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

Physician: Doctor, Example

Patient: Patient, Example

DOB	2/24/2019
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

Ordering Physician Name	NOT PROVIDED Performed by: GeneDx 207 Perry Parkway Gaithersburg, MD 20877 Anne Maddalena, Ph.D.,FACMG,
Ordering Physician Phone Number	NOT PROVIDED Performed by: GeneDx 207 Perry Parkway Gaithersburg, MD 20877 Anne Maddalena, Ph.D.,FACMG,
Shwachman-Diamond Syndrome (SBDS)	NEGATIVE Date Test(s) Started: 7/21/2021 10:27:51 Test(s) Requested SBDS Gene / Shwachman-Diamond Syndrome (SDS) Result(s): NEGATIVE Result Paragraph No reportable variant was identified in exons 1-5 of the SBDS gene of the submitted specimen. Interpretation Boocock et al. identified pathogenic variants in 240 of the 316 alleles in 158 unrelated patients with Shwachman-Diamond syndrome. Approximately 74% of those pathogenic variants were attributable to gene conversion events whereby certain pathogenic variants are copied into the SBDS gene from a nearby nonfunctional copy of the SBDS gene. Other pathogenic variants described include various isolated missense and frameshift variants. Over 90% of patients with Shwachman-Diamond syndrome are expected to have at least one detectable pathogenic variant in the SBDS gene. Recommendation(s) Genetic counseling is recommended. 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

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

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
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 the reported variant(s) may
be re-classified according to the AMP/ACMG guidelines for variant classification (Richards et al. 2015), which may lead to re-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 with this test.
This test cannot reliably detect mosaicism. Capillary sequencing cannot
reliably detect exon-level deletion/duplications, if present, nor will it
detect variants deep within the introns or in the regulatory regions unless
specifically indicated in the methods. If performed, copy number assessment may
not reliably detect chromosomal aberrations and deletions/insertions of less
than 500 bp, and may incidentally reveal large chromosome rearrangements
outside the gene of interest. Some genes have inherent sequence properties (for
example: repeat, homology, or pseudogene regions, high GC content, rare
polymorphisms) that may result in suboptimal data, potentially impairing
accuracy of the results. False negative results may also occur in the setting
of bone marrow transplantation, recent blood transfusion, or suboptimal DNA

quality. 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. The chance of a false positive or false negative 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);Boocock GRB et
al., (2003) Nat Genet. 33: 97-101.
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S S S
Report electronically signed by: Gabriele Richard MD, FACMG
Performed by: GeneDx
207 Perry Parkway
Gaithersburg, MD 20877

Anne Maddalena, Ph.D.,FACMG,

EER Shwachman-Diamond Syndrome (SBDS)

See Note

Access ARUP Enhanced Report using the link below:

-Direct access: [redacted]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Ordering Physician Name	21-197-105883	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Ordering Physician Phone Number	21-197-105883	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Shwachman-Diamond Syndrome (SBDS)	21-197-105883	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Shwachman-Diamond Syndrome (SBDS)	21-197-105883	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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