

Carrier Screening ACT Sheet

CFTR Mutations except R117H

Cystic Fibrosis (CF)

Why Genetic Carrier Screening is Done: Carrier screening is done to identify individuals without a family history of CF or a known family mutation in the *CFTR* gene who may be at increased risk of having children affected with CF. (Individuals with a family history of CF should always be offered cystic fibrosis transmembrane conductance regulator (*CFTR*) gene testing.)

Condition Description: CF is an autosomal recessive inherited disease caused by mutations in the *CFTR* gene. The cystic fibrosis transmembrane conductance regulator (*CFTR*) protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands and genitourinary tract. CF affects multiple body systems and is associated with progressive damage to respiratory and digestive systems. About 1 in 25 (~4%) Caucasians and 1% or higher in other population groups are carriers.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Inform carrier of screening result.
- Refer to genetic evaluation and counseling.
- Offer partnership testing for *CFTR* mutations.

Reproductive Implications: Once one member of a couple has been shown to be a carrier, the other member of the couple should be offered carrier screening. If both are carriers the risk of CF to any offspring is 1 in 4 (25%). The mutations that are recommended to be included in screening panels are relatively common and often associated with classical CF in affected individuals. Because over 1500 mutations have been described in *CFTR*, a more comprehensive genetic test may be considered. Genetic counseling should be offered to all carriers.

Additional Information:

[Gene Tests/Gene Clinics](#)
[Genetics Home Reference](#)
[Cystic Fibrosis Foundation](#)
[American College of Medical Genetics](#)

Referral (local, state, regional and national):

[Testing](#)
[Clinical Services](#)
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Tests/Gene Clinics

<http://www.genetests.org/query?dz=cf>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=cysticfibrosis>

Cystic Fibrosis Foundation

<http://www.cff.org/>

American College of Medical Genetics

http://www.acmg.net/StaticContent/StaticPages/CF_Mutation.pdf

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2220?db=genetests

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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