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

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Initial Assessment[1,2]

Blood Tests

PRIMARY TESTS

- Hemoglobin
- Platelet Count
- Biochemical Markers[3]
  - Chitotriosidase
  - ACE
  - Acid Phosphatase, tartrate resistant (TRAP)
- Mutation Analysis
- Antibody Sample[4]

ADDITIONAL TESTS AS INDICATED[5]

- AST and/or ALT
- Alkaline Phosphatase
- Calcium
- Phosphorus
- PT
- PTT
- WBC
- Total and Direct Bilirubin
- Albumin
- Total Protein
- Serum Immunoelectrophoresis
- Iron
- Iron Binding Capacity
- Ferritin
- Vitamin B_{12}

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 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 Months	Every 12-24 Months	Every 3 Months	Every 12 Months	Every 12-24 Months	
Blood Tests						
Hemoglobin	X		X		X	X
Platelet Count	X		X		X	X
Biochemical Markers[3] • Chitotriosidase • ACE • Acid Phosphatase, tartrate resistant (TRAP)	X		X		X	X
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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References:
1. Pastores, et al. Seminhematol 2004; 41 (suppl 5): 4-14
2. Weinreb, et al. Seminhematol 2004; 41 (suppl 5): 15-22