type 2 Gaucher disease

information booklet

Gauchers ASSOCIATION

contents

Thank you to Takeda and Prevail Therapeutics for their contributions to the creation of this booket

Front cover picture: Summer Smith

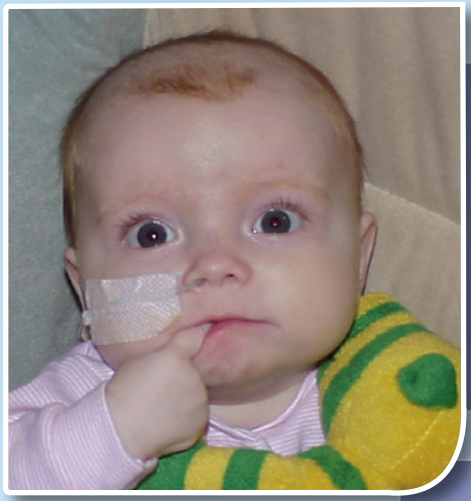
The Gauchers Association would like to thank Aimee Donald, Anupam Chakrapani and Uma Ramaswami for their contributions to this publication.

Gauchers ASSOCIATION

introduction

Type 2 Gaucher disease is a very rare, rapidly progressive form of Gaucher disease which affects the brain (central nervous system) as well as the spleen, liver, lungs and bones.

It is characterised by severe neurological (brain) involvement in the first year of life. It is also called acute neuronopathic Gaucher disease.



Ellie Carter

Fewer than 1 in 100,000 newborn babies have Type 2 disease and this form of the disease is not associated within any particular ethnic group.

what is type 2 Gaucher disease?

Babies usually appear healthy at birth but develop neurologic and other symptoms by the age of 3 to 6 months. Type 2 is almost always apparent by 6 months of age. Many children die in infancy and survival beyond 2 years is rare. In some exceptional cases, the disease course may be prolonged over a number of years.

Early signs and symptoms include slow development, squint (strabismus), poor feeding and slow weight gain.

In the subsequent months, developmental milestones may be lost (regression), there may be rigidity of the neck and limbs (hypertonia), back arching, abnormal head posturing, and noisy breathing (stridor), swallowing problems and recurrent vomiting may become apparent. The abdomen may appear very swollen due to enlargement of the liver and spleen.

As the disease progresses, other difficulties such as throat (laryngeal) spasm, seizures, low blood counts, bleeding and a failure to shake off colds and other infections may complicate the course. The lungs may also be affected and the bones may show signs of disease.

In the later stages of the disease, the infant may show signs of pain and distress that may arise from spasms, seizures, choking, breathing difficulties, infections, bleeding and bone pain. It is very important to recognize and manage these symptoms with appropriate measures and pain relief in order to keep the child as comfortable as possible. Sudden death may occur, or in some cases the baby may eventually 'switch off', not reacting to parents or stimulus, for a period before death.

how is type 2 diagnosed?

Type 2 (also called acute neuronopathic) Gaucher disease is quite distinct from the chronic neuronopathic Gaucher disease (type 3) and non-neurological Gaucher disease (type 1).

It is important that the infant is referred to one of the specialist centres for careful evaluation and initial assessment (see contact list at the end of this booklet).

What is the cause?

Gaucher disease is an inherited disorder. Children with type 2 have a severe deficiency of an enzyme called glucocerebrosidase (or GCase) which is important in maintaining the structure and integrity of all cells in the body.

The enzyme deficiency results in the accumulation of fatty substances (glucocerebroside and related chemicals) which are normally produced during the recycling of cells in the body and are then broken down by the enzyme. Babies with type 2 Gaucher disease are unable to break down glucocerebroside and related chemicals. Instead, these substances remain stored within cells of the body, preventing them from functioning normally and eventually leading to their destruction. The cells affected include those found in the bone marrow, spleen, liver, lungs and brain.

The diagnosis is usually made on the basis of the clinical features, enzyme analysis in blood and/ or genetic (mutation) analysis.

is there a treatment?

