Newborn TREC Screening in Louisiana
to detect severe T cell lymphopenia in newborn infants

Follow up of abnormally low TREC results

Communication of an abnormal result

A low Trec result will be communicated to the primary care physician or designated referral physician to inform them of the newborn screening results and ascertain current status and if there is a history of SCID in the family.

Two basic actions will then be recommended depending on the gestational age of the newborn:

1. If the newborn is premature, additional Guthrie card samples will be requested every 2-3 weeks until TRECS normalize. The gestational age of a newborn is recorded at the time of obtaining the Guthrie card sample. If TRECS remain low by 37 weeks of gestational age, further testing and/or referral to an immunologist is recommended (see next).

2. If the newborn is a term baby with low TREC results, two options will be presented to the primary care physician:

   Obtain a whole blood sample for a CBC with differential, flow cytometry (enumeration of CD3+ T cell CD4+ T helper cells, CD8+ T suppressor cells, CD19+ B cells and CD16/56+ NK cells). A copy of the test results will be requested regardless of the outcome of the test. If this second tier tests show T cell lymphopenia (*or low percentage of naïve CD4 or CD8 T cells)*, referral to an Immunology center for further evaluation, diagnosis and treatment is indicated.

Refer directly to an immunology center for evaluation. This option will be encouraged if the newborn does not appear to clinically healthy.

The family will not be contacted directly unless the primary care physician requests it. Contact with the family will be done by one the immunologists on a list provided to the primary care physician (see list of immunologists and referral centers, below).

Differential diagnosis of low TRECSs:

- Prematurity,
    - Severe combined immune deficiency (SCID),
    - Complete DiGeorge syndrome,
    - Idiopathic T cell lymphopenias,
    - Congenital HIV infection,
- Intestinal lymphangiectasia
- Other disorders resulting in a profound loss of lymphocytes.

Brief condition descriptions

Prematurity. TRECS mature at different rates in different babies. Some premature babies may have very low TRECS that will increase with time. When the baby reaches 37 weeks of gestational age, low TRECS become clearly abnormal and additional testing is required.

SCID is an inherited (X-linked or autosomal recessive) condition causing a deficiency in T cell lymphocytes, with or without an associated defect in B cells and/or NK cells. Affected infants are susceptible viral, fungal and bacterial infections. Patients become symptomatic only after onset of infections or other complications like graft versus host disease.

DiGeorge Syndrome (22q11.2 deletion syndrome) is a rare congenital disease whose symptoms in the newborn period vary greatly but may include hypocalcemic titanic seizures, heart defects, and characteristic facial features. In severe (or "complete") cases (<1%), the presentation can share features of SCID. These infants may be candidates for thymic transplantation.

Idiopathic T cell lymphopenias are relatively frequent. They may have different causes and their prognosis is still under evaluation.

Referral summary for confirmed low TREC results

1. Initial Testing: Confirmatory testing: CBC with differential, Flow cytometry can be performed at local or commercial laboratories
2. Advanced immunology evaluation: lymphocyte proliferation, identification of molecular abnormality is recommended to be done by Clinical immunologists.

Referral information for Centers and Clinical Immunologists

New Orleans/Baton Rouge/Covington/Lafayette
Jeffrey Modell Diagnostic Center (JMC) for Immunodeficiency at LSU Health Sciences Center New Orleans/Children's Hospital. The JM Center has outreach clinics in Baton Rouge, Covington, and Lafayette. The JM Center team can be reached by calling 504-896-9589 during working hours. After hours, the team can be reached by calling 504-899-9511 requesting to talk to the LSU allergy/immunology fellow on call. For more information, visit the Jeffrey Modell Diagnostic Center for Primary Immunodeficiencis website at **http://www.jmcnola.org** or the Children's Hospital Allergy & Immunology website at **http://www.chnola.org/Allergy.**

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Stem cell transplantation (bone marrow or cord blood) in Louisiana is available at Children's Hospital in New Orleans through the hematology/oncology division and clinical immunologists at LSU Health Sciences Center. The Hematopoietic Stem Cell Transplant (HSCT) program is the only pediatric program recognized by COG as well as the National Marrow Donor program in Louisiana. The HSCT is affiliated with the Center for International Blood and Marrow Transplant Research (CIBMTR) , Pediatric Blood and Marrow Transplant Consortium, and the Primary Immunodeficiency Treatment Consortium (PIDTC).