

Living with a diagnosis Tay-Sachs & Sandhoff

A practical guide for
parents and carers

Produced by
The CATS Foundation



The CATS Foundation
94 Milborough Crescent
Lee
London
SE12 0RW

(e) info@cats-foundation.org
(w) www.cats-foundation.org

Registered charity in
England & Wales - 1144543

***Living with a diagnosis**
***Tay-Sachs & Sandhoff**

01

Physical care

How to perform some important physical tasks.

02

Communication and play

How to communicate and play with a loved one.

03

Feeding

The feeding process and other feeding methods.

04

External help

The external service industry can help with providing care.





A challenge

A new diagnosis of Tay-Sachs or Sandhoff disease will have a dramatic effect on you and your family and you will encounter many challenges.

**Being a carer
for someone
affected by
Tay-Sachs
or Sandhoff
disease can be
both a rewarding
and demanding
experience.**

***Introduction**

***Living with a diagnosis**

Beatriz

—
“Yes, it changed my life. It would do for anyone and I actually think it has changed a lot for me as my priorities have completely changed.”

As both Tay-Sachs and Sandhoff progress, those individuals affected by the diseases develop complex needs that can change rapidly. Parents and carers have one thing in common, and that is the need for practical solutions that will enable them to provide a high level of care for their child.

—

This booklet contains useful information on how to care for someone affected by Tay-Sachs and Sandhoff disease. It is important to remember that every child is different with their own personal needs.

***Daniel Lewi - Charity Director**

01

***Physical care**

***How to provide care**

Deborah

—
“We don’t get lie-ins, we take it in turns to sleep in Isabella’s room with her and our weeks are always dotted with appointments for this and that. But none of it is a burden or a hardship; it has just become a different way of life.”

There are certain tasks you can perform which will have a huge impact on a loved ones quality of life. However, it can be difficult to understand how these tasks can be performed as the disease progresses.

—

It is important that you feel comfortable in providing care so we have put together some useful tips and advice on how to perform these tasks. We hope this will make you more confident as their needs develop over time.

***Be confident in providing care**

*Bathing and skin care

*Warm baths and massage

The importance of skin care

Looking after the skin is important because over time your loved one will become prone to bed sores as they lose the ability to move on their own.

