

A practical guide to Acid  
Sphingomyelinase Deficiency  
Niemann-Pick disease type B

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Published by:

**The Niemann-Pick Disease Group (UK)**

Suite 2, Vermont House

Concord

Washington

NE37 2SQ

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

t 0191 415 0693

e [niemann-pick@zetnet.co.uk](mailto:niemann-pick@zetnet.co.uk)

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# Introduction

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The NPDG (UK) recognises the value of the knowledge, skills and experience gained by families living with Acid Sphingomyelinase Deficiency Niemann-Pick disease type B (ASMD NP-B), therefore A Practical Guide to Living with ASMD Niemann-Pick disease type B (NP-B) has been developed with the help of parents and carers of children, young people and adults affected by this condition. It aims to provide practical information, reassurance and support to those living with ASMD NP-B.

As ASMD NP-B can be a variable condition, the way you need and use the information contained in this guide will be specific to the individual.

If at any time you wish to discuss the content of this guide, please contact:

## **The Niemann-Pick Disease Group (UK)**

Suite 2, Vermont House

Concord

Washington

NE37 2SQ

Tel/fax: 0191 4150693

Email: [niemann-pick@zetnet.co.uk](mailto:niemann-pick@zetnet.co.uk)

The content of this guide has been reviewed and approved by **Dr Simon Jones, Medical advisor** to the Niemann-Pick Disease Group (UK).

# My thoughts on living with ASMD Niemann-Pick disease type B

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## Praying for a cure

A swollen stomach was the only clue to Gina's son's rare problem.

I rushed to the loo — at six weeks pregnant, I was suffering awful sickness. But I couldn't fuss about myself — my son James, two, had begun vomiting as well.

Soon he had a high fever and wouldn't drink, so my husband and I took him to our local hospital. James was admitted with pneumonia, but a doctor noticed his tummy looked swollen. 'His liver and spleen are inflamed,' he said.

James recovered from the pneumonia with intravenous antibiotics, but his tummy was still big. Six months of hospital tests were all inconclusive.

Two months on, I gave birth to William, but we still didn't know what was wrong with James.

Finally, James was referred to Great Ormond Street Hospital in London. Six months later, a specialist diagnosed ASMD Niemann-Pick disease type B.

'It's a rare inherited disease that can cause the major organs to malfunction,' he explained.

My husband and I sat terrified. 'There is no treatment and no cure,' I remember hearing.

Later, I surfed the web. There were different types of Niemann-Pick disease — types A and C were more serious. James had type B — the disease wouldn't affect his brain.

It was a comfort, but then came the next shock. My husband and I discovered we were both carriers of the disease. It's how we'd passed it on. We'd simply had no idea.

Heart banging, I wanted more information and rang the support group — the Niemann-Pick Disease Group (UK). Luckily, there was a Niemann-Pick Family Conference being held in Telford, Shropshire, that weekend, so I decided to go along.



I remember walking into the hall. There were children in wheelchairs, some of whom were very seriously affected by Niemann-Pick disease. There were parents of kids who had died of the disease.

I almost ran away. But I forced myself to stay. For James' sake, I had to find out all I could. It was a positive meeting — the parents and families I met were so supportive.

'Lots of research is being done,' I told my husband when I got home, 'and time is on James' side.'

With the help of the NPDG (UK), we gradually found out more and more about the disease. With every year that's passed, my husband and I have grown more hopeful.

Gene therapy could offer a cure in the distant future. And closer than that, enzyme replacement therapy is being trialled in America. It's something the support group has been pushing for and a real breakthrough in combating this awful disease.

James is now a teenager, a football-loving lad who doesn't look or feel ill. The only signs something is wrong are that he's small for his age and his tummy is still big and hard.

It's difficult for him to sit cross-legged and he avoids contact sports in case he gets knocked. Thankfully, his brother William is clear of the disease.

We know James' condition may deteriorate as he grows older and he could develop breathing difficulties. But I refuse to think of that. As I told my husband right back in the beginning, time is on our side. A cure will be found.

**Gina, mum to James and William**

# The Niemann-Pick Disease Group (UK)

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The Niemann-Pick Disease Group (UK) was formed in 1991, as the 'Niemann-Pick support group' with assistance from Contact a Family and The Research Trust for metabolic disease in Children (now Climb). In 1996 the group was granted charitable status as an independent charity 'The Niemann-Pick Disease Group (UK)'. In January 2012, whilst remaining a registered charity, the NPDG (UK) changed its unincorporated charitable status to a charitable company limited by guarantee.

The NPDG (UK) is entirely supported by voluntary donations and fundraising, and is managed by a Board of Directors primarily consisting of people who have family members or friends directly affected by this group of diseases. The Board of Directors each takes on responsibilities within the group to assist in the achievement of the aims and objectives of the organisation.

The main aims and objectives of 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. We focus on three areas: care and support, information and research.

The NPDG (UK)'s employees are central to achieving our objectives; providing support, information and advocacy services, and raising much needed awareness of Niemann-Pick diseases and the social and economic challenges faced by those affected.

- since 1999, the group has funded the salary of a full time clinical nurse specialist, 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, providing non-clinical advice, information and support
- since 2005, the group's executive director and information officer have managed the NPDG (UK) Central Office, operating a 24 hour helpline and ensuring the smooth running of the group's day to day activities

## Care and support

The NPDG (UK) provides a unique care and support service to families affected by the Niemann-Pick diseases; our aim is to ensure that each family or individual receives the level of support they require to meet their needs:

- our families officer provides non-clinical advice, information and support on an individual basis, including home visits and assistance at clinic appointments
- through our clinical nurse specialist, families can access expert advice on all aspects of the Niemann-Pick diseases, plus genetic counselling and help in navigating the NHS



- the NPDG (UK) Central Office operates a 24 hour helpline providing advice, reassurance or just a friendly voice at the end of the phone
- the Interactive Family Care and Support Project, supported by the Big Lottery fund, uses new technologies and secure video links to enhance access to advice and information and to enable families to communicate face to face with each other

“I decided to contact the NPDG (UK) to meet other families living with the same medical problem, also for my daughter to know she is not alone with ASMD Niemann-Pick Disease Type B, and to give support to other members of the group.”

## Information

The NPDG (UK) provides an information and advice service to families and carers, plus health and social care professionals. We develop and distribute educational information that will contribute to the greater understanding of the Niemann-Pick diseases and assist in family support. We maintain active links, both nationally and internationally, with other support organisations, enabling us to share information and to stimulate interest in this group of diseases.

- accurate and up-to-date information about Niemann-Pick disease and its management is available to families, carers and professionals through our website
- a regular newsletter, e-bulletins and use of social media, such as Facebook, Twitter and Linked-In.
- we produce comprehensive Information packs and publications specifically for each type of Niemann-Pick disease
- the organisation of an Annual Family Conference provides the opportunity for families and professionals to share information and experiences, and to highlight the most recent advances in research and clinical care
- the production of a Family Directory encourages the formation of mutual support networks

“The NPDG (UK) Conference weekend is something I look forward all year because it is one of the few times that I get to see people like me, I could hang out and play football with people that were going through the same sort of thing I was. I have some of the best times of my life at the conferences, like going to the cinema, bowling and trying Archery for the first time.”

## Research

Facilitating progress towards therapeutic interventions for the Niemann-Pick diseases is central to everything we do. Over time the NPDG (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 kept up to date with research that is taking place, its status and where it is happening. Over the years the NPDG (UK) has played a key role in supporting and facilitating many research projects.

- we nurture the interest of young researchers through the annual Peter Carlton Jones Memorial Award for original scientific submissions that contribute to the scientific or public understanding of Niemann-Pick diseases
- we promote relevant research through the relationships we foster and the networks we contribute to – such as the International Niemann-Pick Disease Alliance
- we encourage and support NPD research in the UK through the provision of grants to laboratories working on this group of diseases
- we actively pursue opportunities to support UK based clinical trials, and where possible, to facilitate the extension of US based trials to the UK
- we monitor scientific advances and communicate news in a timely way and in different formats, through our newsletter, website, bulletins and social media

# Acid Sphingomyelinase Deficiency

## Niemann-Pick disease type B -

### The Basics

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#### 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. 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 disease 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

ASMD Niemann-Pick disease types A and B are very rare genetically inherited lysosomal storage disorders in which harmful quantities of a fatty substance called sphingomyelin build up in the body's organs.

Over the last decade it has become apparent that Niemann-Pick types A and 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 (ASM).

Many variations exist within this spectrum, in terms of clinical symptoms and rate of progression, for this reason they may be described or diagnosed separately as ASMD NP type A (NP-A) or ASMD NP type B (NP-B). In ASMD NP-B, the build-up of sphingomyelin mainly occurs in the liver, spleen and lungs, however in ASMD NP-A, storage also occurs in the brain leading to aggressive neurological problems.

- in ASMD Niemann-Pick type A, the accumulation of materials occurs very quickly, an affected child will usually die before reaching three years of age
- ASMD Niemann-Pick type B does not usually affect the brain and, although growth may be slow, those affected will usually survive into adulthood, with many being able to lead a full and normal life
- some patients may be described as having A/B variant, falling in the middle of the spectrum and exhibiting neurological problems which may become more apparent over time

In Niemann-Pick disease type C, the brain and other organs are affected, leading to progressive intellectual decline, loss of motor skills, seizures and dementia. Speech can become slurred and swallowing problems may develop. The rate at which the disease progresses can vary greatly between patients; children who develop neurological symptoms in early childhood are thought to have

a more aggressive form of the disease and may not survive to adolescence, whilst others may remain symptom free for many years.

It is important to remember that, despite some similarity in symptoms, Niemann-Pick Type C and ASMD Niemann-Pick disease are biochemically and genetically different disorders, therefore one will not develop into the other.

Niemann-Pick diseases are genetically caused Lysosomal Storage Disorders (LSD's). Lysosomal storage disorders (LSD's) are a group of approximately 40 rare inherited metabolic disorders that result from defects in lysosomal function. Lysosomal storage disorders result when a specific organelle in the body's cells – the lysosome – malfunctions. All lysosomal storage disorders share a common pathogenesis: a genetic defect in a specific lysosomal enzyme, receptor target, activator protein, membrane protein, or transporter, leading to accumulation of substrates in cell lysosomes.

### Why is it called Niemann-Pick?

The Niemann-Pick diseases are quite separate in terms of the fundamental cause, but the similarities in clinical presentation (the way that they affect the body) have resulted in the naming of the diseases as Niemann-Pick, after two doctors who described the symptoms in the early part of the 20th century.

In 1914, a German paediatrician, Dr Albert Niemann, described the clinical presentation of children with the disease, but at that time little was known regarding what was happening inside the body's cells and molecules.

