04

Glossary of terms
Tay-Sachs & Sandhoff

A practical guide for parents and carers

Produced by
The CATS Foundation

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*Living with a diagnosis
*Tay-Sachs & Sandhoff

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Glossary of general terms
Explanations of common terms you will hear as a carer.

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Glossary of disease terms
Detailed explanations of common terms used for Tay-Sachs and Sandhoff.

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Explanations of all the support services you may have access to.

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Final words
A summary of our carer booklets and how they can help you.

Never to old to learn

There is a whole new language to learn in understanding Tay-Sachs and Sandhoff disease.

The language used by health care professionals can be very confusing so it is important that you understand these terms.

*Introduction
*Understanding the language

Katherine
—
"Without charities we would not have been able to give Jack the quality of life we did."

As a carer for someone affected by a life limiting illness you and your family will have regular visits from lots of different people from various support services. At first the language they use can be confusing and this may cause you to come uncomfortable during appointments.

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This booklet contains explanations of many of the most common terms used. This will enable you to understand the disease and also the roles that the various support services can provide you and your family.

*Daniel Lewi - Charity Director

01

*Glossary of general terms
*How to provide care

Nikki
—
"I always feared how much time we had with Ruby, but I didn't let that fear define our life. The love we had for Ruby defined us."

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

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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.

*Understanding the doctors

*Glossary of terms
*Useful information

Write things down

—

If you are confused about anything write it down so you can research and discuss it with your doctors at a later date.

Discuss things at home

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You and your family should discuss phrases or terms you do not understand at home.

During the course of your loved one's illness it is likely that you will encounter many of the terms listed in this glossary. They will probably be unfamiliar, but as time goes by you will begin to understand them. These terms describe the genetic components and medical terms relating to Tay-Sachs and Sandhoff disease and you may eventually find that you know more about the diseases than many of the non-specialist healthcare professionals you meet.

Alle

These are a particular form of gene and alleles occur in pairs, one on each chromosome inherited from each parent.

Amino acids

Organic molecules that link together to form proteins.

Autosomal

This refers to chromosomes 1 to 22 (i.e. a chromosome other than the sex chromosomes).

Blood brain barrier

A highly selective barrier formed by blood vessels and a type of brain cell called astrocytes that allow only appropriate molecules to cross from blood to brain with its function to keep harmful molecules out of the brain.

Catalyst

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

Daniel

—

"We would discuss things we did not understand over dinner and it really helped. We started to learn all the terminology about Tay-Sachs which was important."

*Common phrases
*Things to know

“I honestly didn’t think I would be able to learn everything but you pick up all the complex names and phrases. You actually become the expert.”

***Patricia**

Cells

The basic unit for life, bound by a protective membrane. Cells contain the information which allows them to replicate and function within the body.

Cerebellum

Part of the brain located behind the brain stem, under the main mass of the brain. It is known to be responsible for the coordination of movement.

Chromosome

This is a very long, super coiled DNA molecule that carries the information template and enables replication of a cell and it is located in the cell’s nucleus.

Common mutation

A mutation in a gene that occurs in a high percentage of patients affected by a specific disease.

Cytosol

The internal liquid/gel content of a cell that surrounds the sub-cellular and contains many of the simple and complex molecules required for the cell to function.

Diploid

These are cells containing two sets of chromosomes and therefore have two copies of all genes. Most cells of an organism, except the sex cells and red blood cells, are actually diploid cells.

DNA (deoxyribonucleic acid)

A very long molecule and the main component

of chromosomes. DNA contains a code which, when processed by the cell's production machinery, produces proteins. It also contains information relating to the timing and life cycle of the cell.

Dominant allele

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

Endosome

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

Enzyme

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

Fibroblasts

These are cells found in connective tissue associated with healing wounds. Skin fibroblasts are often grown to be used extensively in laboratory testing.

Gametes

These are sex cells including sperm cells and ova.

Gangliosides

A form of glycolipid, they are a component of cell membranes which are especially abundant in nerve cells which are found throughout someones body.

Genes

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

Genome

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

Genotype

The genetic make-up of each individual person which is completely unique to themselves.

Glycolipids

A group of lipids which all contain carbohydrate.

Glycosphongolipids

These are sphingolipids with one or more carbohydrate attached to them.

