

Patient ID SA00172629	Patient Name AGDD, NORMAL	Birth Date 1985-11-25	Sex F	Age 38
Order Number SA00172629	Client Order Number SA00172629	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 30 Oct 2024 08:00		

Alpha Globin Cluster Locus Del/Dup

Result Summary

MCR

NEGATIVE

Result

MCR

No deletions or duplications within the alpha globin gene cluster were identified.

Interpretation

MCR

This result reduces the likelihood but does not rule out a diagnosis of or positive carrier status for alpha thalassemia.

This assay does NOT detect non-deletion types of mutations such as point mutations other than the alphaT Saudi and Hb Constant Spring. Therefore, this result should be interpreted in the context of clinical presentation and results of other laboratory tests [e.g. hemoglobin electrophoresis and mean corpuscular volume (MCV)].

A genetic consultation may be of benefit.

Specimen

MCR

WB Whole Blood

Method

MCR

Dosage analysis (PCR and MLPA) was used to detect deletion and duplication-type mutations. This method uses multiple probes that hybridize throughout the alpha-gene locus on chromosome 16 from the HS-40 regulatory region through the 3' hypervariable region (3'HVR).

Disclaimer

1 MCR

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

One or more in silico tools were used to assist in the interpretation of these results. These tools are updated regularly and predictions for a given variant may change. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

Released By

MCR

LINDA HASADSRI

Received: 31 Oct 2024 11:05

Reported: 05 Nov 2024 16:04

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.

Performing Site Legend

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