**Section 661.50 Diagnosis and Treatment**

The Department will also maintain a registry to record the results of diagnosis and treatment for all diagnosed cases identified. Ongoing evaluation of the newborn screening program is imperative. This process includes outcome evaluation of children diagnosed through newborn screening. The Department will annually request updated information from the medical specialist or primary care provider concerning developmental milestones for each child diagnosed with a disorder for which the Department screens. The Department will maintain confidentiality at all times with regard to patient information.

a) Phenylketonuria and Hyperphenylalaninemia. The Department will supply the necessary medically prescribed treatment formulas for diagnosed cases as long as medically indicated. Long-term follow-up of children with phenylketonuria or hyperphenylalaninemia is necessary to adjust diet and to assess growth and development. Medical management by a designated medical specialist is required in order for a patient to receive treatment formulas from DPH. The administration of treatment formulas shall not be instituted until a complete amino acid analysis to corroborate the positive screening test has been performed, under the direction of a designated medical specialist, to establish the diagnosis of phenylketonuria.

b) Primary Hypothyroidism. Medical management by a designated pediatric endocrinologist is highly recommended. Replacement therapy with thyroid hormone is currently the standard treatment. Long-term follow-up of children with primary hypothyroidism is necessary in order to adjust medication and to assess growth and development.

c) Galactosemia. 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 in order to ensure proper growth and development.

d) Congenital Adrenal Hyperplasia. Medical management by a designated pediatric endocrinologist is highly recommended. Replacement therapy with glucocorticoids and, in some cases, mineralocorticoids is currently the standard treatment. Long-term follow-up of children with congenital adrenal hyperplasia is necessary in order to adjust medications and to assess growth and development.

e) Biotinidase Deficiency. 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 in order to ensure proper growth and development.

f) Sickle Cell Disease. Medical management by a designated pediatric hematologist-oncologist is highly recommended. Antibiotic prophylaxis and immunization to prevent pneumococcal infections are currently the standard treatment after a designated medical specialist has made a definitive diagnosis of a sickling disease. Long-term follow-up of children with sickle cell disease is necessary in order to assess growth and development.

g) Other Amino Acid, Organic Acid and Fatty Acid Oxidation Disorders. The Department will supply the necessary medically prescribed treatment formulas for diagnosed cases as long as medically indicated. Long-term follow-up of children with these metabolic disorders is necessary to adjust diet and to assess growth and development. Medical management by a designated medical specialist is required in order for a patient to receive treatment formulas from DPH. 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.

h) Cystic Fibrosis. 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.

i) Lysosomal Storage Disorders. 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 lysosomal storage disorders is necessary to monitor treatment and to assess growth and development.

j) Severe Combined Immunodeficiency (SCID) and T Cell Lymphopenia. Medical management by a designated medical specialist is highly recommended to confirm the diagnosis of SCID or other cause of T cell lymphopenia and to start therapy as soon as possible. Adenosine deaminase-deficient SCID can be treated by enzyme replacement and immunoglobulin replacement therapies. All forms of SCID can be treated by stem cell transplantation, while a few forms of SCID can be treated by gene therapy. Complete DiGeorge syndrome can be treated by thymic transplantation. Long-term follow-up is necessary to document immune reconstitution and to assess growth and development.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)