Stepping Up

Your stepping stone forward

By young adults with Gaucher disease type III

Dedicated to the memory of

Laurenna Simpkin

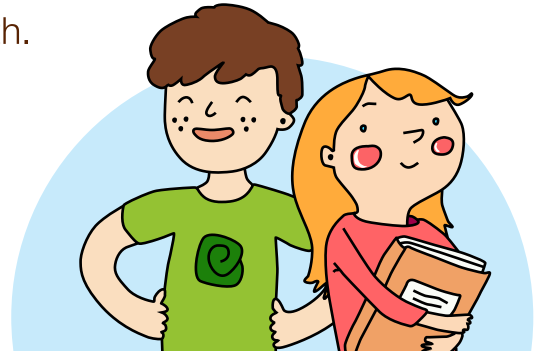
1984–2013

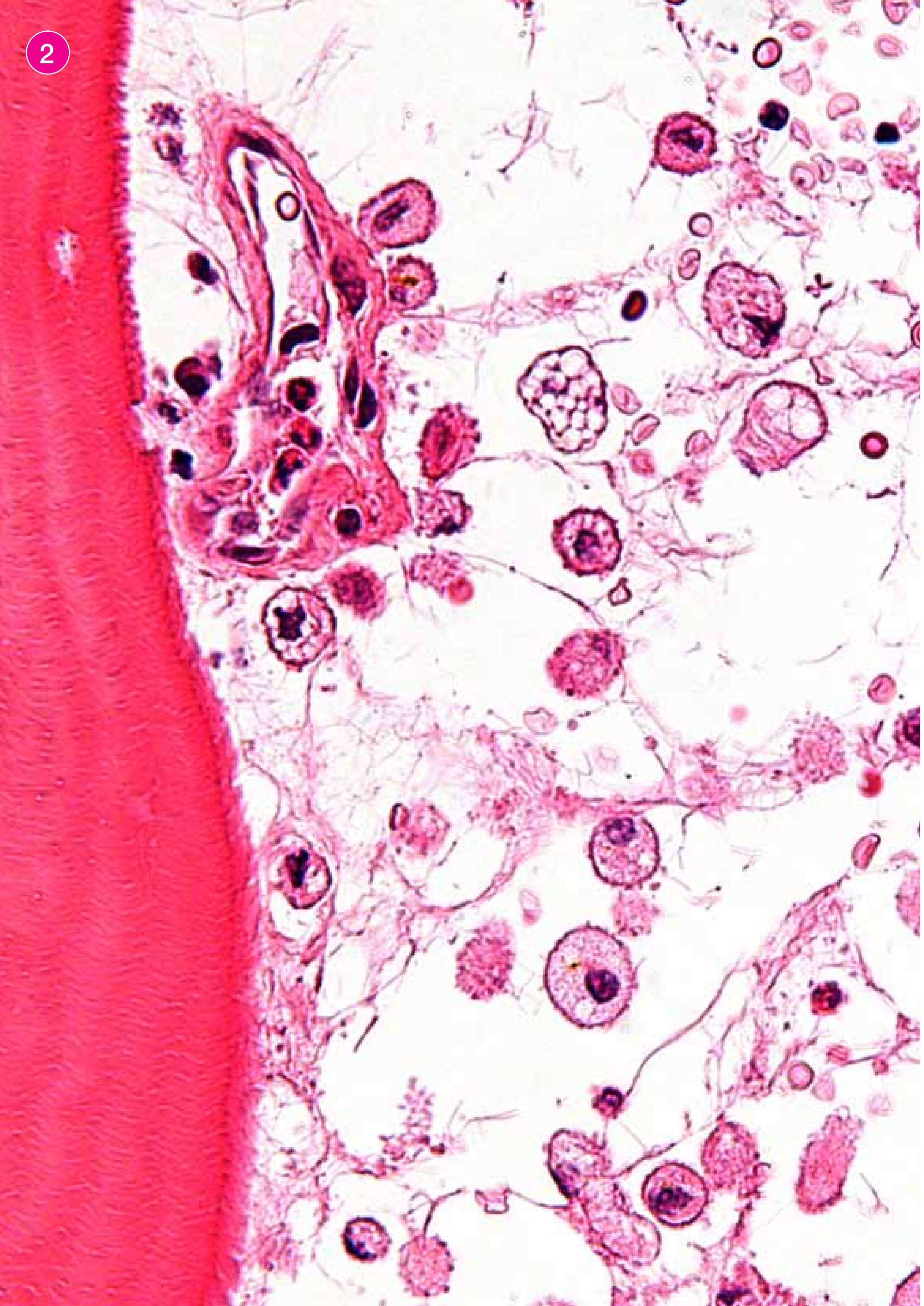
This booklet is dedicated to our friend Laurenna,
she was a true inspiration and we will miss her terribly.

From left to right:
Saphia, Maddie, Sara Khan,
Sara Pickton, Nadia and Irma

This booklet has been written by young adults ages 16-28 who have grown up with type III Gaucher disease. The aim of this booklet is to help you understand what to expect while going through transition into adult services.

We are hoping our experiences, stories and information will guide you through the process and make it easier to cope with.





What is Gaucher disease type III?

by Sara Khan

Gaucher is a genetic disease in which a fatty substance (lipid) accumulates in cells and certain organs. The disorder is characterized by bruising, fatigue, anemia, low blood platelets and enlargement of the liver and spleen. It is caused by a hereditary deficiency of the enzyme glucocerebrosidase (breaks down a fatty substance called glucocerebroside). The enzyme is defective particularly in white blood cells. Glucocerebroside can collect in the spleen, liver, kidneys, lungs, brain and bone marrow.

Symptoms may include enlarged spleen and liver, skeletal disorders and damage to the bones that may be painful, severe neurologic (nervous system) complications, swelling of joints, distended (painful) abdomen, jaundice (a yellowish pigmentation to the skin), anemia, low blood platelets and yellow fatty deposits on the white of the eye (sclera). Persons affected more seriously may also be more at risk of infection. Some forms of Gaucher disease may be treated with enzyme replacement therapy.

Type III (the chronic neuronopathic form) is normally diagnosed in early childhood and occurs in approximately 1 in 100,000 live births. It is characterized by slowly progressive but milder neurologic (nervous system) symptoms, other major symptoms include an enlarged spleen and/or liver, seizures, poor coordination, skeletal irregularities, eye movement disorders, blood disorders including anemia and respiratory (organs) problems.

Routine Gaucher Tests

by Madeline Collin

Blood Test

Bone marrow

1. Haemoglobin – indicates the levels of iron in the blood, too low can indicate anaemia.
2. Platelets – too low can cause bleeding; too high can cause the blood to clot.
3. White cells – too high can indicate an infection

Chitotriosidase

This shows the number of active Gaucher cells – high is bad.

Liver

to measure its size and to see if there is any scar tissue.

Clotting

to see if the blood clots.

Kidney Function

to see if they are working properly.

MRI

of the bones

to look at the skeletal system.

of the head

to look at the brain.

EEG

electricity in the brain (in some people).

Various tests

to look at how electricity is conducted.

Ultrasound

to see if the liver and spleen are enlarged.

Auditory

hearing tests.

Auditory Processing

to see how sounds are interpreted by the brain for Type III.

Lung Function

to look at how efficient the lungs are at taking in and breathing out air

Visual Tests

How well you can see.

Eye Movements for Type III

to look at the eyes, horizontal (sideways) and vertical (up and down).

Bone Density Exam

to see if the bones are hard.

Neuropsychology

tests to see how the brain is working.

Echocardiogram

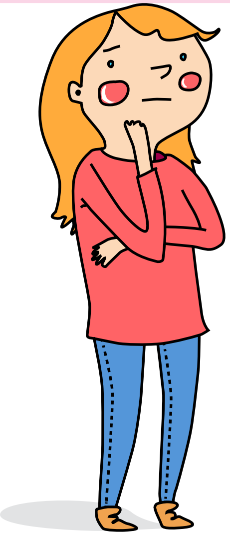
to look at the heart; its blood flow, shape, size and tissue damage.

What is meant by adult transition?

by Irma Shah

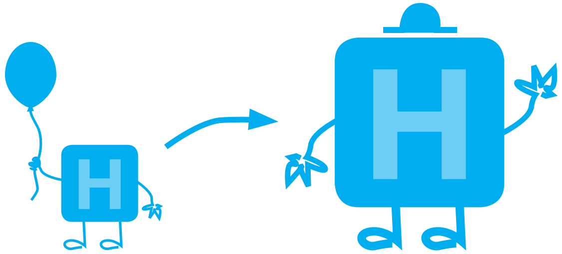
When a person is diagnosed at a very young age with their illness they visit the children's hospital which means they are under the paediatric consultant. As the person is growing up into an adult there comes a time when they become 18 and it is time for them to be transferred to an adult hospital.

Deciding which hospital to be transferred to...



When deciding on which hospital you would like to be transferred to you may need to think about some things such as

- Do the consultants have knowledge and experience about your illness?
- Are the consultants and staff friendly?
- How far away is the hospital going to be?



Moving to an adult hospital

advantages

- You are an adult so you will be able to talk to the doctors by yourself.
- You have the opportunity to make your own appointments and travel to your appointments independently.

disadvantages

- Moving from paediatric hospital can be emotional as you will lose contact with all the doctors and nurses who have been looking after you for a number of years.
- The hospital is designed for adults so you won't find any toys or books.

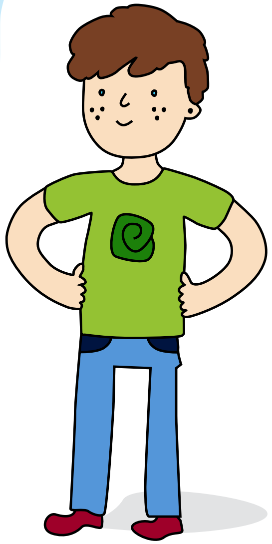
DON'T BE SCARED

BE BRAVE

Moving to an adult hospital may be scary, but it's also very exciting because you are being treated like an adult

Treatment management with enzyme replacement therapy (ERT)

by Karl Phillips



Most people manage their infusions at home. Your medication and all your medical equipment are delivered to your home.

Your ERT has to be kept in a fridge which is also supplied.

Your medication has to be reconstituted with sterile water which you will be shown by your specialist nurse the exact amount of water to use.

Your ERT then has to be put into a bag of saline.

Some patient's parents do the infusions which means you can do it when it's convenient to you. Others have a nurse who comes out to their home, once again at a time convenient to you. Some patients just have a nurse to cannulate them, then the patient or parent does the rest of the infusion. Most infusions are between 1-2 hours.

Having your treatment at home means no more travelling to the hospital, trying to find somewhere to park, then waiting for your infusion to be made up, having the infusion, then having to travel back home.

You will have training to do your treatment at home so there's nothing to worry or stress about.

You can organise your ERT for when you want it to fit around your life.

Managing other areas of care

Appointment choices

- Thinking about who you want to bring e.g. parent, friend partner etc...
- Making your own appointments
- File your clinic letters so you know what is happening
- Take letters or notes from other appointments with you
- Prepare questions in advance for clinic
- When you get your clinic letter if you have any questions then please contact your doctor or nurse

Laurenna Simpkin

My Story

Gaucher disease is damaging to the spleen, the enzymes and the immune system. Most patients today are treated with Enzyme replacement therapy. During the mid 1980's there was very little knowledge of how to treat Gaucher Disease and there wasn't a treatment. I had to have chemotherapy and a bone marrow transplant by a blood related sibling. They mainly used this therapy for leukemia sufferers. In my case, my siblings were not a match and I had to find a suitor. This was very hard for me because there are big risks.

