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 SCANS[1]

Bone marrow infiltration and bone disease can be assessed with:

- **MRI**, particularly bone infarction and necrosis[1]
- **DEXA**, the gold standard method for assessing BMD[1]

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 GOALS[1]

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 GOALS[1]

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.