

Patient ID SA00100509	Patient Name TESTINGRNV, REPORT	Birth Date 1999-11-11	Gender M	Age 17
Order Number SA00100509	Client Order Number SA00100509	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Nov 2017 06:00		

Gamma Globin Full Gene Sequencing

Gamma Globin Gene Sequencing Result

MCR

see interpretation

Gamma Globin Interpretation

1 MCR

G-gamma gene seq (HBG2): Negative. See below.

A-gamma gene seq (HBG1): Positive. See below.

Gene: HBG1

Legacy nomenclature: A-gamma 5, GAG>AAG, Glu>Lys

HGVS nomenclature: c.16G>A, p.E6K

Allele status: Heterozygous

Classification: Hb F-Texas-I mutation

These results do not exclude large deletions or duplications involving the gamma globin genes. For more information about the 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/protein analysis 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 gamma-1 hemoglobin (HBG1) and gamma-2 (HBG2) genes with reported mutations. HGVS mutation nomenclature is based on human assembly GRCh37 (hg19) and RefSeq accession numbers NM_000559.2 (HBG1) and NM_000184.2 (HBG2).

Received: 08 Nov 2017 16:05

Reported: 08 Nov 2017 16:35

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