PUBLIC HEALTH DIVISION
Oregon State Public Health Laboratory - Newborn Bloodspot Screening
Kate Brown, Governor

Oregon Health Authority

7202 NE Evergreen Parkway, Suite 100
Hillsboro, OR 97124-7251
Voice: 503-693-4100
FAX: 503-693-5600
TTY: 971-673-0372

Date: April 29, 2022

To: Oregon birth facilities and health care providers

From: Patrice Held, Newborn Screening Program Manager

Subject: Addition of Spinal Muscular Atrophy (SMA) to the Newborn Screening Panel

Beginning on June 1, 2022, the Oregon State Public Health Laboratory (OSPHL) will add Spinal Muscular Atrophy to its screening panel and initiate testing for all newborns.

What is changing?
All first specimens received for newborn screening at the OSPHL beginning on June 1, 2022 will be tested for spinal muscular atrophy, in addition to the other disorders already on the screening panel.

The practitioner's manual has been updated to include SMA. This will be available closer to the implementation date at www.bitly.com/nbs-resource.

How will the result report change?
For first screen specimens, the result table on the report will include Spinal Muscular Atrophy. Below is an example of the result table. The screening test specifically looks for the presence or absence of exon 7 of *SMN1*.

- If exon 7 is **present**, this is a **normal** result and the baby is NOT at risk for SMA.
- If exon 7 is **absent**, this is an **abnormal** result. The baby is at high risk for SMA and will require immediate referral to a pediatric neurologist.

Screening Test	Analyte Result	Disorder Evaluation	Reference
Congenital Hypothyroidism	T4=	Normal	T4= 5.0- 35.0 ug/dL
Congenital Adrenal Hyperplasia	17OHP=	Normal	17OHP <= 90 ng/mL
Cystic Fibrosis	IRT=	Normal	IRT < 60.0 ng/mL
Hemoglobinopathies	FA	Normal	FA= Normal
Biotinidase	Has color	Normal	Normal Has Color
Galactosemia	GALT Normal	Normal	Normal
Amino Acid Profile (Includes PKU)	Normal	Normal	Normal
Fatty Acid Oxidation Profile	Normal	Normal	Normal
Organic Acidemias	Normal	Normal	Normal
Severe Combined Immunodeficiency	Normal	Normal	Normal
Spinal Muscular Atrophy	Exon 7 SMN1 Present	Normal	Normal

The screening test for SMA will not be performed on a second screening specimen, unless the first specimen is unsatisfactory or abnormal. Therefore, there will be no change to the report for second screen specimens unless repeat testing for SMA was requested.

How do birth facilities and health care providers implement this change?
There is no change to the process for collection of the first and second newborn bloodspot specimens. Birth facilities and health care providers should continue to collect specimens and review result reports for each infant.

If there is an abnormal result, you will be given instructions on the report for additional actions that should be taken immediately. A phone call by the newborn bloodspot screening program will also accompany this abnormal result report.

What is Spinal Muscular Atrophy?
Spinal muscular atrophy (SMA) is a genetic disorder with an incidence of approximately 1 in 10,000 live births. SMA is caused by a lack of the spinal motor neuron (SMN) protein, which is essential for muscle development. Individuals affected with SMA have impaired nerve cells in the brain stem and spinal cord leading to difficulty in speaking, walking, breathing, and swallowing. The OSPHL will screen for SMA using a molecular test targeting exon 7 in *SMN1* which is absent in 95% of all affected individuals. Newborns identified to be at risk for SMA will be referred to pediatric neurologists for immediate evaluation and diagnostic testing. Treatment will be initiated as needed. There are currently FDA-approved therapies for SMA, enabling substantial improvements to the quality of life for affected individuals.

Please refer to this the ACMG fact sheet for more information about newborn screening for Spinal Muscular Atrophy. https://www.acmg.net/PDFLibrary/SMA-ACT-Sheet.pdf

For more information about the Oregon Newborn Bloodspot Screening Program, please visit https://www.oregon.gov/oha/ph/laboratoryservices/newbornscreening/Pages/index.aspx

Why did OSPHL decide to add this test to their screening panel?
The U.S. Secretary of Health and Human Services approved the addition of Spinal Muscular Atrophy to the Recommended Uniform Screening Panel for Newborn Bloodspot Screening in 2018. The Northwest Regional Newborn Bloodspot Screening Advisory Board, who advises the OSPHL on their screening panel, reviewed the condition and unanimously decided to add SMA in 2020. In order to meet national standards and comply with the recommendations of the advisory board, the OSPHL will perform testing for SMA starting on June 1, 2022.

If you have any questions regarding details for change, you may contact our NBS program at 503-693-4172 or at patrice.k.held@dhsoha.state.or.us.

Sincerely,

Patrice Held

Patrice K. Held, PhD, FACMG
Newborn Screening Program Manager
Oregon State Public Health Laboratory
Oregon Health Authority, Public Health Division