

Generalised lymphatic anomaly (GLA)

A close-up photograph of a young boy with light brown, spiky hair and freckles. He is smiling broadly, showing his teeth. He is wearing a dark-colored hoodie. The background is a dark, possibly black, fabric, likely a chair or sofa. The overall lighting is soft and natural.

Information for families

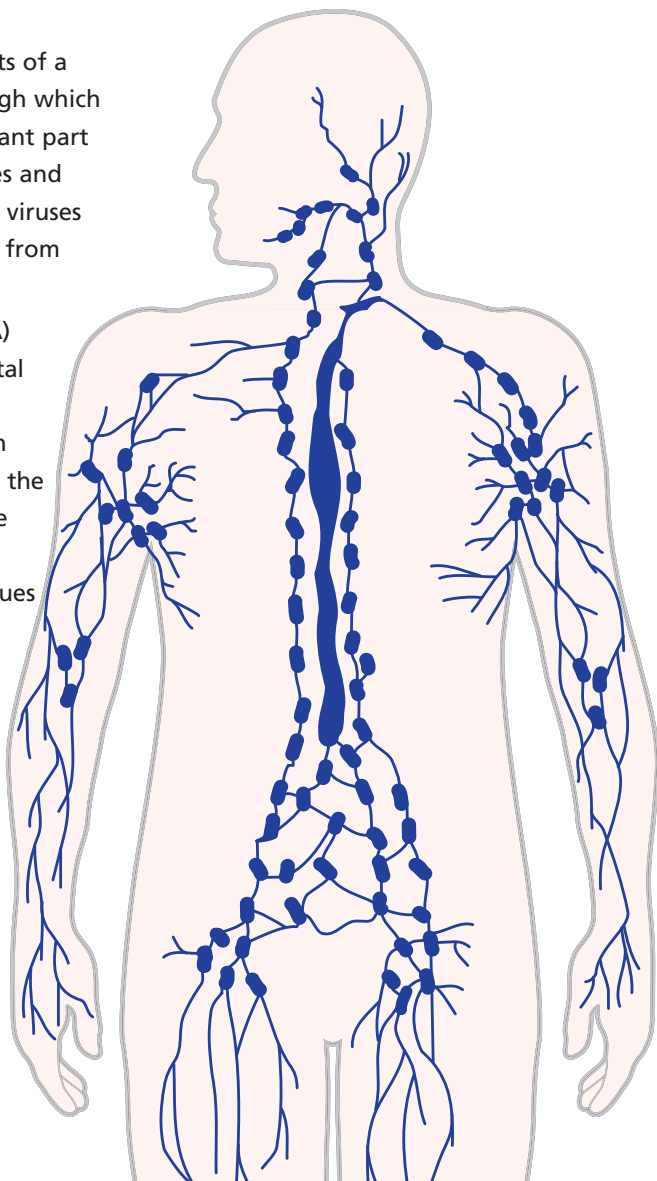
Great Ormond Street Hospital
for Children NHS Foundation Trust

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of generalised lymphatic anomalies (GLA) – previously known as lymphangiomatosis – and where to get help.

The normal lymphatic system consists of a network of vessels and nodes through which lymph circulates. It forms an important part of the immune system that identifies and fights off invaders, such as bacteria, viruses and tumour cells. It also absorbs fat from the digestive system.

Generalised lymphatic anomaly (GLA) is the name given to a rare, congenital (present at birth), and progressive disorder of lymphatic channels which can affect different organs including the bones and the intestines. It can cause problems if the abnormal lymphatic tissue develops within important tissues and structures.

It does not 'spread' from one area of the body to another. However, the affected area can get worse over time. If a new affected area is diagnosed, it is highly likely the anomaly was always there but may not have been identified initially.



What causes GLA?

It is thought that GLA is caused by an error in the development of lymphatic tissue in early foetal life. It does not appear to be inherited (passed from parent to child), although it may develop as a result of a genetic mutation (change) that occurs out of the blue (sporadically). Further research is needed to better understand the causes of GLA. It affects males and females in equal numbers.

