MAYO CLINIC

1-800-533-1710

Hereditary Renal Cancer Panel

RENCP

PATIENT NAME				ORDER NUMBER
RENCP1, TESTING				**K704028073**
PATIENT ID M195599206	DATE OF BIRTH 05/15/1990	AGE 34 Y	SEX Male	REQUESTED BY CLIENT UNKNOWN
COLLECTED 12/4/2024, 7:30 AM	RECEIVED 12/5/2024, 7:50 AM	REPORTED 12/16/2024, 12:45 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL Rochester Campus Rochester MN 55901				CLIENT ORDER NUMBER M195599206 CLIENT MRN C7028846-0002033

TEST DESCRIPTION

Evaluation of 19 genes associated with hereditary renal cancer

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Pathogenic Variant Detected

RESULT

Gene (Transcript)	Variant	Variant Type	Zygosity	Classification
FLCN NM_144997.7 GRCh37	Exon 11 Chr17 :g.17119716dup c.1285dup p.His429Profs*27 (p.H429Pfs*27)	Duplication	Heterozygous	PATHOGENIC

The following PATHOGENIC variant was detected:
FLCN (NM_144997.7), chr17(GRCh37):g.17119716dup, c.1285dup, p.His429Profs*27 (p.H429Pfs*27), heterozygous

INTERPRETATION

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

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

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coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known pathogenic variants. NGS, multiplex ligation-dependent probe amplification (MLPA) and/or a polymerase chain reaction (PCR)-based quantitative method was performed to test for the presence of deletions and duplications in the genes 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 over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for indels less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. See the Genes Analyzed field for a list of genes tested.

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 RENCP) for details regarding genes with regions not routinely covered.

GENES ANALYZED

BAP1, DICER1, FH, FLCN, MET, MITF (c.952G>A p.E318K variant only), PTEN (including promoter), SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TSC1, TSC2 and VHL

DISCLAIMER

Clinical Correlations

The gene(s) analyzed by this test may have more than one associated phenotype or inheritance pattern. Additionally, the genetic variants detected may have reduced penetrance, variable expressivity, or different classifications based on disease state. Classification and reporting of the variants detected is based on the intended disease state of the test ordered. For more information regarding the phenotypic spectrum which may be involved for a specific gene, see OMIM (www.ncbi.nlm.nih.gov/omim) or GeneReviews (www.genereviews.org).

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

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diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

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.

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.

Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the Hereditary Renal Cancer Panel in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

Reclassification of Variants Policy
See www.mayocliniclabs.com (TEST ID RENCP) 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
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