Then, in the 1920's, the studies of Ludwig Pick provided evidence of a new disorder, one distinct from storage disorders previously described. Further investigations using cells taken from the tissues of affected individuals in the mid and latter years of the century, resulted in an improved understanding of the diseases and their cause.

Since then there has been a considerable amount of investigation into these and other inherited diseases of metabolism. It was not until 1958 that the disease presentations were classified into type A, B and C. In 1966 types A and B were identified with a lysosomal enzyme, acid sphingomyelinase.

### What causes Niemann-Pick disease?

ASMD Niemann-Pick Type A and B are genetically inherited conditions caused by an enzyme deficiency, which results in a build-up of toxic materials in the body's cells. In ASMD NP-B, the enzyme deficiency arises from mutations in a gene on chromosome 11. To have the disease a person or child will have inherited two faulty genes, one on each chromosome 11. This inheritance pattern is called **autosomal recessive**. All types of Niemann-Pick disease are acquired through **autosomal recessive inheritance**; this means that both parents have to be carriers of the faulty gene (mutation). Parents seldom know that they are carriers of the disease and have no control over whether the disease will be transmitted to their child. In each pregnancy of a carrier couple, there is a 25% chance that they will both pass on this gene mutation to their child. A mutation is a change or fault on a normal gene meaning that it does not perform the function that it should do.

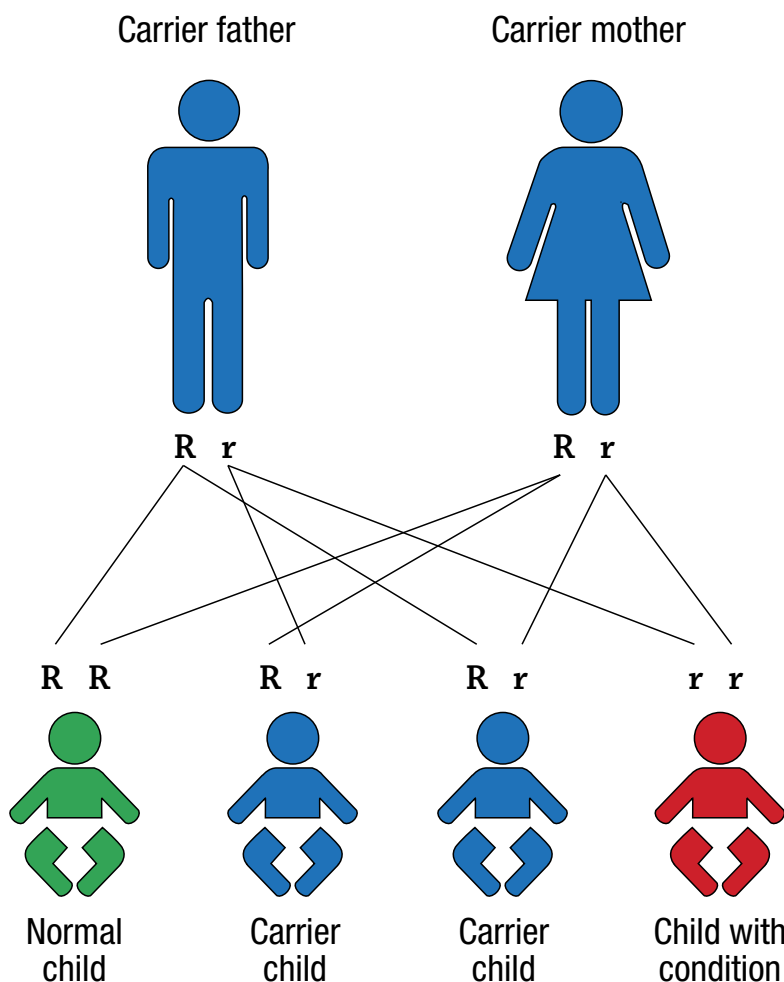
## What is autosomal recessive inheritance?

Autosomal recessive inheritance means that the effects of the disease-causing gene are hidden. With a recessive condition, a person may be a carrier of a disease gene, but with no noticeable affect in their everyday lives and health. A positive diagnosis of ASMD Niemann-Pick type B disease in a child means that each parent is a carrier of a disease causing mutation on this gene.

The child may inherit identical mutations from each parent; this would be referred to as **homozygous**. The homozygous condition may arise through intermarriage although this may not be obvious over many generations. In other instances the parental mutations may be different, but still disease causing; this would be referred to as **heterozygous**.

Genes are found in pairs within the body. When a child is conceived each parent passes just one gene from every pair of genes to their child. The diagram below shows the combinations of a gene that may result if both parents are carriers of one copy of a faulty gene. In this condition each parent is described as being heterozygous for the disease gene.

## Autosomal recessive inheritance



## How is ASMD Niemann-Pick disease type B diagnosed?

ASMD NP-B is diagnosed by measuring the acid sphingomyelinase (ASM) activity in white blood cells or cultured cells. These are obtained from a small blood sample taken from individuals who are suspected of having the disease.

This enzyme test is not reliable at detecting carriers of the condition, and at present it is not possible to accurately predict the severity of the disease by enzyme testing. The gene for ASM has now been cloned and many mutations have been identified so it is possible to detect NP-B by DNA testing. The analysis is complex and consequently can take a long time as it is only performed in one or two centres with a research interest.

The symptoms of ASMD Niemann-Pick type B are variable. Abdominal enlargement may be detected in early childhood but there is usually no neurological involvement, such as loss of motor skills. Growth may be slow and some patients may develop repeated respiratory infections.

Pre-natal testing is available for ASMD Niemann-Pick disease A/B. Cells can be grown from samples taken at around 11 weeks of pregnancy (chorionic vil-lus sampling – CVS) or an amniotic fluid specimen/enzyme specimen can be analysed during the 15th to 20th weeks of pregnancy. If the DNA mutations are known, this process is much quicker.

The incidence of ASMD NP-B is estimated as 1:250,000 in the general population. The number of UK patients, known to the Niemann-Pick Disease Group (UK), is 29 (data correct August 2013).

“My niece underwent 3 weeks of intensive testing before a diagnosis was given. Her mum and dad were told that she had something called Niemann-Pick Disease Type B. They were told it was progressive and degenerative and there was no treatment available. Essentially that day, they had all hope ripped away from them and were shown the door. They were told that they would see a consultant in a couple of months, but when you are given such devastating news, minutes seem like days and a couple of months seems like a long, long time. That was it -we were in no man’s land - numb. We received no contact details, no support, nothing. Instead we had “Google” and we found the NPDG (UK) website and my sister emailed their clinical nurse specialist and within days she had arranged a visit. Her visit breathed life back into the family, she gave them information about ASMD type B and most importantly gave them back hope, we will always be grateful for her support.”



## What are the signs and symptoms of ASMD Niemann-Pick disease type B?

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ASMD NP-B is biochemically similar to ASMD NP-A, however the symptoms usually present later and are less severe. Abdominal enlargement due to an enlarged liver or spleen, may be detected in early childhood but there is usually no neurological involvement, such as loss of motor skills.

Other symptoms can include a progressive enlargement of liver and spleen, poor growth, delayed puberty, susceptibility to respiratory infections, bleeding problems, bone pain and increased stress on the heart

The enlarged spleen often seen in ASMD NP-B is due to storage of the enzyme sphingomyelin. This can cause slow growth, tiredness and increased infections. In addition, it may cause pressure on other organs, bruising and nosebleeds. An enlarged spleen needs to be recognised prior to any surgical procedure, however minor.

Most patients with ASMD NP-B will survive into adulthood, but not without experiencing health problems.

### Is there any treatment for ASMD Niemann-Pick disease type B?

There is no specific treatment for ASMD NP-B but recent advances in medical science have brought the hope of some possible treatments. Patients will benefit from symptomatic treatments - individual medications that can help to ease the symptoms of the disease.

Genzyme, a Sanofi Company, completed a Phase 1 clinical trial in 2009 that was designed to evaluate the safety of single-dose administration of recombinant human acid sphingomyelinase (rhASM) as a potential treatment for ASMD Niemann-Pick disease type B. Genzyme initiated a Phase 1b clinical trial in May 2013 to evaluate the safety and tolerability of rhASM when administered once every two weeks for 6 months. The next step after the Phase 1b will be a Phase 2 trial, which is expected to commence early in 2014.

### What, typically, is the age when ASMD Niemann-Pick disease type B becomes apparent?

ASMD NP-B is present in an individual from the moment of conception but is not necessarily apparent. The symptoms are often related to the age when the disease takes hold and, this is in itself an extremely variable factor ranging from birth to old age. Similarly, people with very late onset may be incorrectly diagnosed.

With rising disease awareness and improving diagnostics, knowledge and understanding of the disease is increasing. Natural History information – that is information on the number of people affected, age of onset and progression – is currently being collected in a number of countries; this will contribute to the greater understanding of this condition and provide valuable data for scientists and researchers working in this field.

## Prenatal testing and pre-implantation genetic diagnosis

Once a child has been diagnosed with ASMD NP-B, it is usually possible to test future pregnancies at 11 weeks (chorionic villus sampling - CVS) or at around 13 weeks by amniocentesis. Ideally it is best to have looked at the affected child's DNA to identify the mutations first, as this will enable the prenatal test results to be confirmed within a shorter time frame.

If none of the mutations have been identified, or only one has been found, skin biopsies are carried out on both parents - this gives a specific picture for the affected child and the parents, enabling the comparison of samples from future pregnancies. This method involves growing the sample from the placenta of the baby, which takes around 6 weeks from the date the sample is taken. A result will therefore not usually be available until 16-20 weeks into the pregnancy, when the decision on how to proceed has to be made.

Both CVS and amniocentesis do carry a slight risk of miscarriage, so it is important for parents to discuss their options prior to having the test. If parents have decided they would not wish to terminate a pregnancy under any circumstances, they may then feel that putting that pregnancy at risk, however small that risk, would not be an option.

Pre-implantation genetic diagnosis (PGD) is now available in the UK. PGD is based on IVF-in vitro fertilisation and the genetic mutations from each parent need to be known in order to proceed. The principle behind it is that very early embryos, soon after the early divisions, are checked for the known mutations, and only an unaffected or possibly carrier embryo would then be implanted. This procedure is in its infancy and may not be possible for all parents; please speak to our clinical nurse specialist for further information.



# Coping with a diagnosis

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When your child or loved one is diagnosed with ASMD Niemann-Pick disease type B it can adversely affect your whole family. For many families, a diagnosis of ASMD NP-B does not happen instantly, and can come at the end of weeks or months of tests and investigations. This can be, however, the point from which you begin to contemplate the future. During this period of adjustment families can face what can feel like a never ending round of appointments with the long list of professionals that will now be involved in caring for their loved one. You may find yourself having to tell your story time and time again.

