FirmaLab™
Bio-Diagnostics

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Patient Name
Jane Doe

DOB	Gender	Ethnicity
04/27/2003	F	Unknown

Test Ordered	Test Result	Expected (Negative) Value
LAL (LIPA) Enzyme Assay Dry Blood Spot - RESEARCH	***Positive***	Reference range 0.29 - 2.30 nmole/punch/hr

Result Note: -0.0068 nmol/punch/hr

Result Interpretation: LAL enzyme activity is below normal reference range. This result is consistent with the diagnosis of LAL deficiency.

Method of Analysis: The measurement of lysosomal acid lipase (LAL) in dried blood spots (DBS) is done using a fluorimetric substrate, 4-methylumbelliferyl palmitate (4mU palmitate), with cardiolipin present as an activator of LAL. The presence of other forms of lipase in whole blood will interfere with the measurement of LAL. Since Lalistat 2 is a specific inhibitor of LAL, measuring the total lipase activity and lipase activity in the presence of Lalistat 2 will allow for determination of LAL in DBS. If an individual has been the recipient of bone marrow transplantation, or if he/she has undergone any blood transfusion procedure within the past six months, blood may not be tested reliably. This test was developed and its performance characteristics determined by the laboratory. The principle of the assay was based on studies described by Hamilton J, et al, Clinica Chimica Acta (2012), doi:10.1016/j.cca.2012.03.019.

The assay has not been cleared or approved by the U.S. Food and Drug Administration (FDA), or the FDA has determined that such clearance of approval is not necessary. The laboratory is accredited by College of American Pathologists (CAP), and regulated under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity molecular testing.

LIPA Sequence (Wolman Disease)	***Cancelled***	Negative (No variations found)

Result Note: Exon DNA amplification and cycle sequencing failed 07/05/2013 and 07/08/2013.
Unable to obtain to new specimen, ordering physician reported that the patient passed away prior to testing.

Electronically signed by: Daniel Darvish, MD, on 07/16/2013 13:37
Report Type: Complete, Result correction

Specimen ID	Specimen Type	Collection Date Time	Date Received	Report Date
9001009	Dry Blood Spot	09/13/2009 10:00	06/18/2013 10:00	07/16/2013 13:30