LabLink

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Director, Bureau of Laboratories
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this issue...

Bureau Vision

The Bureau of Laboratories is a stronger, more diverse team within an integrated public health system. We utilize advanced technology and innovative leadership to provide comprehensive public health services in our dynamic global community.

Bureau Mission

We are dedicated to continuing leadership in providing quality laboratory science for healthier people and communities through partnerships, communication and technical innovation.

cap ACCREDITED

MDHHS
Michigan Department of Health & Human Services

Dr. Marty K. Soehnlen, National Award Honoree

The Association of Public Health Laboratories (APHL) awards and honors the Unsung Heroes of Public Health. The Awards Program highlights outstanding achievements in laboratory science, creative approaches to public health challenges, and exemplary support of laboratories serving public health and their communities.

Dr. Marty K. Soehnlen PhD, MPH, PHLD(ABB), Infectious Disease Division Director for the Bureau of Laboratories (BOL) was an award honoree. Dr. Soehnlen received the Emerging Leader Award, that honors a laboratorian whose leadership has been instrumental in one or more advances in laboratory science, practice, management, policy, or education within five to ten years of working at a publicly-funded laboratory that conducts testing of public health significance. This recipient must also be employed by an APHL member institutional laboratory. Dr. Soehnlen was nominated by three peer public health laboratories located across the United States and was voted to receive this award by an APHL Awards Vetting Committee.

Dr. Marty Soehnlen began her career as a medical technologist and molecular epidemiologist, followed by an Emerging Infectious Disease fellowship with the CDC Rabies Unit, and then earned a PhD within the Department of Veterinary and Biomedical Sciences at Penn State University. She served as Deputy Chief of Veterinary Pathology within the US Army Public Health Command stationed in Europe and later became their Chief of Microbiology and Molecular Biology. In 2015, she became the Microbiology Section Manager at the Michigan Department of Health and Human Services Bureau of Laboratories (MDHHS BOL) where she provided critical leadership, technical support, and oversight of laboratory operations during the Flint, Michigan water crisis and Legionnaires' disease outbreak. Her leadership and commitment afforded her promotion to Division Director of Infectious Disease in 2017.

Dr. Soehnlen's contributions have enabled MDHHS BOL to serve as the PulseNet USA Midwest Regional Laboratory and also as the National TB Molecular Surveillance Center for Whole Genome Sequencing. She provides testing expertise, training, and support of outbreak investigations across the Midwest and the United States. She is passionate, committed, energetic, and a source of inspiration for those interested in scholarly and public health laboratory pursuits. Dr. Soehnlen is a graduate of the APHL Emerging Leaders Program Cohort 8, Vice-Chair of the APHL Infectious Disease Committee, a member of the APHL Workforce Development Committee, and Chair of the APHL Next Generation Sequencing Subcommittee. She also spent time in Uganda assisting the Ministry of Health set-up their National Public Health Laboratory.

Other accomplishments include Director for the South Central Association of Clinical Microbiology, Co-Chair of the 2019 HIV Diagnostic Conference, an invited member to both the 2018 CLIAC NGS Workgroup and the 2017 Syphilis National Guidelines Workgroup. She is currently a member of the CDC NGS Quality Management Technical Coordinating Committee, the ABB/ABMM Test Prep Workgroup, and serves on the Doctor of Public Health Lecture Development Workgroup for the Public Health Laboratory Molecular Diagnostics and Molecular Biology course.

Spinal Muscular Atrophy Added to the Newborn Screening Panel

Newborn Screening is a public health program required by Michigan law to find babies with rare but serious disorders that require early treatment. All babies need to be tested to find the small number who look healthy but have a rare medical condition. Each year more than 250 Michigan babies are found to have a disorder detected by newborn bloodspot screening. As of March 2020, the Newborn Screening Program added Spinal Muscular Atrophy (SMA) to the screening panel.

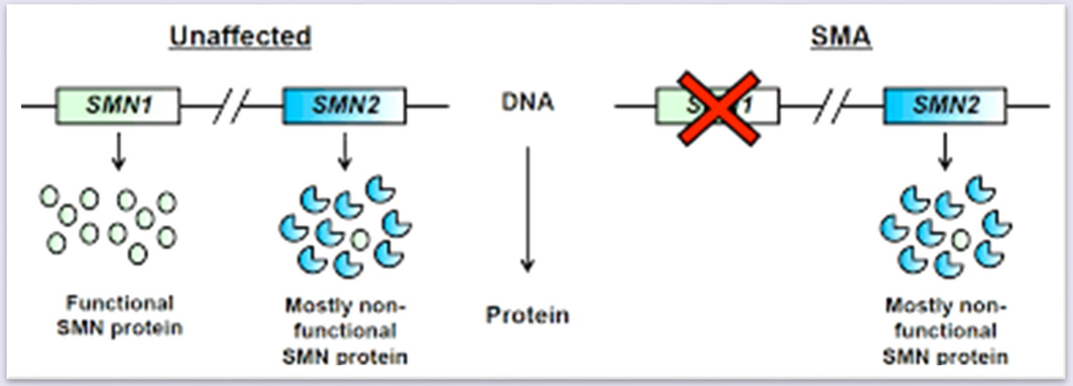
SMA is characterized by muscle weakness and atrophy of the skeletal muscles through the deterioration of motor neurons that control muscle movement. A child will be noticeably weak and delayed in developmental milestones. Symptoms also include trouble breathing, coughing, and swallowing. There are five types of SMA with symptoms ranging from moderate to severe.

