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

ACMG
American College of Medical Genetics and Genomics
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LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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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