No specific curative treatment for type 2 Gaucher disease is available at present.

Although enzyme replacement therapy and substrate reduction therapy has been found to be effective for type 1 and in some cases type 3 Gaucher disease, these therapies have not worked in treating type 2 infants.

Like in most conditions with brain involvement, once neurological damage has occurred, this cannot be reversed.

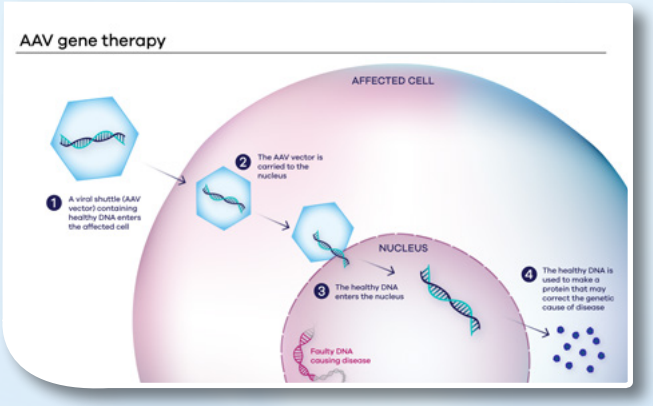
In type 2 Gaucher disease, brain damage starts while the baby is within the womb and to date, no treatment has been effective in preventing this; however, recent advancements in research and understanding of the disease have opened up new possibilities for future treatment options. Currently there is research being conducted to explore the application of an experimental, potential one-time gene therapy intended to slow or stop disease progression in type 2 Gaucher disease. Details of this research can be found below:

Investigational clinical trial for type 2 Gaucher disease

Gene Therapy

Gene therapy can be defined as the introduction of genetic material to cells of patients for therapeutic benefit. While there are several different approaches to gene therapy, in simple terms, it involves providing a healthy functional copy of a gene to the patient's cells to compensate for a defective copy that causes the disease. Ideally this treatment would only need to be administered once in the lifetime of the patient. Even though the concept underlying gene therapy is straightforward, delivering genes into cells of a living organism is a very challenging process; however, researchers are exploring established approaches along with novel techniques to overcome some of these challenges. For example, one approach being studied is called AAV (adeno-associated virus) gene therapy, which utilizes a modified virus or "vector" to deliver genetic material into cells.

Prevail Therapeutics - a biotechnology company focused on developing disease-modifying gene therapies for people living with neurodegenerative conditions - is currently conducting clinical trials to assess the safety and efficacy of an experimental, potential one-time AAV gene therapy called PR001 intended to slow or stop disease progression in children affected by neuronopathic Gaucher disease (nGD).



Source: Prevail Therapeutics, a wholly owned subsidiary of Eli Lilly & Co.

Patients with Gaucher disease have mutations in both copies of the GBA1 gene, which contains the instructions to produce a protein called beta-glucocerebrosidase or GCase. GCase works in a compartment inside of cells called the lysosome, where it is responsible for the disposal and recycling of glycolipids (a type of fat). When there is an insufficient amount of active GCase, the lysosome does not function normally, which can lead to the symptoms seen in nGD. To deliver a healthy copy of the GBA1 gene to the cells of the brain, PR001 uses a viral vector (or shuttle) called AAV9. AAV9 is a well-studied vector used by a number of companies developing gene therapy products, particularly those that target disorders of the central nervous system. The therapy is designed to enable brain cells to produce enough active GCase for lysosomes to properly function, and potentially slow or stop the disease from progressing.

In clinical trials, PR001 is administered by a one-time injection into the cerebrospinal fluid in an area above the spinal cord called the cisterna magna.

This type of injection is a direct, non-surgical technique that has been used safely for other purposes for many years.

Information about Prevail's current clinical trials for nGD can be found at **www.ClinicalTrials.gov** or by visiting their website at **www.prevailtherapeutics.com**.

what are my options?

Professional help and support is available, whether the child is being looked after in hospital or at home. It is important to talk to the doctors and nursing staff especially if parents are finding it hard to deal with a particular issue.

It is worth considering support from a local hospice as they can offer respite and support for the whole family

(please see contacts at end of this booklet)

Although there is currently no cure for type 2 Gaucher disease, the symptoms can be managed with appropriate measures and specific medications.

With help and support, parents can deal with many unfamiliar situations, in particular medical techniques to care for their child. Doctors will outline, at the time of diagnosis, how the disease will progress. It may be difficult at that time for parents to fully anticipate the practical difficulties which may arise, for which the medical team will give ongoing support.

Management of the symptoms and problems that arise as the disease progresses is important in order to ensure a good quality of life and may include:

- Poor feeding and weight gain
- Infections
- Seizures
- Spasms
- Pain
- Bleeding
- Excessive secretion
- Breathing difficulties

Measures such as nasogastric tube feeding, antibiotics, anticonvulsants, pain relief, regular suction and oxygen may be required. Some children develop spasm of the vocal cords (laryngospasm) that can result in choking and may require specialized assessment and management.

Ellie Carter

Input from a range of health professionals is usually necessary and it is important to have a coordinated approach to multidisciplinary assessment and day-to-day management.

This may include the GP, local paediatricians, specialized and community nursing, neurologists, dieticians, physiotherapists, speech and language therapists (SALT) and ear nose and throat (ENT) specialists.

One of the major contributions given by nursing staff is moral and emotional support – this cannot be under-estimated. Social workers can help by giving advice and information on financial allowances and benefits available for parents.

Relatives and friends can give invaluable support. If possible enlist their help in the everyday caring of the child. They often want to help and this can give parents a much needed break, even if just for the evening, or spending more time with another child.

Contacting parents who are, or have been, in a similar situation can also be useful – they alone can understand the pressures on parents who are caring for a sick child. See the contact list at the end of this information booklet.

Is type 2 Gaucher disease an inherited disorder?

Type 2 Gaucher disease is inherited. Both parents must be carriers of the disease in order for there to be a risk of them having an affected child.

For a couple who are both carriers, there is a 1 in 4 (25%) chance with each pregnancy that the child will have the disease, a 1 in 2 (50%) chance the child will be a carrier and a 1 in 4 (25%) chance that the child will neither be a carrier nor have the disease.

Much of a person's make-up is a result of what is inherited from each parent. Many characteristics, such as eye colour, blood groups and genetic conditions, are passed from parents to children through their genes. The genes for these characteristics are organised on 23 pairs of chromosomes, one of each pair coming from the mother and one from the father. Each chromosome carries thousands of genes.

The gene which instructs the body to make the enzyme glucocerebrosidase is also passed on from both parents to their children. In Gaucher disease, this pair of genes is defective. As a result, the enzyme produced from the defective pair of genes, one gene inherited from each parent, is unable to perform its normal function.

Gaucher disease is an autosomal recessive disorder. Autosomal describes the type of chromosome on which the gene is carried. Recessive refers to the fact that in order to develop the disease, a child must inherit two defective genes, one from each parent.

A person with one functional gene and one defective (GBA1) gene is a GBA1 mutation carrier and never develops Gaucher disease. Carriers are perfectly healthy and will not develop any symptoms of Gaucher disease.

As long as one of the pair of genes is functional, enough enzyme can be produced to prevent abnormal chemicals from accumulating within the cells of the body.

If only one parent is a carrier of Gaucher disease, none of the children will have Gaucher disease but there is a 1 in 2 chance (50%) of the child being a carrier.

It must be emphasised that the chances in each pregnancy, of the child inheriting Gaucher disease, are totally independent of whether or not a previous child has the disease. Having had one child with Gaucher disease does not mean that the next three children cannot inherit the disease.

