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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/20/1954
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Mitochondrial Disorders (mtDNA) Sequencing and Deletion Analysis by NGS

ARUP test code 3001965

Ordering Physician Name

Ordering Physician Phone Number

EER Mito Disorders, mtDNA, Seq/Del

See Note

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

[redacted]

Mito Disorders, mtDNA, Seq/Del

Negative

Date Test(s) Started: 15-MAY-2023 10:25:09
Sample Source: Blood in EDTA Date Collected: 05-MAY-2023 Date Received:
10-MAY-2023 Testing Date Started: 15-MAY-2023 Date Reported: 07-JUN-2023
Provider Account #: A.R.U.P Laboratories Additional Provider:
Test(s) Requested Mitochondrial Disorders / Sequence Analysis and Deletion
Testing of the Mitochondrial Genome
Result: Negative
No pathogenic, likely pathogenic, or variants of uncertain significance were
identified by this analysis.
Interpretation This negative result does not exclude a genetic basis for this
individual's clinical features and/or family history. Variants in nuclear genes
important for normal mitochondrial function would not be detected by this
analysis. Furthermore, the percentage of mutant mtDNA (the degree of variant
heteroplasmy) varies among tissues so that mtDNA variants may be detected in
some tissues, but not others. Therefore it is usually best to test an involved
tissue, such as muscle or liver.
Recommendation(s) Genetic counseling is recommended to discuss the implications
of these results.
The level of variant heteroplasmy may differ among tissues so

that mtDNA
variants may be detected in some tissues, but not others. Therefore it is
usually best to test an involved tissue, such as muscle or liver. If clinically
indicated and not already performed, full sequence analysis and deletion
testing of the mitochondrial genome can be performed on a muscle biopsy
(approximately 50mg).The MitoXpanded panel, which includes concurrent patient
and parent sequencing of approximately 1800 genes associated with mitochondrial
disorders or a similar phenotype is also available. Whole exome sequencing
could also be considered. Please visit our website for additional information:
http: //www.genedx.com.
Resources MyGene2 is a portal through which families with rare genetic
conditions who are interested in sharing their health and genetic information
can connect with other families, clinicians, and researchers. If you are
interested in learning more and/or participating, please visit www.mygene2.org.
GenomeConnect is an NIH initiative created to enable individuals and families
with the same genetic variant or medical history to connect and share
de-identified information. If you are interested in participating, please visit
www.genomeconnect.org.
Additional Comments This individual's haplogroup and a table of observed
variants are also provided.* The observed variants have not been reported to be
associated with a disorder of mitochondrial metabolism when present in
association with this individual's specific haplogroup.
Genes Evaluated Mitochondrial Genome
Methods Genomic DNA was extracted from the submitted specimen, and the entire
mitochondrial genome was amplified and sequenced using Next Generation
sequencing. DNA sequence was assembled and analyzed in comparison with the
revised Cambridge Reference Sequence (rCRS GeneBank number NC_012920) and the
reported variants listed in the MITOMAP database (http: //www.mitomap.org).
Next-generation sequencing may not detect large-scale mtDNA deletions present
at 5% heteroplasmy or lower or mtDNA point variants present at 1.5%
heteroplasmy or lower. Alternative sequencing or other detection methods may be
used to analyze or confirm mtDNA variants. Reportable variants include
pathogenic variants, likely pathogenic variants and variants of uncertain
significance. Likely benign and benign variants, if present, are not routinely
reported but are available upon request. Available evidence for variant
classification may change over time and the reported variant(s) may be
reclassified according to our mitochondrial variant classification aligned with
the ACMG/AMP Standards and Guidelines for variant classification (PMID:
25741868), which may lead to issuing a revised report.

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

Disclaimer Genetic testing using the methods applied at GeneDx is expected to
be highly accurate. Normal findings do not rule out the diagnosis of a genetic
disorder since some genetic abnormalities may be undetectable by this test.
Unless indicated in the test methods, the following may not be detected: mosaic
variants, pure heterodisomy, balanced chromosomal aberrations, nucleotide
repeat expansion/contraction, abnormal DNA methylation, and variants located in
regions not evaluated by this test or that cannot be detected by the
methodology used. Regions of certain genes have technical limitations and
inherent sequence properties that yield suboptimal data, potentially impairing
accuracy of the results (for example: repetitive DNA, homology or pseudogene
regions, high GC content). Unless otherwise indicated, sequence analysis cannot
reliably detect deletions of 20bp to 500bp in size, or insertions of 10bp to
500bp in size; deletions/insertions of less than 500 bp cannot be reliably
detected by exon-level array. Rarely, incidental findings of large chromosomal
rearrangements outside the gene(s) of interest may be identified. The zygosity
reported reflects the presumed germline status of this individual, but may be
limited by depth of read coverage and/or parental genotype data at the time of
reporting. The chance of a false positive or false negative result due to
laboratory errors incurred during any phase of testing cannot be completely
excluded. As the ability to detect genetic variants and naming conventions can
differ among laboratories, rare false negative results may occur when no
positive control is provided for testing of a specific variant identified at
another laboratory. False negative results may also occur in the setting of
bone marrow transplantation, recent blood transfusion, or suboptimal DNA
quality. DNA extracted using external methodologies may negatively affect test
performance. In individuals with active or chronic hematologic neoplasms or
conditions, there is a possibility that testing may detect an acquired somatic
variant, leading to a false positive result. The clinical sensitivity of this
test depends in part on the patient's clinical phenotype, and is expected to be
highest for individuals with clearly defined disease and/or family history of
disease. Interpretations are made with the assumption that any clinical
information provided, including family relationships, is accurate. Consultation
with a genetics professional is recommended for interpretation of results. This
test was developed and its performance characteristics determined by GeneDx.
This test has not been cleared or approved by the U.S. Food and Drug
Administration. The FDA has determined that such clearance or approval is not

necessary. The test is used for clinical purposes and should not be regarded as
investigational or for research. The laboratory is certified under the Clinical
Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform
high-complexity clinical testing.
References Lek et al. (2016) Nature 536 (7616): 285-91 (PMID: 27535533);Stenson
et al. (2014) Human genetics 133 (1): 1-9 (PMID: 24077912);Landrum et al.
(2016) Nucleic Acids Res. 44 (D1): D862-8 (PMID: 26582918);Lott et al. (2013)
Curr Protoc Bioinformatics 44 : 1.23.1-26 (PMID: 25489354);Richards et al.
(2015) Genetics In Medicine: 17 (5): 405-24 (PMID: 25741868);
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Report electronically signed by: Erin Wakeling PhD, FACMG
Performed by: GeneDx
207 Perry Parkway
Gaithersburg, MD 20877

Anne Maddalena, Ph.D.,FACMG,

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Ordering Physician Name	23-125-132459	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ordering Physician Phone Number	23-125-132459	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Mito Disorders, mtDNA, Seq/Del	23-125-132459	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Mito Disorders, mtDNA, Seq/Del	23-125-132459	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