

Gaucher Disease: Setting goals to help maximize your health

**7 TESTS:
To help you get the most from your treatment**

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- ◆ **Important to have specific goals and targets to help you and your doctor know how well the treatment is working**
- ◆ **Tests and treatment goals come from a set of guidelines developed by Gaucher disease experts**
- ◆ **Playing an active role in your treatment can help you maximize your health**

Treatment, goals, and monitoring

Setting treatment goals

- ◆ Individual treatment goals based on experience with many patients and treatment guidelines

Monitoring your progress

- ◆ Series of *7 tests* to check whether you are on track to meet your treatment goals
- ◆ Good check to confirm status of your disease and effectiveness of treatment plan
- ◆ Timing: comprehensive monitoring tests at least once a year

Checking your bones

Treatment goals for bone disease

- ◆ **Lessen or eliminate bone pain**
- ◆ **Prevent bone crises**
 - ◆ Severe pain, usually with fever, caused by a sudden lack of normal blood flow and oxygen to affected bone
- ◆ **Treatment response is patient dependent**
 - ◆ Patients with advanced bone disease may require additional treatments
 - ◆ E.g. physical therapy, orthopedic care etc.
- ◆ **Patients should talk with their doctor about individual treatment goals**

Checking your bones – TESTS 1, 2 and 3

TEST 1 – X-ray

- ◆ **Detects fractures and late bone problems**
- ◆ **Limited in assessing changes in:**
 - ◆ Bone marrow
 - ◆ Strength of bones
 - ◆ Early signs of bone disease

Checking your bones – TESTS 1, 2 and 3

TEST 2 – Magnetic resonance imaging (MRI)

- ◆ **Uses magnets and radio waves to make pictures of parts of the body**
- ◆ **Most helpful test to assess:**
 - ◆ Extent of Gaucher cells buildup in the bone marrow
 - ◆ Extent of changes in the bone due to this buildup
- ◆ **Powerful and sensitive tool for ongoing monitoring of bone**
- ◆ **Does not involve use of X-rays**

Checking your bones – TESTS 1, 2 and 3

TEST 3 – Dual Energy X-ray Absorptiometry (DEXA)

- ◆ Buildup of Gaucher cells in bones can make the bones thinner, weaker than normal, and prone to fracture
- ◆ DEXA scan measure bone mineral density (BMD)
 - ◆ Measure of bone strength

Checking your bones – TESTS 1, 2 and 3

Take action TIP

- ◆ Important to have all 3 tests – x-ray, MRI and DEXA
- ◆ Offers most complete information about bone health
- ◆ Tests should be performed at medical centers with healthcare providers experienced in Gaucher disease
- ◆ To find a healthcare provider or treatment centre visit:

www.gaucherconnection.ca

Click “Treatment Centres”

Checking your blood

Treatment goals for blood

- ◆ Monitor hemoglobin levels
- ◆ Eliminate the need for blood transfusion
- ◆ Reduce fatigue related to anemia
- ◆ Maintain platelet counts to reduce or prevent bleeding problems
- ◆ Remember patients should talk with their doctor about individual treatment goals

Checking your blood – TESTS 4, 5 and 6

TEST 4 – Hemoglobin test

- ◆ Measures the total amount of hemoglobin in the blood
- ◆ Hemoglobin – part of red blood cells and carries oxygen
- ◆ Low hemoglobin level is a sign of anemia

Checking your blood – TESTS 4, 5 and 6

TEST 5 – Platelet count

- ◆ Measures the number of platelets in the blood
- ◆ Platelets are needed for clotting blood
- ◆ Low platelet count may be cause bruising and bleeding

Checking your blood – TESTS 4, 5 and 6

TEST 6 – Biochemical evaluations

- ◆ **Special blood tests that can pick up subtle changes in the disease**
 - ◆ Useful for some patients

- ◆ **Help check progress toward achieving goals of the treatment plan**

- ◆ **Some biochemical markers include:**
 - ◆ Chitotriosidase (*Kl-toe-try-O-sih-days*)
 - ◆ Angiotensin-converting enzyme (*ACE*)
 - ◆ Tartrate-resistant acid phosphatase (*TRAP*)

Checking your blood – TESTS 4, 5 and 6

Take action TIP

- ◆ All 3 tests equally important
- ◆ Offers most complete information about blood health
- ◆ Tests should be performed at medical centers with healthcare providers experienced in Gaucher disease
- ◆ To find a healthcare provider or treatment centre visit:

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Click “Treatment Centres”

Checking your liver and spleen

Treatment goals for the liver and spleen

- ◆ **Reduce and maintain liver volume to 1 to 1.5 times its normal size**
- ◆ **Reduce and maintain spleen volume to 2 to 8 times its normal size**
- ◆ **Lessen symptoms, such as abdominal pain, feeling full and bloated**
- ◆ **Remember patients should talk with their doctor about individual treatment goals**
- ◆ Note – the size of the liver and/or spleen may not decrease to normal in patients with very enlarged organs, because of scarring

Checking your liver and spleen – TEST 7

TEST 7

MRI of the liver and spleen

- ◆ Painless test produces pictures of the inside of the abdomen
- ◆ Shows the size and structure of the spleen and liver

OR

Computed tomography (CT) of the liver and spleen

- ◆ Painless x-ray test shows a cross-sectional picture of liver and spleen
- ◆ 3-D picture allows measurement of these organs

7 TESTS – to help you get the most from your treatment

Take action TIP

Important Questions to Ask

- ◆ **When discussing results:**
 - ◆ How are these results different from past results?
 - ◆ What do these results mean for my treatment?
 - ◆ How close am I to my treatment goals?
 - ◆ Does my treatment need to be adjusted?

Summary

Treatment, goals, and monitoring

Personal Plan of Action – *Stay Involved*

- ◆ **Follow infusion schedule**
- ◆ **Be involved in setting treatment goals**
- ◆ **7 tests completed at least once a year to monitor the disease and assess treatment goals**
- ◆ **Depending on health and disease progression more tests may be required**
- ◆ **For more information visit: www.gaucherconnection.ca**