

LASTNAME, FIRSTNAME

DOB: mm/dd/yyyy

Account Number:00000000



Patient ID:

Age: 00

Ordering Physician:

Specimen ID: 000-000-0000-0

Sex: Female

Date Collected: mm/dd/yyyy

Date Received: mm/dd/yyyy

Date Reported: mm/dd/yyyy

Date Entered: mm/dd/yyyy

Specimen Type: Cultured Amniotic Fluid Cells Ethnicity: Not Provided

Indication: Prenatal Test / Family history - both parents carriers

Cystic Fibrosis (CF), 97 Variants, Fetal Analysis

Summary: ■ POSITIVE

SAMPLE REPORT

Variants Detected

Disorder (Gene)	Result	Interpretation
Cystic fibrosis (CFTR) NM_000492.3	POSITIVE: PREDICT AFFECTED Homozygous for c.1521_1523delCTT (p.Phe508del), pathogenic, CHR7:117199645-117199648.	This fetus is predicted to be affected. Genetic counseling is recommended.

Comparison of maternal and fetal DNA markers indicates that maternal cell contamination is unlikely to have interfered with the reported fetal result (maternal specimen # 000000000000). The result obtained from a multiple gestation pregnancy depends on the successful sampling of the intended fetus.

Recommendations

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of positive results, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Labcorp Genetic Counselors please visit <https://womenshealth.labcorp.com/genetic-counseling> or call (855) GC-CALLS (855-422-2557).

Additional Clinical Information

Cystic fibrosis (CF) is an autosomal recessive disorder with variable severity and age at onset. Signs and symptoms of classic CF may include elevated sweat chloride levels, progressive lung disease, pancreatic insufficiency, and male infertility. Symptoms of mild CF may include pancreatic sufficiency. Symptoms of CFTR-related disorders may include pancreatitis, bronchiectasis, and isolated male infertility due to congenital absence of the vas deferens (CBAVD). Treatment is dietary and supportive. Genotype-targeted therapies may be available for some individuals. In severely affected individuals, lung transplantation may be indicated. (Ong, PMID:20301428).

Comments

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder(s) tested. Information about the disorder(s) tested is available at <https://womenshealth.labcorp.com>.

Methods/Limitations

Maternal Cell Contamination Analysis: DNA is isolated and amplified by the polymerase chain reaction (PCR). Fifteen polymorphic markers from 14 chromosomes are analyzed by capillary gel electrophoresis and fluorescence detection. Markers analyzed include TPOX, D3S1358, FGA, D5S818, CSF1PO, D7S820, D8S1179, THO1, vWA, D13S317, Penta E, D16S539, D18S51, D21S11, and Penta D. The analytical sensitivity of the assay is approximately 10%; maternal cell contamination present at a lower percentage may not be detected.

Next-generation Sequencing: Genomic regions of interest in the CFTR gene are selected using the Twist Biosciences® hybridization capture method and sequenced via the Illumina® next generation sequencing platform. Sequencing reads are aligned with the human genome reference GRCh37/hg19 build. Regions of interest include genomic regions encompassing targeted variants. Analytical sensitivity at 30X coverage is estimated to be >99% for single nucleotide variants, >99% for insertions/deletions less than six base pairs and >96% for insertions/deletions between six and forty-five base pairs. Variant detection is performed by QIAGEN CLC Genomics and in-house algorithms. Confirmatory testing is done by Sanger sequencing. Variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, <http://www.hgvs.org/>). Variant classification and confirmation are consistent with ACMG standards and guidelines (Richards, PMID:25741868; Rehm, PMID:23887774).

Analysis is restricted to 97 targeted CF variants, listed below.

c.54-5940_273+10250del21kb, c.178G>T (p.Glu60*), c.223C>T (p.Arg75*), c.254G>A (p.Gly85Glu), c.262_263delTT (p.Leu88Ilefs*22), c.273+1G>A, c.273+3A>C, c.274-1G>A, c.274G>T (p.Glu92*), c.313delA (p.Ile105Serfs*2), c.325_327delinsG (p.Tyr109Glyfs*4), c.349C>T (p.Arg117Cys), c.350G>A (p.Arg117His), c.366T>A (p.Tyr122*), c.442delA (p.Ile148Leufs*5), c.489+1G>T, c.531delT (p.Ile177Metfs*12), c.532G>A (p.Gly178Arg), c.579+1G>T, c.579+5G>A, c.580-1G>T, c.617T>G (p.Leu206Trp), c.803delA (p.Asn268Ilefs*17), c.805_806delAT (p.Ile269Profs*4), c.935_937delTCT (p.Phe312del), c.948delT (p.Phe316Leufs*12), c.988G>T (p.Gly330*), c.1000C>T (p.Arg334Trp), c.1013C>T (p.Thr338Ile), c.1040G>A (p.Arg347His), c.1040G>C (p.Arg347Pro), c.1055G>A (p.Arg352Gln), c.[1075C>A;1079C>A] (p.[Gln359Lys;Thr360Lys]), c.1155_1156dupTA (p.Asn386Ilefs*3), c.1364C>A (p.Ala455Glu), c.1438G>T (p.Gly480Cys), c.1477C>T (p.Gln493*), c.1519_1521delATC (p.Ile507del), c.1521_1523delCTT (p.Phe508del), c.1545_1546delTA (p.Tyr515*), c.1558G>T (p.Val520Phe), c.1572C>A (p.Cys524*), c.1585-1G>A, c.1624G>T (p.Gly542*), c.1646G>A (p.Ser549Asn), c.1647T>G (p.Ser549Arg), c.1652G>A (p.Gly551Asp), c.1654C>T (p.Gln552*), c.1657C>T (p.Arg553*), c.1675G>A (p.Ala559Thr), c.1679G>C (p.Arg560Thr), c.1680-1G>A, c.1721C>A (p.Pro574His),

