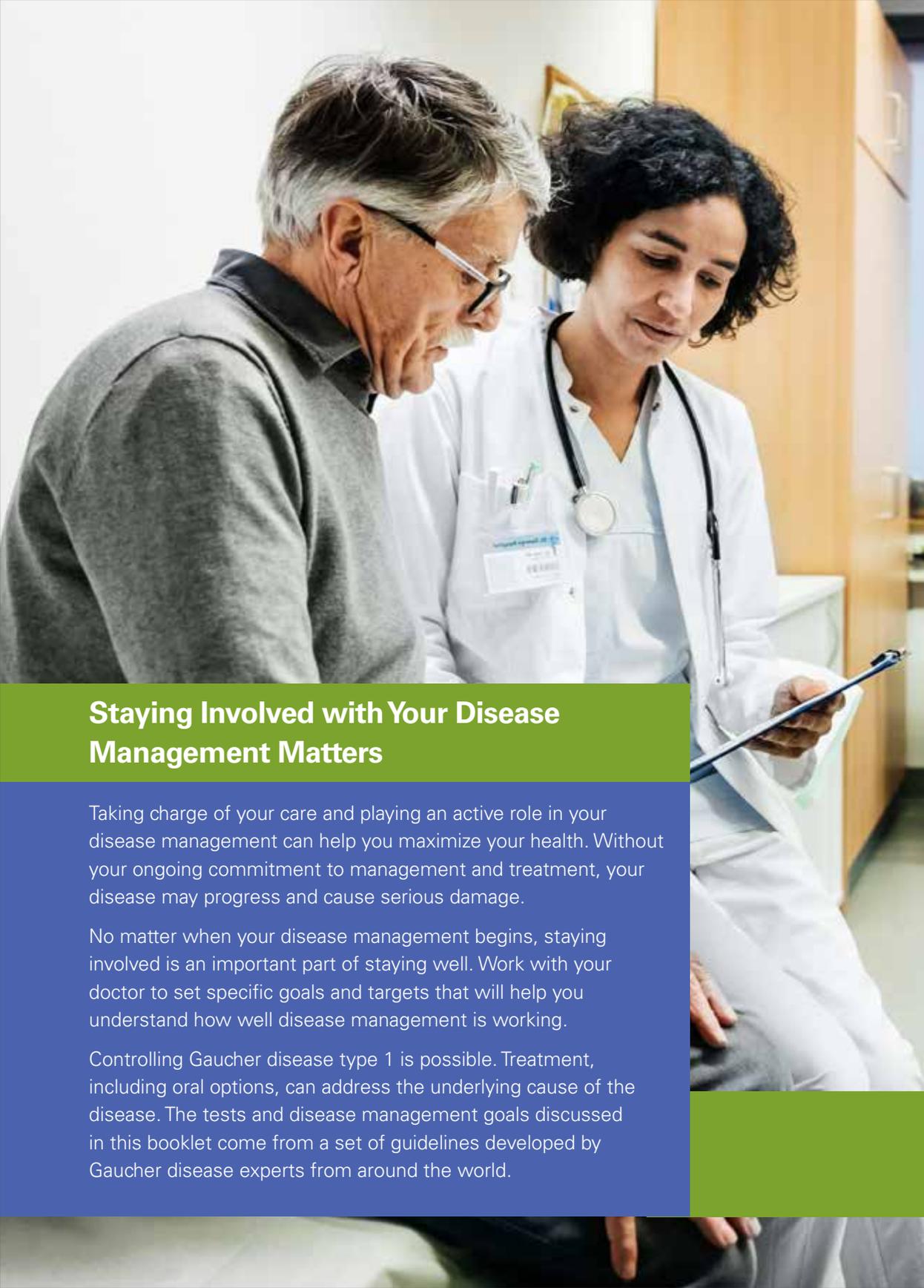
A middle-aged man with short, graying hair is standing outdoors in a park-like setting. He is wearing a light blue, long-sleeved button-down shirt and blue jeans. He is looking directly at the camera with a slight smile. The background is a blurred green landscape with trees and a path.

# Disease Management Starts with You

Understanding your part in disease management,  
monitoring, and testing for Gaucher disease type 1

SANOFI GENZYME 



# A Lifelong Commitment for a Lifelong Condition

Gaucher (pronounced go-shay) disease is a rare, progressive, inherited, genetic condition that causes a fatty substance, called glucosylceramide (gloo-ko-sil-sara-mide, also called GL-1), to build up in the spleen, liver, and bone. As GL-1 builds up, people with Gaucher disease type 1 may experience excessive bruising and bleeding, as well as protruding abdomens caused by swelling of the liver and/or spleen.

