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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/28/1996
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Expanded Carrier Screen by Next Generation Sequencing with Fragile X

ARUP test code 2014677

EER Expanded Carrier SCRN NGS w/FRAG X

See Note

Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

Expanded Carrier SCRN NGS w/FRAG X

See Note

Disease	Result
6-pyruvoyl-tetrahydropterin synthase deficiency	Negative
adenosine deaminase deficiency	Negative
alpha thalassemia, HBA1/HBA2-related	POSITIVE
Positive result: -alpha3.7 [chr16:g.(?_226678)_(227520_?)del] heterozygote (deleterious). This individual is a carrier of alpha thalassemia. Carriers do not experience symptoms, but may have hematologic abnormalities. -alpha3.7 is a pathogenic deletional alpha thalassemia variant. Based on this result, the patient's alpha globin status is -a/aa (alpha+ carrier), where "-" indicates a deleted or nonfunctional alpha globin gene.	
alpha-mannosidosis	Negative
alpha-sarcoglycanopathy	Negative
Alport syndrome, COL4A3-related	Negative
Alport syndrome, COL4A4-related	Negative
Alstrom syndrome	Negative
Andermann syndrome	Negative
argininemia	Negative
argininosuccinic aciduria	Negative
aspartylglucosaminuria	Negative
ataxia with vitamin E deficiency	Negative
ataxia-telangiectasia	Negative

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

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 globin-related hemoglobinopathy (including beta thalassemia and sickle cell disease)	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 deficiency	Negative
cartilage-hair hypoplasia	Negative
cerebrotendinous xanthomatosis	Negative
citrullinemia type 1	Negative
CLN3-related disorders	Negative
CLN5-related neuronal ceroid lipofuscinosis	Negative
CLN8-related neuronal ceroid lipofuscinosis	Negative
Cohen syndrome	Negative
combined pituitary hormone deficiency, PROP1-related	Negative
congenital adrenal hyperplasia, CYP11B1-related	Negative
congenital adrenal hyperplasia, CYP21A2-related	Negative
congenital disorder of glycosylation type Ic	Negative

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

congenital disorder of glycosylation, MPI-related	Negative
congenital disorder of glycosylation, PMM2-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 hyperinsulinism, ABCC8-related	Negative
familial hyperinsulinism, KCNJ11-related	Negative
familial Mediterranean fever	Negative
Fanconi anemia complementation group A	Negative
Fanconi anemia, FANCC-related	Negative
FKRP-related disorders	Negative
FKTN-related disorders	Negative
fragile X syndrome Negative result: Normal: 28 and 31 repeats.	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 Positive result: NM_004004.5(GJB2):c.139G>T(E47*) heterozygote (deleterious). This individual is a carrier of GJB2-related DFNB1 nonsyndromic hearing loss and deafness. Carriers generally do not experience symptoms.	POSITIVE
GLB1-related disorders	Negative

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

glutaric acidemia, GCDH-related	Negative
glycine encephalopathy, AMT-related	Negative
glycine encephalopathy, GLDC-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
hereditary fructose intolerance	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, LAMB3-related	Negative
junctional epidermolysis bullosa, LAMC2-related	Negative
Krabbe disease	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

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

methylmalonic acidemia, MMUT-related	Negative
methylmalonic aciduria and homocystinuria, cblC type	Negative
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
muscular dystrophy, LAMA2-related	Negative
MYO7A-related disorders	Negative
NEB-related nemaline myopathy	Negative
nephrotic syndrome, NPHS1-related	Negative
nephrotic syndrome, NPHS2-related	Negative
neuronal ceroid lipofuscinosis, CLN6-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 lipofuscinosis	Negative
primary carnitine deficiency	Negative

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

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: Negative for g.27134T>G SNP and SMN1: 2 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 adrenal hypoplasia congenita	Negative
X-linked adrenoleukodystrophy	Negative
X-linked Alport syndrome	Negative
X-linked juvenile retinoschisis	Negative
X-linked myotubular myopathy	Negative
X-linked severe combined immunodeficiency	Negative
xeroderma pigmentosum group A	Negative

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

```
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: Karla Bowles, PhD, FACMG, CGMB
```

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
EER Expanded Carrier SCRN NGS w/FRAG X	23-342-136370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Expanded Carrier SCRN NGS w/FRAG X	23-342-136370	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