What are the signs and symptoms of GLA?

The symptoms of GLA vary depending on the extent of the abnormal lymphatic structures, and on the areas affected.

- When bones are affected, there may be bony pain and risk of fractures if the bone structure is weakened.
- When the digestive system is involved, a swollen tummy, diarrhoea, nausea and problems with nutrition may arise.
- The spleen may be affected, though this is often without symptoms.
- The kidneys can be affected, which may lead to a rise in blood pressure and loss of protein in the urine (proteinuria). This is caused by damage to the tiny filtering units in the kidneys, which can be monitored using urine and blood tests.
- Coagulation (clotting) problems including recurrent bleeding or bruising.

- Lung involvement can lead to breathing difficulty. If the space between the lungs and ribcage is affected, this can lead to pleural effusion.

How is GLA diagnosed?

As GLA is such a rare condition, diagnosis will usually only be possible at a specialist centre with input from different medical teams including dermatology (skin specialists), paediatrics (children's specialists), radiology (imaging specialists), haematology (blood specialists), orthopaedics (bone specialists), but also cardiology (heart specialists), gastroenterology (digestive system specialists) and urology (genitourinary system specialists), depending on the site of the lesions.

Imaging scans such as magnetic resonance imaging (MRI) or ultrasound will be used to work out which areas of the body are affected. A biopsy (small sample of tissue) may be taken to examine under the microscope to confirm the diagnosis and check for specific markers to guide treatment.

How is GLA treated?

The aim of treatment is to improve symptoms and to reduce adverse effects on the different body organs, and will depend on the specific features of each case.

Standard treatments include sclerotherapy in case of lesions made of big lymphatic spaces (macrocytic). Sclerotherapy involves injecting a medicine into the cysts, which irritates them encouraging them to scar and shrink. Surgical removal (debulking) of the lesion is another approach but can lead to scarring and leakage of fluid (lymph) for prolonged periods after the operation. All these approaches are invasive and have very high recurrence rates.

Medical treatments such as sirolimus (rapamycin), are increasingly being used. There is still not a great deal of high quality data regarding its long term use in GLA.

Trouble shooting

In most cases, the affected areas just need looking after carefully.

- **Pain** – the affected areas can be painful. In some cases, regular pain medicines such as paracetamol are enough to deal with any discomfort but occasionally stronger pain medicine will be needed.
- **Infection** – it is important to check for signs of infection such as cellulitis (inflammation of the skin) as this can lead to sepsis (widespread life-threatening infection) if untreated. In many cases, a short stay in hospital with intravenous (into a vein) antibiotics will be needed.
- **Bleeding** – if an affected area starts to bleed, apply pressure over it with a clean handkerchief, cloth or tissue for at least five minutes. If blood soaks through the handkerchief, cloth or tissue, put another one on top and keep up the pressure. Do not take it off to have a look as this could start the bleeding again. If the bleeding continues, even after pressing down on the area for five minutes, go to your nearest NHS Walk-In Centre or Accident and Emergency department. The team at GOSH can usually be contacted for advice if needed.
- **Skin care** – the skin over the affected area can be dry so we recommend avoiding using bubble bath, rinse any soap or shampoo off carefully and pat the area gently afterwards. You can ask for special moisturising cream to put in

the bath. We also recommend using a bland (non-scented) moisturiser over the skin. Sometimes, an antiseptic cream will be recommended.

If you have any concerns about your child, please seek medical advice. Some parents have found it helpful to keep a file of their child's information, as GLA is not a well-known condition. See if our 'family file', available online at www.gosh.nhs.uk/parents-and-visitors/coming-hospital/family-file will be helpful.

What happens next?

GLA can be an extensive condition, affecting many areas of the body. The outlook for children and young people with this condition varies depending on the area of the body affected and its severity. If the spinal area is affected, it may limit the sports and activities that can be done safely. It does not, however, affect neurological (brain and nervous system) development, so children are usually able to attend mainstream school with support and go on to live a near-normal daily life.