Haploid

These are cells that contain one set of chromosomes, examples are sex cells or gametes and red blood cells.

Heterozygous

This is a condition that exists if the corresponding genes on each of the related pair of chromosomes are different to each other. The different genes can be normal or disease causing.

Homozygous

This is a condition that exists if the corresponding genes on each of the related pair of chromosomes are the same. The genes may both be normal or disease causing.

*Lots to learn
*Test yourself

“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**

Lipid

An organic molecule that is insoluble in water. This property is important for the formation of membranes that allow separation of different biologically active solutions.

Lysosomes

These are membrane-bound organelles within the cell whose task is to recycle biochemical molecules for reuse.

Metabolism

All processes taking place in living cells. The main processes are anabolic which is the building up of complex molecules from simpler ones. The other process is catabolic and this is the breaking down of complex molecules into simpler ones.

Mitochondria

These are cellular organelles that produce much of the cell’s energy requirements. Mitochondria contain their own DNA which is inherited only through the female line.

Mitosis

A complex process of cell division whereby two cell’s are produced from one, each with its own complement of chromosomes, membranes, organelles and cytosol.

Neurodegenerative

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

Neuropathology

The structure of the nervous system.

Nucleus

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

Organelle

A sub unit of the cell usually bound by a membrane and distinct in its composition and functional behavior.

Plasma membrane

Also know as the cell membrane, this is the outer boundary of the cell.

Polymorphisms

These are variations of genes (DNA) that may improve, degrade or leave unchanged, the capability of the associated protein. They contribute towards the difference between individuals in a population.

Proteins

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

RNA

This is the ribonucleic acid which is a chemical found in the nucleus and cytoplasm of cells. It plays an important role in the formation of proteins and other chemical activities of the cell.

Recessive

The effects of a gene that are masked by the activity of the same gene on the other chromosome.

Ribosomes

Small cellular components composed of specialized ribosomal RNA and protein, the site of protein synthesis.

Sex chromosomes

Non-autosomal chromosomes (X and Y) inherited from parents. XX is female and XY male.

Sex-linked

Traits that may be either normal or disease causing and are derived from the genes or DNA of the X and Y chromosomes or mitochondrion.

Sphingolipids

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

Substrate

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

Transcribe

In terms of the cell, the process of turning the information contained on a RNA template into a protein. This task is conducted by organelles called ribosomes.

X-linked

Traits associated with the X chromosome which may be normal or disease carrying for an individual.

02

*Glossary of disease terms
*More detailed questions

Daniel
—
"I remember the first time I explained Tay-Sachs to someone. They gave me a blank look and I realised that it was such a complex disease not everyone is able to fully understand why and what the disease does to someone."

There are many terms that your loved child's health care professionals will use regarding their symptoms and treatments which is why it is an important step to begin understanding what these all are.

—

Without a basic understanding of all of the symptoms and how they affect an individual it can be difficult to know how to treat them. We suggest that you begin to learn about these symptoms so you are prepared when they start.

***Never be afraid to ask**

*Phrases about the diseases *Tay-Sachs and Sandhoff

Challenging times

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It does take time to fully understand the diseases, but once you have made the effort to learn you will be more confident at providing care to your child.

Plan ahead

—

Before you attend apppointments take time to learn why your loved one is seing that specilast as it will make the appoitment a lot easier for you to understand.

There are many terms you will hear regarding your child's illness and symptoms they may suffer from. The following terms are all specific to Tay-Sachs and Sandhoff disease and there is a brief explanation of each individual term.

Adult stem cells

Adult or somatic stem cells exist throughout the body after embryonic development and are found inside of different types of tissue. These stem cells have been found in tissues such as the brain, bone marrow, blood, blood vessels, skeletal muscles, skin, and the liver. They remain in a state of dormancy or non-dividing state for years until activated by disease or tissue injury.

Aspiration pneumonia

This is a bronchopneumonia that develops due to the entrance of foreign materials into the bronchial tree, usually oral or gastric contents (including food, saliva, or nasal secretions). Depending on the acidity of the aspirate, a chemical pneumonitis can develop, and bacterial pathogens (particularly anaerobic bacteria) may add to the inflammation.

Ataxia

Ataxia is the name given to a group of neurological disorders that affect balance, coordination, and speech. There are many different types of ataxia that can affect people in different ways. Individuals with Tay-Sachs or Sandhoff have a progressive form of ataxia which becomes more obvious as the disease develops. They lose muscle co-ordination, have gaint problems (more prominent in the Juvenile and LOTS forms of Tay-Sachs) and lose the ability to function independently.

Nikki

—

"I challenged everything; I spent every day making Ruby's life as comfortable and as fulfilling as possible. Just because she was losing the ability to express herself, didn't mean she wasn't there on the inside."

*Lots of symptoms *Challenging needs

“Learning about one symptom was quite easy. Two was a bit tougher. But then I had to learn about how they all interact with each other and the effect they have on Haylie. It has been very important to do this it’s easier to work out when she has a problem.”

***Eva**

Bronchiolitis

This is inflammation of the bronchioles, the smallest air passages of the lungs. It usually occurs in children less than two years of age with the majority being aged between three and six months. It presents with coughing, wheezing and shortness of breath which can cause some children difficulty in feeding. This inflammation is usually caused by respiratory syncytial virus (70% of cases) and is much more common in the winter months. Treatment is typically supportive and may involve the use of nebulized epinephrine or hypertonic saline.

Bronchitis

This is an infection of the main airways of the lungs (bronchi), causing them to become irritated and inflamed. The main symptom is a cough, which may bring up yellow-grey mucus (phlegm). Bronchitis may also cause a sore throat and wheezing. Acute bronchitis often occurs during the course of an acute viral illness such as the common cold or influenza where viruses cause about 90% of cases.

Cherry-red spot

A cherry-red spot is a finding in the macula of the eye in a variety of lipid storage disorders and in central retinal artery occlusion. It describes the appearance of a small circular choroid shape as seen through the fovea centralis. Its appearance is due to a relative transparency of the macula; storage disorders cause the accumulation of storage material within the cell layers of the retina, however, the macula, which is relatively devoid of cellular layers, does not build up this material, and thus allows the eye to see through the macula to the red choroid below.

Decerebrate posturing

This symptom is when someone goes into an abnormal posture. It occurs due to damage of the nervous system caused by the diseases.

Dysarthria

This is a motor speech disorder resulting from neurological damage and is characterized by poor articulation. Any of the speech subsystems (respiration, phonation, resonance, prosody, and articulation) can be affected, leading to impairments in intelligibility, audibility, , and efficiency of vocal communication.

Dysphagia

People with dysphagia have difficulty swallowing and may even experience pain while swallowing. Some people may be completely unable to swallow or may have trouble safely swallowing liquids, foods, or saliva. When that happens, eating becomes a challenge and often, dysphagia makes it difficult to take in enough calories and fluids to nourish the body. This can lead to additional serious medical problems for an individual.

Embryonic stem cells

These cells are derived from a four- or five-day-old human embryo that is in the blastocyst phase of development. The embryos are usually extras that have been created in IVF (in vitro fertilization) clinics where several eggs are fertilized in a test tube, but only one is implanted into a woman.

Enzyme replacement therapy

This therapy has been used effectively to treat other lysosomal storage diseases such as Gaucher disease, Fabry disease, and MPS1. However, with those diseases which involve enzymes needed in the brain there has not been as much success. This is due to factors such as the blood brain barrier, the foreign object response of the white blood cells and the quality of enzymes needed and which can be produced by the body.

Gastric feeding tube

A gastric feeding tube is inserted into an individual either on a temporary or permanent basis. The reasons for having a gastric feeding tube include having a condition which makes it difficult to eat properly and is inserted into an individual during a small surgical procedure. The tube goes directly into the stomach and liquid food is pumped into the individual via the tube. The feed itself is a specific type made to be used with gastric feeding tubes. The most common type of gastric feeding tube is the Percutaneous Endoscopic Gastrostomy or PEG for short. This endoscopic medical procedure involves the PEG tube being passed into the individuals stomach through the abdominal wall where the end of the tube is used to administer food and medication.

Gene therapy

The use of nucleic acid polymers as a drug to treat disease by therapeutic delivery into a patient's cells, where they are either expressed as proteins, interfere with the expression of proteins, or possibly even correct genetic mutations. The most common form of gene therapy involves using DNA that encodes a functional, therapeutic gene to replace a mutated gene. In gene therapy, the nucleic acid molecule is packaged within a "vector", which is used to get the molecule inside cells within the body.

