LGD Alliance Europe

Lymphangiomatosis & Gorham's Disease

LGD Alliance Europe helps to educate and support patients, their families and healthcare professionals to improve patient care.

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www.lgda.eu

WHAT IS LYMPHANGIOMATOSIS?

Lymphangiomatosis (spoken as limf-an-jee-oh-mah-TOE-sis) consists of the words:

- lymph: lymph
- angi: vessel
- oma: tumor (benigne)
- tosis: condition

So it is a condition with tumors in the lymph vessels.

Lymph vessels reach almost every part of your body. That's why lymph-angiomatosis can occur anywhere, but the most common location of the disease are the bones and lungs.

The tumors are caused by the size and increasing number of lymphatic vessels.

Lymphangiomatosis is also known as Generalized lymphatic anomaly (GLA).

GLA has many similarities with GSD. 75% of all patients with GLA has bone involvement leading some to conclude that GLA and GSD should be considered as a spectrum of disease rather than separate diseases.

WHAT IS GORHAM'S DISEASE?

Gorham's disease is a rare bone disease in which the bone disappears and vessels and tissue increase in that spot. The disease is called vanishing bone disease or massive osteolysis as well.

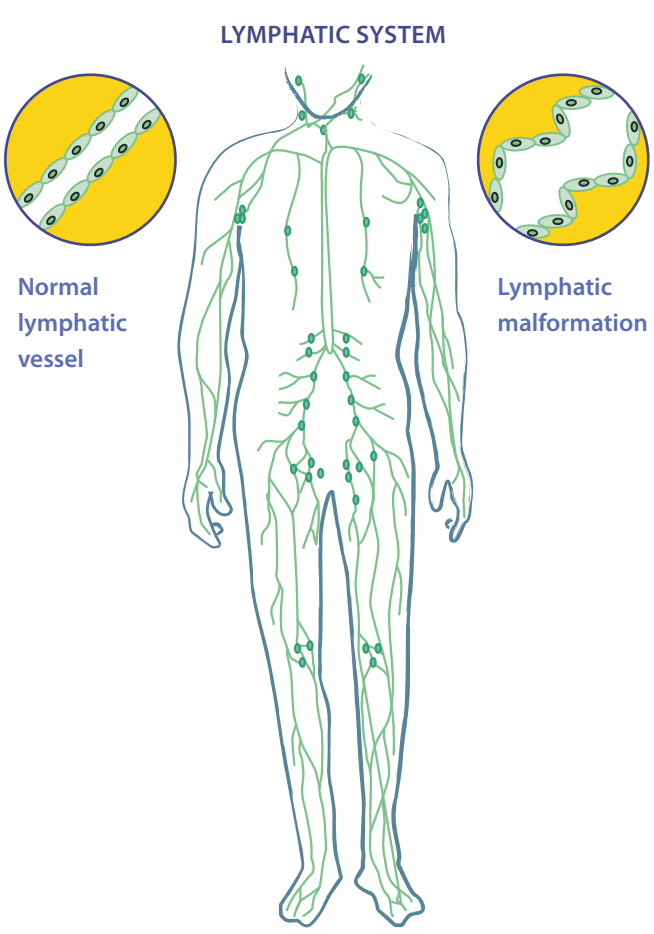
- osteo: bone
- lyse: disappearance

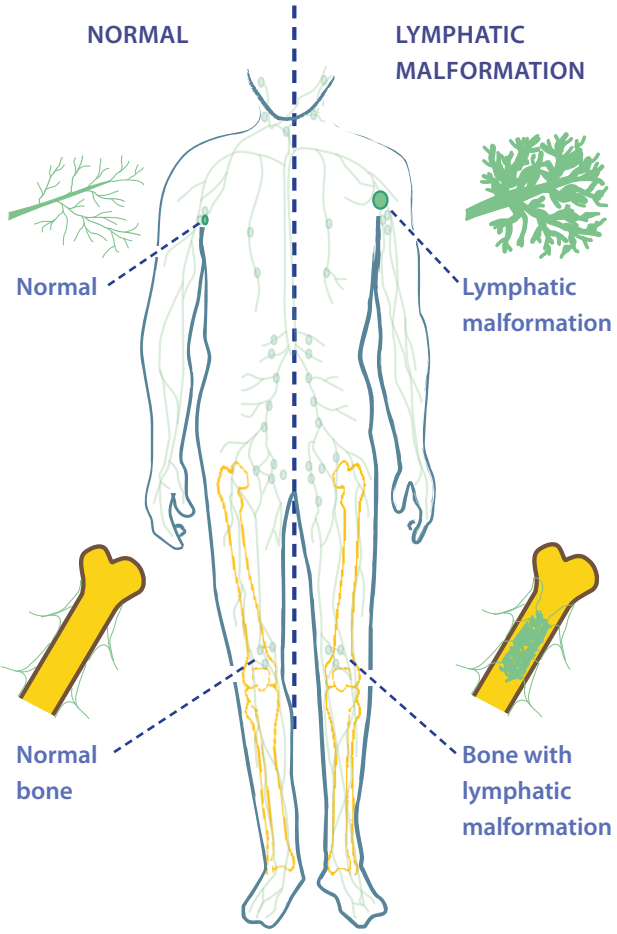
Gorham's disease is also known as Gorham-Stout Disease (GSD).

