VERMONT
AGENCY OF HUMAN SERVICES
DEPARTMENT OF VERMONT HEALTH ACCESS

State of Vermont
Department of Vermont Health Access
280 State Drive, NOB 1 South
Waterbury, VT 05671-1010

Agency of Human Services
[Phone] 802-879-5900
http://dvha.vermont.gov

The Department of Vermont Health Access Clinical Criteria

Subject: Cystic fibrosis transmembrane conductance regulator (CFTR) dysfunction Genetic Testing
Last Review: November 8, 2023*
Past Revisions: January 31, 2022, January 28, 2020, May 10, 2016, June 15, 2015, and July 6, 2017

***Please note: Most current content changes will be highlighted in yellow.**

Description of Service or Procedure

Cystic fibrosis (CF) is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene which is responsible for and contains the information to make the CTFR protein. The CTFR protein works to control water and salt balance on surfaces within the body, such as the lungs and gastrointestinal systems and function as a channel for certain ions such as sodium and chloride across cells of exocrine organs that produce mucus, sweat, digestive enzymes, and tears.

CF is an autosomal recessive disorder, meaning an individual must inherit a copy of a genetic variant from each parent to have the disease. If an individual inherits only one copy, they will not inherit the actual disease but will be a carrier and could pass the disease to their children. If both parents carry the defective gene for CF, there is a 25% chance of producing a child with CF, a 50% chance the child will carry the CF gene but not have CF and a 25% chance the child will not carry the gene and not have CF. Newer estimates of CF incidence derived from newborn screening programs allow for estimate of CF incidence in the range of 1/3000 to 1/6000 live births.

The CFTR gene test uses a blood sample or cells from inside the cheek for DNA analysis to identify the presence of a genetically altered cystic fibrosis transmembrane conductance regulator gene.

Newborn screening for CF is done in the first 2 or 3 days after birth. If screening is positive, a sweat test is performed to confirm the diagnosis.

The severity of clinical symptoms that may occur in individuals with CF cannot be definitively predicted on cystic fibrosis transmembrane conductance regulator gene variant testing alone.

Genetic testing for CF is not available for the general public.

Disclaimer

Coverage is limited to that outlined in Medicaid Rule or Health Care Administrative Rules that pertains to the beneficiary's aid category. Prior Authorization (PA) is only valid if the beneficiary is eligible for the applicable item or service on the date of service.

Medicaid Rule

Medicaid and Health Care Administrative Rules can be found at https://humanservices.vermont.gov/rules-policies/health-care-rules/health-care-administrative-rules-hcar/adopted-rules

7102.2	Prior Authorization Determination
4.101	Medical Necessity for Covered Services
4.104	Medicaid Non-Covered Services
7405	Laboratory and Radiology Services
4.106	Early and Periodic Screening, Diagnostic and Treatment (EPSDT) Services

Coverage Position

Cystic fibrosis testing may be covered for members:

- When the CF gene test is prescribed by a licensed medical provider, enrolled in the Vermont Medicaid program, operating within their scope of practice as described on the Vermont Office of Professional Regulation's website*, who is knowledgeable regarding the CF gene test, and who provides medical care to the beneficiary AND
- When the clinical criteria below are met.

* Vermont's Office of Professional Regulation's website: https://sos.vermont.gov/opr/

Coverage Criteria

CFTR genetic testing may be covered for members who:

1. Have clinical symptoms characteristic of CF (pancreatic insufficiency, lung function abnormalities and intermediate sweat chloride results)
2. Have a family history of CF
3. Are couples planning a pregnancy and wish to know if either is a CF carrier
4. Are reproductive partners of persons with CF
5. Desire a prenatal diagnosis to identify a fetus or embryo with CF for purposes of post-delivery care of the newborn.
6. Members with confirmed or suspected CF, if the genotype is not yet known, as this may affect treatment choices
7. Members with normal sweat chloride results if there is a strong clinical suspicion of CF

*Please review the CF Clinical Care Guidelines for more detailed information

https://www.cff.org/medical-professionals/cf-diagnosis-clinical-care-guidelines#cf-diagnosis-clinical-care-guidelines

https://www.cff.org/sites/default/files/2021-10/Clinical-Care-Guide-for-Diagnosis-of-CF.pdf

Considerations: Providers requesting this test should provide pre- and post-test genetic counseling for the beneficiary and family, if applicable.

