02

Coping with a diagnosis
Tay-Sachs & Sandhoff

A practical guide for parents and carers

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Produced by
The CATS Foundation

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*Coping with a diagnosis
*Tay-Sachs & Sandhoff

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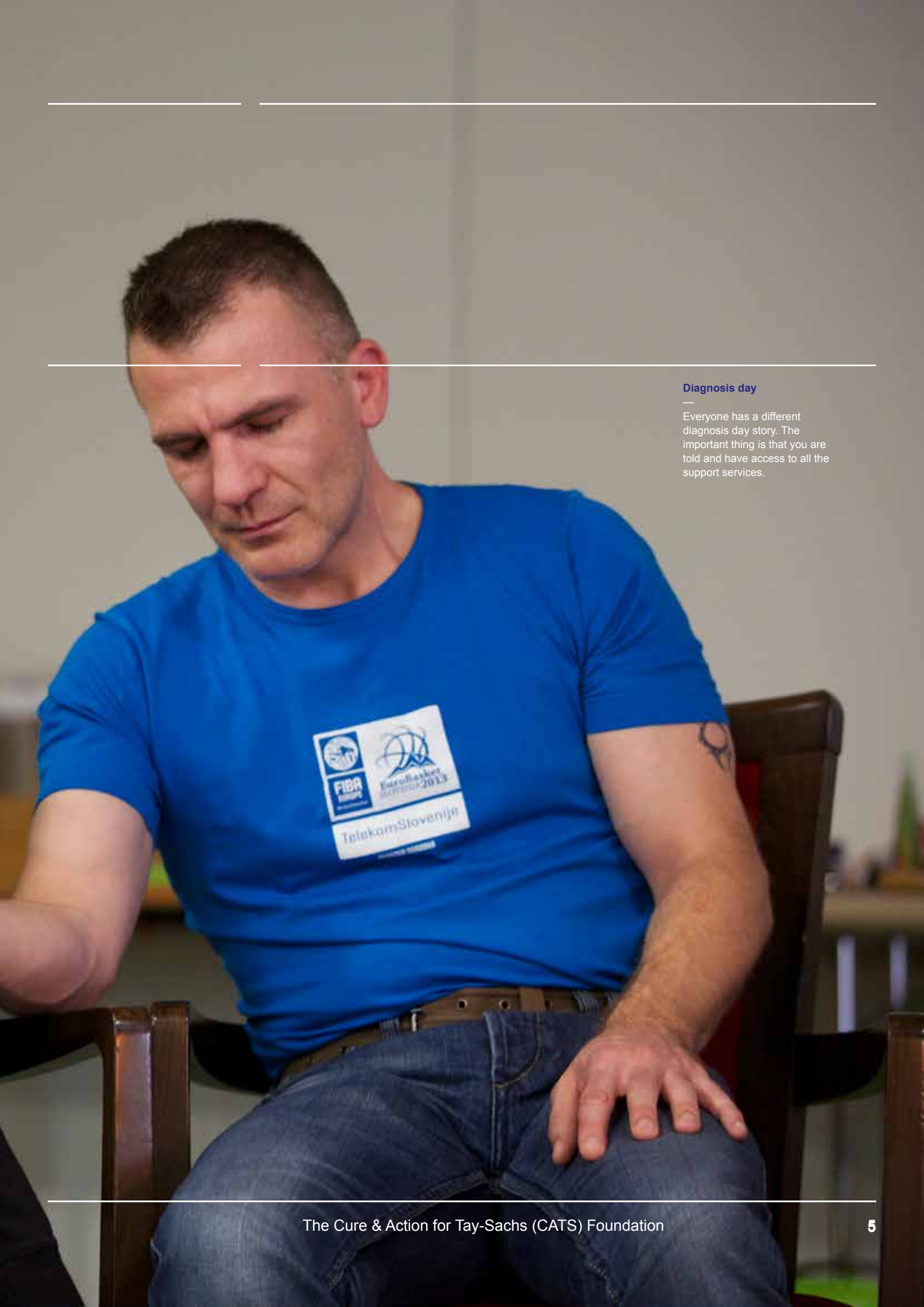

Diagnosis day

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Everyone has a different diagnosis day story. The important thing is that you are told and have access to all the support services.

Hearing the words that your child has been diagnosed with Tay-Sachs or Sandhoff will completely change your family's life.

*Introduction
*Receiving a diagnosis

Deborah
—
"'This is our normal' - we tell people, we don't know any different! Our lives are filled with routines that revolve around medications, treatments and feeds, but in between there is plenty of time for fun."