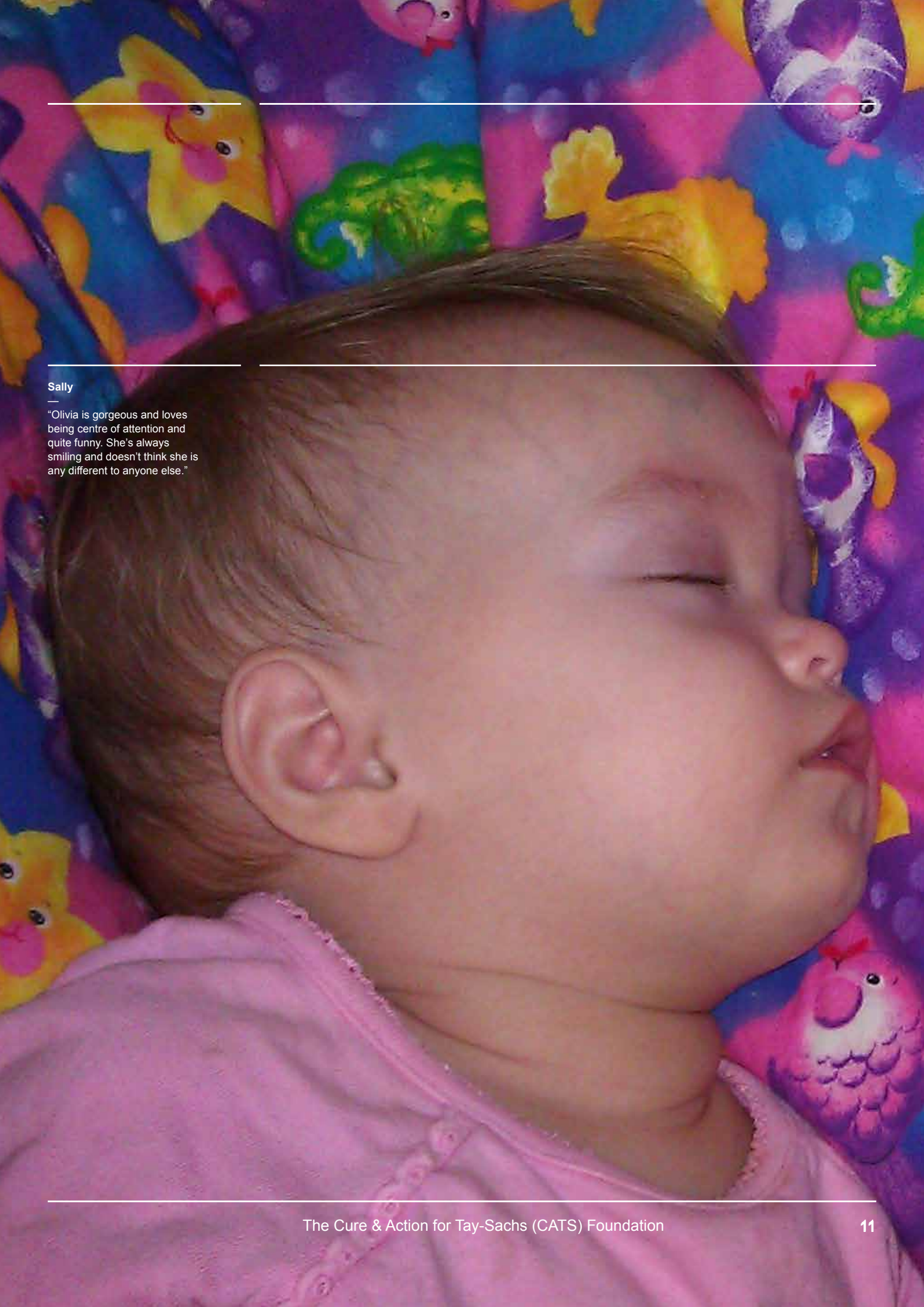
Tay-Sachs and Sandhoff disease will gradually affect your loved one's ability to perform tasks which they may have previously done before. For example, as the diseases progress their physical ability is affected and it will become difficult for them to sit in the bath or on their own. Bath time can not only be a special time, but also a relaxing one and you may have to look into adaptations or equipment that can help with bathing them.

Releasing stress

Massage is a good way to relieve stress and your loved one can benefit from regular sessions as this can help them.

Small children may benefit from having a bath chair and there are many different types available that will fit inside your existing bathtub. These chairs will be height adjustable and some may automatically lift your child out of the water. As your child gets older and heavier, you may want to consider a hoist system or bathing trolley and your occupational therapist will advise you of the products available. It is very important that you remember to consider the safety of both the person and the carer during bath time.

You should try to set aside plenty of time when giving a bath and remember to keep the bathroom warm and test the temperature of the bath or shower in case it is too hot or too cold. Playing in water can provide endless fun for children, and there are plenty of sensory toys you can use at bath time. Water also has therapeutic qualities as a warm bath can relax muscles and more importantly aid sleep. Following a bath, massage can provide further relaxation. Using a good moisturizer 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 in someone who is immobile. Areas that can be prone to this are the shoulders, elbows, bottom, hips and base of the spine and those who cannot move independently should be turned regularly.



Sally

“Olivia is gorgeous and loves being centre of attention and quite funny. She’s always smiling and doesn’t think she is any different to anyone else.”

***Physical comfort**

***Managing health**

“It is only when you become a carer do you realise how important the small things are. Looking after Amélie’s dental hygiene is something we take very seriously to make sure she doesn’t feel uncomfortable.”

***Daniel**

Dental hygiene is very important, even if your 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 and it is advisable to brush teeth at least twice a day. Try using a small amount of mild-flavored toothpaste, or to swab the mouth area with a diluted solution of mouthwash.