Typically, an enzyme called acid glucocerebrosidase (pronounced gloo-ko-ser-e-brosy-daze) breaks down GL-1. People with Gaucher disease may not have enough of this enzyme to keep up with the production of GL-1, or may be missing this enzyme entirely. This causes GL-1 to build up in certain cells, resulting in the signs and symptoms of Gaucher disease.

Gaucher disease type 1 can be effectively managed once a diagnosis is made.

## Disease management, goals, and monitoring can help control Gaucher disease type 1

As a progressive condition, Gaucher disease can get worse with time. That's why it's important to get the most from your disease management by sticking with the treatment regimen as prescribed by your doctor. Talk with your doctor about:



**Setting goals**—your doctor may determine your individual goals based on your history, experience with many patients, or disease management guidelines



**Monitoring your progress**—a series of tests and exams can check the status of your disease, the effectiveness of the disease management plan, and whether or not you are on track to meet your goals

You should have a comprehensive evaluation at least once a year. Your doctor will decide which assessments you need and how frequently you need these tests.

## Staying Involved with Your Disease Management Matters

Taking charge of your care and playing an active role in your disease management can help you maximize your health. Without your ongoing commitment to management and treatment, your disease may progress and cause serious damage.

No matter when your disease management begins, staying involved is an important part of staying well. Work with your doctor to set specific goals and targets that will help you understand how well disease management is working.

Controlling Gaucher disease type 1 is possible. Treatment, including oral options, can address the underlying cause of the disease. The tests and disease management goals discussed in this booklet come from a set of guidelines developed by Gaucher disease experts from around the world.

## Inside This Guide

Learn about the seven tests for monitoring Gaucher disease type 1, why they are important, and how they can help you take charge of your disease management.



## Enlarged organs can cause many complications in Gaucher disease type 1

The accumulation of GL-1 can cause your spleen and liver to become enlarged:

- The spleen can swell to **15 times its normal size**, causing the abdomen to protrude and making people **look overweight or pregnant**
- Increased bleeding, such as nosebleeds, bleeding gums, or heavy menstrual periods can result from the increased activity of an enlarged spleen
- An enlarged spleen can also make a person feel full after eating only small amounts of food
- The liver can also enlarge up to **2½ times its normal size**. The consequences of this are **scarring** (fibrosis) of the liver and the progression of other types of liver dysfunction

Your healthcare provider may recommend tests to help determine if Gaucher disease is affecting your liver or spleen. To best assess your liver and spleen, a physical exam should be combined with these tests.

### Disease Management Goals for the Liver and Spleen

- Reduce and maintain liver volume by 1 to 1½ times its normal size
- Reduce and maintain spleen volume by 2 to 8 times its normal size
- Lessen symptoms, such as abdominal pain and feeling full and bloated

The size of the liver and spleen may not decrease to normal in patients with very enlarged organs due to scarring.

Pain and swelling in your abdomen may be the first symptoms you notice. However, the disease may also be affecting your bones and blood. Talk to your doctor about your symptoms and testing needs.

# Tests to Assess Organ Enlargement



## MRI or CT

An MRI (magnetic resonance imaging) or CT (computerized tomography) scan can be used to produce pictures of the abdomen. **They can show the size and structure of the liver and spleen.**

## Taking an Active Part in Your Care

Regularly scheduled assessments with your doctor are important in managing a chronic, progressive condition like Gaucher disease type 1.

A published Schedule of Assessments is available to your doctor, and includes recommendations on how often each test should be conducted.

Here are some tips for taking an active part in your care:

- Follow your regimen as prescribed by your doctor
- Be involved in setting goals
- Consider asking your doctor which of the tests in this booklet may be of benefit to you and how often you should have them. You should also ask your doctor to explain the risks associated with these tests, which are not covered in this guide

**Take charge! Living with Gaucher disease can be challenging, but the more you understand about the condition and how it affects your body, the more control you'll have over your health.**



## Bone problems are one of the most common complications of Gaucher disease type 1

Nearly all people with Gaucher disease have some issues with their bones. Even if you feel healthy, the damaging effects of Gaucher disease on your bones can be taking place. Without proper management, Gaucher cells can build up in your bone marrow and damage bones in several ways.

There may not be any notable signs or symptoms. However, **the disease can weaken bones and joints**. This can cause **extreme pain** and **problems moving and functioning**. The illness can also cause bones to break more easily.

