

November 30th, 2010

FROM: Neonatal Chemistry Laboratory

Kansas Health and Environmental Laboratories

RE: Two-Tiered Screening Strategy for Cystic Fibrosis is Beginning

Effective December 1st, 2010, the Kansas Newborn Screening Laboratory will begin implementing a two-tiered screening strategy for detecting infants with cystic fibrosis (CF).

A measurement of the immunoreactive trypsinogen (IRT) level from the standard dried blood spot specimen will be used as an initial screen test for cystic fibrosis as is currently done. Specimens with an IRT level ≥ 60 ng/mL will have a DNA mutation test performed on the same dried blood spot card to analyze for the 40 most common CF-causing mutations in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. A list of the mutations tested is shown below.

This will greatly reduce the number of infants needing a repeat screening test for IRT and will also reduce the number of infants needing a sweat test.

If you have comments or questions, please contact Colleen Peterson at 785-296-1650 or email ckpeters@kdheks.gov

Kansas Neonatal Chemistry Laboratory's Cystic Fibrosis DNA Screen

CFTR InPlexTM assay 40-mutation panel

(ACMG recommended mutations in **bold**)

▲ F508	R1162X	2184delA	394delTT	D1152H
G542X	3120+1G>A	3659delC	E60X	1078delT
W1282X	R117H	A455E	Q493X	S549R T>G
G551D	1717-1G>A	R560T	3905insT	Y1092X C>G
621+1G>T	2789+5G>A	G85E	V520F	Y1092X C>A
N1303K	R347P	1898+1G>A	S549R A>C	2183AA>G
R553X	711+1G>T	3849+10kbC>T	Y122X	S549N
▲ I507	R334W	3876delA	R347H	3849+4A>G

To learn more about Cystic Fibrosis visit our Kansas Panel of Newborn Disorders at http://www.kdheks.gov/neonatal/newborn_disorders.htm