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Cystic Fibrosis (CF), 97 Variants, Fetal Analysis

Methods/Limitations (Cont.)

c.1766+1G>A, c.1766+5G>T, c.1820_1903del84 (p.Met607_Gln634del), c.1911delG (p.Gln637Hisfs*26), c.1923_1931delinsA (p.Ser641Argfs*5), c.1973_1985delinsAGAAA (p.Arg658Lysfs*4), c.1976delA (p.Asn659Ilefs*4), c.2012delT (p.Leu671*), c.2051_2052delinsG (p.Lys684Serfs*38), c.2052delA (p.Lys684Asnfs*38), c.2052dupA (p.Gln685Thrfs*4), c.2125C>T (p.Arg709*), c.2128A>T (p.Lys710*), c.2175dupA (p.Glu726Argfs*4), c.2290C>T (p.Arg764*), c.2657+5G>A, c.2668C>T (p.Gln890*), c.2737_2738insG (p.Tyr913*), c.2988G>A (p.Gln996=), c.2988+1G>A, c.3039delC (p.Tyr1014Thrfs*9), c.3067_3072delATAGTG (p.Ile1023_Val1024del), c.3196C>T (p.Arg1066Cys), c.3266G>A (p.Trp1089*), c.3276C>A (p.Tyr1092*), c.3276C>G (p.Tyr1092*), c.3302T>A (p.Met1101Lys), c.3454G>C (p.Asp1152His), c.3472C>T (p.Arg1158*), c.3484C>T (p.Arg1162*), c.3528delC (p.Lys1177Serfs*15), c.3536_3539delCCAA (p.Thr1179Asnfs*12), c.3587C>G (p.Ser1196*), c.3611G>A (p.Trp1204*), c.3659delC (p.Thr1220Lysfs*8), c.3712C>T (p.Gln1238*), c.3718-2477C>T, c.3744delA (p.Lys1250Argfs*9), c.3752G>A (p.Ser1251Asn), c.3764C>A (p.Ser1255*), c.3773dupT (p.Leu1258Phefs*7), c.3846G>A (p.Trp1282*), c.3889dupT (p.Ser1297Phefs*5), c.3909C>G (p.Asn1303Lys)

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants, or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships.

Information Table

Cystic fibrosis, 97 variants, detection rates for prenatal testing

Population	Detection rate
African American	81%
Ashkenazi Jewish	97%
Asian American	55%
Caucasian	93%
Hispanic	78%
Mixed or other ethnic background	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.

References

Deignan JL, Astbury C, Cutting GR *et al.* CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 22, 1288 (2020). PMID: 32404922

Ong T, Marshall SG, Karczeski BA, *et al.* Cystic Fibrosis and Congenital Absence of the Vas Deferens. 2001 Mar26 [Updated 2017 Feb 2]. In: Adam MP, Ardinger HH, Pagon RA, *et al.*, editors. GeneReviews® [Internet]. PMID: 20301428

Performing Labs

Component Type	Performed at	Laboratory Director
Technical component, processing	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC 27709-0150	Laboratory Director
Technical component, analysis	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC 27709-0150	Laboratory Director
Professional component	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC 27709-0150	Laboratory Director

For inquiries, the physician may contact the lab at 800-345-4363

This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration.

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LASTNAME, FIRSTNAME

DOB: mm/dd/yyyy

Account Number: 00000000



Patient ID:

Age: 00

Ordering Physician:

Specimen ID: 000-000-0000-0

Sex: Female

Cystic Fibrosis (CF), 97 Variants, Fetal Analysis

Patient Details

LASTNAME, FIRSTNAME

Phone:

Date of Birth: mm/dd/yyyy

Age: 00

Sex: Female

Patient ID:

Alternate Patient ID:

Physician Details

CLIENT NAME

CLIENT ADDRESS

Phone: 00000000

Account Number: 00000000

Physician ID:

NPI:

Specimen Details

Specimen ID: 000-000-0000-0

Control ID:

Alternate Control Number:

Date Collected: mm/dd/yyyy 0000 Local

Date Received: mm/dd/yyyy 1858 ET

Date Entered: mm/dd/yyyy 0908 ET

Date Reported: mm/dd/yyyy 0954 ET

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