You may feel that your hopes and dreams for the future are shattered: you are suddenly faced with the news that your child or family member has a long term health condition, and no-one can tell you the exact path the disease will take.

“I was diagnosed when I was three years old, and from an early age I knew that I was different, that I was special. Unlike the other kids I was always going into London for tests at Great Ormond Street. I had to have blood tests, flu jabs, x-rays, overnight stays at the hospital for monitoring. But the biggest hint was my large tummy that was noticeable from a toddler.”

## Where do I start?

At this time there is a lot to think about at a point when you may not very capable of thinking clearly or taking information in.

The NPDG (UK) clinical nurse specialist and families officer can help families to make sense of things in the days following a diagnosis. They can work directly with your local health and social care professionals to ensure they understand the medical, clinical and social needs of your child/loved one.

Although the clinical nurse specialist and families officer are available to give advice over the telephone, they will often travel to work with families in their own homes throughout the UK and they would be happy to accompany you to medical and other appointments as required.

The Niemann-Pick Disease Group (UK) can provide information about the disease that you can distribute to your local health care team and also to your family and friends. The group provide emotional, as well as practical support; if you would like to speak to someone about any aspect of the disease, you can call the 24 hour helpline at any time. The group can also put you in touch with other families who know what you are going through.

There may also be many health and social care professionals involved in caring for your loved one, both in your local area and nationally at a designated specialist centre. These may include:

## Your GP

Your GP is often your first port of call and is a valuable source of help and information. Provide them with information about ASMD NP-B (The NPDG (UK) can help you in this), so they can offer appropriate advice and support as and when it is needed.

### **Metabolic consultant**

Your metabolic consultant will have expert knowledge and experience of working with ASMD NP-B patients. Usually seen at your designated specialist centre, your metabolic consultant will also co-ordinate appointments with other specialists, such as cardio/respiratory, haematology and orthopaedics.

### **Dietician**

A dietician will provide advice on nutritional needs, including calories, vitamins and minerals.

### **Cardio/respiratory consultant**

A cardio/respiratory consultant will monitor circulatory systems and respiratory function. Appointments will usually coincide with that of your metabolic consultant at your designated specialist centre.

### **Genetic counsellor**

Genetic counselling can help you and your family to better understand ASMD NP-B and the reasons why it has affected your family. A genetic counsellor will discuss the risk of extended family members developing the condition and provide information about carrier testing. They can also advise on future family planning.

### **Haematologist**

Haematologists are specialists who play an active role in the treatment and care of patients, A haematologist will have a particular interest in diseases of the blood and organs. Your haematologist will monitor blood platelet count and liver function. Appointments will usually coincide with that of your metabolic consultant at your designated specialist centre

### **Orthopaedic consultant**

Orthopaedics is the study of the musculoskeletal system which involves the bones, joints, ligaments and tendons, as well as muscles and nerves. Appointments will usually coincide with that of your metabolic consultant at your designated specialist centre.

### **Physiotherapist**

Your physiotherapist will provide help and advice regarding movement and posture.

### **Social worker**

Social workers are there to help you to access the benefits and services you require, they can also assist you in completing paper work for grants and benefits.

## Occupational therapist (OT)

An OT will assist you in obtaining any practical aids, adaptations or equipment that may be required

## Key worker

A key worker provides a vital link between your family and other health and social care professionals. This person will work closely with you to understand your needs and will liaise with other health and social care agencies to address these needs. Ideally, this role needs to be taken on by someone local to the family, who is familiar with local service provision. Often this person is a social worker or a nurse and will ensure that the team of professionals involved with your loved one work together, are aware of the role each will play and are kept up to date with developments through regular meetings.

## NPDG (UK) clinical nurse specialist

The NPDG (UK) clinical nurse specialist provides expert care and practical advice, plus home visits whenever necessary. Genetic counselling and advocacy services are also provided.

## NPDG (UK) families officer

The NPDG (UK) families officer offers an individual advocacy service, including home visits, assistance at clinic appointments, and providing non-clinical advice, information and support.

**To ensure optimum care, the clinical nurse specialist and families officer will help to arrange multi-disciplinary team meetings, usually annually or more often if need be, to ensure that all health and social care professionals work together**

**The NPDG (UK) can provide an information booklet to all professionals involved in caring for your loved one – please contact us for further details.**

## Developing an emergency health care plan

Once you have established a relationship with your local care team it will be useful to sit down with your consultant and discuss the development of an emergency care plan. This will help you and your care team to understand your/your child's wishes, in the event that a life-threatening situation should arise.

Unless you have discussed an emergency care plan with your consultant, and you have agreed to this plan and put it in writing, your wishes may not be observed.

Whatever decisions you come to, it is very important that everyone knows your wishes, in order that they can provide you with support at times of crisis. Remember – the plan can be reviewed or changed at any time, according to your wishes. Copies should be kept with the person, and also given to others as appropriate, such as schools.

“Although I have had problems in the past, I don’t let my condition rule my life; I skateboard, ski, hang out with mates, and even have a season ticket at Watford FC. The only time I remember that I have what I have is when it is relevant, anytime else I’m just a normal kid, a 1 in 6 million normal kid.”

## Caring for the carer

Carers are people who provide help and support to a family member or a friend who may not be able to manage because of frailty, illness or disability. Carers can be adults caring for other adults, parents caring for ill or disabled children or young people who care for another family member.

Carers are often so preoccupied with the health and well-being of the person they are caring for that their own needs are forgotten. However, it is important not to neglect your own health in the process of care giving. This is, of course, easier said than done - but it doesn’t have to take a lot of time, often just a few minutes of quiet can help. Looking after your own health and emotional needs will mean that you are more able to carry out your role as a carer.

If you are a carer, you may be entitled to receive help and support from social services and other organisations. Your doctor (GP) is a good person to talk to about this or any other issues you may have and will help you to access the information and support that you require.

## Some practical hints and tips

**Eat.** Take time for regular meals.

**Stay healthy.** Although your life is busy and your attention focused on the health of your loved one, don’t ignore your own health needs. Keeping an eye on your health will make a real difference to the way you cope and how you feel. Simple things like eating healthily and taking vitamins can make a big difference.

**Exercise.** You may feel tired, but regular, moderate exercise will actually give you more rather than less energy. As a bonus you’ll sleep better too. Try to make time throughout the day to exercise, if you can create just a twenty-minute gap in the day, it will be worth it. Exercise is not only good for you but can combat depression – a regular walk and some fresh air can do wonders for your well-being. Choose an activity that is right for you – one that you enjoy. Simplest of all is a regular, daily walk – round the block, to the nearest park, or to the shops. The closer to home the exercise is, the easier you will find it to do regularly.

**Sleep.** Try to get enough sleep. Carers often go short of sleep – sometimes they have too much on their minds to be able to relax. Try to establish a good bedtime routine, avoid alcohol, have a warm drink and a bath and make sure the bedroom isn’t overheated. If you like to read before going to sleep, choose something soothing. If you like to listen to the radio, play some music rather than listen to the late-night news. If you wake in the night and find your head is full of worries, try spending an hour or so out of bed, or sitting up with the light on if you sleep alone. You can read, do the ironing, watch television or write down everything that’s on your mind. You’ll find these are preferable to tossing and turning in the dark.

Remember there is no such thing as a perfect caregiver – feelings of anger, frustration and guilt are normal. Share your feelings with a friend, relative, doctor, counsellor or support group.

### Further information for carers

The 'Caring for Someone' section of the **Directgov** website provides detailed information about the help and support available to carers, including carers' rights, employment issues and money matters. For more information visit: [www.direct.gov.uk/en/CaringForSomeone/index.htm](http://www.direct.gov.uk/en/CaringForSomeone/index.htm)

**Carers UK** aims to improve the lives of carers and to ensure that they receive the same rights as everyone else - a fair level of income, adequate support to protect their health and well being and access to the world of work, leisure and education. For more information visit [www.carersuk.org](http://www.carersuk.org)

**The Princess Royal Trust for Carers** is the largest provider of comprehensive carers support services in the UK. The Trust currently provides quality information, advice and support services to carers, including young carers. Visit their website [www.carers.org](http://www.carers.org) for information on all the key issues facing carers.

### Relationships

When a child is diagnosed with a rare condition, such as ASMD Niemann-Pick disease type B (ASMD NP-B), it can affect the whole family. Hopes and dreams for the future are suddenly altered, and many families can struggle to cope with the emotional and financial implications of the diagnosis. As a result of this, the quality of family life can be severely affected.

Relationships, when they are working well, provide us with stability and support. For those living with ASMD NP-B, there may be adjustments to be made, such as new roles, emotional and financial pressures and an astonishing amount of new information to absorb.

You may find that your marriage is more susceptible than usual to these additional challenges and stresses, as you and your spouse adjust to the changes in your lives. Each of you will do this in different ways and often at a different pace – one of you may want to talk openly about the situation whilst the other may need more time. It is normal for feelings of sadness, worry, denial, anger, embarrassment, fear, confusion, guilt, concern, resentment, and shock to occur before a sense of acceptance enters your hearts.

Receiving a diagnosis of ASMD NP-B can represent a loss of control. It takes time to reach acceptance and a willingness to adapt. One of the main challenges that illness may place upon marriage is to find a balance of dependence and independence. It is important for you to talk about the needs of your loved one, plus your individual needs, and how they may be affected, both practically and emotionally. Look at how life has changed, or may change in the future, for each of you and in what ways – ask how you can best support your family, and each other.

### What can you do to strengthen your marriage?

- communicate - talk openly about issues when they occur, listen to each other without criticising or judging, try to understand each other's point of view

- realise that there will be disruptions in the course of your marriage now and then, as there can be in all marriages
- physical intimacy is an important part of a relationship, but tiredness and stress can take its toll. This is a common issue for many parents – talking about your needs and expectations can help
- be patient with one another, each of you will be under pressure, show support for one another in small ways, such as emotionally, by really listening to them or physically, with a hug or a squeeze of the arm
- together, learn all you can about your loved one's condition, and seek help or advice that will provide practical and helpful day to day solutions
- remember to take care of your relationship. Make time for the two of you to be alone whenever you can
- think about your lives and how things have changed, talk about the future, share your hopes and fears

Remember, **all** couples argue, if a conflict occurs, give each other some uninterrupted time to express your feelings. Be prepared to compromise – if you can't reach an agreement – why not agree to disagree? Often re-visiting the issue at a later date will provide time for both of you to consider each others point of view and it will then be easier to address. Try to keep your sense of humour and to understand that some rows often have an underlying cause – tiredness, stress or feelings of resentment. Again, communication is key to working out the real cause and understanding the feelings underneath.