Vascular anomaly research is an area of medicine that is continually advancing. Studies have already given us improved options for treating GLA and continue to tell us more about how and why it develops. We hope that this leaflet has been helpful in giving you more information about your child's condition. If you have any comments about it, please contact the Birthmark Unit.

Further information and support

At Great Ormond Street Hospital (GOSH), contact the Birthmark Unit.

The parents of a child being treated at GOSH have set up a registered charity in the UK, working together with existing organisations to support patients and their families. Visit their website at www.alfiemilne.org.uk or look for their Facebook page – search for Alfie Milne Lymphangiomatosis.

The main organisation offering help and support to families affected by lymphangiomatosis is Lymphangiomatosis & Gorham's Disease Alliance – Europe. Visit their website at www.lgdalliance-europe.org for further information. There is also a similar organisation in the United States, whose website is www.lgdalliance.org.

Everyday life with lymphangiomatosis (GLA)

Tracy's Story (Alfie's mum)

It was very hard in the beginning, after begin given the diagnosis. I felt very alone and very scared. So little was known about the disease and no one could really tell me what the future held for Alfie.

In the beginning there were lots of hospital appointments, hospital admissions, unexplained high temperatures, pain so much pain. Our wonderful little boy was so miserable and there was nothing I could do. After he was started on some medication we saw some changes, the pain was less, he seemed so much happier in himself. This disease is very unpredictable, we have good days when he is happy and full of energy and it is hard to believe that there is this horrible disease inside him and then the bad days come and you realise how quickly it can all change. So fragile. So precious. Over time this has got better, we have more good days than bad and we have started to gain back some control in our life. Have even started to make plans for the future again.

Alfie's mobility has suffered because of the disease and he hasn't walked unaided for many years now. We continue with daily physio in the hope that one day he will get back on his feet. If I ask him if he could change anything what would it be? He'd say, "to be able to run with my friends".

Every day is the same, medicine, stretches, more medicine, school, physio, more medicine and creams to apply before bedtime. Subconsciously, taking note of any new pain he has been experiencing, checking his body over for any signs of further swelling, bruising. If he feels unwell and has a temperature is it just that 'he is feeling unwell' or is it the disease? The logistics of the many hospital appointments. Preparing him for blood test and scans. Being the one responsible for 'knowing it all'. It helps me to document everything, every appointment, result, scan and



pending appointment. It's good to have this just in case I'm not there and someone else has to take him to his appointment.

What scares me is not being there for him, making the wrong decision that will affect the rest of his life. I worry about the amount of time he has off school, playing catch up in the classroom and how this will affect his friendships. I worry because he is different and that he will struggle to be accepted in this society which, more often than not, is based on what you look like.

All of this has a huge impact on family life, siblings, relationships, work. It's not easy to juggle this on top of day to day life but you find a way.

Over time things have got better, easier.



As the years pass, you get to know your child well and you begin to know the signs of when things aren't right and you learn to act quickly.

We have found strength in finding others and I feel it is my job, as a parent, to raise awareness of this very rare disease to help others who are at the beginning of their journey. The only way to help my son, other than love and care for him, is to get involved in any way that I can to help further research into finding better therapy drugs that will give him a better quality of life and hope for the future.

Alfie's Story (aged 8)

I can't ever remember not having GLA. I know that the disease is mainly in my leg and around my tummy. It has something to do with my lymphatic system. I have one fat leg and one normal leg. I don't really understand it but I know that it gives me pain and has stopped me walking – I don't ever remember walking without 'zippie' (walking frame) or my crutches.

I have to go to a lot of different hospital appointments which means I miss quite a bit of school. I'm so glad the hospital has things to do otherwise it would be so boring. I have to have my bloods checked a lot which I hate but the nurses are really good. I take a lot of medicine, which isn't nice, but I know I have to take it to keep the pain away.

I just want to be like my friends. If I could change anything it would be to walk just like my friends.



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