



Welcome to Gaucher Connection eNews

Hello!

If you ever experienced bone pain or the 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. Let's take a closer look at exactly how and why.

Anatomy of a Bone

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.¹

Effects of Gaucher Disease²

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

- **Pain**, which can vary from general dull and achy 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 88% of people with the disease show evidence of bone symptoms—even if they're feeling fine.³ That's because bone changes can start out small and develop slowly and may initially only be detectable with tests like MRIs.

Seemingly mild symptoms can eventually become very serious—sometimes quite suddenly. They can cause pain and physical disabilities that make it hard to do day-to-day activities. If 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 treatment therapy may reverse some bone symptoms. However, it can't reverse the most serious ones—so preventing them is critical.⁴

That means staying steadily on treatment to stop problems from developing, and having regular tests to monitor your 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 and the Seven Tests at the Gaucher Connection website. [Go >>](#)

Patient spotlight: Elizabeth

- 23-year-old schoolteacher
- Living with Gaucher
- Mastered a tough course load at university
- Maintained full control of her treatment success



Read how Elizabeth successfully balanced a full university schedule with her Gaucher treatments. [Go >>](#)

¹ Niederau, C et al., 2009. Gaucher Disease - 2nd Ed. UNI-MED Science. ISBN 978-3-8374-1165-2, page 30.

² Niederau, C et al., 2009. Gaucher Disease - 2nd Ed. UNI-MED Science. ISBN 978-3-8374-1165-2, page 37.

³ Gaucher Registry Annual Report 2010.

⁴ Niederau, C et al., 2009. Gaucher Disease - 2nd Ed. UNI-MED Science. ISBN 978-3-8374-1165-2, page 42.

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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.