If you are worried about your relationship, there are many resources that could provide help. Both women and men can be victims of domestic violence It may be difficult to admit to yourself and others that this is happening, but it's not your fault – and help is available. The police and your local social services department can provide help and advice in this area. You will find their contact details in your telephone directory. You can also call the NPDG (UK) for details of other organisations that can provide support for victims of domestic violence.

## Resources

### **Relate**

Offers advice, relationship counselling, sex therapy, consultations and support face-to-face, by phone and through their website.

Herbert Gray College,  
Little Church Street,  
Rugby CV21 3AP  
Tel: 0845 456 1310  
Web: [www.relate.org.uk](http://www.relate.org.uk)

### **Relate Scotland**

Offers advice, relationship counselling, sex therapy, consultations and support face-to-face, by phone and through their website.

18 York Place  
Edinburgh EH1 3EP  
Tel: 0845 119 6088  
Web: [www.relatescotland.org.uk](http://www.relatescotland.org.uk)

### **Marriage Care**

Marriage care helps people prepare for, achieve and sustain successful marriages and to support them should their marriage break down.

1 Blythe Mews,  
Blythe Road,

London W14 0NW

Tel: 0845 660 6000

Web: [www.marriagecare.org.uk](http://www.marriagecare.org.uk)

### **Parentline Plus**

Parentline Plus is a UK charity offering support to anyone parenting a child. They run a freephone helpline, develop innovative projects and provide a range of information.

520 Highgate Studios,

53-79 Highgate Road,

London, NW5 1TL

Tel: 0808 800 2222 (24hrs)

Web: [www.parentlineplus.org.uk](http://www.parentlineplus.org.uk)

**Further details of organisations and services that may be able to help can be found on the NPDG (UK) website [www.niemann-pick.org.uk](http://www.niemann-pick.org.uk)**

## Siblings

### **Giving siblings information about their brother or sister's diagnosis**

Parents often ask when they should give siblings information, and how much to give them. Parents can worry that giving information to siblings will make them anxious or will be a burden to them. For many, the hardest thing about talking about the illness is their own uncomfortable feelings that can arise when they do so.

Of course the age of the sibling has a big bearing on how much information to give. Think about it like sex education - give matter of fact information with less detail to young children, and more detailed information and discussion of the issues for older children.

In children under 7 years, you will need to gauge how ready they are for information by the questions they ask. When children get to about 7 years, they are likely to be asked questions at school about their brother or sister and this will increase their need for information.

Siblings usually feel less anxious if they are kept informed about what is going on, and if they are not given factual information they may make up their own incorrect story about what is happening. Always answer questions honestly and simply. Even young children will detect if you are not being honest and will lose trust in you as a result.

**“For two years I had hormone injections to help me grow, as I was a little on the small side. My brother was tall for his age and when we spent time together I got a little fed up with being compared to Danny Devito and Arnold Schwarzenegger in the movie ‘Twins.’”**

## Ways to get talking about ASMD NP-B with siblings

### With younger children:

- with very young children, an easy way to start discussions is by looking at a picture book together. Ask what he or she knows about illness in general and start from there – you may find that he or she is well informed or that they need more accurate and age appropriate information
- to help younger children, you can make a poster with the names of the professionals on it and write down, or draw, or stick on pictures about what they do. Ask professionals if you can take a photo of them at their work to help a young sibling remember what they do. This will give you a prop for talking with younger siblings
- encourage them to ask questions that you could ask a professional on their behalf – this will help younger siblings feel a sense of control over the things that are happening in their family

### With older children:

- ask the older sibling if they would like to write down a list of questions for you to answer about the illness, if they seem reluctant to start a face to face talk about it
- you may wish to look at information on the internet together, or at specialist information provided by the NPDG (UK). Always read the information yourself first before you show it to your child
- ask older siblings if they would like to know more about the professionals who are involved with the family. Explain the role of the professionals and the practical ways that they are helping their brother or sister; would they like to meet them and ask any questions?

### Be aware of the sibling issues that may have the most impact at certain stages

Certain issues are particularly relevant for siblings at different stages in their lives. This is a rough guide and these will vary from one child to another and will be affected by each family's own circumstances.

#### Pre-school children 0-5

- less time and attention from parents
- imitation of an older child but not understanding unusual or unfamiliar behaviour
- feeling jealous of the child with the disability or illness
- anxiety through not understanding a brother or sister's condition – child may not be able to verbalise this
- frequent absence of parent and child for hospital stays or visits – most difficult for younger children



## School age children 5-11

- telling friends and others about their brother/sister
- dealing with teasing and embarrassment – tends to happen from 7 onwards
- needing information about their brother or sister's condition
- not having enough time together as a family
- teachers not understanding the pressures at home
- when rules or discipline are different for the sibling and the other child
- frustration if the children are not able to play the same games

## Adolescents 12 +

- dealing with embarrassment
- concern about being like their brother or sister - identity issues
- having enough space and opportunities to develop their own life
- concern about their brother or sister's future
- not wanting to burden parents with their own emotional needs
- dealing with introducing new friends to the family
- responsibility, confusion or guilt about being unaffected by the disease
- concerns about having their own children as they start to learn about genetics at school



## Resources

### Sibs

Sibs is the UK charity who support siblings who are growing up with, or who have grown up with, a brother or sister with any disability, long term chronic illness, or life limiting condition. There are over half a million young siblings and over a million adult siblings in the UK. Siblings have specific needs that require attention at different stages of their lives, including relief from isolation, information, and strategies for coping with the situations they find themselves in.

Sibs aim is to enhance the lives of siblings by providing them with information and support, and by influencing service provision for siblings throughout the UK.

#### Sibs can help in the following ways:

- **young siblings** are offered advice and ideas for coping with difficult situations and the opportunity to learn about the experiences of other siblings
- **adult siblings** can get information on planning for the future and on dealing with issues related to childhood
- **parents** can find out about sibling issues and get tips on supporting siblings at home
- **professionals** can learn more about sibling issues and how to support siblings through their work with families

To find out more about Sibs visit their website [www.sibs.org.uk](http://www.sibs.org.uk) or telephone 01535 645453

### Grandparents

When a child is diagnosed with a life-limiting condition, it can be very hard for grandparents to deal with; most grandparents want to help but don't always know how best to do this. They can also become absorbed by the impact that the diagnosis has on their own children, who are now parents themselves. Most are also concerned about the effects it will have on any other children within the family, and of course, how, as grandparents, they themselves will cope.

Parents of the sick child usually have access to doctors and others who can answer their questions. It is not as easy for grandparents to get information first hand and this can lead to feelings of isolation.

Talk to them about how things are for you and your family, and let them ask questions. If you find this hard, the NPDG (UK) families officer can help you, or can talk to them on your behalf. Stress that whatever information they have heard or read must be taken in context – ASMD NP-B is a variable condition and can be easily confused with other similarly named conditions.

When a family receives a diagnosis of a rare disease such as ASMD NP-B, there is nothing they could have done to prevent it from happening. Most grandparents accept that this is the case, but many still feel helpless, or even guilty, when it is diagnosed, as they see protecting their offspring as their most important role. For some grandparents their faith can be a great comfort, while for others it may be seriously challenged.

Many grandparents play crucial roles in the lives of their grandchildren even before there is illness in the family and they can be a valuable source of support, especially with siblings:

- doing an activity with the affected child or sibling that you can't do as a family (swimming, going to the cinema, having a long walk in the park, help with learning to ride a bike, etc)
- getting one to one time for attention and nurturing – being read a story, getting help with homework, cooking together, etc
- being available on the phone for chats - children will often 'open up' to their grandparents about issues that they feel unable to talk to parents about
- let them know that their contact is really important for you and for the children
- try to be realistic about the kind of help grandparents can offer and remember that they have needs too, including thinking about their own health

### Resources:

**Contact a Family has a dedicated fact sheet for Grandparents:**  
[www.cafamily.org.uk/grandparents.html](http://www.cafamily.org.uk/grandparents.html) or call 0808 808 3555

### Grandparents plus

**Promotes the role of grandparents at all levels.**  
[www.grandparentsplus.org.uk](http://www.grandparentsplus.org.uk) or call 020 8981 8001

### Extended family and friends

Parenting is always easier with support from other people. Receiving a diagnosis of a rare condition such as ASMD NP-B means that you may need additional support now and again. Your extended family and friends can be a great source of support and can offer different types of support for you - when you need a break, someone to talk to, have some quiet time to yourself, or someone to help you do a task like cleaning or shopping.

Help them to understand your situation, how you are feeling and what it means to your family. It can be difficult to find the words to explain a diagnosis to those you are close to: NPDG (UK) can provide specialist materials to help you with this or the NPDG (UK) families officer can visit and chat with your family and friends.

Building an informal support network can be invaluable. If possible, it is better to have a list of people who can all offer different things, rather than expecting one or two people to take on much bigger roles. Some may be available to help you on a regular basis, others may be happy to help when you are stuck or have an emergency. Helping can give others a better understanding of your situation; but sometimes you may need to make the first move and to recognise when you could do with an extra hand.

“I admit that I often tried not to ask people for help even if I needed it – I don’t think this is a good approach. I think it’s important to have the confidence to be honest - with unseen conditions there is often the risk of being viewed as making a fuss over ‘nothing’. I made use of the Disability Services and Counselling services at uni to help me come to terms with these things and deal with them better. I found it was useful for me to rehearse how to explain my condition quickly and clearly, in a way that was easy for people to understand. It took a bit longer for me, but after I learned to be honest about things I made friends for life - and who I knew I could really rely on.”

### Some of the things you can ask for help with:

- transport – by car, taking a child on the bus, bringing a child to or from school, giving you a lift somewhere
- shopping – getting a prescription, getting the large shopping, helping a sibling get a birthday present for a friend
- practical chores, such as housework, cooking or ironing
- helping a sibling with homework
- forge links with people with children the same age so ‘play dates’ can be arranged
- sitting - giving you quality time with a sibling, spouse or partner

# Living with ASMD Niemann-Pick disease type B

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The following information has been generated by a group of experts – parents and carers who have one thing in common; the need for practical solutions that will help them to provide a high level of care for their loved one. By sharing their knowledge and experience of caring, they hope to provide a valuable resource for others.

This information is not a substitute for professional medical care – it is meant to complement the advice and support you receive from your health and social care team.

Please also be aware that ASMD NP-B is a variable disease, meaning that the symptoms experienced by each affected person, and the rate the disease progresses, will be different from person to person.

The NPDG (UK) clinical nurse specialist would be happy to offer further advice and information on specific questions, if you are unable to find answers to your questions in the section below.

“My mum and dad were always truthful with me and always told me that what ever I wanted to do I should try for, whatever the obstacles.”

## Symptoms and solutions to common problems

*Please note; the following is in alphabetical order and not in order of onset or importance*

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### Aids and adaptations

If necessary, there are a number of aids and adaptations that may help to make daily life easier.