When your child or loved one is diagnosed with Tay-Sachs or Sandhoff it can have a profound effect on your whole family. For many, a diagnosis does not happen instantly and can come at the end of weeks or months of tests and investigations.

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This booklet contains useful information on how to cope with a diagnosis and other families diagnosis experiences. At The Cure & Action for Tay-Sachs (CATS) Foundation we hope that the information included in the guide is useful.

***Daniel Lewi - Charity Director**

01

*Where to start
*Getting support

Nikki
—
"It took several months for Ruby to be diagnosed with Sandhoff Disease. I knew it was bad news. At first, I was devastated, but as I continued to look after Ruby I realised she needed me."

In many cases the diagnosis day is the point from which you begin to contemplate the future and the impact the disease will have not just on your loved one but your entire family.

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During this period of adjustment you may face what feels like a never-ending round of appointments as you begin to meet with the many professionals who will form a network of care around your loved one. You may also find yourself having to tell your story time and time again.

***There are many support services**

*Who can help
*A useful list

Learn what they do

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If you research each of the services who can help your family you can make sure you get the most out of them.

Speak with your GP

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Your GP is the person who can put you in contact with many of the services. Do not be afraid to speak with them about what help you need.

After receiving the initial diagnosis of Tay-Sachs or Sandhoff disease there is an awful lot to think about. Diagnosis comes at a point when you may not be capable of thinking clearly or taking information in. After a diagnosis is made it can be a very daunting experience with the range of medical professionals your child will see. We have provided a brief overview of each service which will be available to you below.

The main goal at The CATS Foundation is to ensure that all the families affected by the diseases are supported once a diagnosis has been made. Feeling isolated at such a time can have a dramatic effect on an individual and their family, however you may find the following people involved with your child's care;

- Family doctor (GP)
- Community nurse.
- Social workers
- Portage
- Dietitian
- Speech and language therapist
- Physiotherapist and cccupational therapist
- Lead professional

The lead professional is the first port of call for non-medical problems. If they cannot help they will find someone who can. Many hours can be wasted as families phone around trying to find the right person to speak to and this can be stressful on top of your other commitments and concerns, so is best avoided.

Sharon

—

“At fifteen months we spent a week at a community assessment centre where he was seen by every discipline. Everyone agreed it was serious, but despite large number of blood tests taken, no one had an idea what it was.”

02

*You and your family
*Coping together

Leann
—
"The initial diagnosis for me, was the hardest 'stage' of the disease to deal with. We came away from a meeting with the consultant with no real information about the disease and searches on the internet at that time weren't very helpful either."

Coping with a diagnosis of Tay-Sachs or Sandhoff disease is one of the most difficult things a family must come to terms with. It is important that as a family you are able to support one another so you can make sure your child has as high a quality of life as possible.

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Friends and family will be able to help you with everyday tasks. Do not be afraid to ask for help so you are able to look after yourself as well as the person you are caring for.

*Take your time

*Caring for the carer
*Looking after yourself

Contact your GP

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.

Get enough rest

As a carer you must try to get as rest as possible so you can look after your loved one.

The definition of a carer is someone who provides help and support to an individual who may not be able to manage because of illness or disability. Carers can be anyone, although parents generally tend to be the main carers for their own ill or disabled child.

Being the carer for a child affected by Tay-Sachs or Sandhoff can be very challenging and it is not uncommon for them to become so preoccupied with the heath and wellbeing of the person they are caring for that their own needs are forgotten. It is important that you look after yourself too, although it is common for people to think that this is easier said than done. However, it does not have to take a lot of time and often just taking a few minutes of quiet time to yourself can help. Looking after your own health and emotional needs can help you in your role as a carer and make you more able to carry out your role. Some useful tips are below.

- Get enough sleep

  A common problem for many carers is that they often go short of sleep. This is because the person they care for needs 24 hour care or because they are unable to relax

- Eat regular meals

  If possible, get your family and friends to help with cooking and shopping. Caring for your child may take up a lot of time and they can either help make meals or with caring for your child.

- Look after your own health needs

  As a carer it is important to be as healthy as possible. You will be coping with a lot and keeping an eye on your own health will make a difference to the way you cope and feel.

Sally

—

"My elder daughter Abigail has always been very positive and supportive. She's always curios to know about Olivia's medication as she works within clinical trials."

