Patient ID	Patient Name		Birth Date	Gender	Age
321	TESTRNV, IMPLEMENTATION		1970-11-13	M	47
Order Number	Client Order Number	Ordering Physician	Report Notes		
X100265123	X100265123	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		24 Aug 2018 08:00			

NGS for Myeloid Neoplasms (NGSHM)

Specimen Type

MCR

Bone marrow

Indication for Test

MCR

AML

Pathogenic Mutations Detected

MCR

None. See below for Variants of Unknown Significance and Additional Notes. Please see the section of "Panel Gene List" below for the complete list of genes tested.

Interpretation

MCR

No pathogenic genetic alteration is detected in the listed gene regions. This finding does not exclude the presence of genetic alteration occurring at allele frequency below our established detection limit of 5–10%, or other genetic alterations present in untested gene regions.

Variants of Unknown Significance

MCR

None

The VUS variants listed here (with approximate variant allele %) are not sufficiently characterized in the current literature and are therefore of uncertain clinical significance at this time. They are reported here for future reference in the event they become clinically significant in the light of new scientific data.

Clinical Trials

MCR

Information regarding possible clinical trials for this patient can be found at the following sites:

1). ClinicalTrials.gov:
http://clinicaltrials.gov/ct2/search/advanced

2). Mayo Clinic:
http://www.mayo.edu/research/clinical-trials

3). National Cancer Institute:
http://www.cancer.gov/clinicaltrials/search

4). Molecular Match:
https://www.molecularmatch.com/

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
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Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100265123	X100265123	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		24 Aug 2018 08:00			

Additional Notes

MCR

None

Method Summary

1 MCR

DNA is extracted from the peripheral blood or bone marrow sample and following library preparation by hybrid capture, subjected to next generation sequencing (NGS) with post-sequencing analysis of tumor-associated mutations. Because some regions in the target panel are not consistently resolved by NGS, additional separate clinical laboratory assays may also be used to complete the overall analysis including: CALR (CALR), CEBPA (CEBPA), CSF3R (CSF3R), FLT3 (FLT), JAK2 V617F Mutation Detection (JAK2B, JAK2M, or JAK2V), JAK2 Exon 12 Mutation Detection (JAKXB or JAKXM), KIT (KITB, KITBM, KITAS, or KITE), MPL (MPLB or MPLM), NPM1 (NPM1), and TP53 (P53CA).

Performance characteristics of NGS panel:
Single base substitutions: accuracy >99%; reproducibility 100% (intra- and interassay); sensitivity 5–10% variant allele fraction with a minimum depth coverage of 250X. Insertion/deletion events: accuracy >99%; reproducibility 100% (intra- and interassay); sensitivity 5–10% variant allele fraction with a minimum depth coverage of 250X.

Disclaimer

MCR

CLINICAL DISCLAIMER

Mutation calls detected between 5–10% variant allele fractions (VAF) may indicate low-level (i.e. subclonal) tumor populations, although the clinical significance of these findings may not be clear. Some apparent mutations classified as VUS may represent rare or low frequency polymorphisms. A low incidence of gene mutations associated with myeloid neoplasms can also be detected in hematopoietic cells with advancing age in some individuals without evidence of a hematologic malignancy ("clonal hematopoiesis") and these alterations may not be clearly distinguishable from tumor-associated mutations (PMID 25326804;25426838;25426837). Prior treatment for hematologic malignancy could affect the results obtained in this assay. In particular, prior allogeneic hematopoietic stem cell transplant (HSCT) may cause difficulties in resolving somatic or polymorphic alterations, or in assigning variant calls correctly to donor and recipient fractions, if pertinent clinical or laboratory information (e.g. chimerism engraftment status) is not provided. Correlation with clinical, histopathologic and additional laboratory findings is required for final interpretation of these results. This assay does not distinguish between somatic and germ line alterations in analyzed gene regions, particularly with VAF near 50% or 100%. If nucleotide alterations in genes associated with germ line mutation syndromes are present and there is also a strong clinical suspicion or family history of malignant disease predisposition, additional genetic testing and appropriate counseling may be indicated. This report interpretation is based on current medical and scientific literature, but clinical significance may not be completely established for all reported target gene abnormalities identified. The final interpretation of results for clinical management of the patient is the responsibility of the managing physician.