### What you can do

As an enlarged, distended spleen is often seen in those with ASMD NP-B, the use of a spleen guard for school children may be helpful, as the enlarged spleen is not protected by the rib cage and can be susceptible to injury. However, the NPDG (UK) has had not received reports of children or adults injuring their spleen whilst undertaking normal daily activities and it is always advisable to seek professional advice before using a spleen guard.

A spleen guard is a hard piece of moulded plastic, individually fitted over the abdomen to protect an enlarged spleen. The moulded piece of plastic may be held in place by straps or a wide band of elastic that wraps around the abdomen, fastened by Velcro secured on the plastic mould. It is sometimes lined with soft material. This can help to protect the spleen from injury during play sessions.

For those who experience severe respiratory issues or tiredness, the use of an electric wheelchair may help with mobility, and the consideration of

adaptions to the home may be necessary. As going up and down the stairs may be an issue, adapting your home to enable permanent use of the ground floor can be helpful.

“As I progressed into my teens, that’s when I started to take more notice of what was wrong with me. I was and still am noticeably shorter than the other boys and due to my over-sized, liver and spleen I had to sit out of PE lessons whenever we did full contact sports such as rugby. I had to quit playing football in my team as I was in danger of getting hurt as the other teams didn’t know what was wrong and I was an easy target due to my size.”

### Who can help?

In the first instance, discuss your needs with your GP, he will usually refer you to an occupational therapist (OT) who can assist you in obtaining any practical aids, adaptations or equipment that may be required. For further information or advice, you can contact the NPDG (UK) clinical nurse specialist

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### Body image

Body image refers to how people see themselves. ASMD NP-B can cause slow growth in children and teenagers, meaning they can be shorter in stature and have an enlarged spleen, which can impact the way in which children see themselves and also their relationship with their peers, causing them to have a negative body image and low self-esteem.

If you notice your child scrutinising themselves in mirrors, making disparaging comments about their body or frequently comparing themselves to others - perhaps to a friend or a celebrity, these can be signs of negative body image.

Weight and body shape can also impact on body image. A child with ASMD NP-B may have a normally healthy weight range for their age, but have an enlarged stomach due to splenomegaly.

In addition, the late onset of puberty can have an effect on a child who sees their peers ‘growing up’ whilst he or she remains ‘a child’. When changes to the body do happen, this can also lead to insecurity and negative body image in formerly confident, self-assured children.

“When I first started my new school people made up a rumour that I was pregnant because of the size of my belly; but no-one says it now because everyone knows me.”

### What you can do

The first step is to ensure open communication with your child, in regard to their feelings about their body image. Try to counter the effects of the media culture of perfection, explaining in realistic terms that everyone is different. Keep an eye out for good role models — for instance, those who aren’t unnaturally slender or who have confidence in their own beauty, underlining the message that real-life people come in all shapes and sizes and lead successful and interesting lives.

Secondly, you can take active steps to teach your child healthy ideas about food and eating. Try to create lifestyle changes that foster a relaxed relationship with food, free of bingeing, restricting, and emotional eating. Never portray any food as an enemy. Educate your child about the health benefits of high-nutrition foods, but don't place junk food completely off-limits, as we can 'crave;' what is off limits. Treat all family members equally with regard to food. Never single out a child based on his or her condition/weight/body shape.

In addition, talking freely about the changes that may happen during puberty, and reassuring a child who is not yet going through these changes can help. The age at which puberty usually occurs varies in girls from between 7 to 13 and boys 9 to 15. In ASMD NP-B it is more likely to be at the later end of the scale, but it will happen. Help children to recognize and prepare for this by discussing the facts about differences in body size, shape and rate of development.

Clothing can also present an issue – teens do like to dress in the latest trend or in a similar way to their friends. With an enlarged spleen, finding clothes that reflect this and also fit can be challenging. Try to find appropriate clothing that your child is happy to wear and is comfortable.

“The first year of living with this strange medical problem was very, very difficult, coming to terms with the sudden change in our life. I felt my daughter was the only person with ASMD Niemann-Pick Type B. As she grows older she is becoming more aware of her body, and would like a flat tummy like her friends. She is also aware of people looking and pointing, this happens in the summer when she wears her beautiful summer dresses. My way of helping her to cope with this is letting her know how special and beautiful she is. My daughter is a confident and bright child, her attitude is positive and we aim to live a normal and happy life.”

## Who can help?

Recognizing, acknowledging and talking about their feelings about body image will help children to become more comfortable in their body and to understand why changes are happening/not happening. If you are struggling to engage your child, speak to your GP or to a trusted member of your health care team, who will be able to offer advice and information.

You might also be referred to a paediatric endocrinologist, a doctor who specialises in treating children and teens who have growth problems, or to another specialist for further tests or treatment.

In some cases, doctors may offer teens a short course (usually a few months) of treatment with hormone medications to get the changes of puberty started. Usually, when the treatment is stopped a few months later, the teen's own hormones will take over from there to complete the process of puberty.

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## Bones

ASMD NP-B can affect your bones. Bone is a living growing tissue that turns over at a rate of about 10% per year. It is made of collagen – the protein that gives bone its strength – and calcium phosphate which hardens the framework. Bone formation happens faster in children and teenagers and begins to slow

in your twenties. After that, small amounts of bone are absorbed by the body (bone resorption) and this happens faster than the formation of new bone.

### What may happen

Osteoporosis, or porous bone, is a disease in which there is a loss of bone mass and destruction of bone tissue. Osteoporosis develops when bone formation slows and resorption occurs too rapidly. This process causes weakening of the bones and makes them more likely to break, meaning you may be at greater risk of fractures to the hip, spine and wrist.

Osteoporosis is often called the “silent disease” as symptoms may not be noticeable.

Some people may experience pain in their bones and muscles, particularly in their back. Occasionally, a collapsed vertebra may cause severe pain, decrease in height, or deformity in the spine.

Osteopenia is another bone condition characterized by a decreased density of bone, which leads to bone weakening and an increased risk of fracture. Osteopenia and osteoporosis are related conditions. In osteopenia, however, the bone loss is not as severe as in osteoporosis. That means someone with osteopenia is more likely to fracture a bone than someone with a normal bone density but is less likely to fracture a bone than someone with osteoporosis.

### What you can do

There are many steps you can take to help keep your bones as healthy as possible. To help keep your bones strong and slow down bone loss, you can:

- eat a diet rich in calcium and vitamin D
- exercise
- not drink in excess or smoke

A healthy diet with enough calcium and vitamin D can help to keep your bones as strong as possible. Good sources of calcium are:

- low-fat milk, yogurt, and cheese
- foods with added calcium such as orange juice, cereals, and breads

Vitamin D is also needed for strong bones. Some people may need to take vitamin D supplements – consult your doctor for further advice.

“Suddenly, one day I got strong problems when I was walking. Both feet hurt whenever I took a step. At first I did not take it serious and thought I had worn uncomfortable shoes. But unfortunately the pain became worse and both feet got swollen and red on their sides. At this point I could hardly walk and my shoes didn’t fit anymore. On the day of the appointment suddenly things happened quickly: again a lot of blood testing was done, I got anti-rheumatic medication and the swelling had slowly receded. I was so happy that finally something worked! My feet slowly became normal again but the pain stayed – however not as strong as before. Since I now look after myself and only do what my body allows I can bear the pain. They still haven’t found the cause for all my pain. But it is assumed that the pain is due to the ASMD Niemann-Pick disease type B.”



## Who can help?

Your consultant will refer you to a bone specialist (usually an orthopaedic consultant) who will check your bone density using a DEXA scan – this is a special X-ray of the bones.

Monitoring your bone density at regular intervals can help, so that the appropriate medical therapy and treatment can be implemented if necessary. Early treatment can help to prevent future bone fractures.

The advice of a dietician could also be helpful. They can advise you in regard to nutrition and a healthy balance diet.

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## Delayed puberty

Puberty is the time when your body develops from a child's to an adult's. You'll know that you are going through puberty by the way that your body changes.

If you're a girl, you'll notice that your breasts develop and your pubic hair grows, that you have a growth spurt, and that you get your period (menstruation).

If you're a boy, your voice will change, you'll start growing pubic and facial hair, have a growth spurt, and your testicles and penis will get larger.

These changes are caused by the sex hormones (testosterone in boys and estrogen in girls) that your body begins producing in much larger amounts than before.

Puberty takes place over a number of years, and the age at which it starts and ends varies widely. It generally begins somewhere between the ages of 7 and 13 for girls, and somewhere between the ages of 9 and 15 for boys, although it can be earlier or later for some people. For children with ASMD NP-B, it is more likely to occur at the later end of the range but continue well into the twenties.

## What may happen:

Sometimes, though, people pass this normal age range for puberty without showing any signs of body changes. This is called delayed puberty.

Medical problems also can cause delays in puberty. People affected by ASMD NP-B may go through puberty at an older age because their condition can make it harder for their bodies to grow and develop.

## What you can do

It can be really hard to watch your friends grow and develop when the same things are not happening to you. You may feel like you're never going to catch up. People at school may joke about your small size or your flat chest. Even when the doctor or your parents reassure you that things will be OK eventually — and even when you believe they're right — it's difficult to wait for something that can affect how you feel about yourself.

If you're feeling depressed or having school or other problems related to delays in your growth and development, talk to your parents, your doctor, or another trusted adult. This person can help you sort out your feelings and suggest ways to cope with them.

Delayed puberty can be difficult for anyone to accept and deal with — but it's a problem that usually gets solved. Ask for help if you have any concerns

about your development. And remember that in most cases you will catch up with your peers.

“My nephew is now a teenager and has realised that he is different from his friends, whilst they have all grown he has remained small and has a bigger stomach. He is also asking questions about why he has to attend hospital appointments and not play contact sports. He may be the smallest child in his class but he is certainly the brightest - he has a zest for life, and his creativity and imagination know no bounds!”

### Who can help?

Your consultant might refer you to a paediatric endocrinologist, a doctor who specialises in treating children and teens who have growth problems, or to another specialist for further tests or treatment.

Some teens may have a difficult time waiting for the changes of puberty to finally get going — even after a doctor has reassured them that everything will be fine. In some cases, doctors may offer teens a short course (usually a few months) of treatment with hormone medications to get the changes of puberty started. Usually, when the treatment is stopped a few months later, the teen’s own hormones will take over from there to complete the process of puberty.

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### Fatigue

Fatigue is frequently reported in patients with ASMD NP-B for several reasons. Most patients have a large spleen which causes increased breakdown of blood cells. Blood is made of plasma, a fluid which contains red and white blood cells, platelets and proteins. Red blood cells take oxygen around the body, whilst white blood cells, which form part of the immune system, defend the body from infection. Platelets help the blood to clot and the proteins have various other functions.

