

Niemann-Pick type C disease

A practical guide for
parents and carers



03:

LIVING WITH NIEMANN-PICK TYPE C DISEASE

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Introduction

As the disease progresses, those affected by NP-C can develop complex needs that can change rapidly. Parents and carers 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. The information in this pack has been generated by a group of experts. By sharing their knowledge and experience of caring, they hope to reduce the feelings of isolation that are often experienced by those new to this disease, and to help you realise that you are not alone.



Bathing and skin care

NP-C will gradually affect your loved one's ability to perform tasks which they may previously have found easy. For example, as their physical abilities are affected, it may become difficult for them to get into or out of a bath.

As bath time can be a special time for children, and a relaxing one for adults, you may want to look at adaptations or equipment that can help.

For a small child, a bath chair may be of help. There are many different types available that will fit inside your existing bath tub. They will be height adjustable and may automatically lift your child out of the water. As your child gets older, you may want to consider a hoist system or a bathing trolley. Your occupational therapist will advise you of the products available and the NPDG (UK) has a record of the items that have been successful for other families.

For adults, you may want to consider creating a wet room, or using a shower chair. You may also need to install a hoist system to ensure safety of the affected person and the carer.

Generally, try to set aside plenty of time, keep the bathroom warm and remember to test the temperature of the bath or shower in case it is too hot or too cold. Have everything you need to hand, including towels and nightwear. Playing in water can provide endless fun for little ones, and plenty of sensory toys are available for bath time. Water also has therapeutic qualities – a warm bath can relax muscles and aid sleep.

Following a bath, massage can provide further relaxation. Using a good moisturiser can help prevent skin problems and dryness and this is also a good opportunity to check the skin for signs of redness or breakdown (pressure sores) which can occur if the person is immobile. Areas that can be prone to this are the shoulders, elbows, bottom, hips and base of spine. If you find anything which concerns you, contact your local healthcare team as soon as possible.

If the person is immobile, they must be moved regularly to avoid pressure sores. Your physiotherapist can provide advice on positioning, and offer advice regarding aids that can provide comfort.

Those affected by NP-C do not tend to have skin problems other than the more common issues such as eczema or acne as they get older. However, care should be taken if the person is incontinent, as the skin around the lower body is at greater risk of soreness. Good hygiene and frequent changing of nappies or pads can avoid problems in this area.



Dental care

Dental hygiene is very important, even if your child or loved one is not being fed by mouth. Teeth and gum problems can develop in those who do not regularly use the muscles in their mouth and throat. It is advisable to brush teeth at least twice a day if this is possible, using a small amount of mild-flavoured toothpaste, or to swab the mouth area with a diluted solution of mouthwash (non-alcohol).

If your child or loved one is not being fed by mouth, remember to moisten the mouth at regular intervals. Your community nurse will be able to advise you how to do this. Stimulating the mouth in this way will provide comfort and assist in maintaining the health of the mouth and swallowing reflex.

As the disease progressed in our daughter, it was a testament to her character that she never lost her sense of humour or her stubbornness if she did not want to do something. Cleaning her teeth could be a real trial if she decided she was not going to open her mouth!

A common symptom of NP-C can be increased secretions, such as saliva. Medication is available to ease this symptom. Speak to the NPDG (UK) clinical nurse specialist, or your local community nurse for advice.

Certain anticonvulsant medications can cause enlargement of the gums. These medications will be prescribed if the person suffers from seizures as a symptom of NP-C. Gentle brushing can help in this instance. Again, if you are concerned seek advice from a member of your healthcare team.

Continence

As the disease progresses, it is likely that the person will no longer realise when they need to go to the toilet. At first this can be very embarrassing and frustrating, especially for older children and adults. While the person is mobile, regular trips to the toilet can avoid accidents. Continence and dignity can be maintained for a long time just by using this approach.

Regular bowel habits are important for health and wellbeing. Normal bowel and bladder habits vary from person to person, and changes in health, diet or exercise can make a difference. Restricted mobility can cause constipation, which can be very uncomfortable and distressing. Problems in swallowing can lead to changes in diet, including a reduction in fluid or fibre intake, which again can be a cause of constipation.

Speak to your physiotherapist for advice on exercises that may help; your dietitian or nutritionist can advise regarding diet. Medication can also help. Speak to your consultant, GP or community nurse about the available options.