*Relationships
*Coping together

When a child is diagnosed with a rare disease like Tay-Sachs or Sandhof disease it can have a huge effect on the whole family. For parents caring for a disabled child there are many changes and none more so than your own future. Your own plans and dreams for the future may suddenly be dramatically changed as you try to face the fact that your loved one will lead a very different life to the one you had planned.

As the diseases progress, those affected by Tay-Sachs or Sandhoff develop complex needs that change rapidly. It is not uncommon for families to struggle to cope with the emotional and financial implications that either condition brings and this can lead to the quality of family life being affected drastically.

A stable relationship provides us all with stability and support but after a diagnosis you find that you have to take on new roles. You will need to deal with emotional and financial pressures and absorb an astonishing amount of new information. These stresses can put a strain on your relationship due to both you and your partner adapting to the changes in your lives.

You may find that each of you adjust to these changes at a different pace, where one of you may want to talk openly while the other may need more time. Feelings of sadness, worry, denial, anger, embarrassment, fear, confusion, guilt, concern, resentment, and shock are normal and occur before a sense of acceptance.

It is common that having an ill or special needs child or loved one can represent a loss of control. It is important for you and your partner to talk about your individual needs and how they are affected in both a practical and emotional sense.

“I would be lying if I said our relationship didn’t change. But, Amelie’s diagnosis has actually brought us much closer together and we feel we can get through anything together.”

***Daniel**

*Siblings
*The family

A common question parents often ask is when they should give siblings information and how much to give them. The main concern is that giving information to siblings will make them anxious or it will be a burden to them. It is not unusual to find that the opposite is the case and if they are not given factual information they may make up their own incorrect story about what is actually happening. What cannot be ignored is the age of the sibling as this has a big bearing on how much information to give. You can begin by giving less detail to young children and more detailed information and facilitate the discussion of various issues to older children. In children younger than seven it is important to gauge how ready they are by the questions they may ask you.

Many parents find that the hardest thing about talking about their loved one's condition is actually dealing with their own feelings when doing so. You should try to be comfortable with these emotions as young children will be unlikely to attach these feelings you may have when talking about their sibling. When children get to around seven years of age they are more likely to be asked questions about their brother or sister at school so will need more information.

There are certain issues which are particularly relevant for siblings at different stages in their lives. Pre-school children may have feelings of anxiety of not understanding what is wrong with their older sibling while school age children may have to contend with being teased and embarrassment of the condition. Older children tend to worry about their sibling and the effect the condition has on their family and may not want to burden their parents with their own problems.

"Being Ruby's twin, Sam was too young to remember his sister without symptoms. As he grew more aware, he accepted Ruby exactly for the person she was, with limitations and an illness."

***Nikki**

Leann

—

“I think we lived in hope that some miracle cure would appear from nowhere, but in the back of minds we knew this would never happen during Lydia and Ella’s lifetime. Having an amazing network of family and friends was a great help too.”

*The extended family
*Disscussing your needs

Speak to each other

It is important that you and your partner are able to discuss how you feel and the impact the disease is having on you.

Speak to others

Your wider family will also be effectd by a diagnosis. Speak to everyone to find out how they are coping as they may be able to help you.

One of the most valuable resources for parents is to have information on the support network of people in the community who can help. This list includes people who can offer different types of support, such as when you need a break, someone to talk to, or even someone to help you do tasks like shopping. It is better to have a list of people who can help take on much bigger roles for you and your family.

There may be some people who are available to help you on a regular basis or other people may be happy to help you when you are stuck or have an emergency. You may find other parents; friends and older people are often more than wiling to help you and your family.

Many people now live away from their extended family or do not live in a community where people automatically support one another. However, you should not worry about doing something in return and for most people being able to help is reward in itself.

There are some people who find it hard to approach a family, and although they may be only too pleased to help they may need to be asked. Helping other people in need gives some people a sense of purpose and belonging and it may also help them get a better understating of your situation. However, be prepared that sometimes you may need to make the first move and one way to achieve this is to ask people round to get to know them. See if there is any small thing you can do for them and whether you are able to ask them if you can call on them if things are difficult. The worst that can happen is that they say no and if they come round and want to be sociable, it is quite likely they will be happy to help you and your family.

03

*Diagnosis stories
*Family experiences

James
—
"For families that are in the same situation than us, we hope they can make the most of everyday with their child. Every child is different; using the experience of other families to help making informed decisions about the child's care is key."

Every family has a different diagnosis experience where some families receive a quick diagnosis while others have to wait years. Diagnosing Tay-Sachs or Sandhoff disease is a very complex procedure which is why it can take some time.

