Ohio | Department of Health

Mike DeWine, Governor
Jon Husted, Lt.Governor

Bruce Vanderhoff, MD, MBA, Director

October 14, 2022

To: Primary Care Providers
From: Ohio Department of Health Laboratory

**RE: Spinal Muscular Atrophy added to the Ohio Newborn Screening Panel**

Effective Monday October 24, 2022, the Ohio Newborn Screening Laboratory at the Ohio Department of Health will be screening for Spinal Muscular Atrophy (SMA). SMA is a progressive neuromuscular condition that can be diagnosed through newborn screening. Infants born with SMA appear normal at birth. Symptoms of SMA affect both males and females and appear weeks or months after birth in infantile cases, or even years after birth in mild cases. The most common and most severe form of SMA (SMA type 1) results in progressive weakness and loss of motor skills in the first months of life. Newborn screening detects nearly 95% of cases of SMA by identifying babies with homozygous deletions of exon 7 of the SMN1 gene. Infants identified early through newborn screening can begin effective treatment before they develop muscle weakness.

SMA is an autosomal recessive disorder due to mutations in the SMN1 gene that cause death of motor neurons in the brain stem and spinal cord. Motor neuron loss results in weakness affecting swallowing, breathing, head control and limb movement. Males and females are equally likely to be affected. Several treatment options are available that prevent or lessen weakness.

When a baby is identified as at-risk for SMA by newborn screening, the primary medical provider must contact a Neurology and Genetics Service Team to arrange for an evaluation of the infant **within 2-5 days** of being notified of the abnormal screening results. This multidisciplinary evaluation must be done by physicians with expertise in neuromuscular diseases. Included is a list of SMA Service Teams in Ohio. Confirmatory testing ordered by the specialty center includes genetic testing to confirm the SMN1 mutation and to evaluate the number of copies of another gene, SMN2, which supports the function of SMN1 and modifies the severity of the disease.

Please contact the Newborn Screening Laboratory if you have any questions.

Ohio Department of Health Public Health Laboratory
Newborn Screening Program
8995 East Main Street, Building 22
Reynoldsburg, Ohio 43068 U.S.A

888-634-5227
FAX 614-644-4648
CLIA ID# 36D0655844
www.odh.ohio.gov/newbornscreening

# Ohio Department of Health
# Newborn Screening Program
# Spinal Muscular Atrophy (SMA) Service Teams

**Akron**
Akron Children's Hospital
Genetics Center
One Perkins Square
Akron, OH 44308
P (330) 543-8792 F (330) 543-3677

**Columbus**
Nationwide Children's Hospital
Neuromuscular Clinic
700 Children's Drive
Columbus, OH 43205
Neuromuscular Clinic general line:
P (614) 722-2203 F (614) 355-5247

**Cincinnati**
Cincinnati Children's Hospital Medical Center
Division of Human Genetics
3333 Burnet Avenue (MLC: 4006)
Cincinnati, OH 45229
P (513) 636-4760 F (513) 636-7297

**Dayton**
Dayton Children's Hospital
Medical Genetics Division
One Children's Plaza
Dayton, OH 45404-1815
P (937) 641-3800 F (937) 641-5325

**Cleveland**
University Hospitals Case Medical Center
Rainbow Babies and Children's Hospital
Center for Human Genetics
11000 Euclid Avenue, Lakeside 1500
Cleveland, OH 44106
P (216) 844-3936 F (216) 844-7497
P (216) 292-9597 (after hours)

**Toledo**
ProMedica Ebeid Children's Hospital
Northwest Ohio Comprehensive Genetics Ctr
2150 W. Central Ave
Toledo, OH 43606
P (419) 291-2334 F (419) 291-6468

**ClevelandE**
The MetroHealth System
Department of Pediatrics
Division of Genetics and Genomics
2500 MetroHealth Drive
G730A
Cleveland, OH 44109
P (216) 778-4323 F (216) 778-2987

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**RE: X-Linked Adrenoleukodystrophy added to the Ohio Newborn Screening Panel**

Effective Monday October 24, 2022 the Ohio Newborn Screening Laboratory at the Ohio Department of Health will be screening for X-Linked Adrenoleukodystrophy (X-ALD). X-ALD is a rare disorder of fatty acid metabolism that can be diagnosed through newborn screening. Infants born with X-ALD may not look or act differently than other babies. Symptoms can appear in both males and females and may not appear for years or even decades. The most severe form of X-ALD results in a progressive neurodegenerative disease in preschool and school aged boys. Adrenal insufficiency can also develop in boys in infancy and early childhood. Treatment of adrenal insufficiency with corticosteroids can prevent life-threatening adrenal crisis. Monitoring for brain disease can identify children who will benefit from treatment that, if given prior to disability, can prevent disease progression. Stem cell transplant is currently the treatment of choice for progressive cerebral disease if diagnosed early.

When a baby is identified as at-risk for X-ALD by newborn screening, the primary medical provider needs to initiate a referral to an X-ALD specialist within 3 weeks (a list of X-ALD Specialty Teams is attached). While the symptoms of X-ALD do not present in the newborn period, other disorders of fatty acid metabolic can screen positive for X-ALD. As a result, confirmatory testing and evaluation by a specialist with expertise in metabolic disorders are needed. Confirmatory testing may include repeat fatty acid measurement and genetic testing. If X-ALD is confirmed, the infant will be referred to a pediatric endocrinologist for screening for adrenal insufficiency and to a neurologist or other specialist who is familiar with the MRI screening protocols for X-ALD. Current guidelines recommend that the first MRI of the brain for the purpose of monitoring for X-ALD be obtained between 12-18 months of age rather than in the newborn period.

X-ALD is an X-linked disorder that is classified as a peroxisomal disorder due to mutations in the ABCD1 gene. The resulting abnormal protein prevents fatty acid transport into peroxisomes which results in elevated levels of very long chain fatty acids in blood and brain tissue. Males are most often affected. The earliest symptoms can be related to adrenal insufficiency which can present in infancy. Later in early childhood, boys can develop progressive cerebral disease that can initially present with attention deficits and learning difficulties, often between the ages of 4 and 8 years of age, but earlier and later onset have been reported. In adulthood both males and females can development spinal cord and peripheral nerve disease (adrenomyeloneuropathy). Hematopoietic stem cell transplantation can slow and sometimes halt the progression of brain disease if performed before symptoms present. Newborn screening allows identification of boys that require periodic screening for adrenal insufficiency starting in the first year of life and for MRI screening for cerebral disease throughout childhood. Approximately 80% of females with X-ALD who are at risk of adult onset adrenomyeloneuropathy will also be identified by newborn screening.

Because X-ALD is a dominant disorder with delayed onset, identification of infants with the condition may result in identification of other family members who are at risk. Please contact your regional genetics center if you have questions about referral of other family members, particularly older brothers of the newborn.

Please contact the Newborn Screening Laboratory if you have any questions.

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