

Patient ID SA00100507	Patient Name TESTINGRNA, REPORT	Birth Date 1999-11-11	Gender M	Age 17
Order Number SA00100507	Client Order Number SA00100507	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Nov 2017 06:00		

Beta Globin Gene Sequencing, B

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MCR

<http://globin.cse.psu.edu>

see interpretation

Interpretation

1 MCR

Beta globin gene seq: (HBB): Positive. See below.

Gene: HBB

Legacy nomenclature: Beta promoter region -29, A>G

HGVS nomenclature: c.-79A>G

Allele status: Heterozygous

Classification: Beta plus thalassemia mutation

This test does not exclude large deletions or duplications involving the beta globin gene. If multiple alterations are present, this method cannot distinguish between in cis (same allele) or trans (different alleles) configurations. This may have implications for clinical phenotype, inheritance and genetic counseling. For more information about the findings, please see HbVar: A database of Human Hemoglobin Variants and Thalassemias at

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 are not included in this report but are available upon request.

Signing Pathologist: Melissa Tricker-Klar

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 beta hemoglobin gene (HBB) with reported mutations. HGVS mutation nomenclature is based on human assembly GRCh37(hg19) and RefSeq accession number NM_000518.4.

Received: 08 Nov 2017 16:05

Reported: 08 Nov 2017 16:33

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