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

Physician: Doctor, Example

**Patient: Patient, Example**

| | |
|---|---|
| **DOB** | 12/10/1954 |
| **Gender:** | Male |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Shwachman-Diamond Syndrome (SBDS) Sequencing

ARUP test code 2006240

EER Shwachman-Diamond Syndrome (SBDS)

See Note

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

[redacted]

Shwachman-Diamond Syndrome (SBDS)

Negative

Date Test(s) Started: 12-SEP-2023 10:39:08
Sample Source: Blood in EDTA Date Collected: 12-SEP-2023 Date Received:
12-SEP-2023 Testing Date Started: 12-SEP-2023 Date Reported: 12-SEP-2023
Provider Account #: A.R.U.P Laboratories Additional Provider:
Test(s) Requested SBDS Gene / Shwachman-Diamond Syndrome (SDS)
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. It is possible this
individual has a pathogenic variant that is not detectable by this analysis or
is in a gene not evaluated by this test.
Recommendation(s) Genetic counseling is recommended to discuss the implications
of these results.
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.
Genes Evaluated SBDS
Methods Using genomic DNA from the submitted specimen, the coding regions and

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

splice junctions of the requested gene were PCR amplified and capillary
sequencing was performed. Bi-directional sequence was assembled, aligned to
reference gene sequences based on human genome build GRCh37/UCSC hg19, and
analyzed for sequence variants. Capillary sequencing or another appropriate
method was used to confirm all potentially pathogenic variants. If present,
apparently homozygous variants were confirmed using alternate primer pairs to
significantly reduce the possibility of allele drop-out. Sequence alterations
were reported according to the Human Genome Variation Society (HGVS)
nomenclature guidelines. Reportable variants include pathogenic variants,
likely pathogenic variants and variants of uncertain significance. Likely
benign and benign variants are not routinely reported but are available upon
request. The methods used by GeneDx are expected to be greater than 99%
sensitive in detecting variants identifiable by sequencing. Available evidence
for variant classification may change over time and reported variant(s) may be
reclassified according to the ACMG/AMP Standards and Guidelines (PMID:
25741868), which may lead to issuing a revised report.
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. The
methods used cannot reliably detect deletions of 20bp to 500bp in size, or
insertions of 10bp to 500bp in size. Sequencing cannot detect low-level
mosaicism. The copy number assessment methods used with this test cannot
reliably detect mosaicism and cannot identify balanced chromosome aberrations.
Regions of certain genes have inherent sequence properties (for example:
repeat, homology, or pseudogene regions, high GC content, rare polymorphisms)
that yield suboptimal data, potentially impairing accuracy of the results.
Inaccurate results may occur in the setting of allogeneic bone marrow/stem cell
transplantation, active or chronic hematologic conditions, recent blood
transfusion, suboptimal DNA quality, or in other rare circumstances. Rarely,
incidental findings of large chromosomal rearrangements outside the gene of
interest may be identified. 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. In addition, the chance of an
erroneous result due to laboratory errors incurred during any phase of testing
cannot be completely excluded. Interpretations are made with the assumption
that any clinical information provided, including family

relationships, are
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);
###

Report electronically signed by: LIMS Cardiology
Performed by: GeneDx
207 Perry Parkway
Gaithersburg, MD 20877

Anne Maddalena, Ph.D.,FACMG,

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| EER Shwachman-Diamond Syndrome (SBDS) | 23-248-106422 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Shwachman-Diamond Syndrome (SBDS) | 23-248-106422 | 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**