ADMINISTRATIVE POLICY STATEMENT
Ohio Medicaid

Policy Name & Number	Date Effective
Cystic Fibrosis Testing-OH MCD-AD-0837	06/01/2024
Policy Type	
ADMINISTRATIVE	

Administrative Policy Statement prepared by CareSource and its affiliates are derived from literature based on and supported by clinical guidelines, nationally recognized utilization and technology assessment guidelines, other medical management industry standards, and published MCO clinical policy guidelines. Medically necessary services include, but are not limited to, those health care services or supplies that are proper and necessary for the diagnosis or treatment of disease, illness, or injury and without which the patient can be expected to suffer prolonged, increased or new morbidity, impairment of function, dysfunction of a body organ or part, or significant pain and discomfort. These services meet the standards of good medical practice in the local area, are the lowest cost alternative, and are not provided mainly for the convenience of the member or provider. Medically necessary services also include those services defined in any Evidence of Coverage documents, Medical Policy Statements, Provider Manuals, Member Handbooks, and/or other policies and procedures.

Administrative Policy Statements prepared by CareSource and its affiliates do not ensure an authorization or payment of services. Please refer to the plan contract (often referred to as the Evidence of Coverage) for the service(s) referenced in the Administrative Policy Statement. If there is a conflict between the Administrative Policy Statement and the plan contract (i.e., Evidence of Coverage), then the plan contract (i.e., Evidence of Coverage) will be the controlling document used to make the determination.

According to the rules of Mental Health Parity Addiction Equity Act (MHPAEA), coverage for the diagnosis and treatment of a behavioral health disorder will not be subject to any limitations that are less favorable than the limitations that apply to medical conditions as covered under this policy.

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A. Subject

Cystic Fibrosis Testing

B. Background

Cystic fibrosis (CF) is a recessive genetic disorder caused by mutations of the cystic fibrosis transmembrane conductance regulator (CFTR) gene. When an individual inherits two abnormal CFTR genes from their parents, they may develop dysregulation of the epithelial lining fluid transport, affecting systemic mucus production. Diagnosis of CF is based upon the finding of genetic and/or functional abnormalities of the CFTR gene (CFTR variants) and is typically associated with progressive lung disease, pancreatic insufficiency, and elevated sweat chloride levels.

Many individuals with CF demonstrate mild or atypical symptoms and can present later in childhood or as adults, while others may test positive for CF, yet remain asymptomatic (CRMS). Untreated CF in some can lead to early mortality and infertility. This makes early CF diagnosis and identification of carriers of CF important for early intervention to mitigate disease progression as well as to allow for more informed health determinations. Advances in CF research has led to steadily evolving tests that can identify abnormal CFTR genes as well as tailored treatments to address the underlying mechanisms leading to disease.

More than 10 million Americans are carriers of a defective CFTR gene and show no symptoms of the disease. Others who do not fit CF diagnostic criteria still go on to develop disease associated with evidence of CFTR dysfunction limited to one organ (CFTR-related disorder). Carrier testing may provide an early indication as to whether a person might develop a CFTR-related disorder. Potential parents who test positive as carriers for CF will be able to make educated reproductive decisions, better prepare for birth, and investigate additional testing for CF-related health conditions. A negative screening result does not completely rule out the possibility that a person is a CF carrier. A negative screening test only rules out the specific CF variants that were part of the screen.

Diagnosis of CF is a multistage process. Individuals must have clinical symptoms consistent with CF in at least one organ system, have a positive newborn screen, or have a sibling with CF. In addition, there must be evidence of CFTR dysfunction via elevated sweat chloride, presence of two disease-causing CFTR mutations (one from each parent), or abnormal nasal potential difference (NPD). Newborn screening involves immunoreactive trypsinogen (IRT), sweat chloride testing, and genetic testing.

C. Definitions

- **American College of Medical Genetics and Genomics (ACMG)** – A nationally recognized interdisciplinary organization dedicated to the practice of medical genetics. The ACMG regularly posts technical recommendations for genetic diseases, including cystic fibrosis.

- **Autosomal Recessive** – A trait or disorder requiring a deleterious variant in both copies of the gene to express a phenotype.
- **Carrier** – An individual with a gene variant for a disease or disorder who can pass the variant on to offspring but does not have symptoms or features of the disorder.
- **Cystic Fibrosis Transmembrane Conductance Regulator (CFTR)** – Transmembrane protein that functions as a regulated anion channel to maintain a gradient necessary for normal cell function. Mutations that alter the function of the CFTR protein are associated with CF.
- **CFTR-Related Disorder** – A single pathogenic CFTR variant leading to the development of disease limited to one organ system in an individual who does not fit the CF diagnosis.
- **CFTR-Related Metabolic Syndrome (CRMS)** – Infants and children who are asymptomatic but have positive CF screening results. Found in 3-4% of infants with a positive newborn CF screen, CRMS is also known as CF screen positive, inconclusive diagnosis (CFSPID).
- **Immunoreactive Trypsinogen (IRT)** – A pancreatic enzyme precursor measured in newborns to screen for the presence of CF.

D. Policy

I. Genetic testing for CF should only be performed once in a lifetime and the results documented in the member's health record. All genetic testing for CF should use currently recommended ACMG CFTR panels. Prior authorization is only required for CF genomic sequence analysis.

II. Diagnostic testing is considered medically necessary when the member meets any of the following criteria:
   A. clinical presentation of CF
   B. infertility from oligospermia/azoospermia/congenital bilateral absence of vas deferens (CBAVD)
   C. infant with meconium ileus or other symptoms indicative of CF but unable to produce adequate amounts of sweat for a sweat chloride test
   D. infant with an elevated IRT value on newborn screening and a sweat chloride of at least 60 mmol/L or intermediate sweat chloride (between 30 mmol/L and 59 mmol/L)

III. Carrier screening is indicated when the member meets any of the following criteria:
   A. members who are pregnant or of reproductive age with intent and potential to procreate
   B. members whose partner tests positive while the member is pregnant or intending to become pregnant
   C. members with a family history of CF
   D. members where both parents are CF carriers

IV. The following are not considered medically necessary and will not be covered:
   A. repeat testing
   B. gene sequencing of anything other than the CFTR gene
   C. Fetal testing is generally not considered medically necessary. However, fetal testing is medically necessary with the presence of any of the following:
      1. Both parents have disease-causing mutations of the CFTR gene.
      2. Echogenic bowel is detected on ultrasound examination of fetus during pregnancy.
      3. The mother is a confirmed carrier, and father is unknown or unavailable for testing.

E. Conditions of Coverage

CareSource may request documentation for post-payment review of claims submitted for payment of CF testing. If documentation is not provided, CareSource may recoup previously paid claim(s).

F. Related Policies/Rules

Genetic Testing and Genetic Counseling

G. Review/Revision History

	DATES	ACTION
Date Issued	09/02/2020	
Date Revised	07/20/2022 02/14/2024	Addition of Section D, IV and V. Annual review: changes to title, background, and definitions, expanded policy to include diagnostic testing, and updated references. Approved at Committee.
Date Effective	06/01/2024	
Date Archived		

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