*Technical descriptions
*New terms

Join Facebook

—

If you are not already a member of Facebook you can join the social networking site and communicate with other families online.

Join Twitter

—

If you use twitter you can interact with families all over the world and discuss your the needs of your loved one.

GM2

GM2 is a type of ganglioside. G refers to ganglioside, the M is for monosialic (as in it has one sialic acid), and 2 refers to the fact that it was the second monosialic ganglioside discovered.

GM2 gangliosidoses

A group of related genetic disorders that result from a deficiency of the enzyme beta-hexosaminidase. This enzyme catalyzes the biodegradation of fatty acid derivatives known as gangliosides. The diseases are better known by their individual names of Tay-Sachs, Sandhoff and GM2-gangliosidosis, AB variant.

GM2 ganglioside activator (GM2A)

This is a protein which in humans is encoded by the GM2A gene. The protein encoded by this gene is a small glycolipid transport protein which acts as a substrate specific co-factor for the lysosomal enzyme β-hexosaminidase A. β-hexosaminidase A, together with GM2 ganglioside activator, catalyzes the degradation of the ganglioside GM2, and other molecules containing terminal N-acetyl hexosamines.

Hexosaminidase

This is an enzyme involved in the hydrolysis of terminal N-acetyl-D-hexosamine residues in N-acetyl-β-D-hexosaminides. Functional lysosomal β-hexosaminidase enzymes are dimeric in structure. Three isozymes are produced through the combination of α and β subunits to form any one of three active dimers. The α and β subunits are encoded by separate genes, HEXA and HEXB respectively. Beta-hexosaminidase and the cofactor GM2 activator protein catalyze the degradation of the GM2 gangliosides and

James

—

"Having the support network in place where we can communicate with other families has been invaluable. To be able to talk to other parents and share our experiences and gain knowledge from them has helped as well."

*Look to the future
*Planning

“Through the school, we built up a network of friends who had children with life-limiting diagnosis, some with rare conditions, many with cerebral palsy.

***Katherine**

other molecules containing terminal N-acetyl hexosamines. Gene mutations in HEXB often result in Sandhoff disease; whereas, mutations in HEXA decrease the hydrolysis of GM2 gangliosides, which is the main cause of Tay-Sachs disease.

Hexosaminidase A (HEXA)

This is an enzyme that in humans is encoded by the HEXA gene, located on the 15th chromosome. Hexosaminidase A and the cofactor GM2 activator protein catalyze the degradation of the GM2 gangliosides and other molecules containing terminal N-acetyl hexosamines.

Hexosaminidase B (HEXB)

This is an enzyme that is encoded by the HEXB gene. Hexosaminidase B is the beta subunit of the lysosomal enzyme beta-hexosaminidase that, together with the cofactor GM2 activator protein, catalyzes the degradation of the ganglioside GM2, and other molecules containing terminal N-acetyl hexosamines.

Hypotonia (low tone)

Hypotonia is a state of low muscle tone (the amount of tension or resistance to movement in a muscle), often involving reduced muscle strength. Hypotonia is not a specific medical disorder, but a potential symptom of many different diseases and disorders that affect motor nerve control by the brain.

Infantile Tay-Sachs / Sandhofff

Infants with Tay–Sachs or Sandhoff disease appear to develop normally for the first six months after birth. Then, as neurons become

distended with gangliosides, a relentless deterioration of mental and physical abilities begins. The child may become blind, deaf, unable to swallow, atrophied, and paralytic. Death usually occurs before the age of four.

Juvenile Tay-Sachs / Sandhofff

Juvenile Tay–Sachs or Sandhoff disease is rarer than the infantile forms, and usually is seen in children between two and ten years of age. People with the Juvenile form develop cognitive and motor skill deterioration, dysarthria, dysphagia, ataxia, and spasticity. Death usually occurs between the age of five to fifteen years.

Ketogenic diet

The ketogenic diet is a high-fat, adequate-protein, low-carbohydrate diet that in medicine is used primarily to treat difficult-to-control (refractory) epilepsy in children. The diet forces the body to burn fats rather than carbohydrates. Normally, the carbohydrates contained in food are converted into glucose, which is then transported around the body and is particularly important in fuelling brain function. However, if there is very little carbohydrate in the diet, the liver converts fat into fatty acids and ketone bodies. The ketone bodies pass into the brain and replace glucose as an energy source. An elevated level of ketone bodies in the blood, a state known as ketosis, leads to a reduction in the frequency of epileptic seizures.

Late onset Tay-Sachs / Sandhofff

It is characterized by unsteadiness of gait and progressive neurological deterioration. Symptoms which typically begin to be seen in adolescence or early adulthood and include speech and swallowing difficulties, unsteadiness of gait, spasticity, cognitive decline, and psychiatric illness. It does not usually result in a limitied life span for those individuals affected by LOTS.

Lysosomal storage disease (LSD)

A group of approximately eighty rare inherited metabolic disorders that result from defects in lysosomal function.

Nasogastric tube

Nasogastric intubation is a medical process involving the insertion of a plastic tube (nasogastric tube or NG tube) through the nose, past the throat, and down into the stomach. It is used for feeding and administering drugs and other oral agents.

Seizure

An epileptic seizure (colloquially a fit) is a brief episode of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain. The outward effect can vary from uncontrolled jerking movement (tonic-clonic seizure) to as subtle as a momentary loss of awareness (absence seizure).

Stem cells

Stem cells are a class of undifferentiated cells that are able to differentiate into specialized cell types. Commonly, stem cells come from two main sources; embryos formed during the blastocyst phase of embryological development (embryonic stem cells); and adult tissue (adult stem cells). Both types are generally characterized by their potency, or potential to differentiate into different cell types (such as skin, muscle, bone, etc.).

03

*Glossary of support services
*Who can help

Nikki
—
"You should take advantage of all of the services while you can. There are some which your child can use more before the disease has progressed and you do not want to miss the opportunity."

At first you may be overwhelmed by the quantity of support services which are available to you and your family. Take time to learn who they are and what they do so you can take advantage of the services they offer.

—

We recommend that for some, one services you make appointments with as many as possible on the same day as this will mean you are not travelling every day to different appointments as this can be very exhausting for you and your family.

<u>*There are many support services</u>

*Services who help
*Access to the right people

They are there to help

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All of your support services are there to help you and your child. Make sure that you explain what you want to get out of your appointments so you both are able to manage expectations.

Share your exerpiences

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You can learn about which services are the best to use by sharing your stories with other people who are caring from someone with a life limiting illness.

Your child will have access to a variety of people and institutions who can provide help with their disease. The following provides a brief overview of each service.

Clinical nurse specialist (CNS)