TECHNICAL DISCLAIMER

The depth of sequencing coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria, or for failed regions are noted. Analysis of rare (low allele frequency) polymorphisms may be problematic in some cases. A low tumor cell percentage in the sample may affect the true mutation VAF and/or sensitivity. Suboptimal- performing regions (i.e. less than the expected minimum depth of coverage) may affect analytic sensitivity for detecting lower level mutations. This is a qualitative test. The variant read fractions are provided for information only and represent a relative proportion of mutated alleles, but do not indicate a measure of analytical sensitivity for the given genes; assay sensitivity is as stated in the method summary. Some genetic or genomic alterations, such as large insertion/deletion (indel) events, copy number alterations (CNA) and gene translocation events are not detected by this assay.

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Account Information		**Collected**			
C7028846 DLMP Rochester		24 Aug 2018 08:00			

OncoHeme Panel Gene List

MCR

ANKRD26 (NM_014915.2) 5'UTR, exons 1–4, intron c.172, ASXL1 (NM_015338.5) exons 10–13, BCOR (NM_001123385.1) exons 4–15, CALR (NM_004343.3) exon 9, CBL (NM_005188.3) intron 7 last 110bps before start of exon 8, exon 8, intron 8, CEBPA (NM_004364.4) exon 1, CSF3R (NM_000760.3) exons 14 and 17, DDX41 (NM_016222.2) exons 1–17, DNMT3A (NM_022552.4) exons 8–23, ELANE (NM_001972.2) exons 1–5, ETNK1 (NM_018638.4) exons 2–5, ETV6 (NM_001987.4) exons 3–8, EZH2 (NM_004456.4) exons 2–20, FLT3(NM_004119.2) exons 14–20, GATA1 (NM_002049.3) exons 2 and 4, GATA2 (NM_001145661.1) exons 3–7, intron 5, c.1017+1–1017+730, IDH1 (NM_005896.3) exon 4, IDH2 (NM_002168.3) exon 4, JAK2 (NM_004972.3) exons 12–16, KDM6A (UTX) (NM_021140.3) exons 1–29, KIT (NM_000222.2) exons 8–11 and 17, KRAS (NM_033360.3) exons 2–3, MPL (NM_005373.2) exons 10–12, NPM1 (NM_002520.6) exons 9–11, to -30 before exon 11, NRAS (NM_002524.4) exons 2 and 3, PHF6 (NM_001015877.1) exons 2–10, PTPN11 (NM_002834.3) exons 3–4 and 12–13, RAD21 (NM_006265.2) exons 1, 2, 4–7, 9–11, 13, 14, exon 10 flank 15bp, RUNX1(NM_001001890.2) exons 1–6, intron 4 c.725–13T>A and intron 5 c.886+1–4del, SETBP1 (NM_015559.2) partial exon 4; amino acids 400 - 950, SH2B3 (LNK) (NM_005475.2) exon 2–8, SF3B1 (NM_012433.2) exons 13–16, SRP72 (NM_006947.3) exons 6, 10, SMC3 (NM_005445.3) exons 7, 8, 13, 17, 19, 21, 29, SRSF2 (NM_003016.4) exons 1 and 2, STAG2 (NM_001042750.1) exons 4–34, 12, 17 and 22 flank 15bp, TERT (NM_198253.2) exons 2–16, TET2 (NM_001127208.2) exons 3–11, TP53 (NM_000546.4) exons 4–9, U2AF1(NM_001025203.1) exons 2, 6, and 8, WT1 (NM_024426.2) exons 1–10, and ZRSR2 (NM_005089.3) exons 1–11.

For some genes, the transcript IDs used in this analysis may not be the same as in other cancer mutation databases, such as COSMIC (http://cancer.sanger.ac.uk/cosmic).

Reviewed By:

MCR

Signing Pathologist: BENJAMIN BAILEY

NGSHM Result

MCR

See Interpretation

Received: 24 Aug 2018 14:41 **Reported:** 24 Aug 2018 15:04

Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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Code	Laboratory	Address
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905