

6.1.4 Mailer Wording for Cystic Fibrosis NBS Results (Draft) 2/23/10			
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.		

Follow-up Rider - CF - (negative); elevated IRT and negative mutation panel	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 below).		
CF- positive - 2 mutations found on DNA panel - Interpretation	Consistent with Cystic Fibrosis (_____ and _____ mutations)		
Follow-up Rider - CF positive, 2 mutations found on DNA panel	Cystic Fibrosis (CF): An immediate referral to a CCS approved CF Center is strongly recommended.		
CF - Additional Results Pending - Initial Mailer - Interpretation	Elevated IRT and one mutation (<i>mutation name</i>) found using California CFTR mutation panel.		
Follow-up Rider - CF - Additional Results Pending - Initial Mailer	Cystic Fibrosis (CF): These results are consistent with either CF carrier status or the rare possibility (about 2%)* of CF. Further testing is in process to distinguish between the two. Additional results will be sent in 3-4 weeks. 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).		
CF - positive after DNA sequencing. CFTR DNA Sequencing mailer	Test Results and Interpretation: Mutation(s) Identified:_____ Novel Variation(s):_____ Polymorphisms:_____ Interpretation text from CFTR sequencing lab with references.		
Follow-up Rider - CF - positive after CFTR sequencing	Consistent with cystic fibrosis. An immediate referral to a CCS approved CF Center is strongly recommended.		
CF - carrier - no additional mutation found by CFTR sequencing. CFTR DNA Sequencing mailer	Test Results and Interpretation: Mutation(s) Identified:_____ Novel Variation(s):_____ Polymorphisms:_____ (Interpretation text from CFTR sequencing lab with references.) This patient is a carrier of the _____ mutation in the CFTR gene. No other mutations or novel variants were detected. This result reduces the likelihood that this patient is affected with CF.		

<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 the Ambry Test™:CF. 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.</p>		
	<p>*Changes being discussed based on 2.5 yrs of follow-up which suggests there is about a 34% chance for a second mutation, novel variant, or polymorphism to be found by CFTR sequencing and a diagnosis of CF or CRMS (CF-Related Metabolic Syndrome) to be made.</p>		