MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

PKLRG

PKLR Full Gene and Deletion

Patient ID	Patient Name		Birth Date	Gender	Age
SA00324244	TESTINGRNV, PKLRG		1981-06-02	F	35
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00324244	SA00324244	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 Feb 2017 00:00			

PKLR Full Gene and Deletion

Result Summary

MCR

Positive

Result Details

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The following variant was detected in PKLR:
Genomic position: g.155264430
cDNA change: c.808C>T
Amino acid change: p.Arg270X
The patient is heterozygous for this variant
Classification: Pathogenic

In addition, a heterozygous large deletion was detected in the PKLR gene.

Interpretation

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The c.808C>T (p.Arg270X) variant in the PKLR gene is a known pathogenic variant.

The presence of a large deletion is predicted to be pathogenic.

Pyruvate kinase (PK) deficiency is an autosomal recessive condition. The identification of a both a pathogenic variant and a large deletion in the PKLR gene is supportive of a diagnosis of PK deficiency but must be correlated with clinical findings, family history, and other laboratory testing, especially as this testing method cannot determine if these mutations are in cis (on the same allele) or in trans (on different alleles). PK deficiency is usually associated with markedly decreased PK enzyme activity levels; however, some affected individuals have only mildly depressed or even normal enzyme activity (paradoxically, this can occur in those who are most severely anemic). If this individual's phenotype is consistent with a compound heterozygous genotype, screening and/or management procedures should be considered. A genetic consultation may be of benefit.

Since pathogenic or likely pathogenic variants have been identified in the PKLR gene in this individual, genetic testing for this variant in first-degree relatives is available. Please contact the laboratory at 1–800–533–1710 or use the online test catalog at www.mayomedicallaboratories.com for information about how to order the test for PKLR Full Gene and Deletion, B for a family member of this individual. Please include the variant identified in this individual with your request.

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

References:
1. 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

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Bi-directional DNA sequence analysis was used to test for the presence of variations in the PKLR gene (transcript NM_000298.2) 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

1 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 presence or absence of a large 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.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
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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.

Reviewed by MCR

Mary Karow

Received: 07 Feb 2017 12:39 **Reported:** 07 Feb 2017 12:43

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
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905