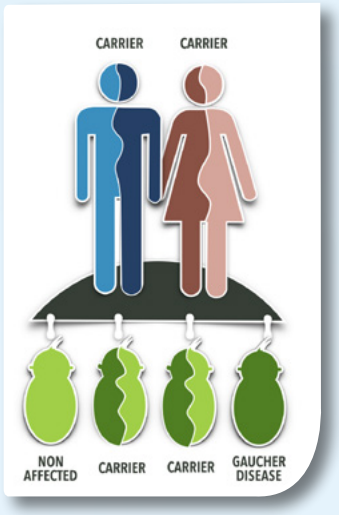
Genetic Counselling

In families where a baby with type 2 Gaucher disease has been born, the parents and their close family may wish to be offered genetic counselling. Your paediatrician or GP can arrange this.

What is genetic counselling?

Genetic counselling is a process of communication on various aspects of genetic information that empowers individuals to make informed decisions and seek further information about a medical condition and future genetic risks.

Genetic counsellors often are part of the multidisciplinary teams caring for rare diseases. Strong communicative and supportive element is key for the genetic counsellor that ensures that those who seek information are able to reach their own fully informed decisions without undue pressure or stress.



For example, during reproductive counselling advice, the genetic counsellor will provide the person/couple all the information they need to arrive at their own informed decision. The genetic counsellor will draw a detailed family tree and quantification of risk, explains the purpose and types of the genetic testing, explains prenatal diagnosis and screening and common diagnostic test and discuss various genetic techniques such as invitro fertilisation (IVF), pre-implantation genetic diagnosis (PGD). The discussions will also include the limitations and risks associated with methods available for prenatal diagnosis. The genetic counsellor role also extends to discussions around ethical, legal, social and psychological considerations; and importantly support for individuals, couples and extended family as required.

The role of the genetic counsellor has expanded significantly over time. They are able to provide information at different stages of life. Some examples include family screening for a rare disease, predictive genetic testing, understanding inheritance patterns, genetic risks during pregnancy, supporting the individual during pregnancy and subsequently the children if they present with clinical manifestations. Appropriate genetic counselling results in positive outcomes in genetic decision making for the individual and their families.

With advances in genetic testing and genomic medicine, the role of the genetic counsellor is increasingly important to enable individuals to adapt to the vast amount of new genetic information that may become available.

The psychological and emotional support provided by the genetic counsellor cannot be underestimated, as often they may have to support the individual and their families during and after counselling for inherited conditions.

In the UK, the Human Fertilisation and Embryology Authority (HFEA) is an organisation that oversees the licensing, monitoring and inspecting fertility clinics. The HFEA has approved over 600 rare diseases for pre-implantation genetic diagnosis, including type 2 and type 3 Gaucher disease.

Early engagement with a genetic counsellor is important before accessing PGD.

Prenatal Testing

In families where a baby with type 2 Gaucher disease has been born, pre-natal testing on subsequent pregnancies is available and can be discussed in advance of a pregnancy.

A chorionic villus sample (CVS) or amniocentesis can be performed to diagnose the disease early in pregnancy. A CVS is carried out at around 10 weeks of pregnancy and an amniocentesis around 16 weeks. In CVS, a sample of cells is taken from the developing placenta under ultrasound guidance and analysed. In amniocentesis, a needle is inserted through the mother's abdominal wall into the amniotic sac holding the baby. A sample of amniotic fluid removed, and cells are separated and tested. Results are usually available within a few days.

PGD

PGD (pre-implantation genetic diagnosis) is licensed by HFEA (Human Fertilisation and Embryology Authority) for use in couples who have a 1 in 4 risk of having an affected child (both partners are carriers of type 2 Gaucher disease. In PGD, the genes of embryos created by IVF (in vitro fertilisation) are checked for the condition before implantation. If you would like to know more please contact your GP.

Carrier Screening

Close relatives of a family where a baby with type 2 has been born and who are of reproductive age or younger may wish to be offered genetic counselling in order to discover if they are a carrier. Carrier screening may then be offered.

The chance of close family members being carriers exists but there is only a risk to their children if their partner is also a carrier of Gaucher disease. The chance of any one person in the general population, outside of an affected family, being a carrier of type 2 is very small.

Harper Dorothy's story starts on the 15th February 2018, she was born via emergency c section at Cramlington hospital weighing a healthy 8.01oz. It is written by her mum, Carly Edminson.

The first time I held her was bliss, taking in the sight of this stranger I'd been growing for 9 months. Her perfect blue eyes and long hair, her tiny toes and her squeaky little cry.

Nurses gathered round to do routine checks and to figure out why she was so squeaky, they consulted the ward sister then took our little one to the special care unit to provide her with some oxygen. I was taken to recovery. After a little while I was taken back to meet harper for the second time. I still couldn't believe this little angel was mine. We bonded, and she fed. She struggled to get the hang of it at first but then again we were both new to it so I wasn't worried.

The nurses told me they were going to keep Harper in for a little while just observe her as her SATs would sometimes drop and she was holding her breath when she would cry. They reassured me that it would be OK.

A week passed, I would make my way daily to the SCBU to see Harper and wouldn't leave her side until late into the night. At the end of the first week my husband, who had been by our side throughout, took the opportunity to go home, tidy the house and get a good nights sleep. His sister took his place by my side.

That night as I went to sleep after a day of cuddles, feeds and observations a Dr came rushing into the room to talk to me.

Harper Dorothy

He informed me that they were becoming increasingly concerned about Harper's breathing, her breath holding and the squeaky little breaths she would do whilst crying.

Harper Dorothy

He informed me that they were going to transfer her to the Freeman, a heart specialist hospital, to check on her heart then transfer her straight to the Royal Victoria Infirmary, a children's hospital in Newcastle. There they would check her over and observe her, and had access to more technical equipment.

I rang my husband who arrived within minutes, his mother just behind to take his sister home. We watched as they took her away in the ambulance. We followed behind in our car.

Upon arrival at the RVI they informed us that her heart seemed healthy however her breathing rate is extraordinarily slow but they were no further forward to finding out why. At this time in Harper's life she was doing only 6 breaths per minute compared to the 25-30 she should be doing.

I barely remember that week; all I remember is being so desperate to get our Little Mouse home. I barely slept.

As the week went on an ear nose and throat doctor came to see us several times. On the 3rd occasion he came to us with an initial diagnosis. Laryngomalacia. A floppy larynx. Something that would get better as her larynx matured and her throat strengthened. He told me she would be fine. Our only concern at the time was her weight, no matter how many feeds, whether breastfeeding, bottle or NG feeds. She just didn't seem to gain weight, certainly nothing significant. By the end of that week we had battled and worked furiously to get Harper to gain weight, the goal was to get her up to the 9 pound mark. We got her to the 8.11oz mark and were finally discharged to the community.

Coming home was amazing but nerve-wracking. Within days of arriving home our entire family had been to meet her and fallen in love.

We settled into our routine; Dad would leave for work early in the morning, me and Little Mouse would lie in bed watching tv and she would feed. We would pull ourselves out of bed and play downstairs or go for walks, see family, attend clinics, meet with health visitors and nurses. Every day was full.

Two weeks flashed by. We did our usual routine, I put my sleepy, full baby down for a nap and stayed with her for 20 minuntes to check she was really asleep. I turned the baby monitor on and nipped out the room for a moment, just a moment. I heard noise over the monitor and dashed back in to see Harper was choking and wasn't breathing. I jumped into action, I cleared her airways and started CPR. She took in the biggest breath and started breathing again. I had called an ambulance and it was there in moments. By this time Harper was fine of course.

The ambulance took us to the local and A&E where my husband met us. They gave her oxygen and sent us on to the children hospital.

We were there for 2 and a half months.

Whilst in the children's hospital they ran tests, they observed and tried desperately to figure out what was making our Little Mouse struggle so much.

Harper still wouldn't put on weight, she was now completely NG fed due to her choking and was losing some of her cognitive abilities. She struggled to track, her eyes becoming more permanently glued upwards. Her smile was rare as it would cause a great deal of strength. Her breathing still very slow and her arms and hands would grip tightly.

About a month into our time in the RVI hospital, Dr Ramesh, came to us with a diagnosis: Congenital myasthenia, a condition that affects the nerves and breathing. He explained its rarity, and that with medication it can be managed. He even put us in touch with a family with a daughter that had the condition. The last person he ever

diagnosed with the condition. She was 13 years old, she rode horses, and attended school. A normal child. She had a tracheostomy, something they had mentioned Harper may need.

They called a meeting of all the experts; Doctors and nurses involved with Harper's care, along with myself, Harper's dad and my mother in law. They explained the next steps of treatment and explained they believed it necessary to fit Harper with a trache.

I was devastated but placed my trust in the experts knowing it was for the best.

And it was; over the next few weeks Harper made massive improvements. Her trachy made it possible for her to take 15 breaths per minute and the salbutamol and pyridostigmine medication helped her gain some cognitive abilities and relaxed her muscles that were always so tense.

We were trained up and given all the equipment we would need for at home care. In June 2018 we were finally discharged.

Adjustment at home was difficult but worth it. We were able to have the comfort of our own home, Harper had her own bed (not that she ever slept in it) we had suction machines, feeding machines, trach kits, catheters, SATS machines, medication and all the syringes we could possibly need.

We settled into our new normal. Learning how to change Harper's trachy, clean it and monitor it.

Night-time was always the nerve wracking part, even though we had a SATS monitor I could never help but to count her breaths, check her heart rate.

The nursing team, physio and health visitors were amazing; they would come and meet with us nearly every day to check how we were managing, making sure we never felt alone or struggled. They were our guardian angels and our life lines.

A few weeks went by and arranged an appointment to get a larger trachy as even though Harper wasn't putting on weight she was growing. They asked me to come in the following day. I travelled

to the RVI children's hospital they showed me to the Doctor's room where a trolley with glass vials lay. They wanted Harper's blood. I asked why as I thought this was a trachy appointment. The doctor informed me that they believe her initial diagnosis was wrong and it could be another condition, Gauchers disease. But it could also not be. They needed to run more tests.

Harper Dorothy

We returned home where I stupidly *Googled* the condition researching the likely hood etc. And thus ensued the longest few days of my life, waiting to hear back to see if Harper was to live months or years.

Whilst at home waiting, Harper developed an infection which meant being rushed to the RVI for treatment. After the first dose of antibiotics and a stint on the oxygen, Harper's temperature started to come down and she stabilised. We were called for a meeting.

Me, Harper's dad, her paediatrician, her nurses, her ENT and airways doctor were all there. They broke the news to us. Harper has type 2 Gauchers disease. The way she is presenting shows us she will most like not make it to 6 months.

I grieved, my 4 and half month old Little Mouse... my little girl so tiny and precious. A lifetime worth of memories I had planned out during the 9 months I had grown her flashed before my eyes. Birthdays, weddings, parties, Christmas, Halloween, swimming and holidays....

I pushed the grief to one side. My stubborn nature and determination came out. I wasn't going to let this condition steal it all away from me, from her, from us.

I began to form the beginning of a plan.

The day after we were discharged, I sat with my Little Mouse and me and my husband began to plan all the things we could fit in to this tiny amount of time.

We used social media to ask for suggestions - I didn't want to miss a thing. We raised money so we could do it all for her.

We started with the aquarium, we went to the beach, we went to a butterfly house, she met her favourite book character *The Gruffalo*, we threw a massive birthday party (a very early birthday party) we went swimming and took her to our family holiday spot in Scotland. Every single day was full of something new.

Then in October she was there for our wedding. Our perfect flower girl. The day was flawless; she behaved all day. One of the best days of my entire life, second only to the day she was born.

A week later as her health declined, she was only weighing 6.1lb at this point, very frail and would sleep a lot. We knew it was time. We hosted Halloween on the Saturday and Christmas on the Sunday. She met Santa, we sang songs, had a real Christmas tree, watched Christmas films... there was even snow. As the night faded Harper began to struggle, I managed to attach the ambi bag and get her to keep breathing. Her Dr arrived and told us she was fading. And if her heart stopped it would be too hard for Harper to keep it going.

