ARUP LABORATORIES | 800-522-2787 | aruplab.com
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	8/20/1993
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Cystic Fibrosis (CFTR) Expanded Variant Panel, Fetal

ARUP test code 2013662

Cystic Fibrosis, Allele 1	Negative
Cystic Fibrosis, Allele 2	Negative
Cystic Fibrosis 5T Variant	Not Applicable
CF, Expanded Var Pan Fetal, Interp	0 variants

Specimen: Direct Amnio
Symptoms: No
Family History: No

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Cystic Fibrosis (CFTR) Expanded Variant Panel, Fetal
CHARACTERISTICS OF CYSTIC FIBROSIS (CF): Chronic sinopulmonary disease, gastrointestinal malabsorption/pancreatic insufficiency, and obstructive azoospermia. Symptoms of a CFTR-related disorder include: pancreatitis, bilateral absence of the vas deferens, nasal polyposis, and bronchiectasis.
INCIDENCE: 1 in 2,300 Ashkenazi Jewish, 1 in 2,500 Caucasians, 1 in 13,500 Hispanics, 1 in 15,100 African Americans, 1 in 35,100 Asians.
INHERITANCE: Autosomal recessive.
PENETRANCE: High for severe pathogenic variants and variable for variants of varying clinical consequences.
Cause of CF: Two severe pathogenic CFTR variants on opposite chromosomes.
CAUSE OF CFTR-RELATED DISORDERS: Two pathogenic CFTR variants on opposite chromosomes, at least one of which is classified as mild or a variant of varying clinical consequences.
VARIANTS TESTED:
*Note: variants are listed by standard nomenclature. Legacy names are also provided for the 23 recommended ACMG variants.
c.1A>G, p.Met1Val; c.54-5940_273+10250del21kb, Exons 2-3del; c.115C>T, p.Gln39X; c.178G>T, p.Glu60X; c.200C>T, p.Pro67Leu; c.223C>T, p.Arg75X; c.254G>A (Legacy G85E), p.Gly85Glu; c.262_263delTT, p.Leu88IlefsX22 (aka p.Leu88fs); c.273+1G>A, Intronic; c.273+3A>C, Intronic; c.274-1G>A, Intronic; c.274G>A, p.Glu92Lys; c.274G>T, p.Glu92X; c.292C>T, p.Gln98X; c.313delA, p.Ile105SerfsX2 (aka p.Ile105fs); c.325_327delTATinsG,

