**Section 661.310 Biotinidase Deficiency**

a) Interpretation of Results. Although the majority of infants affected by biotinidase deficiency 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 biotinidase deficiency are designed to detect a deficiency of the biotinidase enzyme. Normal test results indicate the presence of the enzyme. Test results are abnormal when the presence of the enzyme is not detected.

2) When the determination of the enzyme is deemed abnormal, the Department will recommend a repeat newborn blood spot screening test or referral of the newborn to a designated medical specialist for a quantitative determination of the biotinidase enzyme and 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 biotinidase deficiency shall possess certification by the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics, Medical Biochemical Genetics or certification 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 biotinidase deficiency and inborn errors of metabolism. 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 pharmacological doses of biotin is required. Long-term follow-up of children with biotinidase deficiency is necessary to ensure proper growth and development.