MAYO CLINIC LABORATORIES

1-800-533-1710

PKLRG

PKLR Full Gene and Deletion

Patient ID	Patient Name		Birth Date	Gender	Age
SA00082430	TESTINGRNV, PKLRG NEG		1981-06-02	F	35
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00082430	SA00082430	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Dec 2016 00:00			

PKLR Full Gene and Deletion

Result Summary

MCR

Negative

Result Details

MCR

No sequence variants or large genomic deletions were detected in the PKLR gene.

Interpretation

MCR

This result greatly decreases the likelihood of, but does not completely exclude, pyruvate kinase (PK) deficiency.

This result does not exclude the presence of another variant that may be responsible for the clinical features present in this individual. Some individuals who have features of PK deficiency may have a pathogenic variant in the PKLR gene that is not detectable by the described testing methodology. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant(s) in another gene(s).

This result should be interpreted in the context of clinical findings, family history, pyruvate kinase enzymatic levels, and other laboratory testing. If testing was performed due to a family history of PK deficiency, genetic testing of an affected family member is necessary to determine whether this test is of predictive value for this family. Identification of the pathogenic variant in this family would allow for more accurate risk assessment of at-risk family members.

A list of common, benign variants identified for this patient is available from the lab upon request.

Reference: Zanella A, Fermo E, Bianchi P, Chiarelli LR, Valentini G. Pyruvate kinase deficiency: the genotype-phenotype association. Blood Rev. 2007 Jul;21(4):217–31. Epub 2007 Mar 13. Review. PMID: 17360088.

Method

MCR

Bi-directional DNA sequence analysis was used to test for the presence of variations in the PKLR gene (transcripts NM_000298.2 and NM_181871.3 for all exons and intron-exon boundaries). Fragment analysis was used to detect large deletions within intron 2 up through the 3' untranslated region of the PKLR gene.

Disclaimer

MCR

CAUTIONS:

Rare variants may be present that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Deletion testing for this assay will report the size the deletion but does not yield information about the location of the deletion in the PKLR gene.

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

Rarely, individuals may have a variant in the gene tested that is not identifiable by the described testing methodology. In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing. 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

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Reviewed by MCR

Mary Karow

Received: 08 Dec 2016 13:23 **Reported:** 08 Dec 2016 13:29

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