Gaucher Disease Enzyme Analysis (Glucocerebrosidase)

Order Name: **Gaucher Enzyme**
Test Number: 5194842
Revision Date: 12/12/2022

TEST NAME	METHODOLOGY	LOINC CODE
Gaucher Disease Enzyme Analysis (Glucocerebrosidase)	See Test Notes	

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	**See Instructions**	**See Instructions**	**See Instructions**	**Refrigerated**

Instructions

Specimen Type: Adult: Two 10 mL Whole Blood from lavender-top (EDTA) tube(s). Pediatric: One lavender-top (EDTA) tube.
Specimen Storage: **Refrigerate after collection.** Transport to the testing laboratory using cool packs. **Do NOT allow the sample to freeze.** **Time-sensitive; specimens must be received within four days of collection; please collect/ship to Labcorp Oklahoma Inc. in Tulsa Monday through Wednesday ONLY.**
Special Instructions: Testing referred to Integrated Genetics TC# 370 Specimens must arrive at testing laboratory within four days of collection. Collect Monday through Wednesday ONLY. While enzyme testing is offered as a standalone test.

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GENERAL INFORMATION

Expected TAT	3 - 13 days
Clinical Use	Gaucher disease occurs at an increased frequency in individuals of Ashkenazi Jewish descent, with a carrier frequency of approximately 1 in 15. It is a lysosomal storage disorder with variable severity that, if untreated, may result in anemia, hepatosplenomegaly, nosebleeds, and fractures. In the more severe and rare form, the brain and nervous system are involved. Enzyme analysis is performed in individuals suspected of having the disease, who may need enzyme replacement therapy.
Notes	Labcorp Test Code: 451780 Methodology: Glucocerebrosidase (leukocyte) is measured by fluorometric enzyme assay using 4-methylumbelliferyl-beta-D-glucosida as substrate, with/without glucocerebrosidase-specific inhibitor.
CPT Code(s)	84155, 82963
Service Provided By	labcorp Oklahoma, Inc.