She was always a fighter. Our little wonder woman. She lasted another week. The seizures started on the last day, getting longer and longer as the day went on.

Then on the 11th of October 2018, Harper Dorothy finally finished her fight. She passed away surrounded by her family and in my arms. She was finally at peace having lived a full and wonderful life.

Harper's story, whilst a difficult one to hear sometimes is one of hope. I share her story far and wide to show that whilst Harper's life was tragically short it was filled with magic, wonder, love and happiness.

A lifetime full of beautiful memories, endless hugs and bravery.

"What do we do when our hearts hurt?

We wrap them in love, with friendship, shared tears and time... till they wake hopeful and happy again"

Charlie Macksey

specialist centres

Specialist Paediatric Lysosomal Storage Disorder Centres in the UK:

Birmingham Children's Hospital
Steelhouse Lane, Birmingham B4 6NH Tel: 0121 333 9907/08 (Secretaries)
Fax: 0121 333 9998
Consultants:
Dr Suresh Vijay Dr Si Santra

Great Ormond Street Hospital for Sick Children
Great Ormond Street
London WC1N 3JH
Tel: 020 7405 9200 (ext. 0075) Fax: 020 7813 8258
Consultants:
Dr Spyros Batzios, Dr Anupam Chakrapani, Dr James Davison,
Dr Emma Footitt, Dr Maureen Cleary, Dr Mildrid Yeo, Prof Paul Gissen,
Dr Julien Baruteau, Dr Stephanie Grunewald

Royal Manchester Children's Hospital
St Mary's Hospital, Oxford Road Manchester M13 9WL
Tel: 0161 701 2137/2138
Fax: 020 701 2303
Consultants:
Dr Simon Jones; Dr Alex Broomfield; Dr Bernd Schwahn, Dr Arunaba Ghosh,
Dr Sergei Korenev, Dr Andrew Morris

University Hospital of Wales
Heath Park, Cardiff CF14 4XW Tel: 02920 747 747
Consultant: Dr Graham Shortland

Organisations that can offer help and support to families:

The Gauchers Association
www.gaucher.org.uk
The Gauchers Association, 8 Silver Street, Dursley, Gloucestershire, GL11 4ND,
Tel: 01453 549 231

Information on Gaucher disease. Have a Patient & Family Support Worker to help with non-clinical support needs.

Contact a Family
www.contact.org.uk
209 City Road, London EC1V 1JN Free helpline: 020 7608 8700

Supports families whatever the disability or health condition. Offers local support, resource library and connects families.

The Compassionate Friends
www.tcf.org.uk
53 North Street, Bristol BS3 1EN Tel: 0845 123 2304

Supporting bereaved parents and siblings. Has a helpline, online forums and offers local support.

Child Bereavement UK
www.childbereavement.org.uk
Child Bereavement UK,
Unit B Knaves Beech Way, Knaves Beech Industrial Estate,
Loudwater, High Wycombe, Bucks HP10 9QY
Helpline: 0800 02 888 40

Supporting families with bereavement.

Together for Short Lives
www.togetherforshortlives.org.uk
New Bond House,
Bond Street, Bristol, BS2 9AG.
Helpline: 0808 8088 100

The UK voice for children and young people who are not expected to reach adulthood and their families. Families can search for palliative care services and children's hospices in their local area.

Winstons Wish
https://www.winstonswish.org
17 Royal Crescent, Cheltenham, GL50 3DA
Helpline: 08088 020 021

Supporting families with bereavement.

Rainbow Trust
https://rainbowtrust.org.uk
Cassini Court
Randalls Way, Leatherhead, Surrey, KT22 7TW
Tel: 01372 363438

Emotional and practical support for families whose child has a life-threatening illness

Emma Hall

Contact us

For more information contact:

The Gauchers Association,
8 Silver Street, Dursley,
Gloucestershire, GL11 4ND

Tel/Fax: 01453 549 231
Email: ga@gaucher.org.uk

www.gaucher.org.uk

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