

## Cystic Fibrosis Newborn Screening

DNA Panel as of April 2, 2009

Oklahoma newborn screening for cystic fibrosis begins with measuring the immunoreactive trypsinogen (IRT) level from the dried blood spot. If the IRT level is elevated, a DNA mutation test is performed on the dried blood spot to analyze for some of the most common mutations in the cystic fibrosis gene - cystic fibrosis transmembrane conductance regulator (CFTR). As there are >1,200 known CF mutations, it is not feasible to test for each mutation. The following chart represents the mutations currently included in the Oklahoma newborn screen for cystic fibrosis:

|                                                    |           |              |                      |
|----------------------------------------------------|-----------|--------------|----------------------|
| ΔF508                                              | A455E     | 3849+10kbC>T | 2183AA>G             |
| ΔI507                                              | 1717-1G>A | W1282X       | 2307insA             |
| G542X                                              | R560T     | N1303K       | Y1092X               |
| G85E *                                             | R553X     | 394delTT *   | M1101K               |
| G551D                                              | Y122X     | S1255X       | 1898+1G>A            |
| R347H                                              | 3876delA  | 621+1G>T     | 2184delA             |
| V520F                                              | 3905insT  | 711+1G>T     | 2789+5G>A            |
| A559T                                              | 3120+1G>A | S549N        | R334W                |
| R1162X                                             | S549R T>G | 1078delT     | R347P                |
| 3659delC                                           | 1898+5G>T | R117H        | I148T – Not in Panel |
| <b>Reflex:<br/>F508C,I507V,I506V,<br/>5T/7T/9T</b> |           |              |                      |

\* CFTR 40+4: 40 mutation panel with reflex analysis for I506V, I507V, F508C, 5, 7, and 9t as appropriate.

As understanding of CF mutations, their associated phenotypes and prevalence in certain populations is continually evolving, the panel of mutations used for newborn screening is expected to change. Any questions regarding the mutation panel used currently or in the past should be directed to the Newborn Screening Program- (405) 271-6617 or (800) 766-2223.

Please note, screening for CF results in more false negatives than are documented for other established newborn screening tests. Additionally, healthy carriers of CF may be detected. Any infant who is symptomatic (recurrent cough, wheezing, chronic abdominal pain, loose stools and failure to thrive) should be referred to a CF Center for clinical evaluation.