

Minimum Recommendations for Monitoring Patients with Non-Neuronopathic (Type 1) Gaucher Disease

Initial Assessment^{1,2}

Blood Tests	
PRIMARY TESTS	ADDITIONAL TESTS AS INDICATED⁵
Hemoglobin	AST and/or ALT Albumin
Platelet Count	Alkaline Phosphatase Total Protein
Biochemical Markers ³	Calcium Serum Immunoelectrophoresis
• Chitotriosidase	Phosphorus Iron
• ACE	PT Iron Binding Capacity
• Acid Phosphatase, tartrate resistant (TRAP)	PTT Ferritin
Mutation Analysis	WBC Vitamin B ₁₂
Antibody Sample ⁴	Total and Direct Bilirubin
Visceral ⁶	
Spleen Volume (Volumetric MRI or CT)	
Liver Volume (Volumetric MRI or CT)	
Skeletal	
MRI (coronal; T1- & T2-weighted) of entire femora ⁷	
X-ray: AP view of entire femora ⁷ and lateral view of spine	
DEXA: lumbar spine and femoral neck	
Bone Age (for patients age 14 years or less) ⁵	
Pulmonary ⁸	
ECG, Chest X-ray, and Doppler Echocardiogram (right ventricular systolic pressure) for patients >18 years old	
Quality of Life	
Patient-reported functional health and well-being (SF-36 Health Survey)	

Ongoing Monitoring²

	Patients Not on Enzyme Therapy		Patients on Enzyme Therapy		
	Every 12 Months	Every 12-24 Months	Not Achieved Therapeutic Goals	Achieved Therapeutic Goals	At Time of Dose Change or Significant Clinical Complication
Blood Tests					
Hemoglobin	X		X	X	X
Platelet Count	X		X	X	X
Biochemical Markers ³	X		X	X	X
• Chitotriosidase					
• ACE					
• Acid Phosphatase, tartrate resistant (TRAP)					
Visceral ⁶					
Spleen Volume (Volumetric MRI or CT)		X	X	X	X
Liver Volume (Volumetric MRI or CT)		X	X	X	X
Skeletal					
MRI (coronal; T1- & T2-weighted) of entire femora ⁷		X	X	X	X
X-ray: AP view of entire femora ⁷ and lateral view of spine		X	X	X	X
DEXA: lumbar spine and femoral neck		X	X	X	X
Quality of Life					
Patient-reported functional health and well-being (SF-36 Health Survey)	X		X	X	X

1. A complete patient and family history, preferably including a pedigree, should be conducted.
2. A comprehensive physical examination should be performed at least annually.
3. One or more of these biochemical markers should be consistently monitored at least every 12 months and in conjunction with other clinical assessments of disease activity and response to treatment.
Of the three recommended markers, chitotriosidase, when available as a validated procedure from an experienced laboratory, may be the most sensitive indicator of changing disease activity, and is therefore preferred.
4. A baseline sample will be drawn and stored at Genzyme. A subsequent sample is suggested to be drawn at 6 months after starting ERT but is optional. The baseline and additional samples will be tested only if clinically indicated, such as for a suspected immune-mediated adverse event, prior to a switch to home therapy, or for suspected loss of ERT effectiveness.
5. These should be followed appropriately if abnormal based on each patient's age and clinical status.
6. Obtain contiguous transaxial 10 mm thick sections for sum of region of interest.
7. Optimally, obtain hips to below knees.
8. Pulmonary assessments are recommended every 12-24 months for patients with borderline or above normal pulmonary pressures at baseline.

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