

6.1.4 Mailer Wording for Cystic Fibrosis NBS Results Updated 2/21/12

Test Type/Result	Wording for Interpretation or Follow-up
General	
General Disclaimer on all Mailers	Test interpretations are based on the Birth/Collection Information provided above and subject to disclaimer below.
General Disclaimer on all Mailers	Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does <u>not</u> rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should <u>not</u> be considered diagnostic and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.
Multiple Positives	ATTENTION: THIS CHILD HAS TESTED POSITIVE FOR MORE THAN ONE TEST.
Contact Coordinator	If you have questions regarding these results, please contact the Newborn Screening staff at [xxx] Area Service Center [phone #].
Inadequate	
Inadequate Specimen	This specimen was inadequate for testing for the following reason: [Inadeq. Code#] [Reason] THE PHYSICIAN LISTED ABOVE HAS BEEN NOTIFIED
Cystic Fibrosis (CF)	
CF - (negative); elevated IRT, no mutations on DNA panel	Interpretation: No follow-up required. Elevated IRT and no mutations found using California CFTR mutation Panel.

<p>Follow-up Rider - CF - (negative); elevated IRT and negative mutation panel</p>	<p>Cystic Fibrosis (CF): No mutations identified using the California CFTR panel. However this does not exclude the presence of rare mutations. If this baby has symptoms of CF, both parents are carriers of a CF mutation or there is a family history of CF, call your NBS Coordinator for assistance (Phone number listed below).</p>
<p>CF- positive - 2 mutations found on DNA panel (Initial Mailer)</p>	<p>Interpretation: Consistent with Cystic Fibrosis (<i>mutation name</i> and <i>mutation name</i> mutations)</p>
<p>Follow-up Rider - CF positive, 2 mutations found on DNA panel</p>	<p>Cystic Fibrosis (CF): An immediate referral to a CCS approved Cystic Fibrosis Center for evaluation and additional testing is strongly recommended.</p>
<p>CF - Additional Results Pending - Initial Mailer</p>	<p>Interpretation: Elevated IRT and one mutation (<i>mutation name</i>) found using California CFTR mutation panel.</p>
<p>Follow-up Rider - CF - Additional Results Pending - Initial Mailer</p>	<p>Cystic Fibrosis (CF): Further testing by DNA sequencing is in process to determine whether this infant is a CF carrier or may have CF or a CFTR-related disorder. The additional results will be available in approximately 3 weeks. If this baby has symptoms of CF call your NBS Coordinator for assistance (phone number listed below).</p>
<p>CF - positive after DNA sequencing. CFTR DNA Sequencing mailer</p>	<p>Test Results and Interpretation: Mutation(s) Identified:_____ Novel Variation(s):_____ Polymorphisms:_____ Interpretation text from CFTR sequencing lab with references.</p>
<p>Follow-up Rider - CF - positive after CFTR sequencing</p>	<p>Consistent with cystic fibrosis or CFTR related disorders. An immediate referral to a CCS approved CF Center is strongly recommended.</p>
<p>CF - carrier - no additional mutation found by CFTR sequencing. CFTR DNA Sequencing mailer</p>	<p>Test Results and Interpretation: Mutation(s) Identified:_____ Novel Variation(s):_____ Polymorphisms:_____ (Interpretation text from CFTR sequencing lab with references.)</p>

<p>Follow-up Rider - CF - carrier - no additional mutation found by CFTR sequencing.</p>	<p>Results are consistent with CF carrier status. One CF mutation was identified by direct DNA sequencing. However, this does not exclude the rare possibility (less than 0.05%) of CF due to the presence of rare gene alterations. If this baby has symptoms of CF, both parents are carriers of a CF mutation or there is a family history of CF, call your NBS Coordinator for assistance (Phone number listed below). Free information for families is available by calling the Newborn Screening CF Carrier Information Line toll-free at 1-800-793-1313. Family testing may identify the possibility of cystic fibrosis in future pregnancies. A Cystic Fibrosis Carrier notification letter will be sent to the family.</p>
<p>CF - carrier - (TG)11-5T polymorphism found by CFTR sequencing and gender is <u>Male</u>. CFTR DNA Sequencing mailer</p>	<p>Test Results and Interpretation: Mutation(s) Identified: _____ Novel Variation(s):None Detected Polymorphisms:(TG)11-5T/(TG)___-___T (Interpretation text from CFTR sequencing lab with references) and: There is evidence of a small increased risk of male infertility, specifically congenital absence of the vas deferens (CBAVD) in males with one CFTR mutation and the 11TG 5T variant. A genetic counselor will call families of male newborns to discuss this risk. Families of female newborns will not be called. They are encouraged to contact a counselor at the number listed to learn more about CF carriers.</p>
<p>Follow-up Rider - CF - carrier - (TG)11-5T polymorphism found by CFTR sequencing and gender is Male.</p>	<p>Results are consistent with CF carrier status. One CF mutation was identified by direct DNA sequencing. However, this does not exclude the rare possibility (less than 0.05%) of CF due to the presence of rare gene alterations. If this baby has symptoms of CF, both parents are carriers of a CF mutation or there is a family history of CF, call your NBS Coordinator for assistance (Phone number listed below). Free information for families is available by calling the Newborn Screening CF Carrier Information Line toll-free at 1-800-793-1313. Family testing may identify the possibility of cystic fibrosis in future pregnancies. A Cystic Fibrosis Carrier notification letter will be sent to the family.</p>

Revised 2/21/12	

<p>Suggested change of "Free information for families..." to: Please encourage the family to call the Newborn Screening CF Carrier Information Line toll-free at 1-800-793-1313 for more information on carriers and parent testing. (Submitted for GDSP approval 2/14/12)</p>	
<p>Suggested change: The medical literature suggests there is a small increased risk of infertility, specifically congenital absence of the vas deferens (CBAVD), in males who inherit one CFTR mutation from one parent and the 11TG-5T variant from the other parent. Please encourage the parents to contact a genetic counselor at the toll-free number below for information on CF, carriers, and parent testing. (Submitted for GDSP approval 2/14/12)</p>	
<p>Suggested change of "Free information for families..." to: Please encourage the family to call the Newborn Screening CF Carrier Information Line toll-free at 1-800-793-1313 for more information on carriers and parent testing. (Submitted for GDSP approval 2/14/12)</p>	

