



PKLRZ

PATIENT NAME HOSPICES, KAMI Q				ORDER NUMBER O604000509
PATIENT ID 11256341	DATE OF BIRTH 08/30/1951	AGE 73 Y	SEX Female	REQUESTED BY CYBELE ABAD
COLLECTED 9/4/2024, 2:59 PM	RECEIVED 9/4/2024, 2:59 PM	REPORTED 9/5/2024, 1:57 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location.				
RODO6D RST RODO 06 D Rochester MN 55905				

TEST DESCRIPTION

Evaluation of PKLR gene associated with hereditary nonspherocytic hemolytic anemia and pyruvate kinase deficiency

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Negative

RESULT

No reportable variants were detected.

INTERPRETATION

This result decreases the likelihood but does not rule out the presence of a variant in the PKLR gene.

Individuals may have a pathogenic variant in the PKLR gene that is not detectable by the methods utilized. Additionally, pathogenic variants in other genes not interrogated by this assay may cause a similar phenotype.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. (PK) enzyme activity assays (preferably as a part of EEEV1 / Red Blood Cell [RBC] Enzyme Evaluation, Blood)).

Given this result, additional genetic testing, such as (PK) enzyme activity assays (preferably as a part of EEEV1 / Red Blood Cell [RBC] Enzyme Evaluation, Blood), may be considered.

A genetic consultation may be of benefit.

METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the gene analyzed. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth >30X. Sensitivity is estimated at >99% for single nucleotide variants, >94% for indels up to 39 base pairs, >95% for deletions up to 75 base pairs and insertions up to 47 base pairs. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the gene analyzed. See the Genes Analyzed field for a list of gene(s) tested.



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There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID PKLRZ) for details regarding genes with regions not routinely covered.

GENES ANALYZED

PKLR

DISCLAIMER

Clinical Correlations

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

Technical Limitations

Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

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Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Reclassification of Variants Policy



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See www.mayocliniclabs.com (TEST ID PKLRZ) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation

Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

TEST CLASSIFICATION

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.

RELEASED BY
BENJAMIN VANSTEINBURG

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