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p.Tyr109GlyfsX4 (aka p.Tyr109fs); c.328G>C, p.Asp110His; c.349C>T, p.Arg117Cys; c.350G>A (Legacy R117H), p.Arg117His; c.366T>A, p.Tyr122X; c.442delA, p.Ile148LeufsX5 (aka p.Ile148fs); c.489+1G>T (Legacy 621+1G>T), Intronic; c.531delT, p.Ile177MetfsX12 (aka p.Ile177fs); c.532G>A, p.Gly178Arg; c.579+1G>T (Legacy 711+1G>T), Intronic; c.579+5G>A, Intronic; c.579+3A>G, Intronic; c.580-1G>T, Intronic; c.595C>T, p.His199Tyr; c.613C>T, p.Pro205Ser; c.617T>G, p.Leu206Trp; c.658C>T, p.Gln220X; c.680T>G, p.Leu227Arg; c.722_743del, p.Gly241GlufsX13 (aka p.Gly241fs); c.803delA, p.Asn268IlefsX17 (aka p.Asn268fs); c.805_806delAT, p.Ile269ProfsX4 (aka p.Ile269fs); c.935_937delTCT, p.Phe312del; c.948delT, p.Phe316LeufsX12 (aka p.Phe316fs); c.988G>T, p.Gly330X; c.1000C>T (Legacy R334W), p.Arg334Trp; c.1007T>A, p.Ile336Lys; c.1021T>C, p.Ser341Pro; c.1021_1022dupTC, p.Phe342HisfsX28 (aka p.Phe342fs); c.1040G>A, p.Arg347His; c.1040G>C (Legacy R347P), p.Arg347Pro; c.1055G>A, p.Arg352Gln; c.1081delT, p.Trp361GlyfsX8 (aka p.Trp361fs); c.1116+1G>A, Intronic; c.1130dupA, p.Gln378AlafsX4 (aka p.Gln378fs); c.1155_1156dupTA, p.Asn386IlefsX3 (aka p.Asn386fs); c.1202G>A, p.Trp401X; c.1203G>A, p.Trp401X; c.1209+1G>A, Intronic; c.1327_1330dupGATA, p.Ile444ArgfsX3 (aka p.Ile444fs); c.1340delA, p.Lys447ArgfsX2 (aka p.Lys447fs); c.1364C>A (Legacy A455E), p.Ala455Glu; c.1393-1G>A, Intronic; c.1397C>A, p.Ser466X; c.1397C>G, p.Ser466X; c.1400T>C, p.Leu467Pro; c.1418delG, p.Gly473GlufsX54 (aka p.Gly473fs); c.1438G>T, p.Gly480Cys; c.1466C>A, p.Ser489X; c.1475C>T, p.Ser492Phe; c.1477C>T, p.Gln493X; c.1519_1521delATC (Legacy I507del), p.Ile507del; c.1521_1523delCTT (Legacy F508del), p.Phe508del; c.1545_1546delTA, p.Tyr515X; c.1558G>T, p.Val520Phe; c.1572C>A, p.Cys524X; c.1573C>T, p.Gln525X; c.1585-1G>A (Legacy 1717-1G>A), Intronic; c.1585-8G>A, Intronic; c.1624G>T (Legacy G542X), p.Gly542X; c.1645A>C, p.Ser549Arg; c.1646G>A, p.Ser549Asn; c.1647T>G, p.Ser549Arg; c.1651G>A, p.Gly551Ser; c.1652G>A (Legacy G551D), p.Gly551Asp; c.1654C>T, p.Gln552X; c.1657C>T (Legacy R553X), p.Arg553X; c.1675G>A, p.Ala559Thr; c.1679G>A, p.Arg560Lys; c.1679G>C (Legacy R560T), p.Arg560Thr; c.1680-886A>G, Intronic; c.1680-1G>A, Intronic; c.1703delT, p.Leu568CysfsX4 (aka p.Leu568fs); c.1705T>G, p.Tyr569Asp; c.1721C>A, p.Pro574His; c.1753G>T, p.Glu585X; c.1766+1G>A (Legacy 1898+1G>A), Intronic; c.1766+3A>G, Intronic; c.1792_1798delAAAACTA, p.Lys598GlyfsX11 (aka p.Lys598fs); c.1911delG, p.Gln637HisfsX26 (aka p.Gln637fs); c.1923_1931del9insA, p.Ser641ArgfsX5 (aka p.Ser641fs); c.1973_1985del13insAGAAA, p.Arg658LysfsX4 (aka p.Arg658fs); c.1976delA, p.Asn659IlefsX4 (aka p.Asn659fs); c.2012delT, p.Leu671X; c.2051_2052del, p.Lys684ThrfsX4; c.2051_2052delinsG (aka c.2051_2delinsG), p.Lys684SerfsX38; c.2052delA (Legacy 2184delA), p.Lys684AsnfsX38; c.2125C>T, p.Arg709X; c.2128A>T, p.Lys710X; c.2175dupA, p.Glu726ArgfsX4 (aka p.Glu726fs); c.2195T>G, p.Leu732X; c.2215delG, p.Val739TyrfsX16 (aka p.Val739fs); c.2290C>T, p.Arg764Ter; c.2453delT, p.Leu818TrpfsX3 (aka p.Leu818fs); c.2464G>T, p.Glu822X; c.2490+1G>A, Intronic; c.2491G>T, p.Glu831X; c.2537G>A, p.Trp846X; c.2538G>A, p.Trp846X; c.2551C>T, p.Arg851X; c.2583delT, p.Phe861LeufsX3 (aka p.Phe861fs); c.2657+5G>A (Legacy 2789+5G>A), Intronic; c.2668C>T, p.Gln890X; c.2737_2738insG, p.Tyr913X; c.2780T>C, p.Leu927Pro; c.2810dupT, p.Val938GlyfsX37 (aka p.Val938fs); c.2834C>T, p.Ser945Leu; c.2875delG, p.Ala959HisfsX9 (aka p.Ala959fs); c.2908G>C, p.Gly970Arg; c.2988+1G>A (Legacy 3120+1G>A), Intronic; c.2988G>A, Intronic; c.2989-1G>A, Intronic; c.3039delC, p.Tyr1014ThrfsX9 (aka p.Tyr1014fs); c.3067_3072delATAGTG, p.Ile1023_Val1024del (aka I1023_V1024del); c.3140-26A>G, Intronic; c.3194T>C, p.Leu1065Pro; c.3196C>T, p.Arg1066Cys; c.3197G>A, p.Arg1066His; c.3230T>C, p.Leu1077Pro; c.3266G>A, p.Trp1089X; c.3276C>A, p.Tyr1092X; c.3276C>G, p.Tyr1092X; c.3302T>A, p.Met1101Lys; c.3310G>T, p.Glu1104X; c.3472C>T, p.Arg1158X; c.3484C>T (Legacy R1162X), p.Arg1162X; c.3528delC (Legacy 3659delC), p.Lys1177SerfsX15 (aka p.Lys1177fs); c.3532_3535dupTCAA, p.Thr1179IlefsX17 (aka p.Thr1179fs); c.3587C>G, p.Ser1196X; c.3611G>A, p.Trp1204X; c.3612G>A, p.Trp1204X; c.3659delC, p.Thr1220LysfsX8 (aka p.Thr1220fs); c.3691delT, p.Ser1231ProfsX4 (aka p.Ser1231fs);

