



Welcome to Gaucher Connection eNews

Hello

Thank you for signing up for Gaucher Connection eNews, the monthly newsletter designed to help keep you up-to-date and on track with Gaucher disease treatment and management.

You already know that Gaucher disease can cause serious bone problems. And, if you ever experienced bone pain or other skeletal symptoms of Gaucher disease, you know what a serious effect they can have on your ability to stay active and live life to the fullest. In this issue we take a closer look at exactly how and why Gaucher affects bone health with a quick refresher of the science behind the symptoms. We also check in with Joseph and his daughter to hear how regular treatments contribute to their active and rewarding life.

Bone has two major components:

- A strong, hard outer layer made of minerals like calcium
- A spongy inner layer called marrow

Gaucher's bone symptoms are caused by Gaucher cells building up inside the marrow. This can prevent the entire bone from developing properly, and can also cut off the normal blood and oxygen supply, causing bone tissue to weaken and even die.

Effects of Gaucher Disease

Without treatment, Gaucher cells continue to build up and can cause many bone problems:

- **Pain**, which can vary from general dull and achey to sharp and focused
- **Bone crisis**, when blood flow is cut off, causing severe pain that may last from a few hours to several days
- **Osteoporosis**, when bones become porous and can break easily—even from simple everyday activity
- **Poor development**, so bones may not grow to their proper size or shape, and may require surgery

Who's Affected?

Bone problems are very common. In fact, according to the Gaucher Registry, about 83% of people with Gaucher show evidence of bone symptoms¹—even if they're feeling fine. That's because bone changes can start out small, develop slowly, and may initially only be detectable with tests like MRIs.

But seemingly mild symptoms can eventually become very serious—sometimes quite suddenly.² They can cause pain and physical disabilities that make day-to-day activities hard to do. Left unchecked, they can progress beyond the help of treatment.

Prevention Is Key

There's no predicting what bone symptoms might arise from your Gaucher disease. The good news is that enzyme replacement therapy can reverse some symptoms. However, it can't reverse the most serious ones—so prevention is critical.

That means staying on steady treatment to stop problems from developing, and having regular tests to monitor bone health. Other emails in this series offer more information and tips on maintaining a strong and healthy skeleton.

Take Action

At your next doctor visit, ask to review a recent MRI for an up-close look at your own bone health.

Learn more about bone symptoms at the Gaucher Connection website. [Go >>](#)

Did You Know?

83% of people with Gaucher have evidence of bone problems.

Patient spotlight: Joseph

- 37-year-old Accountant
- Father of Amy, newly diagnosed
- Trusts his treatment for himself, and his daughter



Read Joseph's story about how Gaucher treatments let him live an active life. [Go >>](#)

1. Gaucher Registry Annual Report. Genzyme, a Sanofi Company 2013.

2. Pastores GM, Einhorn TA. Skeletal complications of Gaucher disease: pathophysiology, evaluation, and treatment. *Semin Hematol.* 1995;32(Suppl 1):20-27.

Patient Stories are examples based on aggregate medical data. The characters represented are not actual Gaucher patients. Their stories are inspired by those of real Gaucher patients.

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