In the later stages, advice can be sought regarding specialist aids and products to maintain dignity and keep the person as comfortable as possible.



Moving and handling

Those affected by NP-C may develop varying problems with mobility as the disease progresses. Everyday activities such as climbing stairs or getting in and out of the bath, may become more difficult. Taking advice on aids and equipment can help provide individualised solutions to help you or your loved one get the most out of life.

Arrange a meeting with your occupational therapist and physiotherapist to discuss options that may assist you, such as the means of transferring from the bath to a chair or adaptations to your home. It is advisable to do this well in advance of these things being needed, as it can take time to make arrangements.

How can my physiotherapist help?

- they can help to plan an individual exercise programme suitable to the person's needs, with exercises that assist with posture and coordination
- they can help to reduce muscle stiffness and discomfort
- they can give advice about walking aids, splints or braces for support
- they can provide training in safe and comfortable ways to lift or move a person with reduced mobility

How can my occupational therapist help?

- they can give advice on alternative ways of doing day-to-day things
- they can give advice about available equipment to help with everyday tasks and could arrange a trial
- they can advise as to the best way to adapt your home for ease, comfort and safety
- they can provide training in safe and comfortable ways to lift or move a person with reduced mobility

If balance and mobility becomes a problem then safety in and around the home should be of prime importance, to minimise the risk of injury and falls.

Simple precautions can help around the home. Ensure the affected person wears suitable and appropriate footwear; remove any loose rugs or trip hazards such as frayed or raised carpet edges. A walking frame can assist if handrails are unavailable. If stairs are a problem, your occupational therapist can advise and may be able to organise the provision of stair rails and grab rails, or a lift.



Once a person is unable to move around themselves, use of a sling or a hoist can be invaluable, not only for the health and safety of the person, but also the carer. When getting up from sitting or lying is no longer possible without the help of another person, your physiotherapist or your community nurse can offer advice on safe ways of moving and handling. Using incorrect techniques can hurt both the person being moved and their carer so it is important to avoid injury to either party.

Communication

Communication is key to sharing our thoughts and feelings, making sure that our voice is heard and our needs are met. Communication is not just speech; we express ourselves in a variety of ways, through gestures, the written word, facial expressions and emotions such as laughter and tears. A person with NP-C may gradually lose the ability to communicate as effectively as before. Problems in communicating can impact greatly on the psychological wellbeing of the affected person and those caring for them. When the time comes, your speech and language therapist will be able to provide advice and equipment for alternative ways of communicating.

At first speech may become slurred. Those closest to the person should still be able to understand them. However, when speaking to others, you may find that they do not understand, or in some cases, may assume that the person is drunk, which can be hurtful and embarrassing.

Using a pocket-sized notebook can help in some cases, or carrying a laminated card which explains your situation can be useful. An example of this card is available from the NPDG (UK), or you can design your own with a short statement explaining 'What you need to know about my condition'. This can be based on the following:

Although I have Niemann-Pick type C disease the symptoms vary from person to person and are different from one time to another. To help me cope (or while I am in your care) you need to know the following information.

Other important information to include would be:

- emergency contact details
- your name and if you use a shortened version of this
- your likes and dislikes
- details of any medication you may need and at what times
- details of any symptoms you may be experiencing, such as seizures
- how you eat, and what your preferences are
- how mobile you are
- how you prefer to communicate

The card can be adapted for different occasions, such as holidays or hospital stays.

Parents and carers can help in the following ways:

- the person affected by NP-C may not have perfect speech, but this does not mean that they cannot understand what you are saying – always be considerate, polite and do not talk about someone as if they are not there
- you may not need to change the sound of your own voice, or slow your speech – this will depend on the preferences of the person affected
- if the affected person is speaking slowly, be patient and try not to finish their sentences
- if you are having difficulty understanding, sit or stand face to face with the person and watch their lips and facial expressions
- try simple communication cards, with pictures, such as those used in Makaton sign language. You can show these to the affected person, and judge their response by their facial expression. If they are able, perhaps they could give the universally known ‘thumbs up’ or ‘thumbs down’ sign
- use of ‘closed’ questions that only need a ‘yes/no’ answer can be helpful. For example ‘Would you like a cup of tea?’ rather than ‘Would you like tea or coffee?’