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The following pages are the diagnosis stories of some of the families which The CATS Foundation supports. You will notice that everyone has a very different experience.

***No diagnosis story is the same**

*Isabella's story
*A long journey

James

"Just after Isabella's second birthday we had our first appointment with our paediatrician who straight away noticed that Isabella was having absence seizures. "

Deborah

"When Isabella was around twenty months old we noticed she was losing some of the skills that she had previously developed."

"For us, the heartbreak began on the day that a GP told us that Isabella was showing various signs of delay. She had been so ahead of her milestones earlier in her life. That conversation made us fear the worse and we shed many tears of worry. Later, when a neurologist suggested that it was probable that Isabella had a neurological condition, our worse fears were confirmed.

Our world began to feel like it was falling apart and we began to grieve for the loss of the kind of life that we thought Isabella would have had. We were devastated.

I researched every possible neurological condition until I found one that described Isabella perfectly. It was called Tay-Sachs. I knew in my heart that this was the one but tried to remain positive. When the ophthalmologist found the Cherry-red Spots in Isabella's eyes, we knew that whatever the diagnosis, it was bad news. The diagnosis came on the 15th March 2011 and it was not a shock. We were already prepared.

Our grieving, on the whole, had been done during the six months prior to that moment. From that day we vowed to make Isabella's life the happiest it could ever be, filling each day with love, laughter and adventure. A promise we have always kept.

'This is our normal' - we tell people, we don't know any different! Our lives are filled with routines that revolve around medications, treatments and feeds but in between there is plenty of time for fun. 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."

Deborah - Isabella's mum

Deborah

—

"We have done so much as a family! We have gone on trips and had experiences I don't think we would ever of attempted."

Sally

—

"I am so proud of her. She has dealt with the disease so well and manages to do so much."

*Olivia's story
*Living life to the full

Sally

—

"Olivia is a happy little girl who we make sure makes the most of her time."

"My initial reaction to Olivia's diagnosis was shock and thinking why hadn't this been tested for. Behind closed doors I had my private moments of upset. I was strong in front of other people as I felt I had to be to hold it all together. As a single parent it was difficult not having anyone to lean on. However it's made me a stronger person. People ask me how I'm going to cope further down the line. I don't look too far ahead, I live day to day.

Sally

—

"The diagnosis was a shock but is one we have had to come to terms with."

Olivia had a seizure at three years old. After a nine day stay in hospital with numerous blood tests, brain scan and lumber puncture, and 45 seizures 3 days in, epilepsy was diagnosed. We were devastated but we researched the condition and came to terms with it. The usual check ups continued. Then at the age of 13 after a routine blood test Tay-Sachs was confirmed.

My elder daughter Abigail has always been very positive and supportive. She's always curious to know about Olivia's medication as she works within clinical trials. Grandparents were very shocked and upset. They were curious to know if any of us had Jewish descendants that we didn't know of as this was where it was presumed to have come from. They didn't want to accept it and were looking for blame which I suppose is natural. I just wanted to scream accept it, I have to. Some people would say it's unlucky which I can understand considering it's so rare. I think I'm blessed or maybe chosen.

Olivia is gorgeous and loves being centre of attention and is quite funny. She's always smiling and doesn't think she is any different to anyone else. She is very demanding and it is physically and emotionally draining at times. However everyday brings something new and hopefully a cure."

Sally - Olivia's mum

*Ruby's story
*Misdiagnosis

Nikki

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"As a twin we had to make sure that both Sam and Ruby were well looked after."