If your loved one is not being fed by mouth, remember to moisten the mouth a regular intervals. Your community nurse will be able to advise you on how to do this and stimulating the mouth in this way will provide comfort and assist in maintaining the health of the mouth and swallowing reflex. A common symptom of Tay-Sachs and Sandhoff disease is an increase in secretions and there is medication available to ease this symptom.

As the diseases progress, it is likely that your loved one will no longer realise when they need to go to the toilet. In older children and adults this can be very embarrassing and frustrating. 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 while problems in swallowing can lead to changes in diet, including reduction in fluid or fibre intake, which can also cause constipation.

Speak to your physiotherapist for advice on exercises that may help and they can give advice regarding diet. Medication may also help so speak to your consultant, GP or community nurse about the available options.

As Tay-Sachs and Sandhoff disease progress there will be varying problems with mobility. Taking advice on aids and equipment can help provide individualised solutions to help your loved one get the most out of life.

You should arrange a meeting with your occupational therapist and physiotherapist to discuss options that may be of assistance, such as the means of transferring from the bath to a chair. It is advisable to do this well in advance of anything being needed, as it can take time to make arrangements.

The physiotherapist can help by planning an individual exercise program with suitable exercises that assist with posture and coordination. They can also give advice on walking aids, splints or braces for support. The occupational therapist can help by giving advice on alternative ways of doing day-to-day things and also advising on the best way to adapt your home for ease, comfort and safety.

If balance and mobility becomes a problem then safety in and around the home should be of prime importance so that the risk of injury and falls are minimized. Simple precautions can help around the home such as ensuring the affected person wears suitable and appropriate footwear and also removing any trip hazards. If stairs are a problem, your occupational therapist can advise and may be able to organise the provision of stair rails or a lift.

Once a person is unable to move around themselves your physiotherapist or community nurse can offer advice on safe ways of moving or handling. Using incorrect techniques can hurt both the person being moved and their carer so it is important to avoid injury to either.

***Moving** ***Safety**

“Moving Isabella is tougher these days as she is quite big and heavy. We have to make sure that we don’t hold her in ways which could hurt her.”

***James**

02

***Communication and play**

***Having fun**

Deborah

—
“The symptoms started to come thick and fast and we had to shift our focus on to that. Gradually we started to concentrate on quality of life not quantity. Trying to alleviate the symptoms became the everyday reality.”

Communication is key to sharing thoughts and feelings, making sure that our voice is heard and our needs 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.

—

It is important that your child plays and enjoys their life to the full, whatever their physical limitations are. As Tay-Sachs and Sandhoff disease progress you will have to adapt the ways in which you play with your child.

***Be imaginative with play time**

*Behavior and play

*Be patient

Learn new techniques

As the diseases progress you may need to learn new ways to communicate. Be open minded as to what will work for you and your family.

It can take time

It may take longer for you to communicate with your loved one. Do not rush as it can be frustrating for both of you.

A person with Tay-Sachs or Sandhoff disease will gradually lose the ability to communicate as effectively as before. This can impact greatly on the psychological wellbeing of the affected person and also those caring for them. When the time comes, a speech and language therapist will be able to provide advice and equipment for alternative ways of communicating. At first you may notice that speech becomes slurred in those individuals affected by a later onset variant of Tay-Sachs and Sandhoff disease. Although those closest to the person should still be able to understand them, others may not which they could find embarrassing.

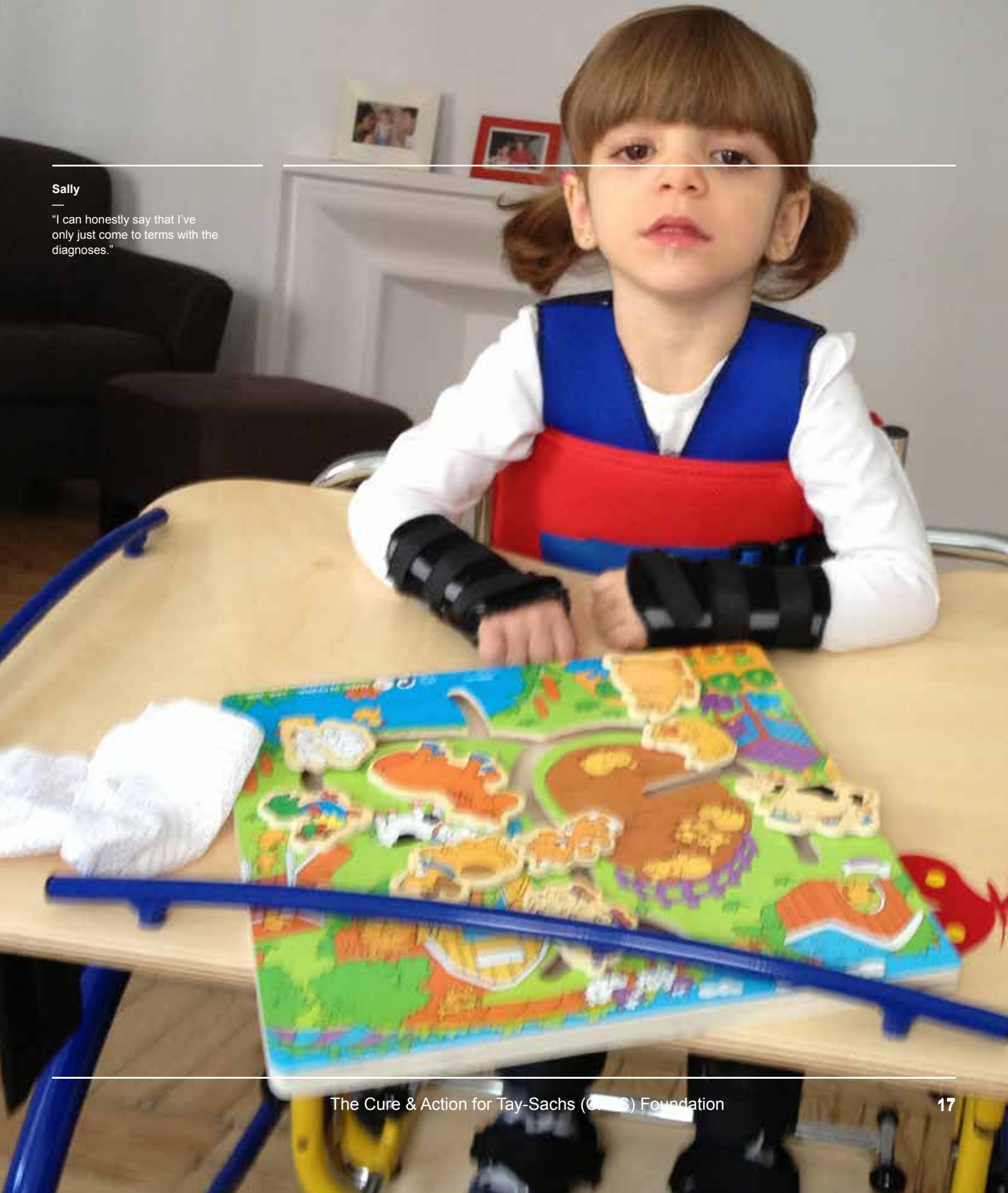
There are many ways in which the parents and carers of someone affected by Tay-Sachs and Sandhoff disease can help with communication. These include always being considerate, polite and do not talk about someone as if they are not there. Speak slowly, be patient and try not to finish sentences and also try using closed questions that only need a yes or no answer.

Behavior can be an issue in children and adults with Tay-Sachs and Sandhoff disease. Sometimes this can be caused by feelings of frustration as a result from losing skills or an inability to do what they want to do. The onset of neurological symptoms in a child affected by Tay-Sachs or Sandhoff may delay the development of play skills. Your child may need many more opportunities to imitate and learn how to play and if communication difficulties are also present, appropriate aids can assist them in inclusive play.

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 there are plenty of appropriate sensory toys they can use.

