

CADHAM PROVINCIAL LABORATORY

Screening for Spinal Muscular Atrophy Added to Manitoba Newborn Screening Program

Date issued: May 25, 2022

To: Manitoba Newborn and Pediatric Care Practitioners & Other Relevant Care Providers

Beginning June 1, 2022, all dried blood spots received for newborn screening (NBS) at the Manitoba Newborn Screening Program at Cadham Provincial Laboratory (CPL) will also be screened for Spinal Muscular Atrophy (SMA).

SMA is an autosomal recessive neurodegenerative condition that occurs in approximately 1:18,000 births (recent Ontario data). The condition causes degeneration of motor neurons that ultimately results in widespread muscular atrophy. Approximately 95 per cent of SMA is due to a deletion of the exon 7 segment of the SMN1 gene, which is critical for motor neuron survival. SMA has 4 types of presentation ranging from severe, in early infancy (type 1), to less apparent adult presentations of disease (type 4). The severity is largely dictated by the number of related SMN2 gene copies that are present.

The Manitoba newborn approach will be similar to other centres using the Centres for Disease Control (CDC) assay which specifically screens for the exon 7 deletion. If there is no exon 7 target detected, then the blood spot will be sent to a reference centre for quantification of SMN2 gene copies. Any positive screens will also be re-confirmed by requesting a second blood spot. Results will be available by faxed report, and in eChart, and positive screens will also provide the SMN2 gene copy number.

Any positives will be immediately referred to Pediatric Neurology for timely assessment and will be coordinated with the infant's primary care practitioner. Effective treatments are available to significantly improve the outcomes of children affected by SMA but rely on very early detection to make the biggest impact. Please direct any screening questions to paul.vancaesele@gov.mb.ca or call the Newborn Screening Section at 204-945-7458.

More Information:

Relevant resources located at this link are in the process of being updated:

<https://www.gov.mb.ca/health/publichealth/cpl/baby.html>

Contact Information:

Dr. Paul VanCaesele, MD FRCPC, Chair, Manitoba Newborn Screening Committee & Medical Director, Cadham Provincial Laboratory, Shared Health