Carrier Screening ACT Sheet
No Mutations Detected by Carrier Screening
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. Only the most common and severe mutations are included in carrier screening panels; therefore not all carriers are detected by screening.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Inform carrier of screening result and residual risk as in report.

Reproductive Implications: A negative screening test result does not exclude the possibility that one of the rarer *CFTR* mutations is present. Over 1500 mutations have been described in CFTR. The mutations that are recommended to be included in routine screening are relatively common and often associated with classical CF in affected individuals. Those screened should be made aware of this limitation to CF carrier screening and given their residual risk of being a carrier.

Additional Information:

Gene Tests/Gene Clinics
Genetics Home Reference
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
Translating Genes Into Health®

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

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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