ensure that each activity is of value and benefit to the community we support. This year we have:

- Provided support and information to families and associated professionals in the form of a 24-hour help line, a comprehensive website, educational literature, regular newsletters and bulletins.
- Continued to fund the post of Clinical Nurse Specialist for Niemann-Pick Diseases, with support from family fund raising, BBC Children in Need and The Big Lottery Fund.
- Offered UK wide networking and mutual support opportunities for families through the organisation of an Annual Family Conference, Clinic Days, a website e-forum, social networking sites and the production of a Family Directory.
- Strengthened our support service by the addition of a Families Officer, offering an individual advocacy service including home visits and assistance at clinic appointments, and providing non-clinical advice, information and support via face to face meetings, the telephone, email or in writing.
- Involved volunteers in working with NP families through the Children and Young Persons' Activity Programme at our Annual Family Conference; ensuring they receive the training and support they need to fulfil this valuable role.

## Information Services

The NPDG(UK) recognises the need to provide current, relevant, information and news regarding all aspects of the Niemann-Pick diseases in a timely manner. This year we have:

- Continued to develop and distribute educational information on Niemann-Pick diseases to support families and professionals.
- Encouraged close working relationships with other Niemann-Pick and associated disease groups to share information, stimulate interest in, and further the knowledge and understanding of this group of diseases.
- 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.

## Supporting and Influencing Research

Facilitating progress towards therapeutic interventions for all of these diseases is central to everything we do in the NPDG(UK) and has been for the past 20 years. This year we have:

- Maintained links with clinicians, scientists and pharmaceutical companies in the development of possible therapies for Niemann-Pick diseases, through support for clinical trials for patients with ASMD Niemann-Pick Type B and Niemann-Pick Type C, both in the UK and in the USA.
- Actively supported and, where appropriate, funded scientific research into the Niemann-Pick diseases.
- Nurtured the interest of young NP researchers through 'The Peter Carlton Jones Memorial Award'.
- Monitored scientific advances and effectively communicated news in a number of different formats – such as through our website, newsletter, social media and bulletins.

**N**iemann-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.

There are two commonly recognised forms of the disease: Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick Disease Type A and Type B represent opposite ends of a spectrum of the same disease, characterised by a deficiency of an enzyme which causes a build up of toxic materials in the body's cells.

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

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

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 (NP-C)**

The presentation of NP-C is very variable and the onset of symptoms may occur at any time from early infancy to adulthood, though it most usually affects children of school age. Life expectancy varies considerably and there are a variety of symptoms. These may include an enlarged spleen and liver and, in newborn babies, there may be prolonged jaundice.

The disease is neurologically degenerative leading to progressive loss of motor skills and



difficulty with walking. Speech can become slurred and swallowing problems may develop. Patients may experience sudden loss of muscle tone, which can lead to falls, also epileptic seizures that are generally difficult to control.

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.

### **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 palliative treatments - individual medication to treat the symptoms of the disease.

### **Research and Treatment**

Over the years the NPDG(UK) has built a strong working knowledge in the field of NP research. By developing and sustaining robust relationships with key figures and organisations across the world, the Group is kept up to date with research that is taking place, its status and where it is happening. During these years the NPDG (UK) has played a key role in supporting and facilitating many research projects.

Recent advances in characterisation and understanding of the disease have led to a number of possible therapeutic targets being identified. Our extensive clinical experience, built up through the existence of a clinical centre and a clinical nurse specialist, makes the UK very well placed to be a part of any future trials; Enzyme replacement therapy is currently being investigated for NP-B and studies are looking at potential therapies that may control the neurological symptoms of NP-C.

## Making a Difference

September 2010 saw our 17th NPDG(UK) Annual Family Conference, welcome the highest number of family and professional delegates on record. The aim of the Conference is to provide information, and to address the needs of all those affected by Niemann-Pick diseases, their families, and the health and social care professionals involved in their care.

