



# Newborn Screening Bulletin 2020-1

January 15, 2020

## SCREENING PILOT FOR SPINAL MUSCULAR ATROPHY

Starting on January 13th, 2020, newborn screening samples are screened for Spinal Muscular atrophy (SMA). SMA is a rare inherited (genetic) disease that results in muscle weakness and atrophy (muscle wasting). Many babies with SMA will not have symptoms at birth. Treatment has recently become available for babies with SMA and is effective. Without treatment the more severe, early onset forms of SMA will lead to death in the first year or two of life.

SMA will be screened for a few months in early 2020 (as a pilot) before it is expected to be added to the list of diseases screened in Ontario. One reason it is being run as a pilot is that effective treatment has recently become available for this condition. Screening babies as soon as possible (in the form of a pilot) means that more babies with SMA can be identified and started on treatment now. As well, phasing in screening allows time to develop regulatory and policy changes required in the province.

Babies who have their blood taken for dried blood spot screening in Ontario will also be screened for SMA. Families that decline dried blood spot screening will not be eligible for SMA screening.

Screen positive results will be reported for SMA directly to the referral centres. Screen negative results will not be reported but would be available upon request by the family or health care provider. **There will be no change to the screening results report during this pilot.**

**Effective immediately, please inform families of this additional screen. Stickers have been printed and mailed to you—please affix these to the Parent Information Sheet attached to the dried blood spot forms so that parents/guardians know where to go for additional information.** We will continue to regularly replenish the supply of stickers during the pilot but please contact us if you require more. If you receive stickers in the mail, please forward them to any departments involved in regular newborn screening collection (i.e. mother baby unit, lab, NICU/SCN, etc).

If you have any questions, please contact us at 613-738-3222 or by email at [NSO@cheo.on.ca](mailto:NSO@cheo.on.ca)