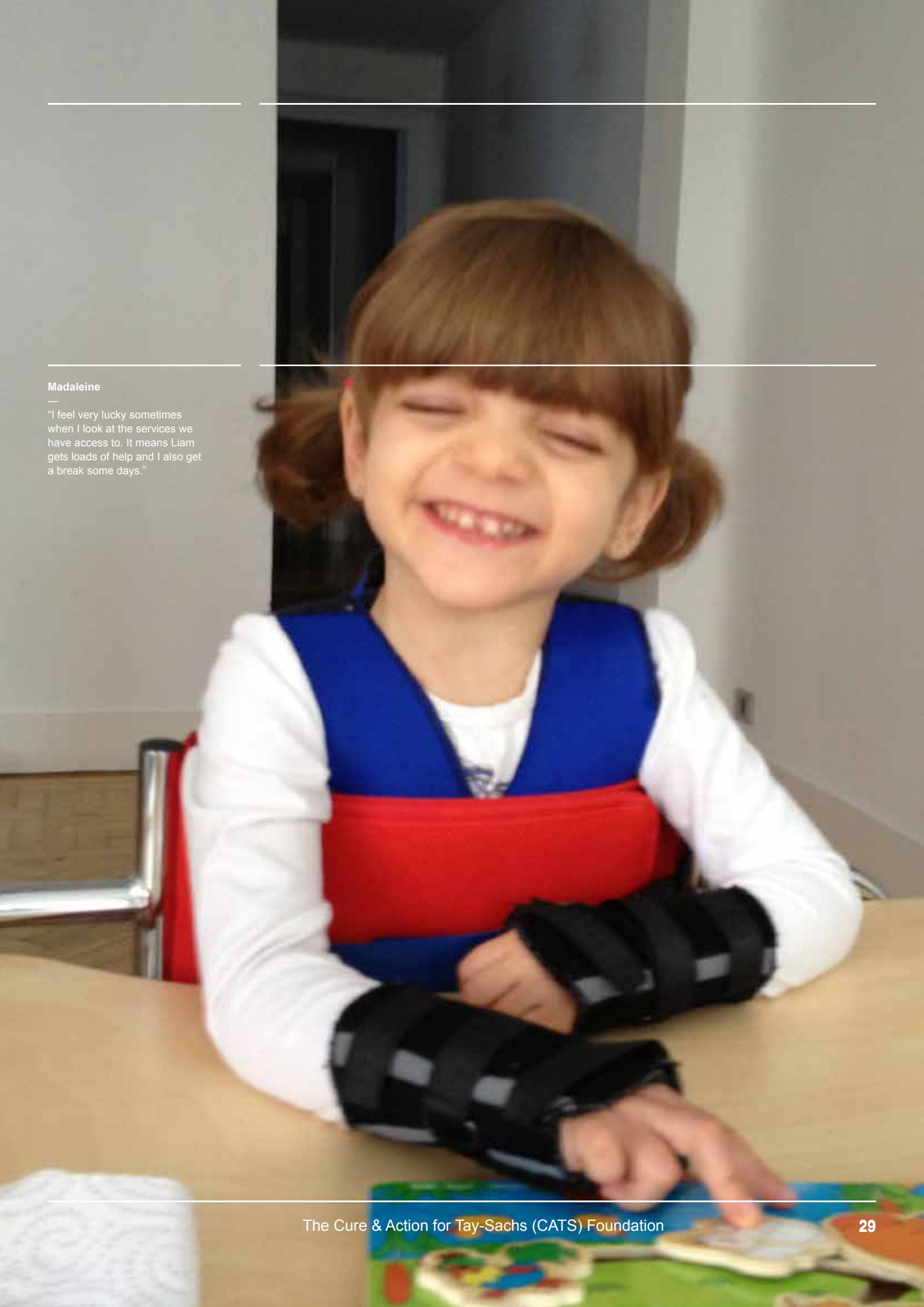
The CNS has a unique advanced practice registered nurse (APRN) role to integrate care across the continuum and through three spheres of influence: patient, nurse and system. The three spheres are overlapping and interrelated but each sphere possesses a distinctive focus. In each of the spheres of influence, the primary goal of the CNS is continuous improvement of patient outcomes and nursing care. Key elements of CNS practice are to create environments through mentoring and system changes that empower nurses to develop caring, evidence-based practices to alleviate patient distress, facilitate ethical decision-making, and respond to diversity. The CNS is responsible and accountable for diagnosis and treatment of health/illness states, disease management, health promotion, and prevention of illness and risk behaviors among individuals, families, groups, and communities.

Community district nursing

Provides a range of high quality and effective nursing care to people in their own homes, care homes and treatment rooms for individuals affected by various conditions.

Community pediatrician

They are a specialist doctor who plays a key role in the identification of children who may have special educational needs. This professional is able to assess your child's needs and provide advice and guidance on various parts of your child's care.



Madaleine

—

“I feel very lucky sometimes when I look at the services we have access to. It means Liam gets loads of help and I also get a break some days.”

*How they help
*Different skills

“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.”

[*Leann]

General practitioner (GP)

They are a doctor who treats acute and chronic illnesses and provides preventive care and health education to patients. A general practitioner manages types of illness that present in an undifferentiated way at an early stage of development, which may require urgent intervention. The holistic approach of general practice aims to takes into consideration the biological, psychological and social factors relevant to the care of each patient’s illness. Their duties are not confined to specific organs of the body, and they have particular skills in treating people with multiple health issues. is

Health visitor

Health visitors are professional individuals engaged in public health work within the domestic setting. They are mainly concerned with helping to ensure that people’s domestic behaviour is sanitary, hygienic, and beneficial to the welfare of themselves and their families, particularly to their children. As their name suggests, they fulfil their role in the community, by visiting family homes, to give advice and support to all age groups. They have a key role with regard to safeguarding vulnerable people, as they are often the first experts to enter the homes of individuals at risk of abuse and neglect, especially children.

