

**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     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 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.**