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

Physician: Doctor, Example

Patient: Patient, Example

DOB	11/8/1995
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	Negative
Ashkenazi Jewish Diseases, Gene 1	N/A
AJP Gene 1, Allele 1	N/A
AJP Gene 1, Allele 2	N/A
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	No
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.

Negative: None of the 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 each disorder, given this negative result. If family history of one of the diseases tested exists, the figures for that disorder do not apply; please contact an ARUP genetic counselor (800-242-2787 x2141) for a risk reduction calculation specific to this individual's family.

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 syndrome (BLM)	p.Y736Lfs (c.2207_2212delins TAGATTC)	1/40,000	1/100	1/3,300
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
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

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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	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Panel Results	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Gene 1	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 1, Allele 1	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 1, Allele 2	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Gene 2	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 2, Allele 1	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
AJP Gene 2, Allele 2	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases Carrier Status	24-019-139728	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ashkenazi Jewish Diseases, Interp	24-019-139728	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