



Phenotype Report

Name: EUR NA12877 Father
Genome ID: NA12877-200-37-ASM

Sequencing Provider: Complete Genomics
Sequencing Type: Whole Genome

Phenotype: Cystic Fibrosis

Description:

CFTR-related disorders include cystic fibrosis (CF) and congenital absence of the vas deferens (CAVD). Cystic fibrosis affects epithelia of the respiratory tract, exocrine pancreas, intestine, male genital tract, hepatobiliary system, and exocrine sweat glands, resulting in complex multisystem disease. Pulmonary disease is the major cause of morbidity and mortality in CF. Affected individuals have lower airway inflammation and chronic endobronchial infection, progressing to end-stage lung disease characterized by extensive airway damage (bronchiectasis, cysts, and abscesses) and fibrosis of lung parenchyma. Meconium ileus occurs at birth in 15%-20% of newborns with CF. Pancreatic insufficiency with malabsorption occurs in the great majority of individuals with CF. More than 95% of males with CF are infertile as a result of azoospermia caused by absent, atrophic, or fibrotic Wolffian duct structures. CAVD occurs in men without pulmonary or gastrointestinal manifestations of CF. Affected men have azoospermia and are thus infertile.

Modes of inheritance*: Autosomal Recessive

Variation Summary:

Variant Classification	Num. Variants Found	Num. Positions Not Called (Missing data)	Num. Positions Matching Reference	Num. Variants Assessed
Pathogenic	<u>1</u>	3 (1.4%)	216 (98.2%)	220
Likely pathogenic	<u>0</u>	2 (4.1%)	47 (95.9%)	49
Risk factor	<u>0</u>	1 (100%)	0 (0.0%)	1

Total number of phenotype variations assessed: 270

Variation Details:

• Chr7: 117,227,792 G>A

Pathogenic ★★★★★[†]

Zygosity: Heterozygous dbSNP ID: rs76713772

Population Allele Frequency: <0.01%

Gene Impact: **CFTR** SPLICE DISRUPT

Supporting Publications: <http://www.ncbi.nlm.nih.gov/pubmed/11280952>
<http://www.ncbi.nlm.nih.gov/pubmed/2236053>
<http://www.ncbi.nlm.nih.gov/pubmed/2210769>

Limitations:

There are several limitations to the results presented here:

- **6** variation(s) known to be related to this phenotype were not called in the sequencing data. This data is missing and would likely require additional sequencing to determine.
- The quality of the sequencing data has not been verified by Enlis. It is possible that variations listed here do not actually exist in the listed genome, and it is possible that positions listed as matching reference are actually variant.
- Knowledge of the genomic factors that relate to this phenotype are incomplete. New variations and genes that relate to this phenotype may be discovered in the future. Variations and genes currently thought to be related to this phenotype may be found to be incorrect. Care should especially be taken in the interpretation of variations with confidence star levels of only 1 or 2.
- The Enlis Genome Research application, and all associated communication (Enlis Products) are for research and educational purposes only. Enlis Products together and separately do not constitute medical advice and should not be used as a source of professional medical advice or diagnosis.

Endnotes

* Mode of inheritance definitions:

Autosomal Recessive

Autosomal recessive inheritance refers to genetic conditions that occur only when mutations are present in both copies of a given gene (i.e., the person is homozygous for a mutation, or carries two different mutations of the same gene, a state referred to as compound heterozygosity).

