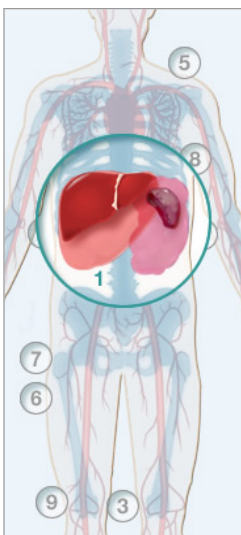


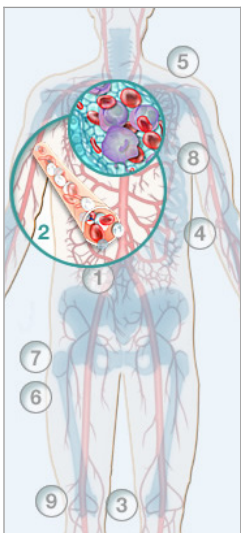
## Type 1 Gaucher Disease Signs & Symptoms

Gaucher disease typically affects the body in many ways, as a result of Gaucher cells accumulating in various organs (especially the liver and spleen) and in the bone marrow (leading to many skeletal complications). The disease's bone-related symptoms can be particularly painful and debilitating, impairing a patient's mobility. Children and adolescents with Gaucher disease may experience a delay in growth and development.



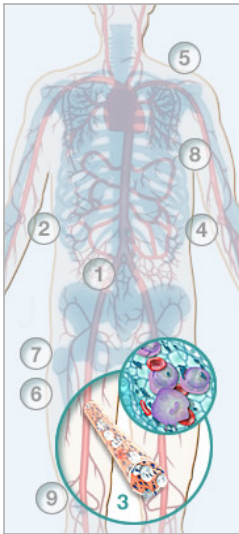
### 1: Enlarged liver and/or spleen

Gaucher patients may have protruding abdomens, caused by the liver and/or spleen swelling to unusual sizes because of accumulated Gaucher cells. The spleen can swell to 5-75 times its normal size, while the liver to 1.25-2.5 times its normal size. These symptoms can have many other effects on patients, including suppressing the appetite (because the enlarged organs press on the stomach, creating a full feeling after eating very little), blood-related problems related to an overactive spleen, more serious liver disease, and low self-esteem related to appearance.



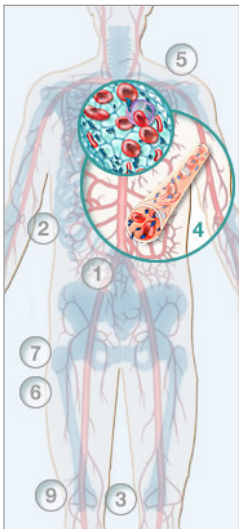
### 2: Low blood platelet count

Platelets are the blood cells responsible for allowing blood to clot; they are formed in bone marrow and then released into the blood. A build-up of Gaucher cells in bone marrow may cause fewer blood platelets to be produced. In addition, an enlarged, overactive spleen (caused by accumulated Gaucher cells) may break down blood cells faster than they are produced, thus also contributing to an overall lower platelet count. As a result, Gaucher patients' blood may not clot well, and they may experience excessive bruising and bleeding, such as frequent nosebleeds, bleeding gums, and longer, heavier menstrual periods.



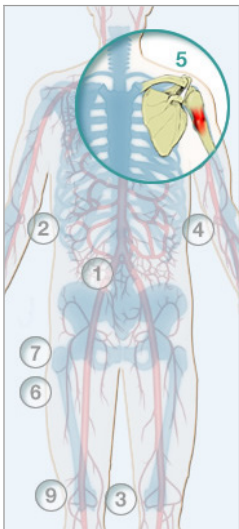
### 3: Low red blood cell count (anemia)

Red blood cells are responsible for carrying oxygen to all cells in the body. The spleen is responsible for breaking down these cells, but when it is enlarged as a result of accumulated Gaucher cells, it may become overactive and break down blood cells faster than they are produced. The resulting red blood cell deficiency is called anemia, and it causes people to feel fatigued because the body is not getting enough oxygen. While anemia is often responsible for fatigue and low stamina in Gaucher patients, these symptoms may also result from a higher-than-normal metabolism found in many suffering from Gaucher.



