

Patient ID SA00154531	Patient Name TESTINGRNV, MDSDF-ABNORMAL	Birth Date 1960-11-02	Sex M	Age 61
Order Number SA00154531	Client Order Number SA00154531	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 20 Sep 2022 08:22		

MDS, Diagnostic FISH

Result Summary

MCR

Abnormal

Interpretation

MCR

The result is abnormal and indicates a 5q deletion in 75% of nuclei.

A 5q deletion is a common chromosome abnormality in both de novo and therapy-related myeloid malignancies, particularly MDS and AML. TP53 mutation status by sequencing should be considered as TP53 mutations are present in a significant subset of MDS cases with an isolated 5q deletion and are associated with increased risk of leukemic transformation and shorter survival (Swerdlow et al., WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC Press:Lyon 2017; page 116).

Clinical and pathologic correlation is recommended.

For monitoring response to therapy, FISH for chromosome 5 is available, test MDSMF [MDS, Specified FISH].

Result Table

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Abnormality Name	Result	Abn%	Cutoff%
-5q31(D5S630x2,EGR1x1)	Abnormal	75	<12.0
inv(3) RPN1/MECOM fusion	Normal		<4.0
-17p13.1(TP53x1,D17Z1x2)	Normal		<15.0
-17(TP53,D17Z1)x1	Normal		<10.0
+8(D8Z2,MYC)x3	Normal		<7.0
-20q12(D20S108x1,20qterx2)	Normal		<12.0
-5(D5S630,EGR1)x1	Normal		<10.0
-7q31(D7Z1x2,D7S486x1)	Normal		<12.0
-7(D7Z1,D7S486)x1	Normal		<10.0

Result

MCR

nuc ish(D5S630x2,EGR1x1)[75/100]

Reason for Referral

MCR

thrombocytopenia

Specimen

MCR

Bone Marrow

Source

MCR

Left iliac crest

Method

MCR

Locus and probes	[Strategy;#Nuclei;Class]
3q21(RPN1[GATA2]), 3q26.2(MECOM)	[DFISH;200;LDT]
5p15.2(D5S630), 5q31(EGR1)	[COPY#;100;AM]
7CEN(D7Z1), 7q31(D7S486)	[COPY#;100;AM]
8CEN(D8Z2), 8q24.1(MYC)	[COPY#;100;AM]
17p13(TP53), 17CEN(D17Z1)	[COPY#;100;AM]
20q12(D20S108), 20qter	[COPY#;100;AM/C]

Probe strategies include:
DFISH=dual color, double fusion;
COPY#=region gain and loss.
Scoring Method: Manual

Probe vendors include:
LDT = Mayo Clinic Developed
AM = Abbott Molecular, Inc (Des Plaines, IL)
C = Cytocell, Inc. (Cambridge, UK)

Disclaimer

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Applicable to Analyte Specific Reagent (ASR) and Laboratory Developed Tests (LDT). This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the U.S. Food and Drug Administration. This FISH test does not rule out other chromosome abnormalities.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



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Released By

MCR

Jess F. Peterson, M.D.

Received: 21 Sep 2022 08:34

Reported: 21 Sep 2022 08:52

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Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292