MAYO CLINIC

1-800-533-1710

CFTR Gene, Full Gene Analysis

CFTRN

PATIENT NAME				ORDER NUMBER
TESTRNV, IMPLEMENTATION				**M702000287**
PATIENT ID X100407759	DATE OF BIRTH 02/19/1968	AGE 54 Y	SEX Male	REQUESTED BY ATLAS TEST
COLLECTED 2/2/2023, 8:33 AM	RECEIVED 2/7/2023, 3:04 PM	REPORTED 2/24/2023, 4:18 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER X100407759 CLIENT MRN 321

TEST DESCRIPTION

Evaluation of CFTR gene associated with cystic fibrosis

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Pathogenic Variant Detected

RESULT

Gene (Transcript)	Variant	Zygosity	Classification
CFTR (NM_000492.3)	c.1521_1523del p.Phe508del (p.F508del) Chr7(GRCh37):g.117199646_117199648del	Heterozygous	Pathogenic

The following heterozygous PATHOGENIC variant was detected:
CFTR (NM_000492.3), p.F508del (p.Phe508del), c.1521_1523del, Chr7(GRCh37):g.117199646_117199648del

Intron 9 poly-T alleles: 7T/9T (c.1210-12T[7]/c.1210-12T[9])

INTERPRETATION

CFTR c.1521_1523del (p.F508del), PATHOGENIC
The heterozygous c.1521_1523del (p.F508del) deletion in the CFTR gene (MIM:602421) was detected and is an established pathogenic variant. This variant is also known as c.1653_1655del. Pathogenic variants in the CFTR gene have been associated with autosomal recessive cystic fibrosis (CF), CFTR-related disorder (CFRD), CFTR-related metabolic syndrome (CRMS) and late-onset CF (1). This result is supportive of carrier status for a CFTR-gene related condition. However, the presence of an undetectable second variant cannot be ruled out. Clinical correlation is recommended.

Individuals may have a pathogenic variant in the CFTR 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. sweat chloride testing).

Of note, current data suggests that some patients with a clinical diagnosis of CF may respond to variant-specific therapies depending on the combination of variants.

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CFTR Intron 9 poly T tract alleles (5T-11, 12 or 13TG/7T/9T) are polymorphic variants. The 5T-11TG, 7T and 9T alleles are considered benign. The 5T-12TG and 5T-13TG alleles are considered clinically significant only when detected in the homozygous state or with a pathogenic CFTR variant.

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family. Genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

REFERENCES:
1) GeneReviews: Cystic Fibrosis and Congenital Absence of the Vas Deferens (https://www.ncbi.nlm.nih.gov/books/NBK1250/) (PMID: 20301428)

ALTERNATE NOMENCLATURE
The intron 9 poly T alleles were previously known as intron 8 poly T alleles, using historic intron/exon numbering.

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.

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

GENES ANALYZED

CFTR

DISCLAIMER

Clinical Correlations

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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.

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.

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

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RELEASED BY

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