In 1985 I went through chemotherapy to weaken my immunity which made me very weak and ill. Then in 1986 I had my first bone marrow transplant which unfortunately my body had rejected my second dose of chemotherapy made me worse and it made me very vulnerable to fight infection and it had affected my reproductive system, my final bone marrow transplant was successful.

My Gaucher Disease has caused me no end of problems, like double vision, balance and hearing problems. I find it hard to lift and carry heavy things because my back is very painful at time. I find school challenging because I have learning difficulties and suffer from epilepsy, despite all this I never gave up I have achieved six GCSE's and I went to college to study many things. I now work at New Look clothes store in East Grinstead on a Saturday morning. The rest of my free time I spend with my boyfriend.

Editors' Note:
Sadly Laurenna passed away in August this year. She was one of the Type III patients who put this booklet together and we wanted to include her story.

Sara Pickton

My Story

My name is Sara and I have Gaucher Disease type III. It was two years that I have been diagnosed at UCL London hospital. It all started when I felt somewhat very ill. At first I had bad flu symptoms such as a bad sore throat and headaches but it had become worse over time and I knew something wasn't at all right. I went to my local doctor and I was told that I had glandular fever and I just had to rest for it to be better. After few weeks nothing changed, after having several tests no one seemed to know what the problem was. I was very anxious at this point. I was told to go to a London hospital where I had more tests, my spleen was large and my blood platelets were too low, the only result I was given was leukemia and that I had to be tested for it many times to be sure. My family was very concerned and I was scared. In London I was sent to see Doctor Hughes at The Royal Free Hospital, there I was tested for Gaucher Disease which was something I had never heard of at the time as it is a very rare condition. It wasn't for certain but I had to be tested for it anyway. The tests came back after some time and I was diagnosed with Gaucher Disease. This wasn't at all easy for me to digest as I was told that this is a lifelong genetic condition but with enzyme therapy I would feel better.

I had so many thoughts in my mind, how would my friends and people treat me? Would I have to change my lifestyle? And what really is Gaucher Disease? I was pretty much in denial and felt alone. Despite this rough journey I have met amazing people along the way that have supported me throughout, like Doctor Hughes, all the medical team and nurses that helped me understand my Gaucher Disease as well as keeping me thinking positive.

Support

by Saphia Shah

Educational

- What levels of support am I entitled to on a day to day basis?
- What levels of support am I entitled to during exams?
- Transferring to Sixth Form
- Applying to college or University
- Moving away from home to study
- Living at home whilst studying
- Are there any grants or bursaries available to me whilst I am studying?

Housing/ Leaving Home

- Do I qualify for any housing benefits?
- Am I entitled to apply for any home efficiency to help me with my bills?
- Am I entitled to apply for any grants to help with home adaptions?
- Do I qualify for any benefits when leaving home e.g. phone, electricity, no of rooms proximity to hospital.
- Finding a new GP when you leave home.

Employment

- What are my rights to time off for hospital appointments?
- What are my rights to time off for treatment?
- What do I have to disclose to my employer
- What are my rights under the Disability Discrimination Act?
- What is the role of a work based occupational health department?
- Are there any types of employment to be avoided?

Who Can help me to get the support and help I need

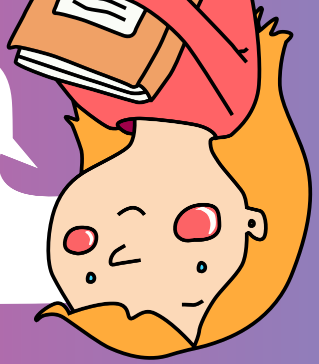
- Social Worker
- Mental Health
- Clinical Nurse Specialist
- Patient Organisation
- General Practitioners (GP)
- College/University support workers?
- Family planning clinics
- Connexions

Benefits

- Am I entitled to Disability Discrimination Act/Personal Independence Payments, who can help me with my forms?
- Am I entitled to Employment Support Allowance, who can help me with my forms?
- Am I entitled to Universal credits, who can help me with my forms?
- Can I top up my wages if I am not able to work full time?
- Am I entitled to Free Prescriptions?
- Am I entitled to any support with travel costs?
- Are there any other personal or carer allowances that I can apply for?
- Do I have any disabled parking entitlements?