Early and Periodic Screening, Diagnostic and Treatment (EPSDT): Vermont Medicaid will provide comprehensive services and furnish all Medicaid coverable, appropriate, and medically necessary services needed to correct and ameliorate health conditions for Medicaid members under age 21.

Please note, Vermont Medicaid Clinical Criteria is reviewed based on available literature, evidence- based guidelines/standards, Medicaid rule and policy, and Medicare coverage determinations that may be appropriate to incorporate when applicable.

Clinical criteria for repeat service or procedure

Repeat testing can be performed if the initial results are inconclusive or ambiguous.

Type of service or procedure covered

Cystic fibrosis testing for the general public is not covered.

Coding guidelines

Please see the Medicaid Portal at http://vtmedicaid.com/#/feeSchedule for fee schedules, code coverage, and applicable requirements.

References

Centers for Medicare and Medicaid Services. (n.d.). *Early and periodic screening, diagnostic, and treatment.* Medicaid.gov. https://www.medicaid.gov/medicaid/benefits/epsdt/index.html

Farrell, P.M., White, T.B., Clement, L.R., Hempstead, S.E., Accurso, F., Derichs, N., Howenstine, M., McColley, S.A., Rock, M., Rosenfield, M., Sermet-Gaudelus, I., Southern, K.W., Marshall, B.C., & Sosnay, P.R. (2017). Diagnosis of Cystic Fibrosis: Consensus guidelines from the Cystic Fibrosis Foundation. *The Journal of Pediatrics,181S,* S4-S15. https://doi.org/10.1016/j.jpeds.2016.09.064

McKone, E. F., Emerson, S. S., Edwards, K. L., & Aitken, M. L. (2003). Effect of genotype on phenotype and mortality in Cystic Fibrosis: A retrospective cohort study. *The Lancet, 361*(9370), P1671-P1676. https://doi.org/10.1016/S0140-6736(03)13368-5

Rohlfs, E. M., Zhou, Z., Heim, R. A., Nagan, N., Rosenblum, L. S., Flynn, K., Scholl, T., Akmaev, V.A., Sirko-Osadsa, D.A., Allitto, B.A., & Sugarman, E.A. (2011). Cystic Fibrosis carrier testing in an ethnically diverse US population. *Clinical Chemistry, 57*(6), 841-848. https://doi.org/10.1373/clinchem.2010.159285

Scotet, V., L'Hostis, C., & Férec, C. (2020). The changing epidemiology of Cystic Fibrosis: Incidence, survival, and impact of the CFTR gene discovery. *Genes, 11*(6). https://doi.org/10.3390/genes11060589

U.S. National Library of Medicine. (2021, July). *Cystic Fibrosis.* MedlinePlus. Retrieved on June 27, 2023, from https://medlineplus.gov/genetics/condition/cystic-fibrosis/

The American Congress of Obstetricians and Gynecologists (ACOG). (2021, December). *Cystic Fibrosis: Prenatal screening and diagnosis.* ACOG For Patients FAQ 171. Retrieved on June 27, 2023, from: http://www.acog.org/Patients/FAQs/Cystic-Fibrosis-Prenatal-Screening-and-Diagnosis

The American Congress of Obstetricians and Gynecologists (ACOG). (2017). Committee opinion no. 691: Carrier screening for genetic conditions. *Obstetrics and Gynecology, 129*(3), e41-e55. https://doi.org/10.1097/aog.0000000000001952

Katkin, J.P. (2023, March 7). Cystic Fibrosis: Clinical manifestations and diagnosis. *UpToDate. Retrieved June 26,2023,* from*:* https://www.uptodate.com/contents/cystic-fibrosis-clinical-manifestations-and-diagnosis?search=cystic%20fibrosis%20genetic%20testing&topicRef=6368&source=see_link

This document has been classified as public information.