Bone problems can become permanent and lead to disability, such as spinal compression, or they may require surgery, like hip and other joint replacements.

This makes it important for you to have thorough tests of your bones at least once a year.

### Disease Management Goals for Bone Disease

- Reduce bone pain
- Prevent bone crises (severe pain, usually with fever, caused by a sudden lack of normal blood flow and oxygen to affected bone)
- Patients with advanced bone disease may need additional management, such as physical therapy, orthopedic care, and medication

Finding and managing bone problems early may help you live with less pain. Talk with your doctor about your individual goals and testing needs.

# Tests to Assess Bone Disease



## X-Ray

An x-ray **detects fractures and advanced bone problems**.

It can help identify changes in bone such as Erlenmeyer flask deformity. However, it is not the best way to assess changes in the bone marrow, strength of bones, or early signs of bone disease.



## MRI

MRI (magnetic resonance imaging) is **a sensitive tool for ongoing monitoring of bone**.

It uses magnets and radio waves to create images to assess the buildup of Gaucher cells in the bone marrow and to what extent they may have caused changes in the bone.



## DEXA

A DEXA (dual-energy x-ray absorptiometry) scan is **the "gold standard" for measuring bone mineral density (BMD)**.

It is used to diagnose bone loss and assess your risk for developing bone fractures.

## TIP

Talk with your doctor about having the above tests. Having all three will offer the most complete information about the health of your bones. Tests should be performed at medical centers with healthcare providers experienced in Gaucher disease. For help finding a healthcare provider or center near you, call Sanofi Genzyme Support Services at 1-800-745-4447 (option 3), or visit [GenzymeSupportServices.com](https://www.genzymsupportservices.com).

## Gaucher disease can affect your blood in many ways

Blood cells are formed in your bone marrow. Because Gaucher cells can build up in the bone marrow, they can interfere with the production of different types of blood cells.

- **Low Red Blood Count**—In Gaucher disease, the spleen becomes enlarged and overactive, breaking down too many red blood cells. This leads to anemia (uh-NEEM-ee-yah). Anemia can make you feel fatigued. People often describe feeling **tired or weak**, being **breathless**, or **lacking energy**.
- **Reduced Platelets in the Blood**—An overactive spleen can reduce blood platelets. This can make it harder for your blood to clot. For this reason, you might **bruise and bleed easily**, including heavy nosebleeds, bleeding gums, and heavier menstrual periods.
- **Low White Blood Cell Count**—An overactive spleen may decrease the number of white blood cells. These cells normally help your body fight infection. **In general, people with low white blood cell counts may experience more infections and become sick more frequently.**

## Disease Management Goals for Blood

- Increase hemoglobin levels to:
  - 11 grams per deciliter or higher for women and children
  - 12 grams per deciliter or higher for men
- Eliminate the need for blood transfusion
- Reduce fatigue related to anemia
- Maintain platelet counts to reduce or prevent bleeding problems

Having regular blood tests can help your doctor find and treat problems early. Talk with your doctor about your individual goals and testing needs.

# Tests to Assess Blood Levels



## Hemoglobin Test

This is a blood test to measure the total amount of hemoglobin in your blood. Hemoglobin is a part of red blood cells that carries oxygen.

**Low hemoglobin levels can be an indicator of anemia, which can lead to fatigue and other problems.**



## Platelet Count

This is a blood test to measure the number of platelets in your blood.

Platelets are needed for normal blood clotting. **Low platelet count (thrombocytopenia) may cause bruising and bleeding.**



## Biochemical Evaluations

These are special blood tests to check substances called biomarkers that **can monitor disease progression and can help check your progress** toward achieving the goals of your disease management plan.

## TIP

Ask when your test results will come back from the lab, and mark your calendar to remember the date. Consider calling to talk about your results. Some questions you may want to ask during the call include:

