



A nonprofit enterprise of the University of Utah
and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

GAUCHER DISEASE TESTING PATIENT HISTORY FORM

Patient Name: _____ Date of Birth: _____
 Sex Assigned at Birth: Female Male Intersex Gender Identity (optional): Female Male _____
 Physician: _____ Physician's Phone: _____
 Practice Specialty: _____ Physician's Fax: _____
 Genetic Counselor: _____ Counselor's Phone: _____

Patient's Ethnicity (check all that apply)
 African American/Black Asian Hispanic White Other: _____

Does the patient have symptoms of Gaucher disease?..... No Yes (check all that apply)

<input type="checkbox"/> Anemia or cytopenia	<input type="checkbox"/> Corneal opacity	<input type="checkbox"/> Oculomotor apraxia
<input type="checkbox"/> Bone disease	<input type="checkbox"/> Hepatomegaly	<input type="checkbox"/> Primary central nervous system (CNS) disease
<input type="checkbox"/> Bulbar signs	<input type="checkbox"/> Ichthyosiform or collodion skin changes	<input type="checkbox"/> Pyramidal signs
<input type="checkbox"/> Calcification of mitral and aortic valves	<input type="checkbox"/> Lung disease	<input type="checkbox"/> Seizures
	<input type="checkbox"/> Nonimmune hydrops fetalis	<input type="checkbox"/> Splenomegaly

Other symptom(s): _____

Laboratory Findings

GBA enzyme testing: Normal Abnormal (result: _____) Not Performed Unknown

Has the patient undergone previous DNA testing for Gaucher disease?..... No Yes Unknown

If yes, describe the test and results: _____

Is there any relevant family history? No Yes Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

Has DNA testing been performed for the family member(s)? No Yes Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing), or indicate the result: _____

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.

