Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: TEST,

Patient: HMYE NGS NEG EXAMPLE,

DOB	
Sex:	Male
Patient Identifiers:	57391
Visit Number (FIN):	57781
Collection Date:	2/20/2024 07:52

Hereditary Myeloid Neoplasms Panel, Sequencing

ARUP test code 3001842

Hereditary Myeloid Neoplasms Specimen	Whole Blood
Hereditary Myeloid Neoplasms Interp	Negative

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INDICATION FOR TESTING
History of Ewing sarcoma and pancytopenia.
RESULT
No pathogenic variants were detected in any of the genes tested.
INTERPRETATION
No pathogenic variants or variants of uncertain significance
were identified by massively parallel sequencing of the coding
regions and exon-intron boundaries of the genes tested. This
result decreases the likelihood of, but does not exclude, a
heritable form of myeloid neoplasm. Please refer to the
background information included in this report for a list of the
genes analyzed and limitations of this test.
RECOMMENDATIONS
Medical screening and management should rely on clinical
findings and family history. Genetic consultation is recommended.
COMMENTS
Likely benign and benign variants are not included in this
report.
Variants in the following region(s) may not be detected by NGS
with sufficient confidence in this sample due to technical
limitations; reportable variants are confirmed by Sanger
sequencing:
NONE
This result has been reviewed and approved by


BACKGROUND INFORMATION: Hereditary Myeloid Neoplasms Panel,
                         Sequencing
CHARACTERISTICS: While the majority of myeloid neoplasms and
malignancies occur sporadically due to somatic mutations, a
portion are due to inherited or hereditary predispositions.
Individuals with an inherited predisposition to myeloid
neoplasms may present at a younger age, with more than one
first-degree relative with myelodysplastic syndrome (MDS) or
acute myeloid leukemia (AML), solid tumors, and/or a family
history of physical findings associated with a known cancer
predisposition syndrome.

EPIDEMIOLOGY: MDS occurs in approximately 4.5 per 100,000
individuals in the general population. MDS is rare in children
and young adults; approximately 50 percent of childhood MDS is
associated with an inherited cause. AML occurs in approximately
3.7 per 100,000 individuals in the general population.

CAUSE: Pathogenic germline variants in genes associated with
predisposition to MDS and/or AML.
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H=High, L=Low, *=Abnormal, C=Critical

INHERITANCE: Variable, dependent on gene/condition.

GENES TESTED: ANKRD26*, ATM, BLM, CBL, CEBPA, DDX41, ELANE, ETV6, GATA1, GATA2, KRAS, NBN, PTPN11*, RUNX1, SAMD9, SAMD9L, SRP72*, TERC, TERT, TP53
*One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

METHODOLOGY: Targeted capture of all coding exons and exon-intron junctions of the targeted genes (unless otherwise specified in the limitations section below), followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants that do not meet acceptable quality metrics. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of cancer nor a heritable form of myeloid neoplasm. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants and deep intronic variants will not be identified unless specifically targeted for their clinical relevance. Deletions/duplications/insertions of any size may not be detected by massive parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay is not intended to detect somatic variants associated with hematologic malignancy, though such variants may be detected incidentally. Though this test is designed to identify germline variants associated with predisposition to myeloid neoplasms, it cannot definitively determine the germline or somatic origin of detected variants when the patient has a hematologic malignancy and the assay is performed on blood or other tissue that may be contaminated by malignant cells. In addition, this assay may not detect low-level mosaic or somatic variants associated with disease, including variants that have undergone somatic reversion. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

The following regions are not sequenced due to technical limitations of the assay:
ANKRD26 (NM_014915) exon 19
PTPN11 (NM_002834) exon 9
SRP72 (NM_006947) exon 19

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Hereditary Myeloid Neoplasms Specimen	24-051-100926	2/20/2024 7:52:00 AM	2/20/2024 7:52:26 AM	2/20/2024 7:56:00 AM
Hereditary Myeloid Neoplasms Interp	24-051-100926	2/20/2024 7:52:00 AM	2/20/2024 7:52:26 AM	2/20/2024 7:56:00 AM

END OF CHART

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