† Confidence Star Levels:



Review Status: Classified by single submitter with no evidence provided, or multiple conflicting interpretations



Review Status: Classified by single submitter with evidence



Review Status: Classified by multiple submitters



Review Status: Reviewed by expert panel



Review Status: Reviewed by professional society

Human genome reference version: [HomoSapiens_GRCh37](#)

Appendix - Full Position List:

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,120,047 >A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,149 >G</u>	rs397508328	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,150 >C</u>	rs397508476	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,162 >T</u>	rs193922501	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,167 >T</u>	rs121909045	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,192 INS TT</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,120,202 >T</u>	rs397508746	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,144,367 >G</u>	rs193922498	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,144,368 >T</u>	rs397508168	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,144,378 >T</u>	rs143456784	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,085 >T</u>	rs200337193	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,094 >A</u>	rs121909025	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,101 >T</u>	rs77284892	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,123 >T</u>	rs368505753	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,143 >T</u>	rs115545701	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,146 >T</u>	rs121908749	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,177 >A</u>	rs75961395	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,182-117,149,183 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,194 >A</u>	rs121908750	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,197 >A</u>	rs121908791	CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,149,199 >C</u>	rs74467662	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,149,200 >G</u>	rs387906374	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,170,952 >A</u>	rs121908792	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,170,953 >T</u>	rs121908751	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,170,953 >A</u>	rs121908751	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,170,971 >T</u>	rs397508461	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,004-117,171,006 >G</u>	rs121908798	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,005 >G</u>	rs121909031	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,007 >C</u>	rs113993958	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,028 >T</u>	rs77834169	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,029 >A</u>	rs78655421	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,034 >G</u>	rs193922518	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,045 >A</u>	rs79660178	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,049 >C</u>	rs193922519	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,058 INS TTA</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,103 DEL</u>	rs387906363	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,104 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,121 DEL</u>	rs121908770	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,138-117,171,155 DEL</u>	rs387906371	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,171,169 >T</u>	rs78756941	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,174,370 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,174,372 >A</u>	rs80282562	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,174,417 >T</u>	rs397508759	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,174,420 >T</u>	rs77188391	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,174,422 >G</u>	rs397508761	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,174,424 >A</u>	rs78440224	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,301 >T</u>	rs121908793	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,314 >A</u>	rs193922529	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,317 >T</u>	rs121908802	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,335 >T</u>	rs121908803	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,339 >G</u>	rs121908752	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,372 >G</u>	rs121909046	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,380 >T</u>	rs397508778	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,442-117,175,463 DEL</u>	rs121908804	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,175,466 >C</u>	rs397508791	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,588-117,176,599 DEL</u>	rs387906367	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,658 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,661-117,176,662 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,683 >G</u>	rs193922532	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,686 >A</u>	rs397508799	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,688 >A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,176,703 INS A</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,704 >T</u>	rs142864834	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,706-117,176,718 >TG</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,711 >T</u>	rs151073129	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,176,715 INS A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,211-117,180,213 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,217 >G</u>	rs121909016	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,230 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,242 >G</u>	rs144476686	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,272 >T</u>	rs79031340	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,281 >T</u>	rs193922533	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,284 >T</u>	rs121909011	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,291 >A</u>	rs397508139	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,297 >T</u>	rs77409459	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,303 INS TC</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,305 >C</u>	rs397508144	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,306 >TC</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,313 DEL</u>	rs121908774	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,324 >C</u>	rs77932196	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,324 >A</u>	rs77932196	CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,180,324 >T</u>	rs77932196	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,330 >T</u>	rs121909021	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,338 >T</u>	rs193922497	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,339 >A</u>	rs121908753	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,359 >A</u>	rs76879328	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,363 >A</u>	rs75053309	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,365 DEL</u>	rs387906361	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,366 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,373-117,180,374 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,180,401 >A</u>	rs397508158	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,182,070 >A</u>	rs556880586	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,182,080 INS A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,182,155 >A</u>	rs397508174	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,182,156 >A</u>	rs397508175	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,182,163 >A</u>	rs397508176	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,188,683-117,188,684 DEL</u>	rs562195055	CFTR	Pathogenic	Cystic fibrosis		Missing Data	
<u>Chr7: 117,188,684 >G</u>	rs73715573	CFTR	Likely pathogenic	Cystic fibrosis		Missing Data	
<u>Chr7: 117,188,811 INS AGAT</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,188,812 >T</u>	rs147422190	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,188,849 >A</u>	rs74551128	CFTR	Pathogenic	Cystic fibrosis		Missing Data	