Treatment should occur as soon as possible, prior to patients becoming symptomatic to prevent non-repairable motor neuron loss. Without treatment, the life expectancy of the most common type of SMA is 2 years of age. There are three FDA approved drugs available for treatment.

MICHIGAN Newborn Screening

	FDA Approved Drug Therapies		
	Spinraza	**Zolgensma**	**Evrysdi**
Age to take	Anytime	< 2 years old	> 2 months old
Dosing	Every 4 Months	Once	Daily
Administration	Intrathecal Injection (IT)	Intravenous Infusion (IV)	Oral
Duration	Lifetime	Once	Lifetime

SMA is caused by mutations in the survival motor neuron (SMN) gene. There are two highly homologous copies that have only five base-pair differences, SMN1 and SMN2. SMN1 produces full-length transcriptional mRNA, and SMN2 produces a small amount of full-length mRNA and due to splicing differences, primarily produces an attenuated mRNA transcript that lacks exon 7 of the gene. Ninety-five percent of SMA individuals are homozygous for the deletion of exon 7 in the SMN1 gene. The severity of the disease correlates to SMN2 copy number and the amount of functional protein present. Treatment options focus on correcting splicing errors on RNA produced from SMN2 gene (Spinraza), gene therapy to replace/repair the SMN1 gene (Zolgensma) and increasing functional SMN protein through the SMN2 gene (Evrysdi).



SMA screening method detection finds the absence of the SMN1 exon 7 deletion by Real-Time PCR (RT-PCR). The SMN1 target was multiplexed with the already established RT-PCR assay detecting Severe Combined Immunodeficiencies (SCID) with RNaseP as a reference gene to ensure satisfactory specimens. The SMN2 copy number is not determined in the newborn screening laboratory.

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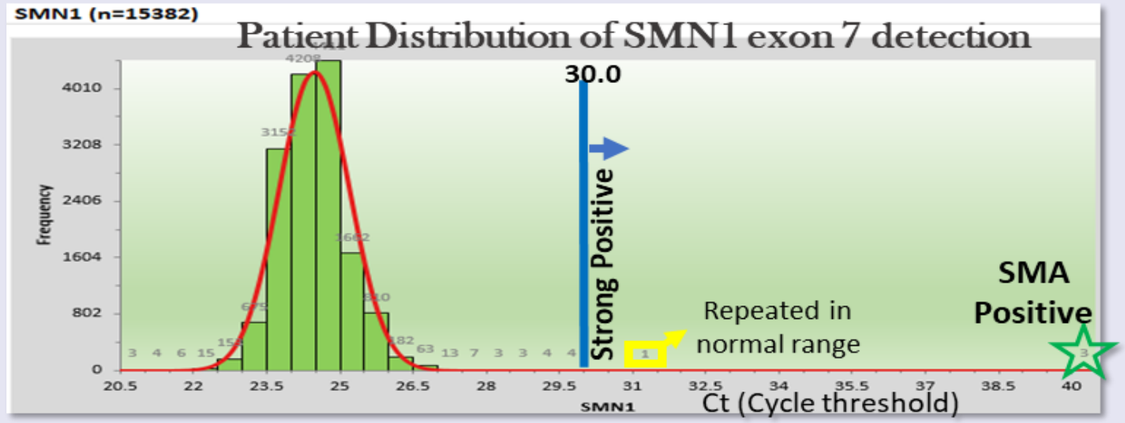
...continued from page 3, **Spinal Muscular Atrophy**



Michigan has screened over 92,000 babies in the past ten months and has identified 10 patients with SMA. All 10 patients were confirmed to have Spinal Muscular Atrophy. Michigan's incidence rate of SMA detection is 1 in 9,243 to date.

Duration (11/4/2019 to 9/22/2020)	
Total Babies Screened (initials)	92,434
Total SMA Positive Screens	10 patients (12 specimens)
False Positive Rate	0% to date
False Negative Rate	0% to date
Positive Predictive Value* (PPV)	100% to date

References:

1) Prior TW, Snyder PJ, Rink BD, Pearl DK, Pyatt RE, Mihal DC, Conlan T, Schmalz B, Montgomery L, Ziegler K, Noonan C, Hashimoto S, Garner S. 2010. *Newborn and carrier screening for spinal muscular atrophy*. Am J Med Genet Part A 152A:1608–1616
2) Pyatt RE, Prior TW. 2006. *A feasibility study for the newborn screening of spinal muscular atrophy.* Genet Med 2006:8(7):428–437.
3) CureSMA.org

LabLink is published quarterly by the Michigan Department of Health and Human Services Bureau of Laboratories, to provide laboratory information to Michigan health professionals and the public health community.

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