For the first time, the Group hosted a workshop exclusively for professionals prior to the start of our Family Conference. The workshop, held on Friday 10th September, brought together scientists, researchers, clinicians and other health and social care professionals with an interest in this group of diseases. Over the course of the day, delegates from the UK, France, Denmark, Austria, Germany and the USA were given the opportunity to share current information regarding all aspects of Niemann-Pick diseases, including clinical management, research and potential therapies.

The Conference, held over three days, had the theme 'Pathway to Progress', to reflect the idea that our shared journey with Niemann-Pick diseases is not an easy one, but with each Conference, we move a small step closer to our common goal – the discovery of effective treatments for this group of diseases. The increasing activity in the field of Niemann-Pick diseases has brought renewed hope, and this year, as always, family delegates had the opportunity to hear the latest developments from around the world, to share their thoughts and experiences and to spend valuable time connecting with professional delegates; previous speakers and professional delegates have told us how much they value the opportunity to meet, network and share their skills with families and colleagues.

### Comments received following Conference 2011

*"Great to have the chance to meet so many doctors and scientists working in the NPD field"*

*"We enjoyed the informal conversation and networking."*

*"The workshop for professionals was a great success and complimented the family conference."*

*"The Conference was very worthwhile and rewarding. It is a milestone event that tracks*

*the progress of children/siblings/parents; research; support services and social and cultural attitudes to palliative care."*

Autumn 2010 also saw the launch of our Big Lottery funded project 'The Interactive Care and Support Service'. The overall aim of the project is to improve outcomes for families with Niemann-Pick diseases, through the use of interactive technology such as hand held video cameras and webcams, to enhance access to support and information services.

Families are provided with a camera and a secure link that enables them to send video clips showing "what is happening" to the NPDG (UK) Clinical Nurse Specialist / and or their local healthcare team at the time of their concern. Families can obtain immediate advice, and provide video evidence of symptoms, behaviour, housing or feeding issues, that cannot always be described or replicated at the next planned clinic appointment.

The project also aims to combat the isolation and despair which so often accompanies a diagnosis of a rare disease, by providing the ability to communicate face to face with other affected families, using webcams, headsets and Skype technology. Five families were recruited to the pilot study which commenced in October 2010, and the project has now been rolled out to all families who wish to participate.

In January 2011 the Group was delighted and honoured to be selected as one of the ten winners (chosen from over 400 charities nationally) of the 2011 GlaxoSmithKline IMPACT Awards, which are managed in partnership with health think-tank The King's Fund. The Group received £25,000 in recognition of its outstanding work as a health care charity.

Lisa Weaks, Third Sector Programme Manager at The King's Fund, said:

*"The Niemann-Pick Disease Group (UK) thoroughly deserves an IMPACT Award. Not only has it taken the lead nationally and internationally in working on this life-limiting disease, but it is very well managed and innovative in its approach. Without the Group's services, there would be a gap in the provision of care and support for those affected and a much higher demand on local health services."*

In April 2011 the NPDG (UK) support service was strengthened by the addition of a Families Officer, providing non-clinical advice, information and support. Working in partnership with families and individuals, the Families Officer will facilitate access to relevant and appropriate support services in their local area. Through the development of professional relationships with health and social care teams, the Families Officer will also build a network of support and information that can be tailored to meet the needs of those we support. Additionally, the post will raise awareness and promote understanding of the Niemann-Pick diseases.

### **Influencing Research**

The NPDG(UK) Board of Trustees is made up of people who have family members or friends directly affected by this group of diseases. We understand the urgency to find effective therapies and the fact that progress is never seems to be fast enough - but it is accelerating! We actively encourage Niemann-Pick disease research through the relationships we foster and the networks we are involved in and contribute to. By sharing knowledge and expertise and by working together, this acceleration will be even faster.

Within its capabilities, the NPDG(UK), networks with organisations, universities and pharmaceutical companies, worldwide, and will maximise the benefits of funding, by working in partnership with such organisations in pursuit of a better understanding and a cure for Niemann-Pick disease.

In early 2011, the NPDG (UK) was able to provide grant support to Cardiff University to enable the completion of a study looking at storage in NP-C cells. This is just one of a number of small grants NPDG(UK) has been able make to laboratories here in the UK who are working on this group of diseases.