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,188,852 >C</u>	rs193922500	CFTR	Likely pathogenic	Cystic fibrosis		Missing Data	
<u>Chr7: 117,188,858 >T</u>	rs121909009	CFTR	Pathogenic	Cystic fibrosis		Missing Data	
<u>Chr7: 117,199,517 >A</u>	rs397508200	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,522 >G</u>	rs121908805	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,522 >A</u>	rs121908805	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,525 >C</u>	rs139573311	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,563 >T</u>	rs79282516	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,591 >A</u>	rs397508211	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,600 >T</u>	rs121909017	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,602 >T</u>	rs77101217	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,602-117,199,603 DEL</u>	rs121908775	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,641-117,199,643 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,645-117,199,647 DEL</u>	rs199826652	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,667-117,199,668 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,683 >T</u>	rs77646904	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,199,697 >A</u>	rs121908754	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,785 >A</u>	rs193922503	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,792 >A</u>	rs76713772	CFTR	Pathogenic	Cystic fibrosis	Zygosity 1		
<u>Chr7: 117,227,809 >A</u>	rs387906368	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,832 >T</u>	rs113993959	CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,227,853 >C</u>	rs121908757	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,854 >A</u>	rs121908755	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,854 >T</u>	rs121908755	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,855 >G</u>	rs121909005	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,859 >A</u>	rs121909013	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,860 >A</u>	rs75527207	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,862 >T</u>	rs76554633	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,865 >T</u>	rs74597325	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,866 >A</u>	rs121909044	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,874 >G</u>	rs75789129	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,881 >C</u>	rs193922504	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,883 >A</u>	rs75549581	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,887 >C</u>	rs80055610	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,227,887 >A</u>	rs80055610	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,406 >A</u>	rs121908794	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,409 >A</u>	rs121909047	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,414 >A</u>	rs121909006	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,417 DEL</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,448 >A</u>	rs121908758	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,458 >T</u>	rs55928397	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,480 >T</u>	rs397508296	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,230,494 >A</u>	rs121908748	CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,230,496 >G</u>	rs397508298	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,038-117,232,121 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,074 >C</u>	rs139468767	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,132 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,164 >T</u>	rs121909033	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,232 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,257 >A</u>	rs397508333	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,267 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,267 INS A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,272-117,232,273 >G</u>	rs121908799	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,273-117,232,274 >AC</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,346 >T</u>	rs121908760	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,349 >T</u>	rs75115087	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,367 >T</u>	rs121909023	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,395 INS A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,416 >G</u>	rs397508350	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,436 DEL</u>	rs397508353	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,511 >T</u>	rs121908810	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,512 DEL</u>	rs387906376	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,638 INS AT</u>		CFTR	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,232,664 >T</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,673 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,685 >T</u>	rs397508378	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,700 >T</u>	rs121909018	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,232,712 >A</u>	rs141158996	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,234,984 >T</u>	rs397508387	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,235,030 >A</u>	rs397508393	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,235,031 >A</u>	rs267606722	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,235,044 >T</u>	rs121909012	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,235,072 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,235,090 >A</u>	rs193922506	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,242,919 INS A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,242,922 >A</u>	rs80224560	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,596 >T</u>	rs79633941	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,628 >A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,663 >T</u>	rs121909034	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,665 INS G</u>	rs397508424	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,666 >G</u>	rs121909008	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,696 >A</u>	rs193922509	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,708 >C</u>	rs397508435	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,738 INS T</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,243,762 >T</u>	rs397508442	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,773 >T</u>	rs121909035	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,803 DEL</u>	rs397508447	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,243,836 >C</u>	rs397508453	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,246,751 >T</u>	rs193922514	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,246,807 >A</u>	rs121908797	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,246,808 >A</u>	rs75096551	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,571 >T</u>	rs193922515	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,572 >A</u>	rs397508470	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,606 DEL</u>	rs397508482	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,622 >A</u>	rs193922516	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,647-117,250,652 DEL</u>	rs397508492	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,679 >G</u>	rs144055758	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,250,723-117,250,724 DEL</u>	rs397508505	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,251,609 >G</u>	rs76151804	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,251,656 DEL</u>	rs387906377	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7:</u> <u>117,251,679</u> <u>INS CTATG</u>		CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,689 >C</u>	rs121909036	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,691 >T</u>	rs78194216	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,692 >A</u>	rs121909019	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,694 >A</u>	rs121909020	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,704 >A</u>	rs78769542	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,707 >C</u>	rs121909037	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,717 >A</u>	rs186045772	CFTR, AC000111. 6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,725 >C</u>	rs139304906	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,749 >G</u>	rs79635528	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,761 >A</u>	rs78802634	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,771 >A</u>	rs121908761	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,771 >G</u>	rs121908761	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,797 >A</u>	rs36210737	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,251,805 >T</u>	rs397508538	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,254,665 >G</u>		CFTR, AC000111. 6	Likely pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7:</u> <u>117,254,753 >C</u>	rs75541969	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,254,767 >A</u>	rs139729994	CFTR, AC000111. 6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,556 >C</u>	rs373002889	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,579 >T</u>	rs79850223	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,591 >T</u>	rs74767530	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,597</u> <u>INS T</u>		CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,634</u> <u>DEL</u>		CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,639</u> <u>INS TCAA</u>		CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,694 >G</u>	rs121908763	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,718 >A</u>	rs121908764	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,719 >A</u>	rs121908765	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,766 >T</u>	rs1800123	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,766</u> <u>DEL</u>	rs121908811	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,796</u> <u>DEL</u>		CFTR, AC000111. 6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,807 >G</u>	rs75389940	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,819 >T</u>	rs121908766	CFTR, AC000111. 6	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7:</u> <u>117,267,828 >G</u>	rs387906362	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,267,829 >A</u>	rs193922520	CFTR, AC000111.6	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,280,015 >T</u>	rs75039782	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,491 >A</u>	rs387906369	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,505 >A</u>	rs267606723	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,505 >T</u>	rs267606723	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,518</u> <u>DEL</u>		CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,520 >A</u>	rs121909040	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,526 >A</u>	rs74503330	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,537 >C</u>	rs121909041	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,538 >A</u>	rs76649725	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,541</u> <u>INS C</u>	rs397508605	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,542</u> <u>INS T</u>		CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,620 >A</u>	rs77010898	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,622 >T</u>	rs77902683	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7:</u> <u>117,282,631 >C</u>	rs121909028	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7: 117,282,647 >C</u>	rs121909015	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,282,648 >A</u>	rs143570767	CFTR, AC000111.6	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,900-117,292,903 DEL</u>	rs387906373	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,906 INS T</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,915 >C</u>	rs193922522	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,929 >C</u>	rs121909042	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,931 >G</u>	rs80034486	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,959 >T</u>	rs121909026	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,969 >A</u>	rs121909010	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,979 INS AGGG</u>		CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,292,986 >A</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,304,775 >T</u>	rs193922524	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,304,824 >A</u>	rs193922525	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,304,834 >C</u>	rs113857788	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,304,855-117,304,858 >AA</u>	rs397508668	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,304,889 >T</u>	rs397508675	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,305,619 >T</u>	rs372227120	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,305,620 >C</u>	rs193922526	CFTR	Likely pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,306,969 DEL</u>		CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr7: 117,306,999 >C</u>	rs193922528	CFTR	Likely pathogenic	Cystic fibrosis			Reference

Position and Variation	dbSNP ID	Gene	Clinical Significance	Phenotype	Variation Found	Position Not Called	Position Matches Reference
<u>Chr7:</u> <u>117,307,145 >T</u>	rs374705585	CFTR	Pathogenic	Cystic fibrosis			Reference
<u>Chr19:</u> <u>41,858,921 >A</u>	rs1800470	TGFB1, TMEM91	Risk factor	Cystic fibrosis		Missing Data	