ARUP LABORATORIES | aruplab.com
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108-
USA

Provider: .108 -TEST,

Patient: **AJP, POSITIVE**
DOB:
Sex: Male
Patient Identifiers: 40694
Visit Number (FIN): 41019
Client Supplied ID:

Specimen Collected: 20-Sep-22 16:21

Ashkenazi Jewish Diseases | **Received: 20-Sep-22 16:23** **Report/Verified: 20-Sep-22 16:55**

Procedure	Result	Units	Reference Interval
Ashkenazi Jewish Diseases, Specimen	Whole Blood		
Ashkenazi Jewish Diseases,Panel Results	**Positive** *		
Ashkenazi Jewish Diseases,Gene 1	**GBA** *		
AJP Gene 1,Allele 1	**c.1226A>G** *		
AJP Gene 1,Allele 2	Negative		
Ashkenazi Jewish Diseases,Gene 2	N/A		
AJP Gene 2,Allele 1	N/A		
AJP Gene 2,Allele 2	N/A		
Ashkenazi Jewish Diseases Carrier Status	**Yes** *		
Ashkenazi Jewish Diseases,Interp	See Note f1 i1		

Result Footnote

f1: Ashkenazi Jewish Diseases, Interp
Indication for testing: Carrier screening for genetic disorders common in Ashkenazi Jewish individuals.

Positive: One mild pathogenic variant, p.N409S (c.1226A>G), was detected in the GBA gene; therefore, this individual is a carrier of Gaucher disease type 1. Genetic counseling is recommended. This individual's reproductive partner should be offered screening for the disorder. At-risk family members should be offered testing to determine carrier status for the identified variant. None of the other targeted variants associated with the 16 common Ashkenazi Jewish disorders screened by this panel were identified. If this individual is of Ashkenazi Jewish descent, he/she may use the table below to review the residual carrier risk for the other disorders. If this individual has a positive family history of a disorder covered by this panel, the figures for that disorder do not apply. Gaucher disease affects lysosomal storage and has extreme symptom variability, ranging from perinatal lethality to asymptomatic individuals. Three Gaucher subtypes have been identified based on symptom characteristics. Individuals affected with Gaucher disease type 1 may be asymptomatic. Symptomatic individuals may have bone disease, hepatosplenomegaly, anemia, thrombocytopenia, and lung disease. Affected individuals with at least one copy of the p.N409S variant do not develop primary neurologic disease associated with this disorder.

This result has been reviewed and approved by Yuan Ji, Ph.D.

Test Information

i1: Ashkenazi Jewish Diseases, Interp
BACKGROUND INFORMATION: Ashkenazi Jewish Diseases, 16 Genes

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

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i1: Ashkenazi Jewish Diseases, Interp

OVERVIEW: This targeted panel detects 51 variants common in the Ashkenazi Jewish population associated with 16 disorders, including ABCC8-related hyperinsulinism, Bloom syndrome, Canavan disease, familial dysautonomia, Fanconi anemia group C, Gaucher disease, glycogen storage disease 1A, Joubert syndrome type 2, lipoamide dehydrogenase deficiency, maple syrup urine disease type 1B, mucolipidosis type IV, NEB-related nemaline myopathy, Niemann-Pick disease type A, Tay-Sachs disease, Usher syndrome type 1F and type 3.
INHERITANCE: Autosomal recessive.
CLINICAL SENSITIVITY: Among Ashkenazi Jewish individuals:
- 99 percent for Canavan disease, lipoamide dehydrogenase deficiency, familial dysautonomia, Fanconi anemia group C, glycogen storage disease type 1A, Joubert syndrome type 2, maple syrup urine disease type 1B, and NEB-related nemaline myopathy
- 98 percent for Usher syndrome type 3
- 97 percent for ABCC8-related hyperinsulinism and Bloom syndrome
- 95 percent for mucolipidosis type IV
- 94 percent for Tay-Sachs disease
- 90 percent for Gaucher disease and Niemann-Pick disease type A
- 62 percent for Usher syndrome type 1F

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring. See table below for specific variants tested.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Variants other than those tested on this panel will not be detected. Diagnostic errors can occur due to rare sequence variations.

DISEASE (GENE)	VARIANTS TESTED	ASHKENAZI DISEASE INCIDENCE	ASHKENAZI PRETEST CARRIER RISK	ASHKENAZI CARRIER RISK AFTER NEG RESULT
ABCC8-related hyperinsulinism (ABCC8)	p.F1388del (c.4163_4165del) p.V187D (c.560T>A) c.3992-9G>A	1/7,800	1/52	1/1,700
Bloom	p.Y736Lfs	1/40,000	1/100	1/3,300

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syndrome (BLM)	(c.2207_2212delins TAGATTC)			
Canavan disease (ASPA)	c.433-2A>G p.Y231X (c.693C>A) p.E285A (c.854A>C) p.A305E (c.914C>A)	1/10,000	1/50	1/4,900
Familial dys-autonomia (ELP1)	p.R696P (c.2087G>C) c.2204+6T>C	1/3,600	1/32	1/3,100
Fanconi anemia group C (FANCC)	p.D23Ifs (c.67delG) c.456+4A>T	1/32,000	1/89	1/8,800
Gaucher disease (GBA)	p.L29Afs (c.84dupG) c.115+1G>A p.N409S (c.1226A>G) c.1263_1317del55 p.V433L (c.1297G>T) p.D448H (c.1342G>C) p.L483P (c.1448T>C) p.R535H (c.1604G>A)	1/900	1/15	1/141
Glycogen storage disease type 1A (G6PC)	p.Q27Rfs (c.79delC) p.R83H (c.248G>A) p.R83C (c.247C>T) p.Y128Tfs (c.379_380dupTA) p.G188R (c.562G>C) p.Q242X (c.724C>T) p.Q347X (c.1039C>T) p.G270V (c.809G>T) p.F327del (c.979_981delTTC)	1/20,000	1/71	1/7,000
Joubert syndrome	p.R73L (c.218G>T)	1/34,000	1/92	1/9,100

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type 2 (TMEM216)				
Lipoamide dehydrogenase deficiency (DLD)	p.Y35X (c.104dupA) p.G229C (c.685G>T)	1/35,000	1/94	1/9,300
Maple syrup urine disease type 1B (BCKDHB)	p.R183P (c.548G>C) p.G278S (c.832G>A) p.E372X (c.1114G>T)	1/50,000	1/113	1/11,200
Mucolipidosis IV (MCOLN1)	c.406-2A>G g.511_6943del	1/63,000	1/127	1/2,500
NEB-related nemaline myopathy (NEB)	exon 55 del (p.R2478_D2512del)	1/47,000	1/108	1/10,700
Niemann-Pick type-A disease (SMPD1)	p.L304P (c.911T>C) p.F333Sfs (c.996delC) p.R498L (c.1493G>T) p.R610del (c.1829_1831delGCC)	1/32,000	1/90	1/890
Tay-Sachs disease (HEXA)	7.6 kb del p.G269S (c.805G>A) c.1073+1G>A p.Y427Ifs (c.1274_1277dupTATC)	1/3,000	1/30	1/480

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	c.1421+1G>C			
	Pseudodeficiency alleles:			
	p.R247W(c.739C>T)			
	p.R249W (c.745C>T)			
Usher syndrome type 1F (PCDH15)	p.R245X (c.733C>T)	1/20,500	1/72	1/190
Usher syndrome type 3 (CLRN1)	p.N48K (c.144T>G)	1/82,000	1/143	1/7,100

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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