**Section 661.360 Lysosomal Storage Disorders (LSDs)**

a) Interpretation of Results. Although the majority of infants affected by an LSD 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) An LSD can be detected in dried blood spots by using tandem mass spectrometry or other methods. Normal testing parameters shall be established using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When enzyme activity is found to be decreased, thus indicating the possibility of an LSD, 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 LSD shall possess certification by the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics or Medical Biochemical Genetics with at least one year of experience post-training in the diagnosis and treatment of LSDs. Medical specialists should have the capacity to provide enzyme replacement infusion therapies and to provide a multidisciplinary approach to care, including, but not limited to, the availability of pediatric specialists in neurology, cardiology and pulmonology. In addition to the above requirements, for Krabbe disease, medical specialists should be affiliated with a facility that has experience in performing stem cell transplantation.

c) Diagnosis and Treatment. Medical management by a designated medical specialist is highly recommended. Enzyme replacement therapy or stem cell transplant have demonstrated benefits for patients with these disorders. Long-term follow-up of children with an LSD is necessary to monitor treatment and to assess growth and development.