**LETTER OF MEDICAL NECESSITY FOR CYSTIC FIBROSIS DIAGNOSTIC GENETIC TESTING**

**(*CFTR* Gene Sequence and Deletion/Duplication Analysis)**

Date: Date of service/claim

To: Utilization Review Department

Insurance Company Name, Address, City, State

Re: Patient Name, DOB, ID #

ICD-10 Codes:

The ICD-10 codes listed below are commonly received by Ambry from ordering providers for the testing described in this letter. Ambry provides this information as a customer service but makes no recommendations regarding the use of any diagnosis codes. As a reminder, it is the ordering provider’s responsibility to always determine, for the specific date of service, the appropriate diagnostic codes based on the patient’s signs and symptoms.

Code Description

Z80.0 FAMILY HISTORY OF MALIGNANT NEOPLASM OF DIGESTIVE ORGANS

Z80.3 FAMILY HISTORY OF MALIGNANT NEOPLASM OF BREAST

Z80.51 FAMILY HISTORY OF MALIGNANT NEOPLASM OF KIDNEY

Z83.49   FAMILY HISTORY OF OTHER ENDOCRINE, NUTRITIONAL AND METABOLIC DISEASES

This letter is regarding my patient and your subscriber, referenced above, to request full coverage of medically indicated *CFTR* gene sequencing and deletion/duplication analysis to be performed by Ambry Genetics Corporation.

Classic cystic fibrosis (CF) is one of the most common hereditary conditions with a prevalence of approximately 1 in 2500 to 1 in 3300 live births.1 A person develops cystic fibrosis when they inherit two non-working copies of *CFTR*, one from each of their parents*.* Parents of a child with CF are typically symptom-free carriers of the condition because they still have one working copy of *CFTR*.

Pathogenic alterations in the *CFTR* gene are known to cause classic CF, which is inherited in an autosomal recessive fashion. Classic CF is characterized clinically by chronic obstructive pulmonary disease, exocrine pancreatic insufficiency, increased sweat chloride concentrations, and infertility in males.

Other *CFTR*-related disorders can be inherited in an autosomal dominant or autosomal recessive fashion and include non-classic CF, congenital absence of the vas deferens (CAVD), recurrent sinusitis, disseminated bronchiectasis, and idiopathic recurrent-acute or chronic pancreatitis.

**Significant aspects of my patient’s medical history that suggest a pathogenic variant(s) in the *CFTR* gene are as follows** [check all that apply]:

* Pulmonary disease (chronic obstructive)
* Pancreatic insufficiency (exocrine)
* Sweat chloride concentration increased
* Sinusitis (recurrent)
* Bronchiectasis (disseminated)
* Pancreatitis (recurrent acute)
* Pancreatitis (chronic)
* Meconium ileus
* Echogenic bowl
* Male infertility
* Congenital absence of the vas deferens
* Follow-up testing to positive newborn screening
* Family history of cystic fibrosis or other *CFTR-*related disorders
* Family and/or partner history of cystic fibrosis carrier status
* Other: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**The patient’s features and/or family history are suspicious for a pathogenic variant(s) in the *CFTR* gene.** The American College of Medical Genetics and Genomics (ACMG) states that *CFTR* testing is used in the following scenarios1:

* Diagnostic purposes:
* For the molecular confirmation of a clinical CF diagnosis
* For infants with meconium ileus
* For males with CAVD
* For individuals with idiopathic pancreatitis or bronchiectasis
* As a follow-up to newborn screening
* Carrier screening purposes:
* For individuals with a positive family history of CF
* In partners of individuals with a positive family history
* In partners of males with CAVD
* For reproductive age women
* For gamete donors
* Prenatal testing purposes:
* When a pathogenic or likely pathogenic variant is confirmed in one or both partners
* When an ultrasound finding suggests an affected fetus

**Due to the medical complications associated with pathogenic variants in the *CFTR* gene, this genetic testing is medically indicated.** Management modifications may include [check all that apply]:

* Confirm a cystic fibrosis diagnosis
* Aid in diagnosis for patients with an atypical presentation of disease
* Tailor medical treatment based on specific pathogenic variants
* Allow immediate management and treatment to anticipate and control common clinical findings associated with cystic fibrosis
* Assist in long-term management and monitoring of suspected disease progression based on mutations identified
* Guide informed decision making for other family members with similar conditions, or who may be at risk for similar conditions
* Other: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**A positive test result would confirm a genetic diagnosis in my patient and would ensure my patient is being managed appropriately.**

Based on these factors, this testing is medically necessary, and I request that you approve coverage of diagnostic genetic testing for *CFTR* gene sequencing and deletion/duplication analysis in my patient.  Thank you for your time, and please don’t hesitate to contact me with any questions.

Sincerely,

Ordering Clinician Name (Signature Provided on Test Requisition Form)

(MD/DO, Clinical Nurse Specialist, Nurse-Midwives, Nurse Practitioner, Physician Assistant, Genetic Counselor\*)

\*Authorized clinician requirements vary by state

**Test Details**

CPT codes: 81221

Laboratory: Ambry Genetics Corporation (TIN 33-0892453 / NPI 1861568784), a CAP-accredited and CLIA-certified laboratory located at 7 Argonaut, Aliso Viejo, CA 92656

**References**

1. Deignan JL, *et al. CFTR* variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2020 Aug; 22(8):1288-1295.