

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT, FAJFP	SA00005451	45	M	SA00005451
Ordering Phys				DOB 05/25/1966
Client Order #	Account Information			Report Notes
SA00005451				
Collected	C7028846-DLMP ROCHESTER			
05/15/2012 13:00	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 55901			
05/16/2012 15:04				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Ashkenazi Jewish HEXA only Mutation		c.1274_1277dupTATC	REPORTED 05	/16/2012 12:22	¥00 4
Interpretation The mutation, c.1274_12776 c.1278insTATC and c.1277_: exon 11 of the HEXA gene Chem. 1988;263:18587-1858; nucleotides creates a fram signal. This is the most of found in the Ashkenazi Jew This patient is a carrier mutation in the HEXA gene	dupTATC (also 1278insTATC), (Myerowitz R C 9). The duplic neshift and pr common Tay-Sac wish populatio of the c.1274.	known as is located in ostigan FC. J Biol ation of four emature stop hs disease mutation n. 1277dupTATC			¥00 4
No other mutations were de Reviewed by	etected.				Y00 4
Steve Swanson, CGMBS, MB and J. Jennifer Wei, MD, M Review date	(ASCP), Inform PhD, Medical D	ation Specialist Virector 05162012			¥00 4
Methodology: Genomic deoxyribonucleic a patient's specimen using a by agarose gel electrophor regions covering the 7 mu followed by single-strando Pyromark MD sequence analy mutation analysis is reque region(s) of DNA is (are) with dye terminator sequen antisense directions. Disclaimer: This test was developed an	acid (gDNA) is a standardized resis. For Tay tations are PC ed pyrosequenc ysis system (B ested for any amplified by ncing chemistr nd its perform	isolated from the kit and quantified -Sachs, DNA R amplified fing using the fiotage). If specific gene, only specific PCR and sequenced y in the sense and mance characteristics			
were determined by Ambry (laboratory is regulated u Improvement Amendments 20) waived testing. The Ambry Test: Ashkenazi specific mutations listed the analysis cannot rule of tested individual carries The Ambry Test: Ashkenazi validated to be capable of previously described 7 mut Although molecular tests a	Genetics Corpo nder the Clini D3 as qualifie Jewish Panel above. A nega but the possib an unexamined Jewish Panel f detecting >9 tations in the are highly acc	HEXA analyzes the tive result from bility that the mutation. is designed and 9% of the HEXA gene. wrate, ble diagnostic			
errors include sample mix	-up, erroneous	paternity			

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, FAJFP	05/15/2012 13:00	Final
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* Report times for Mayo performed tests are CST/CDT



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		_	_		Reference	Perform
Test		Flag R	esults	Unit	Value	Site*
identification, techn genotyping errors. Ge contamination of PCR contamination in feta that interfere with a other sources. This r advice. Any questions interpretation of res counselor, medical ge evaluating the releva are available upon re	nical errors notyping err reactions, : 1 samples, : nalysis, loo eport does no suggestion sults should eneticist, or not medical is quest.	, clerica from mate from rare w-level m not repre ns, or cc be releg r physici literatur	el errors, and result from tra rnal cell e genetic varian losaicism or fro sent medical oncerns regardin gated to a genet an skilled in re. References	ce ts m g ic		
	Carrier 1	Detection	L			
Disease	Rate	Rate	Risk			
Tay-Sachs Disease	1/28	96%	1/675			

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

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SAMPLEREPORT, FAJFP	05/15/2012 13:00	Final
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