- think about listing the questions that you use every day, turning them into easy to answer questions with a 'yes/no' response, either given verbally or by an alternative signal
- families can often work out their own sign language, or can interpret meaning through a certain look, or way of nodding the head

Your speech and language therapist will advise you on the range of equipment and communication aids that are available, and will point out those most suitable to your needs. Some of the items that are available include:

- portable electronic communication aids
- computers with voice or switch activation
- communication passports, providing information on a person's personal needs
- control switches which can be fitted to most battery operated or electronic devices

Other equipment may also be needed such as arm supports, clamps to hold equipment steady or an adjustable table.




Feeding and nutrition

Eating a healthy diet and staying well nourished is important for all of us; eating and drinking are also social activities, shared and enjoyed by many. As a consequence of the disease, those affected by NP-C are likely to develop problems in swallowing at some stage. However, this varies from person to person, with some continuing to eat and drink normally for many years.

For parents, feeding your child is one of the most basic things that you can do for them, and also with them. Meal times can be a fun family occasion, so when problems do occur, it can be an emotive time for all concerned. In the early stages of the disease, it is important to ensure that your child receives a healthy balanced diet. Advice and daily eating plans can be obtained from your GP, dietitian or nutritionist to assist you.

For adults, enjoying a meal or drink with friends can be a pleasure. But once swallowing problems begin, it can be tempting not to join friends or family in this activity, due to embarrassment or negative feelings about food. Seeking advice from your healthcare team can help you to find the most appropriate solutions for you.

In both children and adults, swallowing and feeding will need to be monitored carefully as the disease progresses. Reduced mobility and lack of physical activity due to the symptoms of the disease can result in muscle wastage and weight loss. By maintaining a healthy diet you can help to preserve any remaining muscle strength. Problems with chewing and swallowing can also mean that less food is eaten, which again can lead to weight loss. Due to the nature of NP-C, improvements in diet will not eliminate the symptoms of the disease, but it will help the person to experience greater wellbeing and quality of life for as long as possible.



When eating and drinking become difficult, there are some practical steps that may help in the short term:

- using a straw to take sips of fluids can prevent too much being taken into the mouth at once
- thickening fluids can make them easier to swallow. Thickening powders are readily available and easy to use – your dietitian or nutritionist will be able to provide advice on the options available
- finely chopping, blending or pureeing food can help
- ensuring that mealtimes are relaxed occasions by taking time to eat and trying to sit upright for short periods following the meal
- taking smaller mouthfuls of food
- having several smaller meals, rather than fewer larger ones
- consulting your speech and language therapist, dietitian or nutritionist for advice

There may come a time when oral feeding is not safe, and the risk of aspiration (when food goes down the wrong way) and chest infections outweigh the wish to keep eating orally. At this point it can help to discuss the available options with your GP or the NPDG (UK) clinical nurse specialist.

Gastrostomy tube feeding is one such option. A minor operation is performed to insert a small feeding tube through the abdominal wall into the stomach and this can be discreetly hidden under your clothes. This procedure may often be suggested for an adult or child before they stop eating orally as it can also assist with the safe administration of medicines or help top up the diet to supplement oral intake.

Discussing the options with professionals can help you decide the right course of action for your child or loved one.

Behaviour

Behaviour can be an issue in children and adults with NP-C. Sometimes this can be caused by feelings of frustration. This can result from a loss of skills or an inability to do what they want to do, or because they find themselves unable to be understood.

Early-onset dementia can cause those affected to become quite obsessive in their behaviour. They often need very ordered surroundings and an unchanging routine. In most cases it is not difficult to work around this, but at times of inevitable change, such as changing schools or going on holiday, you need to anticipate possible problems and try to plan ahead.

If you are caring for a person with dementia, you may sometimes find their behaviour confusing, irritating or difficult to deal with. This can leave you feeling stressed, irritable or helpless. By learning to understand the meaning behind their actions, you will find it easier to stay calm and deal effectively with the challenges that arise. Try to remember that the person is not being deliberately difficult. Ask yourself whether their behaviour really is a problem.

What may happen

- repetitive questioning, actions or movements
- trailing the carer around the house
- hiding and losing things
- shouting and screaming
- restlessness
- night time waking



How to cope

- try to put yourself in your child/loved one's situation. Imagine how they might be feeling and what they might be trying to express
- think ahead and plan for situations that could cause problem behaviour
- explain the situation to friends and neighbours so that they will understand changes in behaviour
- offer as much reassurance as you can by touching and holding
- distract them with calming activities such as playing their favourite music
- try to make sure that you have support for yourself and breaks when you need them

A person with memory loss and general confusion may behave in a way that other people find embarrassing. Try to react calmly.

- if they undress or appear naked in public, they may have forgotten when and where it is appropriate to remove their clothes. Take them somewhere private, and check whether they are too hot or uncomfortable or whether they want to use the toilet
- lifting a skirt or fiddling with flies may be a sign that the person wants to use the toilet
- if they start to stroke their genitals in public, discourage them tactfully and try to distract their attention. If such behaviour is frequent or persistent, consult your GP
- if they behave rudely – for example, by shouting, swearing or spitting – don't attempt to argue or correct them. Just try to distract their attention. You can explain to other people later that their behaviour is due to dementia and is not directed at them personally

If you find the person's behaviour really difficult to deal with, ask for advice from professionals or other carers before you become too stressed.

Play and enjoyment

It is important that your child plays and enjoys their life to the full, whatever their physical limitations. The onset of neurological symptoms in a child may delay the development of play skills, therefore your child may need many more opportunities to imitate and learn how to play. If communication difficulties are also present, appropriate aids can assist them in inclusive play.

You may be invited to take your child to a child development centre, where appropriate toys can be recommended to maintain physical and mental wellbeing. If your child has marked problems because of NP-C, these toys may not be age-appropriate but will be right for their individual needs.

There are many ways to ensure that your child is included in activities that the whole family can enjoy. If, for example, your family likes walking, it may be appropriate to get an all-terrain buggy, so that they can go along with you on days out. Occupational therapists and physiotherapists can provide advice in this area.



A young child can benefit from aids that offer visual stimulation and are calm and soothing. They may not be able to handle and play with ordinary toys, but bubble tubes, fibre optic lights and projected pictures and lights can be fascinating. The NPDG (UK) clinical nurse specialist can assist families to access these aids through grant agencies and other sources.

Many toys specifically designed for 'special needs' are very expensive. When you purchase very similar items from high street shops they are often a fraction of the price. Toys for children with special needs should not focus on a child's disability at the expense of the child. With some modifications, these children can often play with the same toys as other children around the same age, depending on developmental ability. Toys for children with special needs should be safe, durable, versatile, challenging and engaging.

Choose items that will stimulate their senses, such as bells, whistles, and colourful lights that are highly exciting for little ones. Toys that stimulate the imagination are also highly beneficial. Look for items that are easy to handle and grip, have different textures and scents or are unusual shapes. Washable toys are always a good idea.




Mobility aids and adaptations

At some point all children and adults with NPD will probably require some form of aid or adaptation to assist their daily living. These aids may be as simple as special cutlery to make eating easier, or a handrail on the stairs, but, at some point, they may entail major work to adapt the living areas.

It can help to have a full mobility assessment as soon as problems occur, or as early as you feel ready to consider the options. Your occupational therapist will come to your home and discuss the needs of your child or loved one. They will also take into consideration the needs of other family members living in the house. It is important that this assessment not only takes into account the current needs of the affected person, but possible future requirements. It can be useful to have the NPDG (UK) clinical nurse specialist in attendance to help during an assessment, as it is often difficult to contemplate your child/loved one needing hoists or similar aids in the future. It can also be difficult for an affected adult, who feels they can still be independent, to accept that they may be safer with a shower instead of a bath, or indeed are better with a ground floor bedroom.

The occupational therapist and NPDG (UK) clinical nurse specialist have worked with many children and adults and will be able to suggest aids to make life as simple as possible. For instance, a child may need a raised desk at school to angle their books, which makes looking from book to the teacher and the board much easier. An adult may find they may work better on a computer. A person with NP-C may be safer with handrails installed throughout the house, or a ramp to the front door instead of steps. These are just examples – every individual's needs are different.



As the disease progresses you may need more help to adapt your house. As mobility decreases the affected person may need to use a wheelchair. Adaptations may then be needed to access the house, such as widening the doors and fitting a ramp. There may also be issues with access to the bedroom or bathroom if these are upstairs. The occupational therapist can assess your family home and suggest appropriate changes. Suggestions are always subject to your agreement and approval. In some cases a stair lift may be appropriate, in others a full lift. Alternatively the bedroom and bathroom may need to be moved to the ground floor. The aim is to meet increasing needs and to make life as easy as possible for the whole family.

