

Patient ID SA00172960	Patient Name VALIDATIONSOF, TESTING	Birth Date 1980-04-25	Sex M	Age 44
Order Number SA00172960	Client Order Number SA00172960	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 18 Nov 2024 08:00		

Beta Globin Gene Cluster, Del/Dup,V

Beta Globin Gene Cluster Del/Dup

MCR

Negative. See interpretation

Specimen

MCR

WB Whole Blood

Reviewed by

MCR

JENNIFER MAIN

Interpretation

1 MCR

Negative result. No large deletions or duplications were detected in the beta globin cluster locus.

Molecular genetic interpretation seen in consultation with Linda Hasadsri, M.D., Ph.D.

ADDITIONAL INFORMATION

Multiplex Ligation-dependent Probe Amplification (MLPA) was used to detect large deletion and duplication-type mutations within the beta-globin cluster locus on chromosome 11. This method uses multiple probes that hybridize throughout the region including the 5'HS/locus control region (LCR), epsilon

(HBE), G-gamma (HBG2), A-gamma (HBG1), pseudobeta (HBBP1), delta (HBD), and beta (HBB) globin genes to beyond 3'HS1. This assay only detects large deletions and duplications and does not detect point mutations or other small mutations. Test resolution is limited by probe density and copy number variations present in between adjacent probes will not be detected. This test provides a size range and does not confirm breakpoints or definitive size of the mutation; therefore, multiple different mutations can give the same result. Rare polymorphisms exist that could lead to false-negative or false-positive results. Isolated HBG1/HBG2 regional deletions/duplications are considered benign polymorphisms and will not be reported.

It is imperative to interpret the results in the context of hemoglobin electrophoresis and red blood cell indices findings. Clinical and family history correlation are important to establish the significance of test results. If results obtained do not match the clinical findings, additional testing should be considered.

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

Received: 19 Nov 2024 09:29

Reported: 19 Nov 2024 09:52

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