ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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 fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR GAUCHER DISEASE TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Physician: _____ **Physician Phone:** _____

Practice Specialty: _____ **Physician Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

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

☐ Anemia or cytopenia
☐ Bone disease
☐ Bulbar signs
☐ Calcification of mitral and aortic valves
☐ Corneal opacity
☐ Hepatomegaly
☐ Ichthyosiform or collodion skin changes
☐ Lung disease
☐ Nonimmune hydrops fetalis
☐ Oculomotor apraxia
☐ Primary central nervous system (CNS) disease
☐ Pyramidal signs
☐ Seizures
☐ 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 8 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 a pathogenic gene variant(s) previously identified in a family member; a copy of relative's lab result is REQUIRED.

☐ **Other test not listed:** _____

Master Label

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