Big changes to the home take time as plans need to be drawn up and grants or other finance sought. It is therefore advisable to make arrangements ahead of time, implementing the required changes before they become necessary. Accepting these changes can be difficult. If you are worried or have concerns, speak to the NPDG (UK) clinical nurse specialist or your own nursing team about the possible future needs of your family. They will be happy to answer any questions that you have and will ensure that you are able to access everything you may need.

Therapies

There are many different therapies that may help to improve quality of life for those affected by NP-C. You may be able to provide many of these quite easily at home, while others might require the help of a physiotherapist.

A physiotherapist is a healthcare specialist who can help to relieve pain, improve or maintain movement and muscle strength, and help prevent injuries. They will evaluate and diagnose before managing a treatment plan designed to meet the needs of the affected person. They will form part of your healthcare team and work alongside other professionals to assess and treat movement and physical needs.

Some complementary therapies can help make day-to-day life more comfortable by helping to control, or ease, certain symptoms of the disease. Complementary medicine offers a different approach to more traditional methods, involving the body, mind and spirit. This holistic approach can lead to feelings of being 'more in control' of the situation.



Generally, relaxation can help to reduce anxiety and stress. These can be brought on by many of the problems associated with everyday life for those affected by NP-C and their carers.

Massage, aromatherapy and reflexology offer effective means of muscle relaxation. These therapies improve blood and lymphatic circulation. They also relieve pain naturally by encouraging the production of endorphins, which are naturally occurring chemicals generated by the body to fight pain. If you do consider using essential oils for massage, you should speak to your GP or consultant for advice beforehand.

Hydrotherapy

Hydrotherapy is a highly effective form of water-based therapy which works by stimulating endorphins, helping to control pain and alleviating tension. Treatment involves the use of warm water to achieve pain relief and to alleviate symptoms caused by many illnesses. It can effectively ease pain caused by aching joints, back pain, fatigue, insomnia, muscular pain, stiffness or cramp, and can therefore work very well for those affected by Niemann-Pick type C disease.

Hydrotherapy can be a vital tool in helping to improve circulation, relieve stress and assist in physiotherapy treatment. The warm water provides a relaxing environment that enables muscles to be more receptive and responsive to exercise. Hydrotherapy and other water-based activities can be beneficial health and social activities for the whole family, leaving people of all ages and abilities with a feeling of wellbeing and relaxation.





Music therapy

Music therapy is an established healthcare profession that can promote wellbeing, manage stress and alleviate pain. It helps to enhance memory and the expression of feelings, improve communication and promote physical rehabilitation. Music therapy can also support the social, emotional, behavioural and cognitive needs of people of all ages and abilities.

A music therapist uses all aspects of music to help to improve or maintain health and quality of life. Using tailored music experiences such as singing, song writing, playing musical instruments and listening to and moving to music, they can help an individual with cognitive functioning, motor skills, emotional development, behaviour and social skills. Music therapy can be a deeply enjoyable and positive experience that can enhance wellbeing.

Rebound therapy

Rebound therapy describes the use of trampolines to provide therapeutic exercise and recreation for people with a wide range of special needs. It is used to facilitate movement and improve fitness and exercise tolerance. It can also promote balance, an increase or decrease in muscle tone and helps to improve sensory integration. Time spent on the trampoline can also improve communication skills and is an opportunity for relaxation. Rebound therapy is fun, gives confidence in movement and can promote a feeling of wellbeing.

For further information on any of the therapies listed here please contact the NPDG (UK) central office.

At school

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 special educational needs and disabilities, accessing the right level of support and information can prove frustrating. Often the level of support available varies greatly depending on where you live. A child with Niemann-Pick disease can face many challenges in the classroom. These 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 are assessed on an individual basis. One way to achieve this is through a statement of special educational needs. Children with long-term conditions or complex needs often require a statutory assessment before they start school. This process is usually begun by a specialist involved in caring for the child but in some cases it may happen after the child starts to attend school. This can be a very difficult and distressing stage for the family and the Niemann-Pick Disease Group (UK) is available to offer help and support as required. It is important that those involved with a child's education have an understanding of the disease and its progression. 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.