Jim

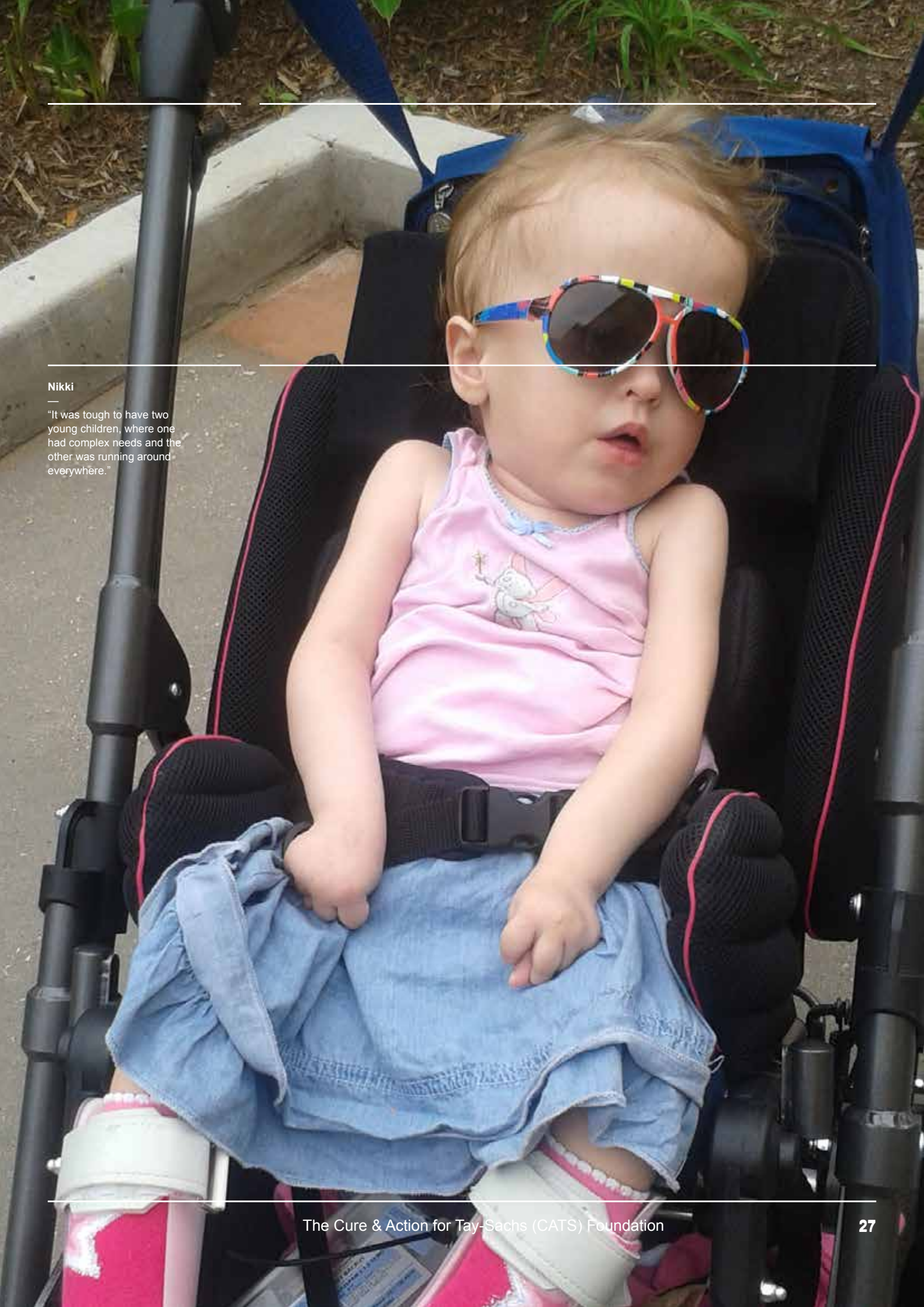
—

"I'm not sure if there is anything else we could have done. We did everything possible to help Ruby during her short life."

"It took several months for Ruby to be diagnosed with Sandhoff Disease, by the time her condition was given a name I knew it was bad news. At first I was devastated, but then as I continued to look after Ruby I realised she needed me. I promised not to give up on Ruby and to be strong for her. I also decided I wasn't going to sit back and accept her prognosis. Just because she was losing the ability to express herself, didn't mean she wasn't there on the inside. I knew she was, I could feel her. So, yes I slowly came to terms with what the future would bring, but I also did my best every single day to make Ruby's life as full as possible. Along the way I found a few lifelines and The CATS Foundation was definitely one of them.

We knew something was different with Ruby from about eight months. She was seen by pediatricians and was labeled as having Global Development Delay. She was put on more frequent observation but the overall feeling from the doctors was that her diagnosis would be mild. Ruby was never quite the same after her first year vaccines, she went quiet and didn't eat much. A few weeks after being in the USA Ruby started having seizures and was rushed to hospital. That was August and it took over a month in hospital to control her seizures. While we were there the doctors did many, many tests and finally said she had a Leukodystrophy, although they didn't know which one. We were devastated but still clinging to hope that they were wrong as so many tests came back negative. We left that hospital and took Ruby to another one where they did more tests; MRI's, Spinal Taps and many more and we were then sent home. Finally, when Ruby was sixteen months old we were called back to the doctor's where Ruby was diagnosed with Sandhoff. My world fell apart."

