



## Welcome to Gaucher Connection eNews

### Hello!

By now you are probably familiar with the signs and symptoms associated with Gaucher disease. But do you know what causes them? For the April issue we spoke with Diana Bolano Del Vecchio, the nurse coordinator for the Quebec Gaucher Program at Montreal's Sainte-Justine University Hospital Centre, to find out what might be behind some of the symptoms. According to Ms. Bolano Del Vecchio, it's important to keep your health care team informed of any changes to the following symptoms:

### Pain in Bones or Joints

Bone pain is the most common symptom experienced by people already diagnosed with Gaucher disease, whether they are being actively treated or simply monitored. The pain is related to the accumulation of Gaucher cells in the bone and is usually felt in the big bones like the thigh or the shin. Joint pain associated with Gaucher disease is less common; adults with joint pain are usually experiencing arthritic pain as they grow older. In all cases, however, pain in the bone and joints can be a sign of changes in the Gaucher disease.

### Bruising and Bleeding

Most people experience nosebleeds from time to time, and we have all had bruises. Frequent nosebleeds, easy bruising, or cuts that take a long time to clot, on the other hand, may signal a low platelet count. Platelets – tiny cell fragments in the blood – are an important element in the formation of blood clots and too few platelets can lead to excessive bleeding, so any changes in bruising or bleeding could be a sign that treatment changes are needed.

### Fatigue

While fatigue might be caused by anything from the onset of a cold to a poor night's sleep, in people with Gaucher disease, long-term increases in fatigue might also be associated with low hemoglobin levels. Low levels of hemoglobin – the protein in red blood cells that transports oxygen throughout the body – occur when the disease leads to an accumulation of Gaucher cells in the bone marrow where red blood cells are produced.

### Loss of Appetite

Reduced appetite is usually a symptom seen prior to the diagnosis of Gaucher disease. It is caused by increases in the size of the spleen and liver that physically limit the amount of space available for food. When people are being treated for their Gaucher disease, changes in appetite are not often seen. People monitoring their Gaucher disease but not undergoing active treatment, on the other hand, usually notice increased pain before diminished appetite if their disease starts to progress.

### Swelling in the Abdomen

Before treatment options were available for people with Gaucher disease, swelling in the abdomen was a common symptom as people's livers and spleens grew to many times their normal size, says Ms. Bolano Del Vecchio. Today, this is far less common in people who have been diagnosed. Enzyme replacement therapy reduces the swelling of these organs in people being actively treated, while those being monitored often have milder forms of Gaucher disease that do not lead to extreme swelling. In addition, changes in these organs occur slowly over time - the size of your liver and spleen is monitored so any changes should be noticed.

### Monitoring Changes

While some changes in your health status may be related to Gaucher disease and signal changes in it, others may be due to aging or a non-Gaucher illness. Keep your health care team informed so your treatment can be adjusted as necessary, and ensure you have access to a regular pediatrician or family doctor to address other health concerns.

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

### Patient spotlight: Linda

- Mother & Grandmother
- Living with Gaucher & Rheumatoid Arthritis
- Pushing Her Luck
- Thankful for the opportunity to grow old



Read how Linda manages her Gaucher at the same time as another chronic illness, and never feels unlucky. [Go >>](#)

Membership in the Gaucher Connection Program you signed up for includes these complimentary eNewsletters. If you do not wish to receive emails and updates from Gaucher Connection in the future, please [unsubscribe](#).

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.

Review our [Privacy Policy](#).