An enlarged spleen results in low levels of all blood cells, including red blood cells which are responsible for the oxygen flow around the body.

Anaemia is a condition in which there are fewer red blood cells than normal, meaning that a reduced amount of oxygen is carried around in the bloodstream, resulting in fatigue.

In addition, storage of fatty substances associated with ASMD NP-B can occur in the lungs, further reducing the flow of oxygen to the body’s cells.

A large spleen also uses up a lot of energy from the calories (food) you consume, so even if you eat a healthy balanced diet, your body may still not have insufficient energy to function and grow normally. A dietician can advise you about adjusting your diet to allow an intake of extra calories - without eating too much at each meal - as an enlarged spleen can press on the stomach and limit intake.

“I found myself tired more than most at school; doing my GCSE’s was particularly difficult”

## What you can do?

Try these simple adjustments that could help to reduce fatigue:

**Eat little and often to beat tiredness.** A good way to keep up your energy through the day is to eat regular meals and healthy snacks every three to four hours, rather than a large meal less often. In addition, cut down on caffeine and alcohol and drink more water to keep you hydrated and boost energy levels.

**Take gentle exercise.** You might feel too tired to exercise, but regular exercise will make you feel less tired in the long run and you'll have more energy. Start with a small amount of exercise, try a short walk, or a gentle swim, and gradually build up your activity to a level that feels right for you.

**Stay within your healthy weight.** If your body is carrying excess weight, it can put extra strain on your heart, which can make you tired.

**Sleep well and rest often.** Try to get a good night's sleep and to rest during the day when possible. Establishing a routine of going to bed and getting up in the morning at the same time everyday can help.

**Reduce stress to boost energy.** Stress uses up a lot of energy! Try to take time out of your day to relax; listen to music, read or spend time with friends.

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## Growth

Most children with ASMD NP-B have delayed growth. This can be particularly noticeable in adolescents and can cause much anxiety and upset. In most cases, although growth is slow, children will continue growing well into early adulthood and usually do attain a normal height by their mid-twenties.

## What may happen

Onset of development, which includes tightly related growth and pubertal development, has a variable timing that depends on genetic and environmental factors. This usually happens in girls between the ages of 7 and 13 and boys between the ages of 9 and 15.

At puberty, girls become more rounded in the hips and their breasts begin to develop. Boys' penises and testicles grow larger they get more muscular. Those who are shorter may feel out of sync with their peers — just as girls who mature earlier may feel awkward if they get their periods before their friends, or boys may feel strange if they grow facial hair and have to shave first.

“I knew from before going into high school that my puberty process will be delayed because I was already one of the smallest in my class at primary school. I did not notice any difference between myself and my peers in year seven due to the fact that everyone was small. As years went on, especially in years nine and ten I started becoming self conscious because all the girls started to grow in all areas. What made me get through it is being happy that I did not have a face full of spots - I also knew that when all my friends had stopped growing, I would continue to grow. Also everyone thinks you are cute when you're small, so I have embraced my height and don't allow ASMD NP-B to take over my teenage years.”

### What you can do

It can be tough having a growth disorder as a teen because it can affect a person's body image and self-esteem. Talking with a trusted friend, adult or a mental health professional is one way to deal with feelings and concerns about growth.

Although no one has much control over the changes taking place in their bodies during puberty, you can do what you can to keep your body in top shape by eating a healthy diet, getting enough sleep, and exercising.

“I was put on testosterone injections to speed up my growth and a year ago I took up archery as a hobby, something I have found that I am quite good at.”

“For a long time the disease was practically unnoticeable. When I was a child my enlarged stomach was very visible but I seemed to grow into it and I wouldn't say that since my teens it has really been very noticeable at all, with the only reminder being that I couldn't do contact sports and had to have regular check ups.”

“The growth thing did bother me and has been a problem, (although I am now nearly 6ft), kids gave me a hard time at school as I was so small and looked so young, before I had a driving licence as proof of age I was always being refused in shops and pubs etc, this part was very frustrating and annoying, although living in a small town where most people know me has helped, having a brother who at 17 was 6ft 5 has not!”

### Who can help?

Your doctor can also be a good resource for advice on your growth pattern and offer suggestions that may be of help. You might be referred to a paediatric endocrinologist, a doctor who specialises in treating children and teens who have growth problems, or to another specialist for further tests or treatment. A dietician can advise on healthy eating and nutrition.

In some cases, doctors may offer a short course (usually a few months) of treatment with hormone medications to help with growth. Usually, hormones will take over from there to complete the growth process.

## Lipid disorders

Most of those affected by ASMD NP-B will also have some form of lipid disorder, such as, but not limited to, Hyperlipidemia or Hypercholesterolemia.

In research studies, it was found that 74% of ASMD NP-B patients were found to be prone to atherosclerosis, a condition in which an artery wall thickens as a result of the accumulation of fatty materials such as cholesterol, and 41% were found to have high cholesterol levels. Children, as well as adults, were found to be affected.

Cholesterol is a fat (also called a lipid) that your body needs to work properly. Cholesterol levels that are too high can increase your chance of getting heart disease, stroke, and other problems. The medical term for high blood cholesterol is lipid disorder, or hyperlipidemia.

## What you can do

Ensure you eat a healthy balanced diet and if necessary seek the advice of a dietician. Regular monitoring of cholesterol and other lipid levels is recommended. Medications, such as statins, may be appropriate – consult your doctor for further information.

## Who can help?

Your GP and metabolic consultant will be able to offer advice and information regarding regular testing and appropriate medications. Your dietician will offer advice regarding a healthy, low fat diet.

## Nutrition

In general, people with ASMD NP-B should follow a normal healthy diet. However, you may need to alter your diet to reflect changes in your condition, such as those associated with lipid disorders (see above). In this case, your consultant will refer you to a dietician for advice regarding diet and nutrition.

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## Respiratory (chest) issues

In ASMD NP-B, the enzyme, Acid Sphingomyelinase (ASM) is not working properly. This can cause storage of sphingomyelin in the lungs. The sphingomyelin storage makes it harder to get oxygen from the lungs into the blood system. When this first occurs, you may not notice any changes or difficulty in breathing, however this can change over time. Respiratory infections are more common in those affected by ASMD NP-B. Many patients also report breathlessness and fatigue.

## What may happen

Breathlessness as a medical symptom is an unpleasant awareness of difficult or uncomfortable breathing that is greater than you would normally expect during exercise. It is sometimes referred to as dyspnoea (pronounced disp-NEE-a), the Greek for difficulty breathing.

“Having ASMD NP-B, you become aware that you are limited as to how far you can walk and activities get cut short due to the fact that you get breathless in a short amount of time. There is no need to stop going on walks or doing activities, you just need to make sure you take breaks when needed to get your breathing back to normal and always take a cool drink with you. All of our bodies are different, so you have to get to know your limits and stick to them to stay healthy and safe.”

### What you can do

To assist with breathing and lung capacity, your Physiotherapist can suggest exercise techniques that may help to maximise lung capacity.

Regular chest X-rays and monitoring of lung function is recommended. Your consultant will usually arrange these for you, and they will usually take place as part of your clinic appointment.

Seek prompt treatment for respiratory infections; keep your GP informed about your condition and the need to monitor respiratory infections.

Medication can help, and in some cases, oxygen therapy may be useful.

For a peaceful sleep, keeping your room well ventilated can also help, as long as the temperature is comfortable. You may also want to consider use of a humidifier, or a vaporiser.

Nutrition is also important. Being overweight makes breathing more difficult. Keeping your weight to within the normal range for your height is important.

Exercise can also help. Light, non-exerting activities such as swimming or walking may be beneficial. Your physiotherapist can provide individualised advice in this area.

In addition, smoking is strongly discouraged; in smokers, the rate of decline in lung function can be three times the usual rate. As lung function declines, it causes breathlessness. If you need help to stop smoking speak to your GP.

“From an early age I was aware of my very large tummy, not least because my trousers wouldn’t stay up! I spent my early years wearing braces and dungarees to save an embarrassing problem like losing my trousers!”

### Who can help?

Your consultant, GP, community nurse or the NPDG (UK) clinical nurse specialist will be able to provide advice regarding medication.

A Physiotherapist can show you a number of exercises that help to maximise lung capacity. You may also be referred to a cardio/respiratory consultant, who will perform lung function test and x-rays. A dietician can offer advice about diet and nutrition.

## Splenomegaly

Splenomegaly is not a disease in itself but the result of an underlying disorder, of which there are many.

The spleen is an organ that is a part of the lymph system. It filters the blood and maintains healthy red and white blood cells and platelets, playing an important role in the immune system. Its main job is to break down blood cells at the end of their life. Due to storage in ASMD NP-B the spleen is enlarged and can be overactive, increasing energy demand and causing the destruction of healthy blood cells in addition to those at the end of their life.

In research studies, it has been found that an enlarged spleen may contribute to poor growth, which is often seen in ASMD NP-B. Other issues may include anaemia, the condition in which the body does not have enough healthy red blood cells, a low white blood cell count and the risk of haemorrhage from thrombocytopenia - a disorder in which there is an abnormally low amount of platelets.

The size of the spleen can also impact on digestion and breathing, causing considerable discomfort. A very enlarged spleen may also press on the lungs, increasing difficulties in breathing.

A low platelet count can lead to bruising and bleeding, whilst low red blood cells can lead to tiredness and anaemia.

Current medical advice is not to remove the spleen, but in certain cases a partial splenectomy might be an option, following consultation with your healthcare team.



## At school

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All children should have the opportunity to learn, play and develop in an environment that is suitable to their needs and abilities.

However, for parents of children with a long term health condition, ensuring your child has access to the right level of support can prove frustrating. Any challenges presented by ASMD Niemann-Pick disease type B can be overcome if parents, teachers and other professionals work together to ensure that the child's needs are met effectively.

It is essential that the needs of each child be assessed on an individual basis. You may feel that this can be achieved through building a good relationship with your child's education providers and by ensuring they have factual and up to date information regarding ASMD NP-B and your child's current health needs. The NPDG (UK) clinical nurse specialist is able to give teaching sessions at a child's school and is also available throughout the school year to answer questions as necessary.

Developing an individual care plan with your child's teachers can be a valuable exercise, not just in helping teaching staff to understand ASMD NP-B but in ensuring your child receives the level of care and support they need to access and achieve to their greatest ability.

Another way to achieve this is through a statement of special educational needs; children with long term conditions often require a statutory assessment to ensure schools are able to meet their needs. It is important to remember that ASMD NP-B is variable, and that all children are individual in their needs. If you feel your child requires additional support to attend school, contact the NPDG (UK) families officer for further advice.

“I suppose school made me realise that I was different, I was made to have an afternoon nap until my mum found out and that soon stopped, both my parents were very keen that I should have as normal life as possible even if sometimes that was hard. I was not allowed however to do contact sports due to my enlarged liver and spleen.”

### Some points to consider at school

- physiotherapy and physical exercise should be encouraged wherever possible to maintain muscle strength and mobility. Again this is according to the child's individual needs and should be correctly supervised
- inclusion as much as possible in the activities of the child's peers, with appropriate supervision and assessment of their particular needs
- rest may be required throughout the day: therefore the school day may need to be adapted to accommodate this
- regular assessment and review to ensure the need of the child needs are met quickly and effectively



## Providing essential information to schools

It is essential to ensure schools and other establishments that may be caring for your child have all of the information they need in order to provide the right environment and equipment to meet any special needs they may have.

Physical activities should be actively encouraged, however, if joining in with certain physical activities is difficult or impossible for your child – the school needs to know. If your child can access certain activities whilst wearing a spleen guard, staff members need to know how to fit it correctly and ensure it is worn at appropriate times.

Educating teaching staff and others that come into contact with your child about ASMD NP-B and its effects upon your child can be invaluable. Arrange a meeting to discuss current symptoms and to develop a strategy that can be initiated at school. This could include advice about the following items:

- nose bleeds and the need for quick action
- information about using a spleen guard
- physical activities – what is possible and what is not
- possible changes to the curriculum to accommodate needs
- transport to and from school and any associated risks
- an emergency contact number for you or your nominated person
- consider developing an emergency health care plan (see page 17)

The NPDG (UK) clinical nurse specialist can visit your child's school to provide information or training sessions if this would be helpful.

“Our son was diagnosed prior to starting Primary School, so before he started I met with the Headmaster and explained my fears about his safety. Being so rare, Niemann-Pick Type B doesn't fit into any of the 'tick boxes' or follow any standard procedures. By getting the Head on side by communicating on a personal level; working with the school nurse on a care plan; giving a little informal talk to all the teachers and by sharing my fears and emotions with his main teacher each year; my son's care at school has been absolutely excellent. From little things like making sure he is always at the front of a queue so his spleen and liver won't get knocked, to putting in a lower urinal because he was shorter than the other boys and couldn't reach! He will move on to High school in a couple of years which will be much more challenging but I'm not going to worry about that just yet. One thing his disease has taught me is to enjoy the moment and to take each day as it comes!”

## Helping your child to cope at school

### Coping with bullying

We all know that bullying can happen in any school but, it's the way in which it is dealt with which can make the difference between a child's life being tolerable or miserable. We also know that children are more likely to be bullied when they are vulnerable in some way. Research suggests that children with disabilities or long term conditions are three times more likely than their peers to be bullied.

This may be due to a lack of understanding of your child's condition or the fact they may be seen as "different". This can happen if they are doing different work, receive special allowances or additional support at school. For this reason it can be harder to make friends and easier for them to feel isolated.

It is not just children who find themselves upset by bullying – parents often feel a range of emotions too – such as anger or guilt.

Schools have a legal duty of care towards their pupils, and must act as any reasonable parent would to keep them safe. They are required to produce an anti-bullying policy which may be part of the overall behaviour policy. It must set out what the school will do to try and prevent bullying, how they will respond to bullying reports, and how they will resolve any situations that arise. You may find it useful to request a copy of the policy.

Don't always assume your child is going to be bullied but do be prepared in case they are. Ask yourself how you can prepare them for school and build their self-confidence and self-esteem.

If you think your child is being bullied, talk to them. This may be a difficult conversation as children can find it a hard subject to discuss or may not recognise that they are being bullied. Actions you may wish to take if they are being bullied, include:

- keep a record of times, dates of any incidents and any injury caused
- contact someone at your child's school and let them know your concerns. This could be the class teacher, your Special Educational Needs Coordinator (SENCO) or the head teacher
- arrange a meeting at the school to discuss your concerns
- agree an action plan and arrange further meetings to discuss progress made

“Being hurt or called a name can make you feel sad, embarrassed or unhappy. As the Families Officer for the Niemann-Pick Disease Group UK, I have supported children and young people who have been bullied at school. What is important to remember is that you are not alone. There are many things that can be done to help you. Talking to your family and teachers is a good idea. You can always contact me and I will help you with this or talk on your behalf if you would like. I can talk to you on the phone or in person if you prefer, but I will always listen and make sure you feel safe.”

### Advice for children

If you are being bullied, tell a friend, tell a teacher and tell your parents. It won't stop unless you do. It can be hard to do this so if you don't feel you can do it in person it might be easier to write a note to your parents explaining how you feel, or perhaps confide in someone outside the immediate family, like a grandparent, aunt, uncle or cousin and ask them to help you tell your parents what's going on.

## Some ways in which you can build your child's self-confidence are:

- praise and encourage your child as much as you can in daily activities
- place a photograph of your child with family members in their room to remind them that they are not alone
- spend time with them and take time to listen
- work on improving social and communication skills; you can do this through play and in everyday activities
- reassure your child that you love them and that you have confidence in them – underline the fact that being bullied is not their fault

## Resources

### Anti-Bullying Alliance

Resources and information to help schools address bullying of children with special educational needs and disabilities. For more information visit: [www.anti-bullyingalliance.org.uk](http://www.anti-bullyingalliance.org.uk)

### Anti-Bullying Network

Scottish organisation providing anti-bullying support to school communities on the internet, with a parents' and young people's section. For more information visit: [www.antibullying.net](http://www.antibullying.net)

### Childline

Helpline for children offering emotional support and counselling on any issue, including bullying. Helpline: 0800 1111 (24 hours). For more information visit: [www.childline.org.uk](http://www.childline.org.uk)

## Transition

Transition takes place between the ages of 14-25, and is the time when young people have to make important decisions about their future. Like all teenagers and young adults, those with a life-limiting condition will inevitably experience the ups and downs of adolescence; however these experiences may be more problematic than their peers, due to additional health problems.

As research has shown that transition can be a difficult time, and in order to ensure the young person's needs are fully met, it is wise to start planning well in advance. Young people may find the decisions they are making about further education, independent living or relationships exciting, but equally they may find the whole experience daunting.

The purpose of planning is to guarantee continuity of care, and to ensure that the young person has access to information and support to assist them in making informed decisions. It is imperative to keep the young person and their family at the centre of the planning and to consider their feelings and wishes throughout the process. Forward thinking will help to reduce feelings of stress and anxiety and will enable the creation of a clear plan to assist all involved in the transition process.

“Do make 100% sure your uni is fully aware and fully understand your condition – I went back to an event at my uni recently and discovered that some of my tutors were barely aware of my health problems while I’d been there, even though I’d informed the department and disability services! In situations like this, at school, uni or work, it might be worth trying to arrange a meeting with everyone you’ll have regular contact with to explain things in person, though it’s a bit of a pain!”

“Nowadays, for me, tiredness and fatigue is been the most noticeable effect of the disease. My endurance is quite limited and my concentration has suffered as a result, though careful planning helps to counteract this. I began noticing the tiredness much more after a serious stay in hospital when I was 17. It was quite jarring to suddenly have to be carted about by my parents in the car all the time at that age, which continued to be necessary for about a year. Shortly after I moved to university where obviously tiredness was much more noticeable having to look after myself - especially in my first year, I had some difficulty socialising and getting necessary tasks/work done. Going to the same university as my boyfriend helped with but also reduced my confidence and sense of independence quite a lot. There were times I felt quite inadequate compared to others and tiredness could often mean a lack of motivation. I found it hard to make friends quickly as I couldn’t join in with things as much, and having to pace myself or ask others who I didn’t know well to make allowances for me did make me feel like a bit of a downer at times.”

## Resources and further information

The Transition Information Network (TIN) is a website for parents, carers and people who work with and for disabled young people in transition to adulthood. TIN is an alliance of organisations and individuals who come together with a common aim: to improve the experience of disabled young people’s transition to adulthood. It is a source of information and good practice for disabled young people, families and professionals. For more information visit: [www.transitioninfonetwork.org.uk](http://www.transitioninfonetwork.org.uk)

Contact a Family has an excellent factsheet which can be downloaded from their website <http://www.cafamily.org.uk/transition.html>

# Financial support

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Asking for help can be difficult for many of us, but particularly when it comes to seeking financial support. Living with a loved one with a life-limiting illness can have a range of financial implications.

Day-to-day living for someone with a disability is often more expensive and adaptations to your home and aids to help daily life can be costly. There are various ways to receive financial support to ease the stress associated with these financial commitments.\*

\*The information in this section is subject to change. For the latest information and advice in this area, please contact Elizabeth Davenport, NPDG (UK) families officer:

Email: [elizabeth@niemann-pick.org.uk](mailto:elizabeth@niemann-pick.org.uk)

Telephone: 01904 438589 or mobile 07896 197576

## Contact a family benefits fact sheets

These fact sheets give an overview of all the main benefits available, including the tax credit and benefits system, and focus on those aspects which affect families with disabled children. For more information visit: [www.cafamily.org.uk/benefits.html](http://www.cafamily.org.uk/benefits.html)

## The Department for Work and Pensions (DWP)

The DWP is responsible for a range of benefits and services for people who are sick or disabled, and their carers. For more information visit: [www.dwp.gov.uk/directgov](http://www.dwp.gov.uk/directgov)

## Tax credits

Tax credits are income-related payments made to people who are responsible for children and to people in relatively low-paid work. The support available to people with children is called Child Tax Credit and the help for those working is called Working Tax Credit. Responsibility for administering tax credits rests with HM Revenue & Customs.

For further information and to assess your entitlements, visit the HM Revenue & Customs website: [www.hmrc.gov.uk/taxcredits](http://www.hmrc.gov.uk/taxcredits)

## Universal Credit

Universal Credit is a new single payment for people who are looking for work or on a low income.

Universal Credit will help claimants and their families to become more independent and will simplify the benefits system by bringing together a range of working-age benefits into a single streamlined payment. Starting in April 2013, Universal Credit will be introduced to claimants within certain areas of the North-West of England. For further information visit: [www.dwp.gov.uk/policy/welfare-reform/universal-credit/](http://www.dwp.gov.uk/policy/welfare-reform/universal-credit/)

## Disability Living Allowance (DLA)

Disability Living Allowance (DLA) is a benefit that is awarded on the basis of the effect that the symptoms of Niemann-Pick disease may have on your daily life. It can be awarded to a child or young adult if they have mobility problems, difficulties with personal care or if they require supervision from another person.