Nikki - Ruby's mum



Nikki

—

"It was tough to have two young children, where one had complex needs and the other was running around everywhere."

Leann

—

“The early diagnosis was a blessing in a disguise, We knew for a long time what the prognosis for the girls was and the journey we had ahead of us.”

*Lydia and Ella's story
*The twins

Leann

"Twins. Both of them affected. It was so cruel to see them both lose their battle to Tay-Sachs."

Leann

"We miss them so much but they lit up our life. Those girls made everyone who knew them smile."

"The initial diagnosis for me, was the hardest 'stage' of the disease to deal with. We came away from a meeting with the consultant with no real information about the disease and searches on the internet at that time (January 2008) weren't very helpful either. We came to the conclusion that whatever was going to happen over the next few years we would deal with as it came up and we would try to carry on as much as normal as possible. I don't think you can accept that your child is going to die until it actually happens.

I think we lived in hope that some miracle cure would appear from nowhere, but in the back of minds we knew this would never happen during Lydia and Ella's lifetime. Having an amazing network of family and friends was a great help too.

Lydia and Ella were born by emergency caesarean at 36 weeks gestation. Lydia had problems after birth with her blood sugar levels so remained on the NICU for three weeks. Ella was 'healthy' and came home with me after a couple of days. Because of Lydia's stay on the NICU she had regular check ups at the hospital. At about 5 months old she developed a squint and our consultant referred us to an opthamologist. It was at this appointment that the cherry red spot was seen on Lydia's eye and blood tests confirmed the diagnosis.

We were devastated to then find out that Ella could too have the same disease. Bloods were taken and Ella's diagnosis was received nine days later. Our journey to diagnosis was fairly early (7 months old) and we had mentioned that they weren't quite meeting the early milestones but were told because they were born early and were twins they would take a little longer."

Leann - Lydia and Ella's mum

04

*Additional help
*Who you can turn to

Patricia
—
"I think one of the best things about the UK is that the NHS gives you access to so many services who are there to help you and your family."

Caring for a child or loved one at home can be a rewarding and positive experience, providing you have the right amount of practical and emotional support, plus the aids and adaptations that will help to make daily living easier.

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It is important that you are aware of all the services which are available to you and your family so you are able to provide as high a level of care as possible.

*A rewarding job

*Who else can help
*Other services

Make adaptations

—

You should make your adaptations to your home as soon as you can.

As a carer of someone with complex needs, there will be times when you may feel overwhelmed, stressed or just tired. At these times you may want to consider enlisting some help that will allow you to have time to relax, catch up with chores or spend some time with your partner and family. There are many options open to you, such as respite care, home carers and your local hospice.

Making life easier

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All the adaptations made to your home will make caring for your loved one easier.

At some point, those affected by Tay-Sachs or Sandhoff disease will require some form of aid or adaptation to assist daily living. The aim is to meet increasing needs and to make life as easy as possible for the whole family. Thinking ahead can be traumatic for a family. However, it is advisable to make arrangements ahead of time, aiming to implement the required changes before they become necessary.

Understandably, many parents want to hold on to ‘normal’ life for as long as possible. Introducing aids and adaptations is a tangible, and in some cases a possibly difficult, step in facing up to the impact of the disease. They are designed to help and planning in advance can mean that you are prepared for your child’s changing needs. Exploring the help available may also provide some comfort and help in dealing with your fears. The priority is to arrange a full assessment as soon as you feel that help would be useful and it is usually the occupational therapist (OT) who will come to the home to discuss your needs

Thinking ahead, especially about the future path of the disease and the increasing needs of your child or loved one is always difficult. But, it is important for the family to talk about future needs with their own nursing team, even if this is just making a list of the problems you fear the most, and asking for the answers and solutions that may be available when they become needed.

Ask for help

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Do not be afraid to ask for help or assistance at home to make things easier for you and your family.

*We are here to help
*Please get in touch

Contact the charity

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We are able to answer the majority of questions you or your family may have in providing care.

It is important t isolated at the t The CATS Foun support team is guide and advis difficult time.

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*We are here for you

that you do not feel
time of diagnosis.
ndation and its
s here to help,
se you in this

Beatriz

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"The European Family Conferences have been a fantastic way for us all to get together in Europe to discuss how we all look after our children."

Eva

—

"As part of a larger family we now speak to our Tay-Sachs friends all the time."

*Acknowledgements
*Final words

Daniel - charity director
—
"The guides produced by The CATS Foundation will enable families to have access to all the informaiton 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 programe 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

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