Sally

"I can honestly say that I've only just come to terms with the diagnoses."



03

***Feeding**

***Changing needs**

Eva

—
"Making a decision on feeding is one of those things that takes a lot of thinking about. Everyone has different views and we did what we thought was best for Haylie."

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 Tay-Sachs and Sandhoff are likely to develop problems in swallowing at some stage.

—

There are many options available to you to help your loved one with their feeding. It is important that you consider all the facts before choosing any feeding intervention.

***Feeding should not be a struggle**

*Nutrition

*Changing needs

Healthy food

While your child can still eat orally your doctors will be able to suggest meals to cook which are healthy. Along with the dietitian, they can give you a list of foods which are easy to swallow and have a high nutritional value.

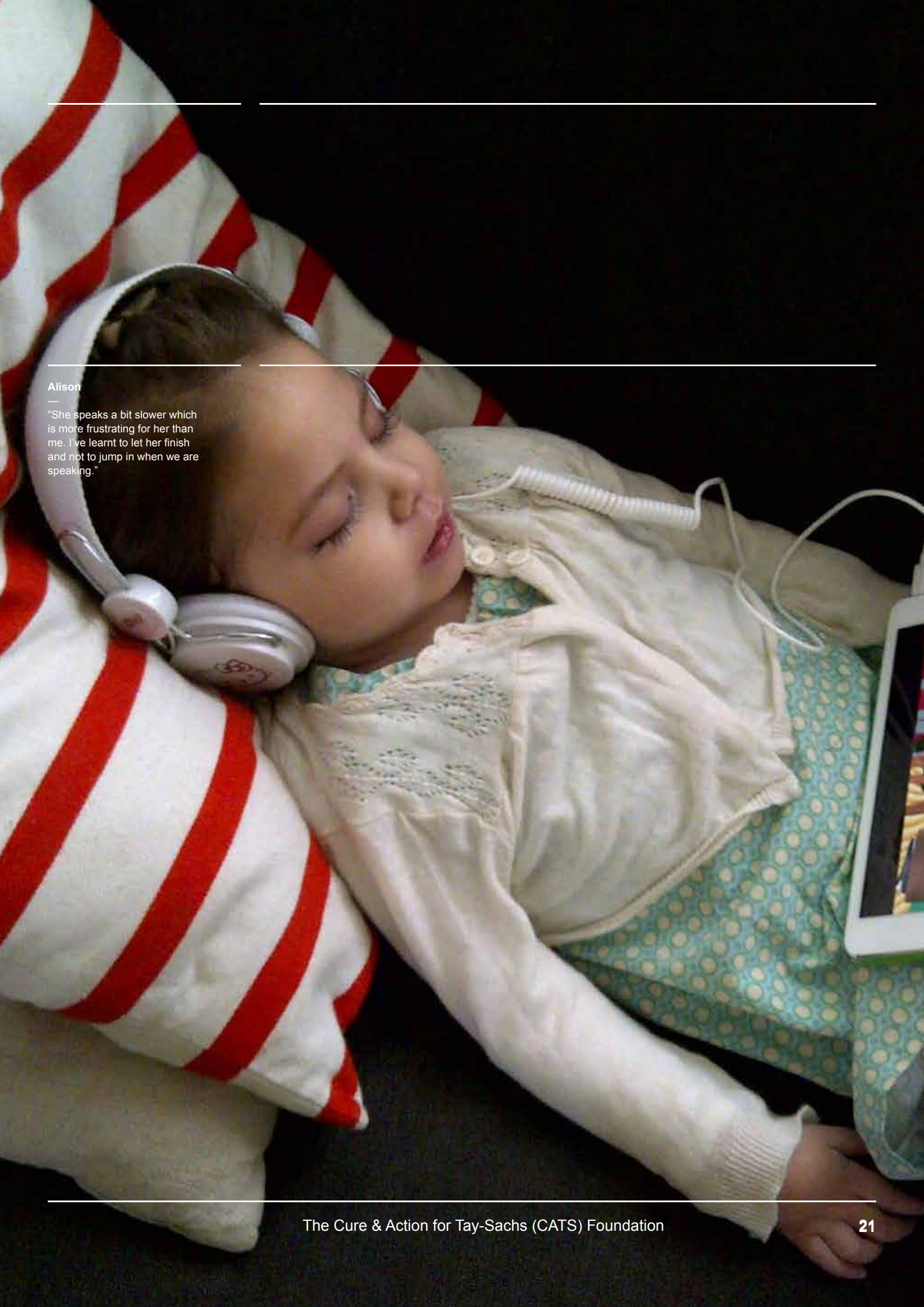
Enjoy meal time

It can become frustrating when your child starts to have problems with their swallowing. Make sure you enjoy your meal times together.

Eating a healthy diet and staying well nourished is important for all of us and they can also be seen as a social activity which are shared and enjoyed by many. As a consequence of the diseases, those affected by Tay-Sachs and Sandhoff are likely to develop problems in swallowing at some stage. Although this varies from person to person with some continuing to eat and drink normally for longer than others you will at some point need to consider some form of intervention. Meal times can be a fun family occasion, so when problems do occur with your child, it can be an emotional time for everyone.

In children and adults, swallowing and feeding will need to be monitored carefully as the diseases progress. Reduced mobility and lack of physical activity due to the symptoms of Tay-Sachs and Sandhoff can result in muscle wastage and weight loss. Problems with chewing and swallowing can also mean that less food is consumed, which can also lead to weight loss. Due to the nature of the diseases, improvements in diet will not eliminate any symptoms, but it will help them experience a higher quality of life.

When eating and drinking starts to become difficult, there are some practical steps that may help. Using a straw to take sips of fluids or thickening them can make it easier to swallow and also making mealtimes relaxed occasions. There may come a time when oral feeding is no longer safe, and the risk of aspiration (when food goes into the airway) and chest infections outweigh the wish to keep eating orally. Gastrostomy tube feeding may become an option where a minor operation is performed to insert a small feeding tube through the abdominal wall into the stomach. This procedure may often be suggested before someone stops feeding orally as it can also assist with the safe administration of medicines.