For an application form, contact the Department for Work and Pensions (DWP) Benefits Enquiry Line on 0800 882200. You can also download a copy at [www.dwp.gov.uk](http://www.dwp.gov.uk)

## Personal Independence Payment

From 8 April 2013 the Government is introducing a new benefit called Personal Independence Payment (PIP) to replace Disability Living Allowance (DLA) for eligible working age people aged 16 to 64. For further information visit: [www.dwp.gov.uk/pip](http://www.dwp.gov.uk/pip)

## Direct payments

### What are direct payments?

Local authorities can give payments, instead of services, to allow disabled people and carers to buy in the services they have been assessed as needing. When a child becomes 16 she or he can receive payments in their own right to allow them to buy in the services they have been assessed as needing.

Find out more by searching 'Direct payments' on the [direct.gov.uk](http://direct.gov.uk) website: [www.direct.gov.uk](http://www.direct.gov.uk)

### Can I get a blue badge?

The Blue Badge scheme provides on-street parking concessions for disabled people in the UK. The badge can also be used in many European countries. You may be eligible if you receive the higher rate component of disability living allowance. To find out whether you qualify you should apply to your local social services department.

## Family Fund

The Family Fund can give lump sums for specific items which help provide care for a child or young person with a life-limiting condition up to the age of 17. Family Fund grants are targeted at low-income families, so social and financial circumstances will be taken into account when an application is assessed. To find out if you qualify you should contact: The Family Fund, Unit 4, Alpha Court, Monks Cross Drive, York YO32 9WN, t: 0845 130 4542 e: [info@familyfund.org.uk](mailto:info@familyfund.org.uk) w: [www.familyfund.org.uk](http://www.familyfund.org.uk)

## Who else can help?

### Government information on rights for people with a disability

For guidance on how to find out about your rights as a disabled person, go to [www.disability.gov.uk](http://www.disability.gov.uk)

### Citizens Advice Bureau

Access up-to-date, independent advice at [www.adviceguide.org.uk](http://www.adviceguide.org.uk)

### Equality and Human Rights Commission

The aim of the Equality and Human Rights Commission is to end discrimination and harassment of people because of their disability, age, religion or belief, race, gender, or sexual orientation. Find out more about the commission at [www.equalityhumanrights.com](http://www.equalityhumanrights.com)

### Benefits Enquiry Line

Claim forms for all benefits can be obtained from your local benefits office or by calling the Benefit Enquiry Line on 0800 882200 (Monday to Friday 8.30am–6.30pm and Saturday 9am–1pm. Customers who have a textphone can call the Benefit Enquiry Line free on 0800 243355).

### The Niemann-Pick Disease Group (UK)

The NPDG (UK) can also provide assistance or advice. Please contact the Central Office on 0191 415 0693 or email [niemann-pick@zetnet.co.uk](mailto:niemann-pick@zetnet.co.uk)

## Where to find additional help and support

A full list of organisations and services that may be of assistance to you can be found on the NPDG (UK) website [www.niemann-pick.org.uk](http://www.niemann-pick.org.uk)

If you do not have access to the internet, please do let us know – we would be happy to send you paper copies of this information. If you are unable to find what you are looking for, please contact us, we will do all we can to assist you.



## Glossary of terms

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During the course of your child/loved one's illness it is likely that you will encounter many of the terms listed in this glossary. Initially they will probably be unfamiliar, but as time goes by you will become familiar with more of them. They describe the genetic components and medical terms relating to Niemann-Pick disease. You may eventually find that you know more about the disease than many of the non-specialist healthcare professionals you meet.

**Acyl-coenzyme A cholesterol acyltransferase.** The gene for this enzyme is on chromosome 1. It is an enzyme located in the membrane of the endoplasmic reticulum (ER) and acts to join cholesterol molecules to long-chain fatty acids for storage purposes.

**Acid sphingomyelinase (ASM).** This is a lysosomal enzyme that breaks down a substance called sphingomyelin. This enzyme is defective to a greater or lesser extent in NP-A/B disease. The enzyme deficiency arises from mutations in a gene on chromosome 11.

**Allele.** A particular form of gene. Alleles occur in pairs, one on each chromosome inherited from each parent.

**Amino acids.** Organic molecules that link together to form proteins.

**Autosomal.** Refers to chromosomes 1 to 22, ie any chromosome other than the sex chromosomes.

**Bases.** In the context of this pack, used to describe a number of chemicals that are used as instruction code by DNA and RNA. These are abbreviated as A, T, C, G in DNA and A, U, C, G in RNA.

**Blood-brain barrier.** A highly selective barrier formed by blood vessels and a type of brain cell called astrocytes that allow only appropriate molecules to cross from blood to brain, eg oxygen and sucrose. It aims to keep harmful molecules out of the brain.

**Calcium.** An essential element of all animals and plants. Vital in all cells.

**Catalyst.** A substance that accelerates a chemical reaction without being changed itself. Enzymes are a type of catalyst.

**Cells.** The basic unit for life, bound by a protective membrane. Cells contain all of the information: lysosome, mitochondria, nucleus, DNA, production mechanisms, material and communication links with other cells needed to replicate and grow into a living creature to allow them to replicate and function.

**Cerebellum.** Part of the brain located behind the brain stem, under the main mass of the brain. It is known to be responsible for the coordination of movement and contains Purkinje cells which are badly affected in NP-C.



**Cholesterol.** A fatty substance known as a sterol that is a major component of cell membranes, especially the main (plasma) membrane. It is also required for digestive bile acids and sterol hormones. Some cholesterol is taken in from certain foods but most is produced by the cells of the body, notably the liver. A constant supply of cholesterol is needed for cell growth and maintenance and is distributed in the blood stream by LDL.

**Cholesteryl esterase.** A lysosomal enzyme that cuts (hydrolyses) LDL cholesterol molecules free from their fatty acid tails. Wolman's disease results from defects in this enzyme.

**Chromosome.** This is a very long, super-coiled DNA molecule that carries the information template and enables replication of cells. It is located in the cell's nucleus.

**Common mutation.** A mutation in a gene that occurs in a high percentage of patients affected by a specific disease, eg I1061T in NP-C.

**Cytosol.** The internal liquid/gel content of a cell that surrounds the sub-cellular organelles and contains many of the simple and complex molecules required for the cell to function. The cytosol is not uniform but varies throughout its extent and is organised by the internal skeleton of the cell.

**Diploid.** These are cells containing two sets of chromosomes and hence two copies of genes. Most cells of an organism except sex cells and red blood cells are diploid.

**DNA (deoxyribonucleic acid).** A very long molecule and the main component of chromosomes. DNA contains a code which, when processed by the cell's production machinery, produces proteins. It also contains information relating to the timing and life cycle of the cell.

**Dominant allele.** Only one copy of the allele is needed to produce a certain characteristic. For instance, if you have one allele for brown eyes and one for blue, brown is dominant and you will have brown eyes.

**Endoplasmic reticulum.** A series of interconnected flattened cavities lined with a thin membrane which is continuous with the nuclear membrane.

**Endosome.** A membrane-bound compartment of the cell; the destination for internalised plasma membrane components and the internalisation of external molecules.

**Enzyme.** A protein that acts as a catalyst which accelerates a chemical reaction.

**Gametes.** Gametes are sex cells including sperm cells and ova. These are haploid cells.

**Genes.** These are regions of DNA that are copied to make proteins.

**Genome.** The total complement of genes on all the chromosomes that define a person or animal's life cycle.

**Genotype.** The genetic make-up of an individual.

**Glycolipids.** A group of lipids containing a carbohydrate.

**Glycosphingolipids.** These are sphingolipids with one or more carbohydrate attached.

**Haploid.** These are cells that contain one set of chromosomes. Haploids are the sex cells or gametes and red blood cells.

**Heterozygous.** A heterozygous condition exists if the corresponding genes on each of the related pair of chromosomes are different to each other. The different genes can be normal or disease-causing.

**Homozygous.** A homozygous condition exists if the corresponding genes on each of the related pair of chromosomes are the same. The genes may both be normal or disease-causing.

**LDL.** LDL is low-density lipoprotein. It is manufactured in the liver and distributed to the other organs and cells of the body via the bloodstream. The LDL particles contain esterified cholesterol within a membrane and also have a protein called Apo B100 which forms a strong bond with the LDL receptors on the surface of the cells. Too much LDL in the blood is considered to be a bad sign and an indicator of heart disease and stroke.

**LDL receptor.** A receptor on the surface of a cell that binds LDL particles prior to transferring them to the interior of the cell. The receptor appears in abundance during cellular cholesterol demand and then diminishes to a low level once the demand has been satisfied.

**Lipid.** An organic molecule that is insoluble in water. This property is important for the formation of membranes (very high lipid content) which allow separation of different biologically active solutions.

**Lysosomes.** These are membrane-bound organelles within the cell whose task is to recycle biochemical molecules for reuse. They contain a variety of enzymes for this purpose.

**Metabolism.** All processes taking place in living cells. The main processes are anabolic – building up of complex molecules from simpler ones – and catabolic – the breaking down of complex molecules into simpler ones.

**Mutations.** These are heritable changes in the DNA of cells. They are often changes that could be detrimental to the operation of the cell.

**Neurodegenerative.** Causing a loss of function of a system due to the loss of, or damage to, cells in the nervous system.

**Neuropathology.** The structure of the nervous system.

**Nucleus.** A membrane-bound organelle within a cell that contains the chromosomes.

**Organelle.** A sub unit of the cell usually bound by a membrane and distinct in its composition and functional behaviour. Examples include: the nucleus, endoplasmic reticulum, Golgi apparatus, mitochondria, lysosomes, and endosomes.

**Plasma membrane.** Also known as the cell membrane, this is the outer boundary of the cell.

**Proteins.** Large organic molecules that perform many of the metabolic activities taking place within a cell. Typical roles are as enzymes, transporters, receptors, sensors, activators and structural elements. The proteins are constructed from chains of amino acids linked together in a sequence defined by the DNA of genes.

**Recessive.** The effects of a gene that are masked by the activity of the same gene on the other chromosome (see dominant).

**Sex chromosomes.** Non-autosomal chromosomes – X and Y – inherited from parents. XX is female, XY is male.

**Sex-linked.** Traits that may be either normal or disease-causing and are derived from the genes or DNA of the X and Y chromosomes or mitochondrion (as opposed to autosomal).

**Sperm.** A male sex cell or gamete containing a haploid (single) set of chromosomes.

**Sphingolipids.** A type of molecule found in all plant and animal cells, and particularly abundant in the tissues of the nervous system.

**Sphingomyelin.** A complex organic molecule that is present in membranes in association with cholesterol. Found in neurons, it is not processed correctly in NP-A/B.

**Steroids.** A type of lipid, eg cholesterol, bile acids, some vitamins (including vitamin D) and steroid hormones.

**Substrate.** A substance on which an enzyme acts to form either a more complex substance or a simpler substance.



**Niemann-Pick**  
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