FOR CARE TEAMS

Diagnosis of Cystic Fibrosis

Case managers will interact with patients already diagnosed with cystic fibrosis, but it is important to be familiar with the diagnostic process of the disease.

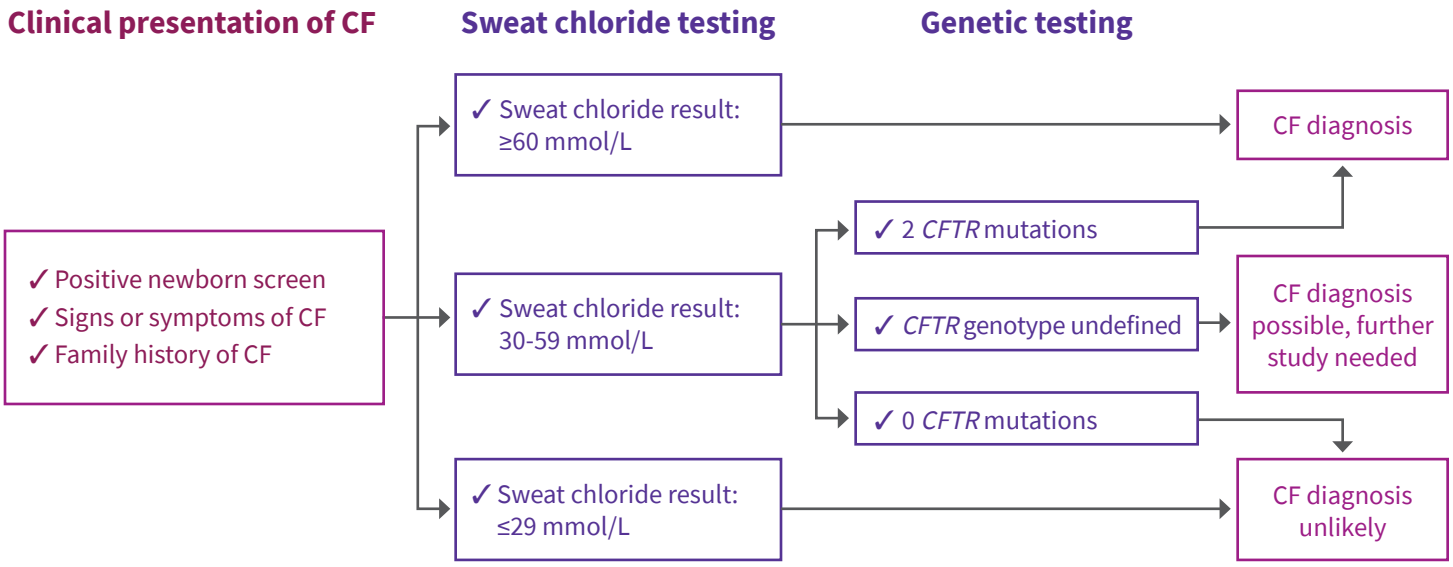
About cystic fibrosis (CF)

CF is a progressive, life-shortening genetic disease that affects the lungs, digestive system, and many other organs.[1,2] CF occurs when a person inherits two defective copies of the cystic fibrosis transmembrane conductance regulator *(CFTR)* gene, one from each parent.[3]

Mutations in the *CFTR* gene can disrupt the normal production and/or function of the CFTR protein, leading to a buildup of thick fluids that are poorly cleared from passageways in the respiratory tract, pancreas, intestines, liver, and other organs.[3,4]

Diagnosis of CF[5]

Diagnosing CF is a multistep process. According to the Consensus Guidelines for the diagnosis of CF from the Cystic Fibrosis Foundation, a CF diagnosis is based on the following measures—clinical presentation, sweat test(s), and genetic testing revealing the presence of 2 *CFTR* mutations.



The benefits of early testing

In 2020, 63% of total new diagnoses and 89% of diagnoses among those younger than 6 months old were reported as being detected by newborn screening. Based on available evidence, individuals diagnosed before showing symptoms have better lung function and nutritional outcomes later in life.[4]

More about sweat chloride testing[6]

The sweat test can be performed on an individual of any age. However, some infants may not make enough sweat for the test to be able to provide an accurate result. Full-term babies usually produce enough sweat by 2 weeks of age. If an infant does not produce enough sweat the first time, the test should be repeated. Sweat chloride testing should be done at a specialized CF care center.

Sweat Chloride Test

Electrode

Mild electrical stimulation pushes medicine into skin to induce sweating

Underside of forearm

Sweat is collected on a piece of filter paper or gauze

A newborn with an inconclusive diagnosis may[5]:

- Screen positive without CF clinical features
- Have a sweat chloride value:
  - ≤29 mmol/L and 2 *CFTR* mutations (at least 1 of which is undefined)
  - 30-59 mmol/L and 0 or 1 CF-causing mutations

Next steps may include[5]:

- At least 1 repeat sweat chloride test
- Extended *CFTR* gene analysis
- Genetic counseling for families considering future pregnancies
- Potential for reclassification to a CF diagnosis if additional diagnostic criteria are met, as determined by an experienced CF physician
- Further research and follow-up

Newly diagnosed patients and patients with established CF can receive specialized and comprehensive care through their CF care team.

References: 1. Cutting GR. Cystic fibrosis genetics: from molecular understanding to clinical application. *Nat Rev Genet*. 2015;16(1):45-56. **2.** Cystic Fibrosis Foundation. About CF. https://www.cff.org/intro-cf/about-cystic-fibrosis. Accessed March 3, 2022. **3.** Cystic Fibrosis Foundation. CF genetics: the basics. https://www.cff.org/intro-cf/cf-genetics-basics. Accessed March 3, 2022. **4.** Cystic Fibrosis Foundation. Patient Registry 2020 Annual Data Report. Bethesda, Maryland. https://www.cff.org/sites/default/files/2021-11/Patient-Registry-Annual-Data-Report.pdf. Accessed March 3, 2022. **5.** Farrell PM, White TB, Reb CL, et al. Diagnosis of cystic fibrosis: Consensus Guidelines from the Cystic Fibrosis Foundation. *J Pediatr*. 2017;181S:S4-S15. **6.** Cystic Fibrosis Foundation. Sweat test. https://www.cff.org/What-is-CF/Testing/Sweat-Test/. Accessed March 3, 2022.

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