**Section 661.350 Galactosemia**

a) Interpretation of Results. Although the majority of infants affected by galactosemia 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) Laboratory tests for galactosemia may be performed by testing for total galactose (galactose and galactose-1-phosphate) or a deficiency of the galactose-l-phosphate uridyl transferase enzyme. Normal test results indicate a normal level of total galactose or the presence of the enzyme. Test results are abnormal when the level of total galactose is above the normal range or the presence of the enzyme is reduced or not detected. Blood transfusion can cause a false negative test result for galactosemia for up to 120 days post-transfusion. Normal ranges shall be established using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When the galactose or enzyme levels are deemed abnormal, recommendations may be given to change the diet of the infant to a galactose free diet. The Department will recommend a repeat newborn screening test or referral of the newborn to a designated medical specialist for further 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 galactosemia shall possess certification by the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics or Medical Biochemical Genetics or by the American Board of Medical Genetics and Genomics in Clinical Genetics with at least one year of experience post-training in the diagnosis and treatment of galactosemia and inborn errors of metabolism. Galactosemia medical specialists should have the capacity to provide a multidisciplinary approach to care, including the availability on site of specially trained metabolic dietitians.

c) Diagnosis and Treatment. Medical management by a designated medical specialist is highly recommended. Therapy with a galactose free diet is currently the standard treatment. Long-term follow-up of children with galactosemia is necessary to ensure proper growth and development.