The Peter Carlton Jones Memorial Award is granted annually 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. In Autumn 2010, the Award was presented to Sarah Pressey for her work in characterizing what happens in the NP-C brain. Sarah has since completed her PhD at The Institute of Psychiatry, King's College London in the lab of Dr Jonathan Cooper, with

additional supervision from Prof Fran Platt of Oxford University.

**With potential therapies** being trialled and research projects actively pursued within the field of Niemann-Pick diseases, there is a need for the NPDG(UK) to understand the process that brings clinical trials to patients and to identify the role that we can play in speeding up this process. Therefore, the NPDG(UK) facilitates and support clinical and scientific research as much as we can, and report the findings to our members. Through our activities this year, the work of our Clinical Nurse Specialist and support for clinical centres of excellence here in the UK, we are supporting the development and progress of such clinical trials. We have worked with specialists to facilitate, when it will benefit progress, the extension of US based clinical trials to the UK. We are also investigating the ways in which we can effectively assist families in preparing for trials and potential therapeutic developments.

### **National and International Collaboration**

Following the formation of the **International Niemann-Pick Disease Alliance** in October 2009, of which the NPDG (UK) is a member, we have participated in regular teleconferences to further develop this organisation, which aims to provide mutual support, to develop the availability of information and to stimulate research into this group of diseases. This has resulted in a number of very practical developments in understanding and research. The Alliance is now developing a website [www.inpda.org](http://www.inpda.org) and an international database to facilitate the collation of information/data on Niemann-Pick diseases in order to accelerate research into potential treatments and therapies. Representatives of twelve countries have so far joined the Alliance, with a further three registering their interest. As understanding increases, the importance of this Alliance will grow.

**As a member of the LSD Patient Organisation Collaboration**, our work has created a strong lobbying and action group for LSD patients and their families in the UK. The group is made up of representatives from the Association of Glycogen Storage Diseases, Batten Disease Family Association, the Gauchers Association, the Society for Mucopolysaccharide Diseases, Save Babies Though Screening Foundation -

representing Krabbe Disease, and the Niemann-Pick Disease Group (UK). As part of this collaboration, the NPDG (UK) has undertaken to jointly promote and share understanding of their diseases to advance standards of care and to enhance the well-being of those affected. The LSD Collaborative has now achieved the status of a recognised forum and was invited to participate in the Brains for Brain European Task Force Meeting held at the European Parliament in Brussels in December 2010.

## Information and Awareness

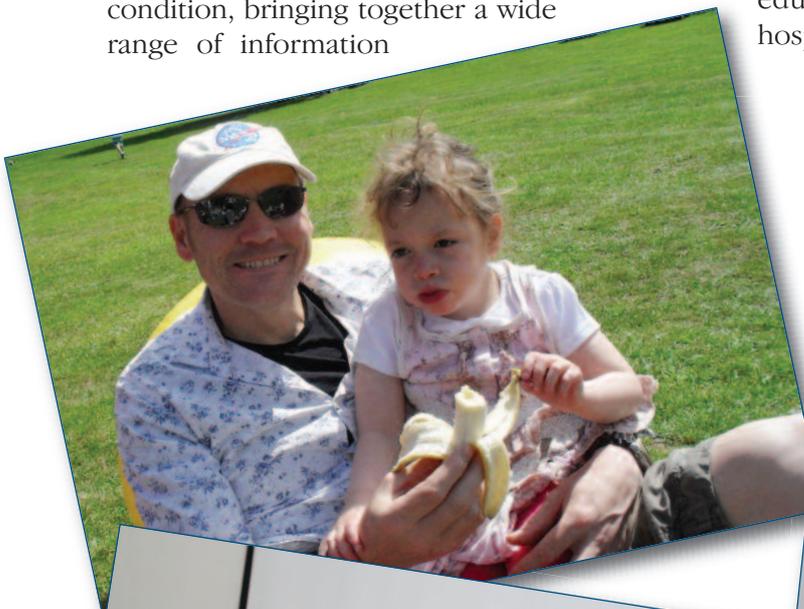
The NPDG(UK) produces a wide range of informative materials and educational resources to assist families and professionals in dealing with all aspects of the Niemann-Pick diseases. These are regularly updated to reflect the changing needs of those we support.