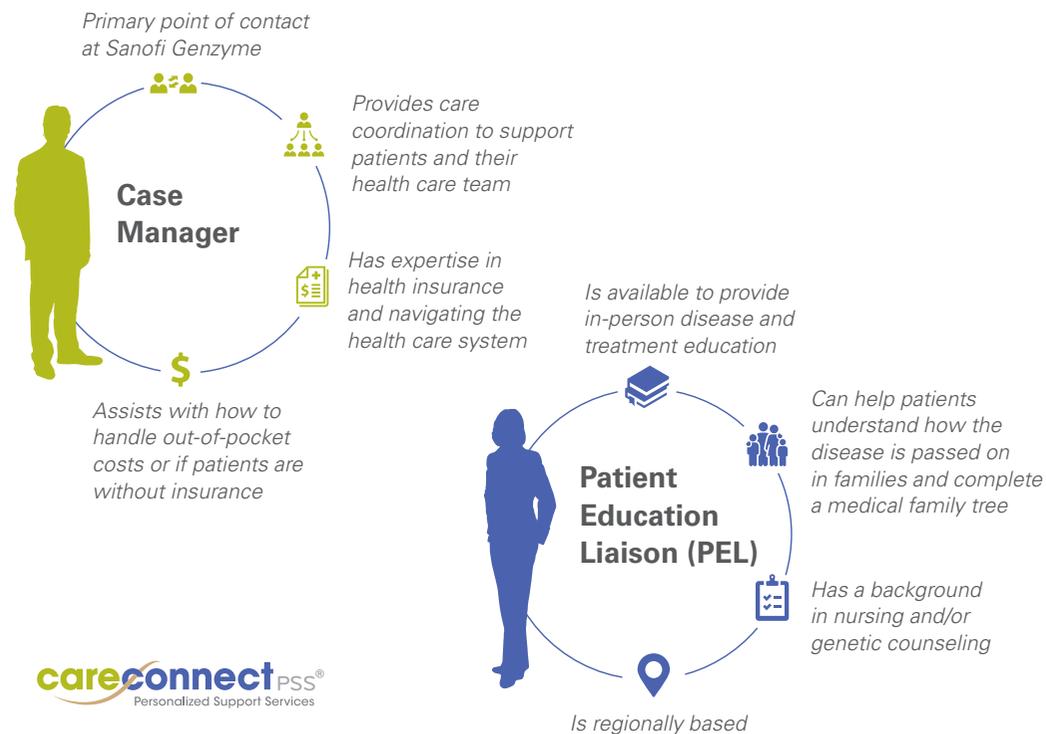
- How are these results different from past results?
- What do these results mean for my disease management?
- How close am I to my goals?
- Does my regimen need to be adjusted?

# Available Resources and Services

## Sanofi Genzyme Support Services

### CareConnectPSS®: Personalized Support Services for rare disease

CareConnectPSS represents Sanofi Genzyme's more than 25-year commitment to supporting the rare disease community. CareConnectPSS is designed to support each patient's unique journey through dedicated Case Managers and Patient Education Liaisons (also called "PELs") who provide one-on-one assistance through a range of resources, programs, and other support offerings. Whether you are newly diagnosed or already receiving treatment, your CareConnectPSS team will work closely with you and your health care providers to give you the confidential and personalized support you need.



Contact a CareConnectPSS Case Manager or Patient Education Liaison by visiting [CareConnectPSS.com](http://CareConnectPSS.com) or call 1-800-745-4447, Option 3

# Taking Charge of Your Well-Being

The consequences of Gaucher disease can affect many aspects of your life. You may find it difficult or painful to do normal activities. You may still feel tired after a full night's sleep. You may also find it difficult to cope with changes to the way you look.

Be proactive about addressing these issues. Your healthcare team needs to know how the disease is affecting your well-being. Have open and honest conversations with your doctor or nurse about changes in the quality of your daily life.

Your healthcare provider may ask you questions in an informal way or give you a paper questionnaire, called an SF-36 Health Survey, to complete. Some of the questions and possible responses include:

- How would you rate your health today? (excellent, good, fair, poor)
- How does your health today compare with your health last year? (same, better, worse)
- How does your health affect your ability to do certain activities? (limited a lot, a little, not at all)

Your answers can help your medical team discuss the impact of the disease with you and suggest strategies to help you cope. If you have any concerns, make a list of your questions to discuss with members of your healthcare team.



Work with your care team to optimize your disease management. Your doctor will decide which tests you need and their frequency based on your individual symptoms and disease management regimen.

## A LONG-STANDING COMMITMENT TO THE GAUCHER COMMUNITY

For more than 30 years, Sanofi Genzyme has partnered with the Gaucher disease community to help address the needs of people living with Gaucher disease and those who care for them. With comprehensive disease awareness and diagnostic efforts, patient services, and innovative therapies, we are leading the advancement of Gaucher care.

**Learn more about Gaucher disease type 1 at [www.gauchercare.com](http://www.gauchercare.com).**

**SANOFI GENZYME** 

50 Binney Street  
Cambridge, MA 02142 U.S.A.

©2019 Genzyme Corporation. All rights reserved. CareConnectPSS, Sanofi, and Genzyme are registered in the U.S Patent and Trademark Office. SAUS.GD.1702.0376(2)