Alison

—
“She speaks a bit slower which is more frustrating for her than me. I’ve learnt to let her finish and not to jump in when we are speaking.”

04

***External help**

***Other services**

Daniel

—
“You can get help from so many people, you just have to know who you can contact. Each provides a different and valuable service.”

At some point children and adults affected by Tay-Sachs and Sandhoff disease will probably require some form of aid or adaptation to assist their daily living. Your healthcare team is best placed to advise you on what changes you can make to your home.

—

There are also many services which you can access to help provide care to your loved one. They all offer very different levels of support but you will find the ones which suit your needs.

***Do not be afraid to ask for help**

*Aids and adaptations

*Making life easier

You can get help

—
There are various aids in the market which can help you and your loved one undertake your day-to-day routine.

Tailor made

—
The majority of aids and adaptations your loved one gets will be tailor made for them so that they fit perfectly.

An occupational therapist can come to your home and discuss the needs of your child or loved one and it can be a good idea to have a full mobility assessment as soon as problems occur, or as early as you feel ready to consider the options. They will also take into consideration the needs of other family members living in the house and it is important that this assessment not only takes into account the current needs of the affected person, but possible future requirements.

As the diseases progress you may need more help to adapt your house. As mobility decreases the affected person may need to use a wheelchair and adaptations to your house may revolve around access to the house, such as widening the doors and fitting a ramp. The occupational therapist can assess your family home and suggest appropriate changes which are always subject to your agreement and approval. In some cases a stair lift may be appropriate while in others a full lift may be better. The aim is to meet any 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.

Deborah

“Some of the equipment we have got for Isabella has made such a huge impact on us and we could not cope without it.”



***School**

***Making friends**

“Jack went to a special school from the age of two. Never more than one day a week and always one-to-one. He loved it and it gave us access to swimming, physio and many things that we would never have thought to do.”