A new booklet providing information about ASMD Niemann-Pick disease types A and B was launched in 2010. The booklet aims to improve understanding of this extremely rare condition, bringing together a wide range of information

useful to all involved in the treatment of patients with ASMD Niemann-Pick diseases.

A **Practical Guide to Niemann-Pick Type C** for parents and carers was produced in 2011. The NPDG(UK) recognises the value of the knowledge, skills and experience gained by families caring for someone with Niemann-Pick type C disease (NP-C), therefore this guide was developed with the help of parents and carers of children, young people and young adults affected by NP-C. The guide aims to provide practical information, reassurance and support that will assist all those caring for an individual with NP-C in dealing with the day-to-day challenges of the disease.

During the year we have also distributed our regular Newsletter, "Niemann-Pick News" plus e-bulletins and a new fundraising bulletin to our members and supporters free of charge. Our Clinical Nurse Specialist, Executive Director and members of the Trustee Board have given talks and presentations at conferences and fundraising events across the UK, including educational talks to schools, hospitals and hospices.



The NPDG (UK) has developed a process to monitor and evaluate our services in an effort to increase our ability to make a difference.

In 2009/10 our 'Awareness Indicators' have shown that the uptake of our services continues to rise, and that there has been increased demand for information regarding the Niemann-Pick diseases. Therefore, we continue to seek ways to sustain and improve our current services, and to allow for future growth.

In 2010/11, the Group have been thankful for the support of the following grant giving bodies:



In 2010, the NPDG (UK) successfully applied to the Big Lottery Fund Reaching Communities Programme, securing £172,601 over three years, giving us the opportunity to enhance our current support service with a new project, entitled the 'Family Care and Interactive Support Service'. This grant is restricted for use as specified by the project criteria.

The Group is grateful for the continuing support of Children in Need, who have provided a grant each year since 1999 to part support the cost of the NPDG (UK) Clinical Nurse Specialist post.

The Group was delighted and honoured to be selected as one of the ten winners (chosen from over 400 charities nationally) of

the 2011 GlaxoSmithKline IMPACT Awards, which are managed in partnership with The King's Fund. The Group received £25,000 in recognition of its outstanding work as a health care charity and also the opportunity to participate in significant and valuable training delivered by The King's Fund.

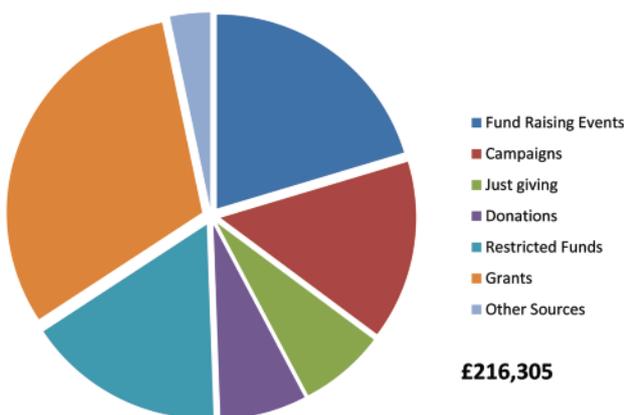
The following charts indicate the year's income and expenditure.

*Please note the displayed figures are subject to Audit.*

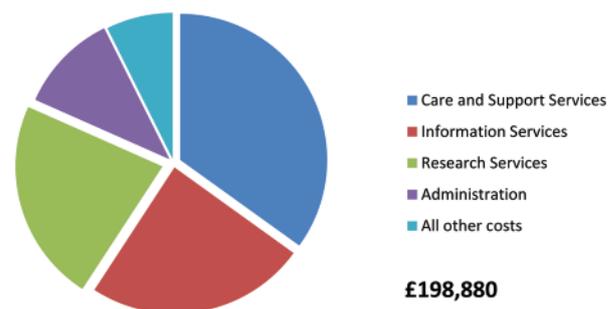
*This Report should always be read in conjunction with the Annual Independently Examined accounts for 2010/2011.*

In the year 2010/2011 Administration and Management costs amounted to 10.1% of total expenditure.