When assessing a child's educational needs the following people may be involved

- teacher
- educational psychologist
- school nurse
- physiotherapist
- social worker
- community nurse
- parents
- GP
- epilepsy nurse specialist
- Niemann-Pick clinical nurse specialist
- specialist consultant

From the onset of NP-C, your child's teachers will play an important role in their care. They will need to share information, concerns and decisions with you, as parents, and also with other health and social care professionals. As NP-C is an extremely rare condition, all parties need to share information about the progression of the disease and their day-to-day support of the young person.

It is important to remember that the progression of NP-C in children is specific to the individual, and that the needs of each child will vary greatly. Initially, the child may experience some learning difficulties and perhaps problems with concentration and short-term memory recall. Speech may begin to appear slurred and the child may struggle to find the appropriate words to communicate. Motor coordination may appear slightly impaired and the child may experience difficulty with upward and downward eye movements (vertical supranuclear gaze palsy) which may make copying from the blackboard difficult.

There are some general points to bear in mind when teaching a child with NP-C

- children with NP-C often have a short attention span so it may be necessary to have frequent breaks from a particular task
- establishing a routine is very important for most children with the disease, as loss of short-term memory can lead to confusion and conflict
- as short-term memory can be limited, repetition of tasks may be necessary
- a sense of achievement is important for all children – it is unlikely that progress will be made within the normal targets of their peer group so these targets need to be adjusted to make them achievable
- 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 supervised
- speech can become affected as the disease progresses. Speech and language therapy should be made available as early as possible
- inclusion, as much as possible, in the activities of the child's peers is very beneficial but needs to be supervised and suitable for the child's particular needs
- rest may be required throughout the day. Your child's school day or timetable may need to be adapted to accommodate this
- regular assessment and review will be required to ensure that the child's needs are met quickly and effectively



Special educational needs

Children with special educational needs all have learning difficulties or disabilities that make it harder for them to learn than most children of the same age. These children may need extra or different help from that given to other children of the same age. They may also need extra help because of a range of needs. These can include reasoning and understanding, physical or sensory difficulties, emotional and behavioural difficulties. They may also have difficulties with speech and language as well as problems with how they relate to and behave with other people.

If you feel that your child needs extra support at school, you can ask for advice from a number of different people

- your GP, social worker or community nurse
- the NPDG (UK) clinical nurse specialist
- your consultant
- your child's teacher, or the head teacher
- the special educational needs coordinator at your child's school (SENCO)

If your child is not of school age or not yet going to an early education setting, such as a nursery, you can talk to your doctor or health visitor.

If you think your child may have a special educational need that has not been identified by the school your child is attending, you should talk to your child's class teacher, to the SENCO or to the head teacher straight away.

If your child is in a secondary school, you should talk to the child's form teacher, SENCO, head of year or head teacher. Your local education authority (LEA) will also be able to help, or you can contact the NPDG (UK) clinical nurse specialist.

Once it has been decided that your child has special educational needs, your child's teachers should take account of the guidance in the SEN code of practice. The school must tell you when they first start giving extra or different help for your child because your child has special educational needs. This could be a different way of teaching certain things or some help from an extra adult, perhaps in a small group. It may also mean the use of particular equipment like a computer or a desk with a sloping top.

Your child's teacher is responsible for working with your child on a day-to-day basis. They may decide to write down the actions or help for your child in an **individual education plan (IEP)**.



The IEP should list:

- the type of special help being provided
- how often your child will receive help and who will provide it
- the targets which have been set for your child
- how and when your child's progress will be checked
- the help you can give your child at home

If possible your child's teacher should discuss the IEP with you and your child. Sometimes the school will not write an IEP but will record how they are meeting your child's needs in a different way, perhaps as part of overall lesson plans. However, the school should always be able to tell you how they are helping your child and the progress they are making.

The teacher or the SENCO may also ask for advice from specialists working outside the school. These can include a specialist teacher, an educational psychologist, a speech and language therapist or other health professionals. You should be included in any discussions, and your views taken into account, before decisions are made about how best to help your child.

If it is then decided that your child needs additional help, the local education authority (LEA) may decide to make an assessment of your child's educational needs, based on specialist advice. They must then write a **statement of special educational needs**, which is usually shortened to '**a statement**'. This will outline all of your child's needs and explain the help and support that they will receive.

A **statutory assessment** is a detailed investigation of your child's special educational needs and the additional support your child requires. A statutory assessment is only necessary if your child's current school or early education setting cannot provide all the help that your child needs.

