**Section 661.370 Amino Acid, Organic Acid and Fatty Acid Oxidation Disorders**

a) Interpretation of Results. Although the majority of infants affected by amino acid, organic acid and fatty acid oxidation disorders 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) The patient metabolite distribution patterns shall be compared to normal populations. Pattern analysis, and internal metabolite ratios relative to normal populations, shall be calculated using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When blood levels or ratios are found to be abnormal, indicating the possibility of a metabolic condition harmful to the infant, the Department will recommend a repeat newborn screening test or 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, the medical specialist shall possess the following:

1) For all disorders of amino acid, and organic acid metabolism, medical specialists designated by the Department to follow-up on a screen positive result 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 amino acid and organic acid disorders. The disorders of amino acid and organic acid metabolism medical specialist shall have the capacity to provide a multidisciplinary approach to care, including the availability on site of specially trained metabolic dietitians and a biochemical genetics laboratory; for citrullinemia and argininosuccinic aciduria, medical specialists should have on-site availability of required medical therapies, such as hemodialysis, that are necessary for the treatment of patients with these disorders.

2) For fatty acid oxidation disorders, medical specialists designated by the department to follow-up on a screen positive result 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 fatty acid oxidation disorders. Fatty acid oxidation disorders 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. The Department *shall supply the necessary* medically prescribed *metabolic treatment formulas where practicable* for Illinois residents *with diagnosed cases of* a metabolic disorder on the Department’s newborn screening panel. Services will be provided *for as long as medically indicated, when the product is not available through other State agencies*, and funding is available. Long-term follow-up of individuals with these metabolic disorders is necessary to adjust diet and to assess growth and development. Annual medical management by a designated medical specialist is required in order for a patient to receive treatment formulas from the Department. Many of these disorders can be properly and supportively managed by dietary therapy. Ongoing care of these children will require long-term follow-up by the medical specialist to ensure proper development. The administration of treatment formulas will not be initiated until further testing has been performed under the direction of a designated medical specialist to establish the diagnosis. The Department will work with the designated medical specialist to provide the metabolic treatment formula required by the affected individual. [410 ILCS 240/2(a)(5.3)]