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|---|--|---|--------------------|------------------|
| Patient ID SA00062408 | Patient Name SAMPLEREPOR, FUCT A | Birth Date 1987-05-22 | Gender F | Age 26 |
| Order Number SA00062408 | Client Order Number SA00062408 | Ordering Physician Client, Client | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 17 Sep 2013 10:27 | | |

Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:15; Test not performed.

Received: 17 Sep 2013 11:15

Reported: 17 Sep 2013 11:15

Fibroblast Culture

MCR

Fibroblast Culture was cancelled on 09/17/2013 at 11:15; Test not performed.

Received: 17 Sep 2013 11:15

Reported: 17 Sep 2013 11:15

Alpha-Fucosidase, Fibroblasts

Alpha-Fucosidase, Fibroblasts

MCR



0.11 nmol/min/mg Prot

Reference Value
≥0.41

Reviewed By

MCR

Daniel Kraft

Received: 17 Sep 2013 11:15

Reported: 17 Sep 2013 13:37

Interpretation (FUCT)

MCR

In this specimen, fucosidase activity is reduced. This is consistent with a diagnosis of fucosidosis (OMIM 230000), a rare autosomal recessive lysosomal storage disorder. If clinically indicated, consider further confirmatory testing by molecular genetic analysis of the FUCA1 gene. A genetics consultation is recommended.

Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

Performing Site Legend

| Code | Laboratory | Address |
|------|--|--|
| MCR | Mayo Clinic Dept. of Lab Med and Pathology | 200 First Street SW, Rochester, MN 55905 |