**Section 661.380 Severe Combined Immunodeficiency (SCID) and T-Cell Lymphopenia**

a) Interpretation of Results. Although the majority of infants affected by SCID or T-cell lymphopenia 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) SCID can be detected in dried blood spots by using DNA-based methods, such as polymerase chain reaction (PCR) 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 screening results indicate the possibility of SCID or other T-cell lymphopenias, 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, the medical specialist designated by the Department to follow-up on a screen positive for SCID shall possess certification by the American Board of Allergy and Immunology with at least one year post-training experience in the diagnosis and treatment of primary immunodeficiency diseases. Medical specialists should have the capacity to diagnose SCID, DiGeorge syndrome or other causes of T-cell lymphopenia and to provide a multidisciplinary approach to treatment, including access to specialists in stem cell transplantation, and 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 to confirm the diagnosis of SCID or other causes 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.