Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/25/1990
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Ashkenazi Jewish Diseases, 16 Genes

ARUP test code 0051415

Ashkenazi Jewish Diseases, Specimen	Whole Blood	
Ashkenazi Jewish Diseases, Panel Results	Positive	*
Ashkenazi Jewish Diseases, Gene 1	FANCC	*
AJP Gene 1, Allele 1	c.456+4A>T	*
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	

H=High, L=Low, *=Abnormal, C=Critical

Indication for testing: Carrier screening for genetic disorders common in Ashkenazi Jewish individuals.

Positive: One pathogenic variant, c.456+4A>T, was detected in the FANCC gene; therefore, this individual is a carrier of Fanconi anemia group C. 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. Fanconi anemia group C presents with short stature, abnormal skin pigmentation and multiple malformations that may affect eyes, ears, heart, oral cavity, thumbs, forearms, kidneys, or urinary tract. Other symptoms may include hearing loss, hypogonadism, and developmental delay. Progressive bone marrow failure occurs during the first decade of life. Hematologic malignancies occur in approximately 20 percent of affected individuals. Nonhematologic malignancies occur in approximately 30 percent of affected individuals.

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Ashkenazi Jewish Diseases, 16 Genes

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 hyper-insulin-ism (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 type 2 (TMEM216)	p.R73L (c.218G>T)	1/34,000	1/92	1/9,100	
Lipoamide dehydro-genase 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	
Mucolip-idosis 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	

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Tay-Sachs disease (HEXA)	7.6 kb del p.G269S (c.805G>A) c.1073+1G>A p.Y427Ifs (c.1274_1277dupTATC) c.1421+1G>C Pseudodeficiency alleles: p.R247W(c.739C>T) p.R249W (c.745C>T)	1/3,000	1/30	1/480
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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Ashkenazi Jewish Diseases, Specimen	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Panel Results	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Gene 1	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 1, Allele 1	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 1, Allele 2	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Gene 2	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 2, Allele 1	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 2, Allele 2	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases Carrier Status	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Interp	24-022-123590	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical