Minimum Recommendations for Monitoring Patients With Nonneuronopathic (Type 1) Gaucher Disease

Initial Assessment [1,2]

Blood Tests		
PRIMARY TESTS	**ADDITIONAL TESTS AS INDICATED**	
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
• TRAP	PTT	Ferritin
Mutation analysis	WBC	Vitamin B_{12}
Antibody sample [4]	Total and direct bilirubin	
Visceral [5]		
Spleen volume (volumetric MRI or CT)		
Liver volume (volumetric MRI or CT)		
Skeletal		
MRI (coronal; T_1- and T_2-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 aged ≥14 years) [5]		
Pulmonary [8]		
ECG, chest X-ray, and Doppler echocardiogram (right ventricular systolic pressure) for patients aged >18 years		
Quality of Life		
Patient-reported functional health and well-being (SF-36 Health Survey)		

Ongoing Monitoring [2]

	Patients Not on Enzyme Therapy		Patients on Enzyme Therapy			
			Not Achieved Therapeutic Goals		Achieved Therapeutic Goals	At Time of Dose Change or Significant Clinical Complication
	Every 12 Mo	Every 12-24 Mo	Every 3 Mo	Every 12 Mo	Every 12-24 Mo	
Comprehensive physical examination	X			X	X (Annual)	
SF-36 (QoL) Survey	X			X	X (Annual)	X
Blood Tests						
Hemoglobin	X		X		X	X
Platelet count	X		X		X	X
Biochemical markers [3]	X		X		X	X
• Chitotriosidase						
• ACE						
• TRAP						
Additional Blood Tests [6]						
Visceral [9]						
Spleen volume (Volumetric MRI or CT)		X		X	X	X
Liver volume (Volumetric MRI or CT)		X		X	X	X
Skeletal [9]						
MRI of entire femora (Coronal; T_1- & T_2-weighted) [7]		X		X	X	X
X-ray [7, 10]		X		X	X	X
DEXA		X		X	X	X
Pulmonary [8]						

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 ot her 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 Cerezyme® (imiglucerase for injection) 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 effectiveness of Cerezyme®.
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. AP view of the entire femora (optimally from hips to below knees), and lateral view of the spine.
8. Pulmonary assessments are recommended every 12-24 months for patients with borderline or above normal pulmonary pressures at baseline.
9. Anatomical sites not included here should be evaluated if symptoms develop in such locations.
10. Optional in absence of new symptoms or evidence of disease progression.

Abbreviations: ACE, angiotensin-converting enzyme; ALT, alanine aminotransferase; AP, anteroposterior; AST, aspartate aminotransfera se; CT, computed tomography; DEXA, dual-energy X-ray absorptiometry; ECG, electrocardiogram; MRI, magnetic resonance imaging; PT, prothrombin time; PTT, partial thromboplastin time; TRAP, tartrate resistant acid phosphatase; WBC, white blood cell

Weinreb et al., Seminars in Hematology 2004, Vol 41 No 4, Suppl 5: 15 - 22