ARUP LABORATORIES

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)

☐ Anemia or cytopenia	☐ Corneal opacity	☐ Oculomotor apraxia
☐ Bone disease	☐ Hepatomegaly	☐ Primary central nervous system (CNS) disease
☐ Bulbar signs	☐ Ichthyosiform or collodion skin changes	☐ Pyramidal signs
☐ Calcification of mitral and aortic valves	☐ Lung disease	☐ Seizures
	☐ Nonimmune hydrops fetalis	☐ 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: _____

Check the test you intend to order.

☐ **2014459 Gaucher Disease (*GBA*), Enzyme Activity in Leukocytes:**
GBA enzyme testing to diagnose Gaucher disease; not accurate for carrier screening.

☐ **0051438 Gaucher Disease (*GBA*), 8 Variants:** Targets eight common pathogenic *GBA* DNA variants for carrier or diagnostic testing in individuals of Ashkenazi Jewish (AJ) descent. Clinical sensitivity 90% in AJ, 55% in other ethnicities.

☐ **3001648 Gaucher Disease (*GBA*), Sequencing:** *GBA* full gene sequencing. Clinical sensitivity ~99%.

☐ **2001961 Familial Mutation, Targeted Sequencing:**
Tests for sequence variant(s) previously identified in the family. Contact an ARUP genetic counselor at 800-242-2787 ext. 2141 prior to test submission to discuss requirements.

☐ **Other test not listed:** _____

Master Label

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