**Section 661.340 Cystic Fibrosis (CF)**

a) Interpretation of Results. Although the majority of infants affected by CF will be identified by this screening, due to genetic variabilities and variations in health status, specimen quality, and timing of specimen collection, not all infants affected by the disorder may be identified. As with any laboratory test, false positive and false negative results are possible. Newborn screening test results are insufficient information on which to base diagnosis or treatment.

1) CF is indicated by elevated neonatal levels of immunoreactive trypsinogen (IRT) that can be detected in dried blood spots. The normal IRT range shall be established using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When elevated levels of IRT are detected, testing by genetic mutation analysis shall be performed as part of the newborn screen, to decrease false positive results. As there are over 1,000 mutations in the CF transmembrane conductance regulator (CFTR) gene, testing will yield only 90 to 95 percent sensitivity.

3) When IRT levels and/or mutation analysis are found to be abnormal indicating the possibility of CF, the Department will recommend referral of the newborn to a designated medical specialist for appropriate definitive testing and diagnostic studies.

b) Designation of Medical Specialist. In addition to the minimum qualifications set out in Section 661.230, medical specialists designated by the Department to follow-up on a screen positive for CF shall possess certification by the American Board of Pediatrics in Pediatric Pulmonology or Pediatric Gastroenterology. CF medical specialists should provide the following: prompt access to quantitative pilocarpine iontophoresis sweat chloride testing in a laboratory that meets all CLSI standards; a multidisciplinary approach to care, including the availability of genetic counselors, dietitians, respiratory therapists and social workers; and access to microbiology laboratories that use CF-specific protocols for detection of respiratory tract infection.

c) Diagnosis and Treatment. Medical management by a designated medical specialist is highly recommended. Prompt evaluation of exocrine pancreatic status coupled with nutritional counseling is recommended after diagnostic confirmation. Close follow-up by a medical specialist is recommended to monitor and treat changes in nutrition and respiratory infection status.