MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FAJFP	SA00005453	45	M	SA00005453
Ordering Phys				**DOB** 05/25/1966
Client Order # SA00005453	**Account Information**			**Report Notes**
Collected 05/15/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 05/16/2012 15:10				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Ashkenazi Jewish Pnl (8 Conditions)			REPORTED 05/16/2012 12:58		
Mutation(s)		See Below			Y004
Result: c.2281del6/ins7 (BLM gene)					
Interpretation					Y004
The results of this test indicate the heterozygous presence of a known mutation, c.2281del6/ins7, at codon 735 of the BLM gene (Ellis et al. Cell 1995 Nov 17;83(4):655-66). This results in a frameshift mutation which leads to the formation of a stop codon, which is deleterious.					
This patient is a carrier of the c.2281del6/ins7 mutation in the BLM gene.					
No other mutations were detected. These results reduce the likelihood this patient is a carrier for the following conditions: Bloom syndrome, Canavan disease, Familial dysautonomia, Fanconi anemia group C, Gaucher diesease, Mucolipidosis type IV, Niemann-Pick disease type A and Tay-Sachs disease.					
Reviewed by					Y004
Steve Swanson, CGMBS, MB (ASCP), Information Specialist and J. Jennifer Wei, MD, PhD, Medical Director					
Review date		05162012			Y004
Methodology: Genomic deoxyribonucleic acid (gDNA) is isolated from the patient's specimen using a standardized kit and quantified by agarose gel electrophoresis. For the 8 conditions, DNA regions covering the 30 mutations are PCR amplified followed by single-stranded pyrosequencing using the Pyromark MD sequence analysis system (Biotage). If specific mutation analysis is requested for any gene, only specific region(s) of DNA is (are) amplified by PCR and sequenced with dye terminator sequencing chemistry in the sense and antisense directions.					
Disclaimer: This test was developed and its performance characteristics were determined by Ambry Genetics Corporation. The laboratory is regulated under the Clinical Laboratory Improvement Amendments 2003 as qualified to perform non-waived testing.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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The Ambry Test: Ashkenazi Jewish Panel analyzes the specific mutations listed above. A negative result from the analysis cannot rule out the possibility that the tested individual carries an unexamined mutation.
The Ambry Test: Ashkenazi Jewish Panel is designed and validated to be capable of detecting >99% of the previously described 30 mutations on the 8 genes listed above. Although molecular tests are highly accurate, rare diagnostic errors may occur. Possible diagnostic errors include sample mix-up, erroneous paternity identification, technical errors, clerical errors, and genotyping errors. Genotyping errors can result from trace contamination of PCR reactions, from maternal cell contamination in fetal samples, from rare genetic variants that interfere with analysis, low-level mosaicism or from other sources. This report does not represent medical advice. Any questions, suggestions, or concerns regarding interpretation of results should be relegated to a genetic counselor, medical geneticist, or physician skilled in evaluating the relevant medical literature. References are available upon request.

Disease	Carrier Rate	Detection Rate	Risk
Bloom Syndrome	1/107	97%	1/3,533
Canavan Disease	1/41	98%	1/2,000
Familial Dysautonomia	1/32	>99%	1/3,100
Fanconi Anemia Group C	1/89	99%	1/8,800
Gaucher Disease	1/15	>95%	1/280
Mucolipidosis Type IV	1/96	95%	1/1,900
Niemann-Pick Disease Type A	1/90	>95%	1/1,780
Tay-Sachs Disease	1/28	96%	1/675

* Performing Site:

Y004	Ambry Genetics 100 Columbia No. 200 Aliso Viejo, CA 92656	Lab Director: