Implementation of Adrenoleukodystrophy (ALD) Screening in Missouri

Dear Practitioner, Laboratory Manager and Newborn Nursery Manager:

The Missouri Department of Health and Senior Services’ Newborn Screening Program is launching the implementation phase to screen all Missouri newborns for Adrenoleukodystrophy (ALD).

Adrenoleukodystrophy (ALD) is an X-linked genetic disease that affects males most severely. ALD primarily affects the nervous system and the adrenal glands. ALD may cause progressive loss of the myelin sheath, which acts as an insulator for the nerves in the brain and spinal cord. This can result in a variety of neurological problems including cognitive, mobility and sensory issues. ALD may also cause a deficiency of certain hormones due to damage to the adrenal glands. Adrenal insufficiency may cause weakness, weight loss, skin changes, vomiting and coma. If left untreated, the severe form of ALD can lead to critical and irreversible disabilities that can ultimately lead to death. Early diagnosis through newborn screening allows for proactive care, consistent monitoring and lifesaving treatment.

Beginning Dec. 1, 2021, all newborn screening specimens received by the Missouri State Public Health Laboratory (MSPHL) will be screened for ALD. The newborn screening dried bloodspot card collection process will not change, as testing will be conducted using current newborn screening samples. All infants with high risk positive screens will be referred for confirmatory testing at one of the four contracted genetic referral centers in Missouri. For infants with low risk positive screens, the Missouri Newborn Screening program will contact the infant’s physician of record to request a second newborn screening specimen. During this implementation phase, ALD screening results will not appear on the standard newborn screening laboratory reports provided by the MSPHL. Once more permanent reference ranges can be established through this initial testing phase, laboratory reports will be standardized to contain this information.

The MSPHL is announcing this pilot phase to make you aware that all newborns will be tested for ALD. If a high risk screening result is detected, the physician of record and the baby’s family will be notified by a genetic specialist with experience in the confirmation and treatment of ALD, so that the baby can benefit from early detection and treatment during this newborn screening implementation phase.

If you have questions or comments, please contact: Tracy Klug, Chief of Newborn Screening Laboratory, 573-751-2662.