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	c.3712C>T, p.Gln1238X; c.3718-2477C>T (Legacy 3849+10kbC>T), Intronic; c.3731G>A, p.Gly1244Glu; c.3744delA, p.Lys1250ArgfsX9 (aka p.Lys1250fs); c.3752G>A, p.Ser1251Asn; c.3763T>C, p.Ser1255Pro; c.3764C>A, p.Ser1255X; c.3773dupT, p.Leu1258PhefsX7 (aka p.Leu1258fs); c.3846G>A (Legacy W1282X), p.Trp1282X; c.3873+1G>A, Intronic; c.3909C>G (Legacy N1303K), p.Asn1303Lys; c.3937C>T, p.Gln1313X; c.3964-78_4242+577del, Exons 22-23del; c.4025_4028dup, p.Cys1344GlyfsX16(aka p.C1344fs); c.4046G>A, p.Gly1349Asp; c.4077_4080delTGTTinsAA, p.Val1360fsX3 (aka p.Val1360fs); c.4111G>T, p.Glu1371X; c.4251delA, p.Glu1418ArgfsX14 (aka p.Glu1418fs). The IVS-8 variant, c.1210-12[5], will be reported only when R117H is detected or in patients who are reported to be symptomatic. CLINICAL SENSITIVITY: Ashkenazi Jewish 96 percent; Caucasian 92 percent; Hispanic 80 percent; African American 78 percent; Asian American 55 percent. METHODOLOGY: Matrix-Assisted Laser Desorption Ionization-Time of Flight (MALDI-TOF) ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent. LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Only the CFTR variants listed above and 5T variant will be interrogated. This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes. Counseling and informed consent are recommended for genetic testing. Consent forms are available online.
Maternal Contamination Study Fetal Spec	Fetal Cells Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cystic Fibrosis, Allele 1	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystic Fibrosis, Allele 2	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystic Fibrosis 5T Variant	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
CF, Expanded Var Pan Fetal, Interp	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	23-233-101599	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical