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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/14/1981
Sex:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Expanded Carrier Screen by Next Generation Sequencing

ARUP test code 2014680

EER Expanded Carrier Screen NGS

See Note

Access ARUP Enhanced Report using the link below:

-Direct access:

Expanded Carrier Screen NGS

See Note

Disease	Result
11-beta-hydroxylase-deficient congenital adrenal hyperplasia	Negative
6-pyruvoyl-tetrahydropterin synthase deficiency	Negative
ABCC8-related familial hyperinsulinism	Negative
adenosine deaminase deficiency	Negative
alpha thalassemia, HBA1/HBA2-related	Negative
alpha-mannosidosis	Negative
alpha-sarcoglycanopathy	Negative
Alstrom syndrome	Negative
AMT-related glycine encephalopathy	Negative
Andermann syndrome	Negative
argininemia	Negative
argininosuccinic aciduria	Negative
aspartylglucosaminuria	Negative
ataxia with vitamin E deficiency	Negative
ataxia-telangiectasia	Negative
ATP7A-related disorders	Negative
autoimmune polyglandular syndrome type 1	Negative
autosomal recessive osteopetrosis type 1	Negative
autosomal recessive polycystic kidney disease, PKHD1-related	Negative
autosomal recessive spastic ataxia of Charlevoix-Saguenay	Negative
Bardet-Biedl syndrome, BBS1-related	Negative
Bardet-Biedl syndrome, BBS10-related	Negative
Bardet-Biedl syndrome, BBS12-related	Negative
Bardet-Biedl syndrome, BBS2-related	Negative
BCS1L-related disorders	Negative
beta-sarcoglycanopathy	Negative
biotinidase deficiency	Negative
Bloom syndrome	Negative
calpainopathy	Negative
Canavan disease	Negative
carbamoylphosphate synthetase I deficiency	Negative
carnitine palmitoyltransferase IA deficiency	Negative
carnitine palmitoyltransferase II	Negative

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

deficiency	
cartilage-hair hypoplasia	Negative
cerebrotendinous xanthomatosis	Negative
citrullinemia type 1	Negative
CLN3-related neuronal ceroid lipofuscinosis	Negative
CLN5-related neuronal ceroid lipofuscinosis	Negative
CLN6-related neuronal ceroid lipofuscinosis	Negative
CLN8-related neuronal ceroid lipofuscinosis	Negative
Cohen syndrome	Negative
COL4A3-related Alport syndrome	Negative
COL4A4-related Alport syndrome	Negative
combined pituitary hormone deficiency, PROP1-related	Negative
congenital adrenal hyperplasia, CYP21A2-related	POSITIVE
Positive result: NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) heterozygote (deleterious). This individual is a carrier of congenital adrenal hyperplasia, CYP21A2-related. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) is a non-classic congenital adrenal hyperplasia, CYP21A2-related mutation.	
congenital disorder of glycosylation type Ia	Negative
congenital disorder of glycosylation type Ic	Negative
congenital disorder of glycosylation, MPI-related	Negative
Costeff optic atrophy syndrome	Negative
cystic fibrosis	Negative
cystinosis	Negative
D-bifunctional protein deficiency	Negative
delta-sarcoglycanopathy	Negative
dihydrolipoamide dehydrogenase deficiency	Negative
dysferlinopathy	Negative
dystrophinopathy (including Duchenne/Becker muscular dystrophy)	Negative
ERCC6-related disorders	Negative
ERCC8-related disorders	Negative
EVC-related Ellis-van Creveld syndrome	Negative
EVC2-related Ellis-van Creveld syndrome	Negative
Fabry disease	Negative
familial dysautonomia	Negative
familial Mediterranean fever	Negative
Fanconi anemia complementation group A	Negative
Fanconi anemia, FANCC-related	Negative
FKRP-related disorders	Negative
FKTN-related disorders	Negative
free sialic acid storage disorders	Negative
galactokinase deficiency	Negative
galactosemia	Negative
gamma-sarcoglycanopathy	Negative
Gaucher disease	Negative
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	Negative
GLB1-related disorders	Negative
GLDC-related glycine encephalopathy	Negative
glutaric acidemia, GCDH-related	Negative
glycogen storage disease type Ia	Negative
glycogen storage disease type Ib	Negative
glycogen storage disease type III	Negative
GNE myopathy	Negative
GNPTAB-related disorders	Negative
HADHA-related disorders	Negative
Hb beta chain-related hemoglobinopathy	Negative

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hereditary fructose intolerance	Negative
Herlitz junctional epidermolysis bullosa, LAMB3-related	Negative
hexosaminidase A deficiency	Negative
HMG-CoA lyase deficiency	Negative
holocarboxylase synthetase deficiency	Negative
homocystinuria, CBS-related	Negative
hydrolethalus syndrome	Negative
hypophosphatasia	Negative
isovaleric acidemia	Negative
Joubert syndrome 2	Negative
junctional epidermolysis bullosa, LAMA3-related	Negative
junctional epidermolysis bullosa, LAMC2-related	Negative
KCNJ11-related familial hyperinsulinism	Negative
Krabbe disease	Negative
LAMA2-related muscular dystrophy	Negative
Leigh syndrome, French-Canadian type	Negative
lipoid congenital adrenal hyperplasia	Negative
lysosomal acid lipase deficiency	Negative
maple syrup urine disease type Ia	Negative
maple syrup urine disease type Ib	Negative
maple syrup urine disease type II	Negative
medium chain acyl-CoA dehydrogenase deficiency	Negative
megalencephalic leukoencephalopathy with subcortical cysts	Negative
metachromatic leukodystrophy	Negative
methylmalonic acidemia, cblA type	Negative
methylmalonic acidemia, cblB type	Negative
methylmalonic aciduria and homocystinuria, cblC type	POSITIVE

Positive result: NM_015506.2(MMACHC):c.271dupA(R91Kfs*14) heterozygote (deleterious). This individual is a carrier of methylmalonic aciduria and homocystinuria, cblC type. Carriers generally do not experience symptoms. R91Kfs*14 may be associated with early onset methylmalonic aciduria and homocystinuria, cblC type.

MKS1-related disorders	Negative
mucolipidosis III gamma	Negative
mucolipidosis IV	Negative
mucopolysaccharidosis type I	Negative
mucopolysaccharidosis type II	Negative
mucopolysaccharidosis type IIIA	Negative
mucopolysaccharidosis type IIIB	Negative
mucopolysaccharidosis type IIIC	Negative
MUT-related methylmalonic acidemia	Negative
MYO7A-related disorders	Negative
NEB-related nemaline myopathy	Negative
nephrotic syndrome, NPHS1-related	Negative
nephrotic syndrome, NPHS2-related	Negative
Niemann-Pick disease type C1	Negative
Niemann-Pick disease type C2	Negative
Niemann-Pick disease, SMPD1-related	Negative
Nijmegen breakage syndrome	Negative
ornithine transcarbamylase deficiency	Negative
PCCA-related propionic acidemia	Negative
PCCB-related propionic acidemia	Negative
PCDH15-related disorders	Negative
Pendred syndrome	Negative
peroxisome biogenesis disorder type 1	Negative
peroxisome biogenesis disorder type 3	Negative
peroxisome biogenesis disorder type 4	Negative
peroxisome biogenesis disorder type 5	Negative
peroxisome biogenesis disorder type 6	Negative
phenylalanine hydroxylase deficiency	Negative
POMGNT-related disorders	Negative
Pompe disease	Negative
PPT1-related neuronal ceroid	Negative

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lipofuscinosis	
primary carnitine deficiency	Negative
primary hyperoxaluria type 1	Negative
primary hyperoxaluria type 2	Negative
primary hyperoxaluria type 3	Negative
pycnodysostosis	Negative
pyruvate carboxylase deficiency	Negative
rhizomelic chondrodysplasia punctata type 1	Negative
RTEL1-related disorders	Negative
Sandhoff disease	Negative
short-chain acyl-CoA dehydrogenase deficiency	Negative
Sjogren-Larsson syndrome	Negative
SLC26A2-related disorders	Negative
Smith-Lemli-Opitz syndrome	Negative
spastic paraplegia type 15	Negative
spinal muscular atrophy	Negative
Negative result: SMN1: 3+ copies.	
spondylothoracic dysostosis	Negative
TGM1-related autosomal recessive congenital ichthyosis	Negative
TPP1-related neuronal ceroid lipofuscinosis	Negative
tyrosine hydroxylase deficiency	Negative
tyrosinemia type I	Negative
tyrosinemia type II	Negative
USH1C-related disorders	Negative
USH2A-related disorders	Negative
usher syndrome type 3	Negative
very-long-chain acyl-CoA dehydrogenase deficiency	Negative
Wilson disease	Negative
X-linked adrenoleukodystrophy	Negative
X-linked Alport syndrome	Negative
X-linked congenital adrenal hypoplasia	Negative
X-linked juvenile retinoschisis	Negative
X-linked myotubular myopathy	Negative
X-linked severe combined immunodeficiency	Negative
xeroderma pigmentosum group A	Negative
xeroderma pigmentosum group C	Negative

This test is performed by Myriad Women's Health
180 Kimball Way, South San Francisco, CA 94080 (888)268-6795
Lab Director: Jack Ji, PhD, FACMG

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
EER Expanded Carrier Screen NGS	20-228-401272	8/13/2020 1:15:00 PM	8/19/2020 11:32:04 AM	8/31/2020 6:21:00 PM
Expanded Carrier Screen NGS	20-228-401272	8/13/2020 1:15:00 PM	8/19/2020 11:32:04 AM	8/31/2020 10:30:00 AM

END OF CHART

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