



STATE OF MICHIGAN

DEPARTMENT OF HEALTH AND HUMAN SERVICES

LANSING

GRETCHEN WHITMER
GOVERNOR

ROBERT GORDON
DIRECTOR

September 2019

**Newborn Screening for
Adenosine Deaminase Deficiency Severe Combined Immunodeficiency**

Dear Practitioner(s):

Following recommendations by the Michigan Newborn Screening (NBS) Quality Assurance Advisory Committee and approval by the Legislature required by law (MCL 333.5431), all Michigan newborns have been screened for most types of severe combined immunodeficiency (SCID) since October 2011.

SCID represents a group of inherited disorders that lack functional T cells and B cells responsible for cellular and humoral immunity, respectively. Newborns with SCID are unable to mount an immune response to infection by viruses, bacteria and fungi. Treatment options for SCID vary and may include preventive medications for certain types of pneumonia, intravenous immunoglobulin, enzyme replacement therapy, hematopoietic stem cell transplantation, and gene therapy.

Currently the screening method for SCID and other primary immune deficiency disorders is the enumeration of T-cell receptor excision circles (TRECs) using real time polymerase chain reaction (PCR). A very low or absent TREC number could indicate SCID or other primary immune deficiency disorders. However, the TREC number is not always a good indicator for one type of SCID, caused by adenosine deaminase (ADA) deficiency.

ADA-SCID is an inherited autosomal recessive disorder of purine metabolism. Nucleosides adenosine (ADO) and 2'deoxyadenosine (dADO) are biomarkers for this disorder and will be elevated in affected newborns. The Michigan Department of Health and Human Services (MDHHS) Newborn Screening Laboratory will use a tandem mass spectrometry method to measure both ADO and dADO using the dried filter paper blood spots collected as part of the current newborn screening process. All Michigan newborns will be screened for ADA-SCID beginning October 7, 2019.

The Children's Hospital of Michigan Coordinating Center for Primary Immunodeficiencies is responsible for the follow-up of all positive SCID screens. Coordinating center staff contact primary care providers to instruct them on the next steps to take after a borderline or strong positive SCID screen result is received. After a careful review of each case, coordinating center staff will work with providers to arrange for either a repeat newborn screen or confirmatory testing.

MDHHS and follow-up coordinating center staff will provide prompt communication and are available to answer questions. Additionally, more resources for providers and families are available on our website, www.michigan.gov/newbornscreening. Please do not hesitate to contact us at 517-335-4181 or newbornscreening@michigan.gov.

Sincerely,

A handwritten signature in cursive script that reads "Mary Kleyn".

Mary Kleyn, MSc
Manager, Newborn Screening Follow-up Program