Hospice

Hospice care is a type of care and philosophy of care that focuses on the palliation of a chronically ill, terminally ill or seriously ill patient’s pain and symptoms, and attending to their emotional and spiritual needs. They can also be used for respite services.

Hydrotherapy

This is a part of medicine, in particular of naturopathy, occupational therapy and physiotherapy, that involves the use of water for pain relief and treatment. The term encompasses a broad range of approaches and therapeutic methods that take advantage of the physical properties of water with a focus on relaxing on an individual.

Metabolic team

The metabolic medicine team have a role which involves the care of patients with a wide range of long-term conditions which demand considerable expertise to achieve disease control and improve quality of life.

Music therapy

Music therapy is the use of interventions to accomplish individual goals within a therapeutic relationship by a professional who has completed an approved music therapy program. Music therapy is an allied health profession and one of the expressive therapies to help people improve both their physical and mental health of an individual.

Occupational therapy

Occupational therapy (OT) is the use of treatments to develop, recover, or maintain the daily living and work skills of people with a physical, mental or developmental condition. Occupational therapy is a client-centered practice that places a premium on the progress towards the client's goals. Occupational therapy interventions focus on adapting the environment, modifying the task, teaching the skill, and educating the client/family in order to increase participation in and performance of daily activities, particularly those that are meaningful to the client.

Portage

Portage is a home visiting educational service for pre-school children with additional support needs and their families. Portage will offer a framework of support with regular home visits, generally weekly or fortnightly, by a trained Portage Home Visitor. Parents share with the home visitor their understanding of their child's individual gifts, abilities and support needs.

Physiotherapy

Physical therapy or physiotherapy (sometimes abbreviated to PT) is the health care profession primarily concerned with the remediation of impairments and disabilities and the promotion of mobility, functional ability, quality of life and movement potential through examination, evaluation, diagnosis and physical intervention of an individual.

Social services

Social services are a range of public services provided by governmental or private organizations. These public services aims to create more effective organizations, build stronger communities, and promote equity and opportunity.

Speech and language therapist

A Speech and Language Therapist is a health care professional who specialises in communication and swallowing difficulties. The main part of their work involves assessing people and planning therapy.

04

*Final words
*Using the booklets

Daniel - charity director
—
"Please get in touch with us if you have any questions about the contents of these booklets."

These booklets have been designed to help you and your family cope with looking after a loved one affected by Tay-Sachs or Sandhoff disease. We hope you find it useful and that it helps your family provide a high level of care.

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If there is anything which you find confusing within the booklets or if you have any questions then please get in touch with us at The CATS Foundation. Your feedback is important to us and we want to make sure you have all the tools to be an effective carer.

*We are here to help

*Contact the charity
*We can help

Always around

—

You can always contact The CATS Foundation and we will get back to you as soon as possible.

A large support network

—

The charity has a large support network around the world and is up to date on all the current treatments for Tay-Sachs and Sandhoff disease.

The content of the booklets have been tailored specifically to the needs of you and your family. The booklets contain information on a range of symptoms that may or may not be relevant to your loved one because the symptoms and disease progression vary between individuals. Our intention is to avoid providing information on symptoms that are not relevant to you and if you are unable to find answers to any questions you may have please contact the charity.

The CATS Foundation was established to provide support to families which is why we are an ideal source of information. At the charity we have contacts throughout the world who are affected by Tay-Sachs and Sandhoff disease so we are uniquely positioned to put you in touch with the correct people. Please do not be afraid to ask us any questions as we are here to help you and your family.

Nikki

"I can honestly say the CATS Foundation have been there through every part of Ruby's journey, helping her, then helping us as a family."

Deborah

—

"When we have struggled to get essential equipment through the NHS to make Isabella's life more comfortable, The CATS Foundation have stepped in and provided it."

Deborah

—

"It saddens me when there are noew cases diagnosed but they can be safe in the knowledge that The CATS Foundation can help them."

*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.

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Genetic Disorders UK

JEANS FOR GENES DAY
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Supporting families affected by genetic disorders

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