Carrier Screening ACT Sheet
Cystic Fibrosis R117H

Carrier Screening: Carrier screening is done to identify individuals with or without a family history of cystic fibrosis (CF) who may be at increased risk of having children affected with CF. Carriers of R117H variant require additional analysis to determine its pathogenicity. When R117H is found in combination with the 5T variant on the same parental chromosome, it is interpreted as a pathogenic mutation for CF. When R117H is present without 5T, it may have implications for infertility in male offspring.

Condition Description: CF is an autosomal recessive inherited disease caused by mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene. The *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 greater in other population groups are carriers.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Inform individual of carrier screening result.
- Refer for genetic counseling.
- Offer partner testing for *CFTR* mutations.

Reproductive Implications: If the R117H/5T combination is identified in an individual, the reproductive partner should be offered carrier screening to determine the risk for having a child with CF. If both are carriers, the risk of CF to any offspring is 1 in 4 (25%). The mutations recommended for inclusion in screening are relatively common and often associated with classical CF in affected individuals. Over 1500 mutations have been described in *CFTR*, and a more comprehensive genetic test may be needed. Genetic counseling should be offered to all carriers.

Additional Information:

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
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:

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&country=United%20States

Clinical Services
http://www.genetests.org/servlet/access?id=8888891&key=ycNiicOsM18KA&fcn=y&fw=z4HV&filename=/clinicsearch/searchclinic.html

Find Genetic Services
http://www.acmg.net/gis