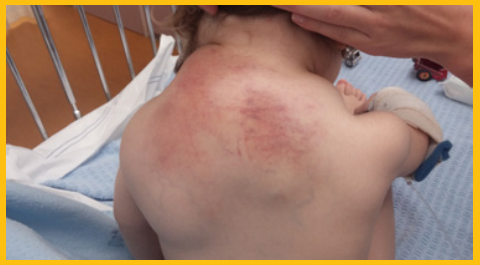
It is thought to be closely related to lymphangiomatosis due to the proliferation of lymphatic vessels in the bone. As these vessels proliferate, they aggressively invade the adjacent bone leading to resorption and replacement of angiomatous tissue.

KAPOSIFORM LYMPHANGIOMATOSIS

Kaposiform lymphangiomatosis (KLA) is distinctive from GLA with abnormal spindle-shaped lymphatic cells. There is limited information on the long-term outcomes of patients with KLA, compared with other patients with generalized lymphatic anomalies (GLA). Patients with KLA may have a worse prognosis, particularly once pulmonary involvement develops.

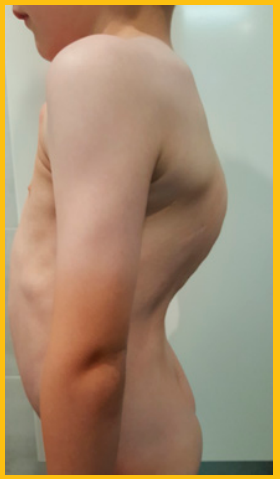

LYMPHATIC SYSTEM
Normal lymphatic vessel
Lymphatic malformation


NORMAL
LYMPHATIC MALFORMATION
Normal
Lymphatic malformation
Normal bone
Bone with lymphatic malformation



Lymphatic malformation in neck, shoulder and back. Pressure in the malformation sometimes causes small blood vessels to rupture.

Lymphatic malformation in a leg.



Lymphatic malformations inside bones can weaken them. The back can get distorted once vertebra are involved.

WHO GETS IT?

It strikes males and females of all races and exhibits no inheritance pattern. It occurs at any age, but the incidence is highest in children and teenagers. Signs and symptoms are typically present before the age of 20 and the condition is often under-recognized in adults.

HOW COMMON IS THIS DISEASE?

Because it is so rare, and commonly misdiagnosed, it is not known how many people are affected by this disease. Estimates are that between 10 and 30 new patients with GLA or GSD are born in the European Union each year.

WHAT ARE THE SYMPTOMS?

Early signs of disease in the chest include wheezing, cough, and feeling short of breath.

The pain that accompanies bone involvement may be attributed to "growing pains" in younger children. With bone involvement the first symptom for disease may be a pathological fracture.

DIAGNOSIS

It's quite challenging to set the right diagnosis and misdiagnosis occur frequently. It is a rare disease with features similar to other more common diseases like asthma or osteoporosis.

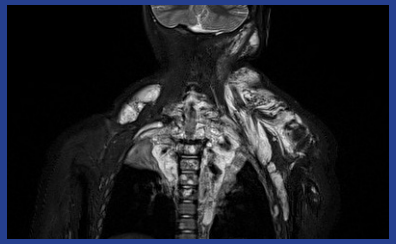
Invasion

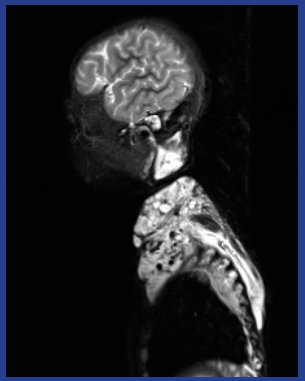
Even though this disease is benign, as these vessels proliferate, they aggressively invade the adjacent tissue, bone or organs leading to resorption and replacement by angiomatous tissue.

Diagnostic tools

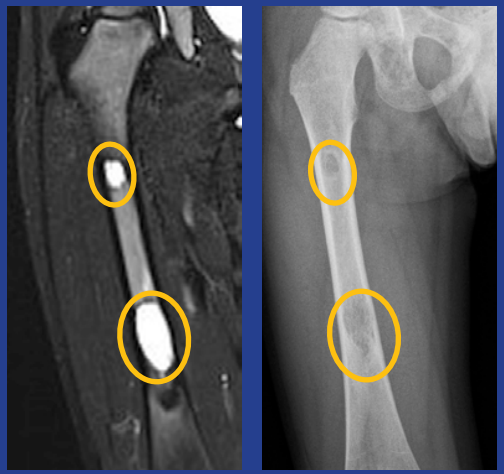
While x-rays, a CT scan, MRI, ultrasound, lymphangiography, bone scan, and bronchoscopy all can have a role in identifying this disease, biopsy remains the definitive diagnostic tool. A biopsy (small sample of tissue) may be taken to confirm the diagnosis and check for specific markers to guide treatment.

However, there are reports of biopsy resulting in serious complications, such as abnormal leakage of chyle in the pleura. Please discuss this with your doctor or consultant.





MRI images of lymphatic malformation in neck, shoulder and back. Most of the bright areas are lymphatic fluid. The brain is bright as well, but is not involved.



Left: MRI image of a bone with lymphatic malformations.

Right: X-ray image of the same bone with lymphatic malformations.

WHAT TREATMENTS ARE AVAILABLE?

There is no standard approach to the treatment of lymphangiomatosis and Gorham's disease.

Treatment is often aimed at reducing and easing symptoms, such as fatigue and immunosuppression.

Removing tumors is often not possible, because the lymphatic vessels are surrounded by blood vessels. Medical therapy, diet, interventional radiology and surgery are possible treatment options. A multidisciplinary approach is needed.

Examples of medical therapy:
- Sirolimus
- Bisphosphonates
- Vincristine (chemo)
- Thalidomide

Diet (necessary on occasion):
- Total parenteral nutrition (TPN)
- Low-fat and high-protein diet

Interventional radiology:
- Sclerotherapy

Surgery options:
- Drainage
- Bone cement
- Transplant

NEED A SECOND OPINION?

Sometimes doctors have a hard time to set the right diagnosis. Or choosing the right treatment is difficult, because your doctor doesn't have any knowledge of this disease. This happens in the case of an extremely rare condition.

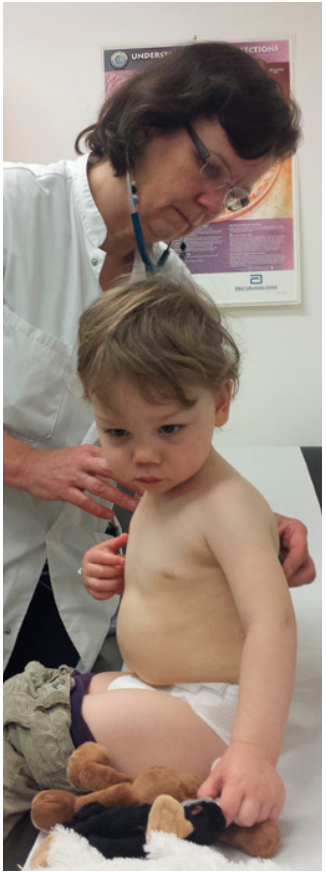
LGD Alliance Europe can help you get in contact with doctors all over Europe who have experience with lymphangiomatosis and Gorham's disease.

PATIENT REGISTRY

Without patients, there would be no information and no scientific research. That's why an international patient registry was established with medical information, family history and other important data. Our hope is that the collected information in the registry will lead to a better diagnosis and treatment for patients to have a better quality of life. Visit www.lgdaregistry.org to sign up. We are happy to help you fill in the registry.

COMMUNITY

Being confronted with a rare disease, it seems like no one understands you. That's why we have two Facebook groups: one for (parents of minor) patients and one for their family and others. Besides this we can also match you with other patients and their family.



CONTACT

Please contact us with questions or requests for assistance. European patients can always reach us via email: patients@lgda.eu

More information on the LGD Alliance Europe and its local partners can be found on our website: www.lgda.eu

SUPPORT US

The local partners of the LGD Alliance Europe are registered charities. The vital support we give to patients, their families and scientific research would not be possible without our generous supporters. Find out how you can help as an individual, a trust or an organisation on our website!

PARTNERS

The LGD Alliance Europe is locally represented by the Alfie Milne Trust in the UK, LGD Alliance Nederland and LGD Alliance Belgium and several patient advocates in other EU countries.

Furthermore, work on Lymphangiomatosis and Gorham's Disease is shared with other organisations like HEVAS, CMTC-OVM, VSOP, RADIORG, Eurordis, the European Commission on rare diseases and the LGD Alliance in the United States.

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Lymphangiomatosis & Gorham's Disease
Nederland

Alfie's Trust
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