

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

PLEASE PRINT

Patient's Name: _____ Date of birth: ____ / ____ / ____

Sex assigned at birth: Female Male Intersex Gender identity (optional): Female Male _____

Physician _____ Provider's Phone: (____) _____

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

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 on 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:

2014459 Gaucher Disease (GBA), Enzyme Activity in Leukocytes:

GBA enzyme testing to diagnose Gaucher disease; not accurate for carrier screening.