MAYO CLINIC LABORATORIES

1-800-533-1710

IDHQ

IDH1 and IDH2, Quant, ddPCR, V

Patient ID	Patient Name		Birth Date	Sex	Age
SA00162550	TESTING, IDHQ ABNORMAL		2001-01-01	F	22
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00162550	SA00162550	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		03 Oct 2023 00:00			

IDH1 and IDH2, Quant, ddPCR, V

Specimen Type

MCR

Bone marrow

Interpretation

[1] MCR

Bone marrow, IDH1 and IDH2 Quantitative detection:
Positive. IDH1 R132C mutation detected, at 5.2% (mutated/total IDH1). See comment.

Comment: IDH1 and IDH2 missense mutations are seen in approximately 5–33% of de novo acute myeloid leukemia (AML), 7–25% secondary AML, and 4–12% of myelodysplastic syndrome (MDS). IDH mutations in AML are predominantly associated with intermediate-risk karyotype and their impact on AML prognosis remains controversial (Patel et al., 2012, 22417203; Papaemmanuil et al., 2016, 27276561; Haferlach et al., 2014, 2420272). The FDA has approved ivosidenib monotherapy and in combination with azacitidine for the treatment of newly diagnosed IDH1-mutated AML (patients ≥ 75 years old or who have comorbidities that preclude the use of intensive induction chemotherapy), and ivosidenib and olutasidenib for relapsed/refractory AML in adult patients. The FDA has approved enasidenib (AG-221) for the treatment of IDH2-mutated relapsed/refractory AML in adult patients.

ADDITIONAL INFORMATION

Method Summary: Hotspot mutations in IDH1 (R132H, R132S, R132C, R132G, R132P, R132L) and IDH2 (R140W, R140G, R140L, R140Q, R172K, R172M, R172G, R172W, R172S) are evaluated by digital droplet PCR on extracted DNA. The analytical sensitivity of the assay is 0.5% (mutated/total IDH1 or mutated/total IDH2 copies).The reproducibility of this assay is such that results within 0.5 log change (3.16 fold) should be considered equivalent.

Signing Pathologist

MCR

LAUREN SCHULTZ

Received: 03 Oct 2023 11:35 **Reported:** 03 Oct 2023 11:38

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.

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