



## Welcome to Gaucher Connection eNews

### Hello !

You know those seven tests that are so critical for monitoring Gaucher disease? MRI, DEXA, hemoglobin... the scientific terms can be very puzzling! In this issue we provide you with a convenient, quick reference guide by walking through exactly what these terms mean, and what each test does:

### Guide to the 7 Tests

You'll notice that while there are seven tests, only six are listed below. That's because MRI counts twice—it's used for testing both the bones AND the liver and spleen.

#### X-Ray

X-rays use **electromagnetic radiation** to penetrate the body and capture images. They're best for analyzing a bone's shape to check for deformities or breaks (although they can't "see through" bones). A special 3-dimensional x-ray technique called a **computed tomography (CT)** is also sometimes used to analyze the liver and spleen.

#### MRI

Using magnets and radio waves, MRI (which stands for **magnetic resonance imaging**) can in fact "see through" bones, making it extremely useful for seeing how much Gaucher cells have built up inside bone marrow. It's also used for checking the size and structure of the liver and spleen.

#### DEXA

DEXA stands for **dual-energy x-ray absorptiometry**. This technology uses two x-ray beams to scan the hard outer part of bones. Based on how the beams are absorbed, the DEXA scan calculates the bone's density to reveal how strong it is.

#### Hemoglobin Test

From a small blood sample, this test measures the amount of hemoglobin, the substance in red blood cells that carries oxygen throughout the body. Low hemoglobin is a sign of **anemia**, meaning the body isn't getting enough oxygen, causing feelings of fatigue, weakness, and/or breathlessness.

#### Platelet Count

Also using a small blood sample, this test measures the number of platelets, the blood cells responsible for **clotting** blood. A low platelet count means blood can't clot properly, causing the body to bruise and/or bleed easily.

### Biochemical Evaluations

Doctors may also analyze a blood sample for certain chemicals that occur naturally in the body. Changes in the activity of these chemicals can indicate changes in health or how the body is responding to treatment.

The enzyme **chitotriosidase** is most commonly evaluated, because it is a particularly sensitive indicator of how well treatment is working<sup>1-2</sup> in many patients. Other enzymes doctors may analyze include angiotensin-converting enzyme (ACE) and tartrate-resistant acid phosphatase (TRAP).

### Take Action

If you have further questions about how these tests work or what they do, be sure to ask your health care providers.

For help keeping track of test results, try this personal treatment tracker, a useful form for recording and reviewing details. Download the [doctor visit tracker](#).

Learn more about the seven tests essential for monitoring Gaucher at the Gaucher Connection website. [Go >>](#)

### Further Reading

Genzyme's "7 Tests" brochure has more information on how these tests are used to measure progress toward treatment goals. Download it [here](#).

### Patient spotlight: Sarah

- 36-year-old pastry chef
- Mother of a teenager with Gaucher



- Works towards her dreams of opening a bakery
- Helps her son achieve his own dreams



**Read how Sarah stays supportive with her teenage son about his Gaucher. [Go >>](#)**

<sup>1</sup> Czartoryska B, Tylki-Szymanska A, Lugowska A: Changes in serum chitotriosidase activity with cessation of replacement enzyme (cerebrosidase) administration in Gaucher disease. Clin Biochem 33:147-149, 2000.

<sup>2</sup> Hollak CE, van Weely S, van Oers MH, Aerts JM: Marked elevation of plasma chitotriosidase activity. A novel hallmark of Gaucher disease. J Clin Invest 93:1288-1292, 1994.

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