During the assessment, the LEA will listen to your views, the school's view, and will also take advice from the specialists mentioned above. You can ask the LEA to contact any person or organisation whose views you feel would be beneficial to the assessment. The LEA will usually tell you their decision within six weeks.

In this modern world we live in people judge success or failure by exam results or by what a person can achieve, however with our daughter this was never going to be a viable option. She fought every day of her life in the early years just to try to achieve what her peers could do with very little effort and this just intensified as her condition deteriorated. If, instead of judging success by schooling, you measure it by courage, determination, strength of character, sense of humour and sometimes downright stubbornness then our daughter had a Masters Degree.

What if I do not agree with the school or LEA's decision?

If you think that your child's needs are not being met, or you do not agree with the outcome of a statutory assessment, you can discuss this with the relevant professionals. You should first talk to the SENCO, the LEA or to the head teacher. If this course of action is unsatisfactory then you have the right to appeal to the SEN Tribunal. Information about how to do this should be available from your local care team. You may find it helpful to talk to other parents who have been in a similar situation or you may wish to speak to the local parent partnership service.

The NPDG (UK) can also provide details of organisations which can give specialist help and advice on the subject of SEN, such as:

Network 81

1–7 Woodfield Terrace
Stansted
Essex
CM24 8AJ
t: 0845 077 4055
e: info@network81.org
w: www.network81.org

Independent Parental Special Education Advice (IPSEA)

t: General advice line 0800 018 4016 (Monday to Friday 10am–4pm, Monday to Thursday evenings 7pm–9pm)
t: Tribunal helpline 0845 602 9579 (Monday, Wednesday and Thursday 10am–1pm)
w: www.ipsea.org.uk

At the time of going to press, the Department for Education is reviewing practices in special educational needs and disability. If implemented, the proposals would change the current system of assessment to a single assessment process and introduce an 'Education, Health and Care Plan'. There will be a period of consultation and testing of proposals before legislative changes are introduced. For the latest news regarding progress of these proposals, please contact the NPDG (UK) central office.

Transition

Transition takes place between the ages of 14–25. It is a period when young people have to make important decisions about their future. Like all teenagers and young adults, those with disabilities will inevitably experience the ups and downs of adolescence. These experiences may be more problematic than those encountered by their non-disabled peers. This is due to the likelihood that they will have additional health problems and may often be involved with a number of additional support agencies.

Research has shown that transition can be a difficult time, so 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. Planning should guarantee continuity of care, and ensure that the young person has access to information and support to assist them in making informed decisions. It is important that the young person and their family's feelings and wishes are central to this process. Forward thinking will help to reduce feelings of stress and anxiety. It will also enable the creation of a clear plan to assist all involved in the transition process.



Resources and further information

The Transition Information Network 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 and can be accessed at www.transitioninforonetwork.org.uk.

Mencap has produced a booklet called Transition: a guide for parents and carers. This can be downloaded free of charge from www.mencap.org.uk.

Contact a Family has an excellent fact sheet which can be downloaded from the publications section of its website at www.cafamily.org.uk.


Hospice and respite care

Children's hospice services

Children's hospice services help the whole family to cope with the emotional and practical challenges of a life-limiting condition such as NP-C. Their aim is to help your child make the most of life, and for all of you to enjoy your time together.

The hospice staff can provide specialist care from diagnosis onwards. At first the staff may visit you at home, and then you will be invited to visit the hospice to look around, before your first residential visit is arranged. A children's hospice is like a home from home. It has comfortable family rooms and open-plan living areas, where you and your child can go for short or long stays, even before they are showing symptoms of the disease.





Staff in the hospice will take care of your child 24 hours a day, with as much or as little help from you as you require. They are dedicated to the care of children with life-limiting illnesses and will therefore be able to help with, or to discuss, any issues that you may have. Hospices work closely with each other and with other professionals involved in supporting the family. This helps provide a continuity of care where all needs are met, whether physical, emotional, social or spiritual.

The children's hospice service offers short breaks, during which the whole family can stay for a weekend or perhaps a week. They are also very happy to care for your child on their own, especially to provide respite that will allow the rest of the family to have some dedicated time together. Even a couple of hours of respite can give parents the opportunity to spend quality time with other children, or leave the house to get some shopping done.

Children's hospice services also provide:

- symptom control
- therapies including play and social activities
- activities for siblings
- 24-hour advisory support to families and professionals
- end-of-life care either at home or in the hospice
- pre- and post-bereavement support for all family members

One family's experience of a children's hospice

I never really knew what a children's hospice was, in fact I didn't know what a hospice was. Now I don't know where I would be without them.

My family and I made our first visit to our local hospice, Martin House in Wetherby, shortly after my son H was diagnosed with Niemann-Pick type A. We attended with the Niemann-Pick clinical nurse specialist, as she had arranged the visit. I remember approaching the hospice feeling quite unsure about what it was going to be like, a hospital or a home? Well, from the minute we entered we were greeted with warm welcomes and smiles.

My other two children felt at home immediately as staff made friendly conversation and introduced them to the play area. On this first day visit, my husband and I were given a chance to look around the buildings and facilities as well as ask questions. Everything was explained well and clearly and I certainly felt my needs were going to be met. The care team were able to offer as much or as little help as I required from them; it was all going to be around what suited me and that is exactly what they do.

So far we have had two stays for all of the family at Martin House for respite, as well as one emergency stay for myself and H. The care team help and support me however they can, whether it's taking care of the children or making me a drink.

Another source of reassurance at the hospice is the medical support provided. The care team includes nursing staff and the doctors are always available no matter what time of day or night. After my first stay at Martin House I immediately felt that these doctors understood my child's illness better than those I had previously dealt with in the hospital or at the surgery and they would be able to offer me the support I needed.

Finally, I would like to add that – particularly for those from a minority ethnic origin like myself – any personal needs and beliefs will be welcomed and respected. Without any doubt I would say to all parents in a similar situation – go and use the hospice. They will help and support you through this difficult time.

For further information contact the NPDG (UK) central office, or Children's Hospices UK direct at:

Children's Hospices UK

4th Floor

Bridge House

48–52 Baldwin Street

Bristol BS1 1QB

t: 0117 989 7820

f: 0117 929 1999

e: (general enquiries) info@childhospice.org.uk

w: www.childhospice.org.uk

Hospice care for adults

Hospice care aims to improve the lives of those diagnosed with life-limiting illnesses such as NP-C, by helping them to enjoy and get the most out of their lives. Families and friends are also supported, both throughout the illness and beyond.

You may be referred for hospice care as soon as a diagnosis is made. The referral could be made by your GP, hospital consultant or the NPDG (UK) clinical nurse specialist. Hospice care is provided completely free of charge.

Hospices provide a comfortable and safe environment. They are not religious places but they do aim to meet the needs of people from all cultures and religions. Care does not just focus on physical needs; emotional and spiritual needs are also considered, and the highest value is placed on respect and choice. The type of care provided by a hospice is called palliative care, often referred to as end-of-life care. Palliative care supports the person with an illness and their family from diagnosis to death and into bereavement by adopting a holistic approach to treatment and care. It can also be provided in your own home, in a hospital or a care home.


The services offered will differ from hospice to hospice but are likely to include:

- medical and nursing care
- pain and symptom control
- rehabilitation
- therapies, including physiotherapy and complementary therapies
- spiritual support
- practical and financial advice
- bereavement care

If you, or your loved one, wish to be cared for at home, this can be made possible by community palliative care nurses and the Hospice at Home service. This specialist care will include advice on pain and symptom control, hands-on nursing, practical advice and emotional support. This service is not just available at the end of life, but can also help at times of crisis.

Palliative care teams also work alongside staff in a hospital, to support them in providing specialist advice on pain and symptom control. They will also support the individual and their family, and can assist in making arrangements to transfer care to a home or hospice setting.





For further information you can contact the NPDG (UK) central office, or Help the Hospices direct at:

Help the Hospices
Hospice House
34–44 Britannia Street
London
WC1X 9JG
t: 020 7520 8200
f: 020 7278 1021
e: info@helpthehospices.org.uk
w: www.helpthehospices.org.uk

The photographs used to illustrate this booklet show people with NPD and their families and friends. They have all been used with permission, but do not generally relate to the text they have been placed alongside.

This booklet forms part of a resource pack published by the Niemann-Pick Disease Group (UK). It is intended to be read in conjunction with the other parts of the pack. If you do not have the other documents in the pack or would like further information, please contact us at the address below.

Note: the information contained in the pack 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.



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