Patient ID	Patient Name		Birth Date	Gender	Age
SA00147546	TESTINGRNV, REPORT		1999-09-09	M	21
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00147546	SA00147546	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		31 Aug 2021 07:00			

Alpha Globin Gene Sequencing, B

Alpha Globin Gene Sequencing, B

MCR

see interpretation

Interpretation

[1] MCR

Alpha-1 Globin Sequencing: Negative
Alpha-2 Globin Sequencing: Positive

Gene: HBA2
Legacy: Alpha2 3'UTR+81, G>T
HGVS: c.*81G>T
Genomic: g.223680G>T
Normal allele absent (Homo/Hemizygous)
Classification: Alteration of uncertain significance

The most common alpha thalassemia mutations are not excluded as this method does not detect large deletion or duplication events. If the normal allele is absent, this method cannot distinguish between a homozygous state (the same alteration on both alleles) and a hemizygous state (the detected alteration present on one allele in combination with the absence of the genomic location, e.g. a large deletion, on the other allele). If multiple alterations are present, this method cannot distinguish between cis (same allele) or in trans (different alleles) configurations. These limitations have implications for clinical phenotype, inheritance and genetic counseling. If alpha globin gene deletion/duplication testing is desired, please order Alpha-Globin Gene Analysis (test code ATHAL). Additional sample required.

Alterations are reported in Legacy and HGVS nomenclatures and by genomic location. For more information about these findings, please see HbVar: A database of Human Hemoglobin Variants and Thalassemias at http://globin.cse.psu.edu.
Classification of hemoglobin disorders by genetic data alone may result in incomplete conclusions which may significantly impact overall disorder classification and expected phenotype. Correlation with hemoglobin electrophoresis results, red blood cell indices and clinical/family history is required. If detected, alterations classified as Benign or Likely Benign that are not expected to result in a hemoglobin variant are not included in this report but are available upon request. Genetic counseling may be of benefit to assist in the interpretation of these results.

Signing Pathologist: BENJAMIN BAILEY

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and non-coding portions of the alpha-1 hemoglobin (HBA1) and alpha-2 hemoglobin (HBA2) genes with reported mutations. HGVS mutation nomenclature is based on human assembly GRCh37(hg19) and RefSeq accession numbers NM_000558.4 (HBA1) and NM_000517.4 (HBA2).

Received: 01 Sep 2021 09:20 **Reported:** 01 Sep 2021 10:59

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