

ONGOING MONITORING

OF TYPE 1 GAUCHER PATIENTS

DISEASE MONITORING

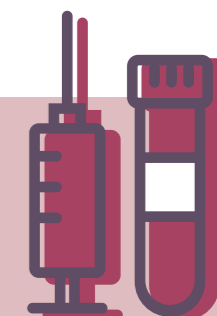
The International Collaborative Gaucher Group (ICGG), a group of physicians with experience in the management of Gaucher disease established in 1991, has developed comprehensive guidelines for the clinical monitoring of adult patients with type 1 Gaucher disease (GD1). The ICGG pooled data from a large observational database registry to create these guidelines for comprehensive evaluation of patients.¹



BLOOD TESTS

Blood tests include hemoglobin count and platelet count. Additionally, the serum levels of several biological markers indicate the severity of GD1, and disease progression^{1,2,3}:

- glucosylsphingosine (**lyso-Gb1/lyso-GL1**)⁴
- chitotriosidase (**CHIT1**)^{1,4}
- chemokine ligand 18 (**CCL-18**)^{1,4}
- tartrate-resistant acid phosphatase (**TRAP**)¹
- angiotensin-converting enzyme (**ACE**)¹



VISCERAL VOLUME

Visceral volumes can be assessed with:

- **volumetric MRI** (since repeat assessment is routine in GD1)^{1,4}
- **CT** or **ultrasound** where MRI is unavailable^{1,4}



SKELETAL SCANS¹

Bone marrow infiltration and bone disease can be assessed with:

- **MRI**, particularly bone infarction and necrosis¹
- **DEXA**, the gold standard method for assessing BMD¹



ACRONYMS

BMD - bone mineral density. **CT** - computed tomography. **DEXA** - dual-energy X-ray absorptiometry. **MRI** - magnetic resonance imaging

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NOT ON ENZYME REPLACEMENT^{1,5}

Assessment	Every 12 months	Every 12–24 months
Comprehensive physical examination and SF-36* survey	○	
Blood tests	○	
Visceral volume		○
Skeletal scans		○

ENZYME REPLACEMENT: NOT ACHIEVED THERAPEUTIC GOALS¹

Assessment	At 3 months	At 6 months	At 9 months	At 12 months
Comprehensive physical examination and SF-36* survey				○
Blood tests	○	○	○	○
Visceral volume				○
Skeletal scans				○

ENZYME REPLACEMENT: ACHIEVED THERAPEUTIC GOALS¹

Assessment	Every 12 months	Every 12–24 months
Comprehensive physical examination and SF-36* survey	○	
Blood tests		○
Visceral volume		○
Skeletal scans		○
Repeat schedule if patient has not achieved therapeutic goals		

*SF-36 - 36-item Short-Form Health Survey

The ICGG was established in 1991 as a group of physicians who are experts in the management of GD1.¹ The ICGG has pooled data from patients in a large observational database registry.¹ The registry is intended to explore and define the natural history of GD1 and characterize patient response to therapy.¹ With the registry information, ICGG has developed a comprehensive evaluation for regular clinical monitoring that is dependent on the circumstances of the patient.¹

REFERENCES

1. Weinreb et al. Gaucher Disease Type 1: Revised Recommendations on Evaluations and Monitoring for Adult Patients. *Semin Hematol.* 2004; 41: 15–22. **2.** Di Rocco et al. A new severity score index for phenotypic classification and evaluation of responses to treatment in type 1 Gaucher disease. *Haematologica* 2008; 93(8): 1211–1218. **3.** Cassinerio et al. Gaucher disease: A diagnostic challenge for internists. *Eur J Int Med* 2014; 25: 117–124. **4.** Stirnemann et al. A Review of Gaucher Disease Pathophysiology, Clinical Presentation and Treatments. *Int J Mol Sci.* 2017 Feb 17; 18(2). pii: E441. **5.** Pastores GM et al. Therapeutic goals in the treatment of Gaucher disease. *Semin Hematol* 2004; 41: 4–14.