**Incoming Funds, Financial Year 2010 / 2011**



**Expenditure, Financial Year 2010 / 2011**



## Reserves Policy

The primary aim of the NPDG (UK)'s Reserves Policy is to ensure that we hold adequate funds to maintain the long term sustainability of the Group's support and advocacy service, to protect commitments during the current financial year, and to manage short-term risks to income.

The level of funding and reserves is reviewed at each Board Meeting, and decisions about future commitments are made against our understanding of the economic climate and the prevailing risks to income.

The net assets at the end of the financial year April 2011 were in the order of £313,000, subject to audit and review. Of this amount, £194,000 is held as an endowment for the purpose of protecting the support and advocacy service, £50,000 has been designated for research, and £37,000 has been designated for other commitments. This leaves a balance in the order of £ 32,000 held as reserves to cover contingencies.

Looking to the future, there is a need to address an estimated shortfall against budget of around £45,000 in 2012/2013 and £105,000 in 2013/2014. Actions to address this funding gap will include a review of the grant application process, a vigorous approach to making applications to grant giving organisations and encouragement to new members to consider fund raising ideas.

## Restricted Funds

The NPDG(UK) is in receipt of grants and donations for a number of its activities, which are considered to be restricted funds.

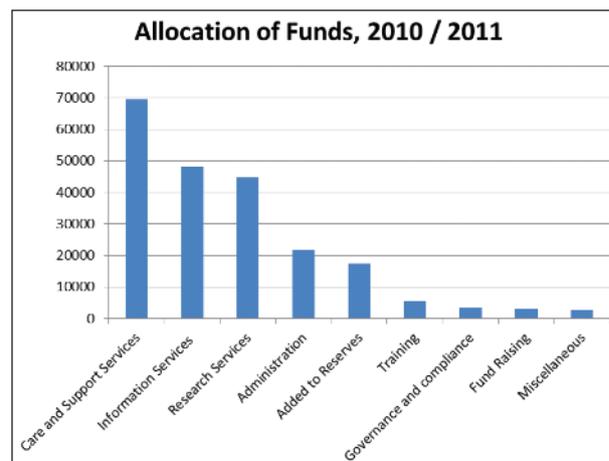
The total amount received during the financial year was £101,922 and the NPDG (UK) confirms that these funds are only used for the purpose for which the funds were given. Further details are contained in the Financial Statement for the year ending 2010/2011, and any underspend is carried forward as restricted funds into the following year.

## Fundraising for the NPDG(UK)

### The Efforts of our Families, Friends and Supporters

The Group would not exist without the fundraising activities by our members, friends and families and the tremendous amount of effort that goes into organising each one of them: – More than half of our operating costs are met by fundraising activities - Recent years have seen an upward trend in the amount raised and we recognize, and are very grateful to, all those who have given their time and effort in support of our work

The following chart shows how the Group uses funds raised to meet its aims and objectives and how your support, and that of our grant funders, is vital to the continuation of our support and services.



As a Charity, we are reliant on fundraising and grants to provide the support and services we offer to families and professionals. Whilst we will strive to reduce our dependence on fundraising, it is vital that we maintain the existing level of income for the benefit of our families.

The NPDG (UK) is a registered charity governed by the Charity Commission for England and Wales, and is therefore required to meet specific criteria in regard to the management of all funds.

**Charity Trustees:**

**Jim Green (Chairman)**  
**Bill Owen (Secretary)**  
**Richard Brooks (Treasurer)**  
**Janice Brooks**  
**Helen Carter**  
**William Evans**  
**David French**  
**Sue French**  
**David Roberts**  
**Richard Rogerson**  
**Prof. Frances Platt (Co-opted)**

**Registered Charity Number: 1061881**

**Medical Advisor**

**Prof J.E.Wraith MB, ChB, FCRP**

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**Website: [www.niemannpick.org.uk](http://www.niemannpick.org.uk)**

**Bankers**

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**Auditors**

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 Edinburgh, EH20 9QH**

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