STATE OF MICHIGAN

GRETCHEN WHITMER
GOVERNOR

DEPARTMENT OF HEALTH AND HUMAN SERVICES
LANSING

ROBERT GORDON
DIRECTOR

September 2019

Newborn Screening for X-linked Adrenoleukodystrophy

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 will be screened for **X-linked adrenoleukodystrophy (X-ALD)** beginning October 7, 2019. In addition, screening will be completed retroactively on NBS specimens that were included in the laboratory's X-ALD validation and population studies.

X-ALD is the most common peroxisomal disorder, affecting approximately 1 in 17,000 people. X-ALD most severely affects males, though females can be carriers and develop some symptoms as adults. Males with X-ALD cannot break down certain fats called very long chain fatty acids. These fats accumulate in the body, causing an inflammatory response that results in demyelination of cerebral white matter and adrenal insufficiency. There are three main forms of X-ALD: childhood cerebral, a rapidly progressive form with onset generally between three to ten years of age; adrenomyeloneuropathy, presenting in older age groups; and Addison disease, or adrenal insufficiency. It is not possible to know which form of X-ALD a male will develop until symptoms arise. Symptoms can be highly variable, beginning anytime from childhood to adulthood. With newborn screening, affected babies can be referred to a specialty center to monitor for early symptoms and ensure prompt treatment. Treatment for X-ALD can include a blood stem cell transplant for the childhood cerebral form, hormone replacement therapy, and physical therapy. Specialized treatment can help those with X-ALD live the healthiest lives possible.

C26:0 lysophosphatidylcholine (C26:0 LPC) is a biomarker for X-ALD and is 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 C26:0 LPC. The newborn will be screened for X-ALD using the dried filter paper blood spots collected as part of the current newborn screening process.

The NBS Program will request a repeat newborn screen for infants with borderline positive or inconclusive results for this disorder. For infants with strong positive results, the NBS Program will notify the primary care provider, as well as the follow-up coordinating center. Follow-up coordinating center staff will contact primary care providers to instruct them on the next steps after a strong positive screen result is received. The baby will be referred to a medical management center with expertise in diagnosing and caring for patients with X-ALD. To ensure the best possible outcome, it is very important to complete the follow-up steps in a timely manner.

The X-ALD follow-up coordinating center in Michigan is:

- X-ALD Follow-up at Michigan Medicine
  University of Michigan (Ann Arbor)
  Phone: 734-647-8938

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,

Mary Kleyn

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