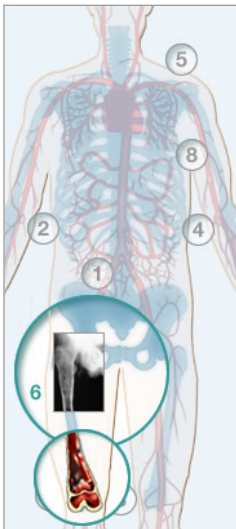
### 4: Low white blood cell count

White blood cells are responsible for helping fight infection, such as when bacteria or viruses enter the body. The spleen is responsible for filtering these cells, but when it is enlarged as a result of accumulated Gaucher cells, it may become overactive and filter white blood cells faster than usual. The resulting white blood cell deficiency makes it harder for the body to fend off infection, so Gaucher patients may get sick more frequently than other people.



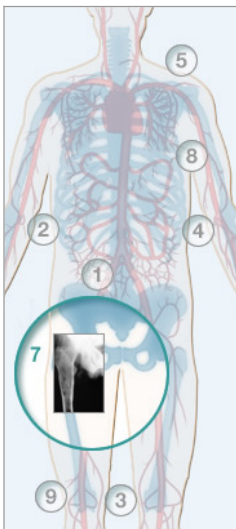
### 5: Bone crisis

Gaucher patients may experience severe bone pain, called “bone crisis,” caused by insufficient blood circulation to the bone due to interference from Gaucher cells and by the local release of chemical factors. The pain is intense, often accompanied by fever, and can last from a few hours to a few days or even weeks, usually rendering patients bedridden during this time.



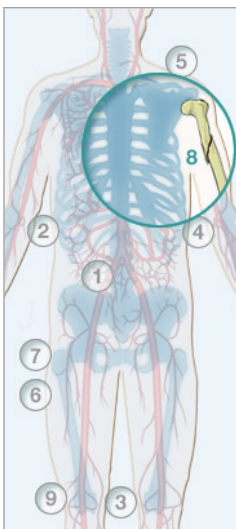
### 6: Bone tissue death

As Gaucher cells accumulate in bone marrow, they can restrict normal blood flow—sometimes to the point that bone tissue dies. This bone destruction causes severe pain and can lead to fractures and joint collapse.



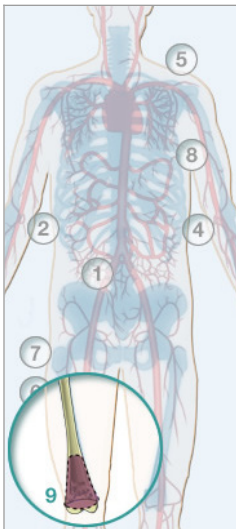
### 7: Bone thinning

Gaucher disease causes reduced mass and density of bone tissue, causing it to thin and weaken and thus be more susceptible to fractures.



### 8: Pathological fracture

Pathological fractures are broken bones caused by disease rather than by trauma (such as impact from a fall or accident). The accumulation of Gaucher cells in bone marrow can weaken bones in various ways (see bone crisis, bone tissue death, and bone thinning on this diagram), making them susceptible to such fractures simply from normal activity.



### 9: Erlenmeyer flask deformity

Gaucher disease includes abnormalities in the way bones develop, or remodel, resulting in irregular shapes. The most common among Gaucher patients is the Erlenmeyer flask deformity (named after the type of laboratory flask whose shape it resembles), in which the ends of the bone (most commonly the femur and tibia leg bones) develop a flared, flattened shape rather than the normal rounded form.

### Other possible signs and symptoms

#### Delayed growth and development

Children with untreated Gaucher disease may experience a temporary delay in growth and development compared with their peers. In addition, puberty may begin later than normal.

#### Loss of appetite

Patients with Gaucher disease may experience a feeling of fullness after eating a small meal. This typically occurs if an enlarged spleen or liver puts pressure on the stomach.

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If you or a loved one has one or more of these signs of symptoms, talk to your doctor about further evaluation.

For more information, visit [Genzyme.com](http://Genzyme.com).