Glossary

by Radhika Dhayatker



Bone marrow
a soft substance found in bones that produce blood cells e.g. red blood cells or white blood cells.

Brain
an organ which controls all parts of the body.

Calcium
a mineral mainly found in hard parts of bones. It is essential for healthy teeth and bones. Calcium is also important for muscle contraction and blood clotting.

Central Nervous System
The brain and the spinal cord make up the central nervous system. The brain lies protected inside the skull and from there controls all the body functions by sending and receiving messages through nerves.

Dexa scan
is a bone density test that determines whether or not you have osteoporosis (means porous bones, which affects the structure, strength, and integrity of bones. Bones afflicted with osteoporosis are subject to becoming brittle which break easily as well as hinder a patient's ability to stand, walk, or be active for long periods of time). This is a test that uses dual x-rays with different energy levels to measure the grams of calcium and other bone minerals packed into a given bone segment.

Enzyme
in your body, chemical reaction rates are controlled by enzymes, these are biological catalysts which speed up chemical reactions. In Gaucher disease there is a deficiency of the enzyme 'Glucocerebrosidase' which is why patients have enzyme replacement therapy.

Enzyme replacement therapy (ERT)
a treatment that replaces or supports specific enzymes in the body so the body functions properly- how it should.

Eye movement
is when the eyes can either move horizontally or vertically.

Gaucher cells
cells which are larger than they should be found 'stuck' in the liver, spleen and bone marrow of people who have Gaucher disease.

Haemoglobin
found in the red blood cell which carries oxygen in the blood to all parts of the body.

Hearing
the ability to recognise sound and voice.

Infusion
an injection of a liquid that carries medicine around the body.

Iron
an essential mineral. Iron is necessary for the transport of oxygen (via haemoglobin in red blood cells).

Kidneys
an organ which filters the blood and removes excess salts and water.

Liver
an organ of the body that helps with digestion, distributes nutrients throughout the body, cleans the blood and helps it clot properly.

Lungs
Either of two spongy, sac like respiratory organs in most vertebrates, occupying the chest cavity together with the heart and functioning to remove carbon dioxide from the blood and provide it with oxygen.

MRI scan
The MRI scanner uses magnetic and radio waves to create pictures of tissues, organs and other structures within the body, which can then be viewed on a computer.

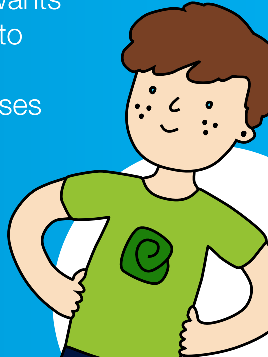
Nerves
one or more bundles of fibres forming part of a system that conveys impulses of sensation, motion, etc., between the brain or spinal cord and other parts of the body.

Platelets
blood cells that allow the blood to clot and that help to form a scab during the healing process.

Spleen
an organ of the body that makes, filters, and stores red blood cells.

Ultrasound scan
An ultrasound scan is a painless test that uses sound waves to create images of organs and structures inside your body. It is a very commonly used test.

X-rays
an image that can be taken of the bones of the body. With Gaucher disease, the doctor wants to take a closer look to see if there are any fractures or weaknesses in the bones.



Gauchers
ASSOCIATION

Registered Charity No: 1095657

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PAL
AWARDS
Patient Advocacy
Leadership Awards

Supported by the UK Gauchers Association through Genzyme's Patient Advocacy Leadership Awards (PALs) – Empowering young type III GD patients to shape the future.