***Katherine**

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 Tay-Sachs or Sandhoff 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.

From the onset of Tay-Sachs or Sandhoff disease in older children, the 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 both diseases are very rare, everyone needs to share information about the progression of the disease and their day-to-day support of the child.

It is important to remember that the progression of Tay-Sachs and Sandhoff disease 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 their vision.

There are many different therapies which may help to improve quality of life for those affected by Tay-Sachs and Sandhoff disease. You may be able to provide many of these quite easily at home, while others require the help of a physio-therapist.

Some complementary therapies can help make the day-to-day life more comfortable by helping to control, or ease certain symptoms of the diseases. 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 the diseases.

Massage, aromatherapy and reflexology offer effective means of muscle relaxation. 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. Some other therapies you may also consider using are hydrotherapy or music therapy which can both promote wellbeing to any individual affected by Tay-Sachs or Sandhoff disease.

Hydrotherapy is a water based therapy where the treatment involves the use of warm water to achieve pain relief and alleviate symptoms caused by many illnesses such as aching joints, muscular pain, insomnia, or cramp.

Music therapy can help manage stress and alleviate pain and is also able to enhance memory and the expression of feelings, improve communication and promote well-being.

***Therapies**

***Other ideas**

“Our lives are filled with routines that can be quite complex and revolve around medications, treatments and feeds, but we do our best to ensure we have quality family time and there is plenty of time for fun in between.”

***Deborah**

*Palliative care

*Hospice or home

Meeting new children

—
Hospices are a great way for your loved one to interact with other children with complex needs.”

Children’s hospice services help the whole family to cope with the emotional and practical challenges of a life-limiting condition such as Tay-Sachs and Sandhoff disease. Their aim is to help your child make the most of life and for all of you to enjoy your time together where they have comfortable family rooms.

Staff in the hospice will take care of your child for twenty-four 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,

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.

If you wish to care for your loved one at home this can be made possible by community palliative care teams and your hospice’s 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 arrangement to transfer care to a home or hospice setting.

Meeting new parents

—
The hospice setting is also a great way for you to meet other parents in a similar position.



Sally

“Hope loves going to school now and she does so much there with the kindest people.”

***Hospice care helps**
***Everything in one place**

What next

—
You can contact The CATS Foundation at any time to find out what the charity does and to have access to its support services.

It can be very difficult for your local children's hospice the first time, but we are a warm and friendly place to help you in a way that we can provide care.

—

***An amazing service**

**daunting to go to
children's hospice for
but they are warm
faces who are there
any way possible to**

Gregor

"All of our Tay-Sachs family friends have really made a big impact on how we can look after our precious boy."





Beatriz

—
"Although it is a club I wish we were not members of, we are so grateful there are other parents out there who we can speak with."

***Acknowledgements**

***Final words**

Daniel - charity director

“The guides produced by The CATS Foundation will enable families to have access to all the information they need when a diagnosis is made.”

This booklet forms part of a resource pack published by The CATS Foundation. It is intended to be read in conjunction with the other parts of the pack.

If you do not have the other documents or would like further information please contact us.

Caroline Harding
CEO of Genetic Disorders UK

“We are delighted to be helping CATS Foundation who support children with Tay-Sachs and Sandhoff disease. Our grant programme is open to all UK support groups and registered charities who work to improve the lives of children and families affected by genetic disorders. In 2014, 25 charities will benefit from the funds raised by the public on Jeans for Genes Day.”

The CATS Foundation would like to acknowledge the contribution made to this guide by parents, carers and medical professionals.

The CATS Foundation would also like to thank Genetic Disorders UK who provided a grant via the Jeans for Genes grant programme to enable the production and printing of the guides.

Funded by a grant from
Genetic Disorders UK



JEANS FOR GENES DAY
GENETIC DISORDERS UK

Supporting families affected by genetic disorders



The CATS Foundation
94 Milborough Crescent
Lee
London
SE12 0RW

(e) info@cats-foundation.org
(w) www.cats-foundation.org

Registered charity in England
& Wales - 1144543

