Title: Current State of Neonatal Testing of Spinal Muscle Atrophy in Canada and Latin America

Spinal muscular atrophy (SMA) is an autosomal recessive condition marked by the irreversible loss of motor neurons, causing increasing muscle weakening and atrophy of the limb, trunk, bulbar, and respiratory muscles, making it difficult to breathe, feed, walk and sit.(Groulx-Boivin et al. 2023; Niri et al. 2023) SMA is one of the leading genetic conditions (1 in 10,000 live-born infants) contributing to infant mortality. (Groulx-Boivin et al. 2023) Severe forms of SMA progress rapidly, early signs are sometimes undiagnostic, and it may take months to diagnose, and treatment may not be entirely possible by then. Before 2016, the main goals of SMA therapy were symptom control and supportive care. Effective disease-modifying treatments (DMTs) have demonstrated significant promise in halting disease development.(Niri et al. 2023) Clinical research has also shown that the medication is most successful when started before the onset of symptoms. Therefore, reducing morbidity and mortality depends on early diagnosis. Early diagnosis is only possible if all infants are screened at birth. With this vision, SMA has been included in many NBS programs. Even the cost-effectiveness analysis has shown that universal NBS for SMA is economically beneficial in regions and nations where disease-modifying treatment is accessible. (Groulx-Boivin et al. 2023)

Spinal muscular atrophy (SMA) is the most recent disease added to NBS programs in Canada.(Groulx-Boivin et al. 2023) As of January 2023, approximately 72% of newborns in Canada are screened for SMA at birth.(Groulx-Boivin et al. 2023) It is not 100% since no national screening panel for inherited conditions exists in Canada, and provincial governments decide which diseases should be included in NBS programs. With Ontario being the first Canadian province to begin screening for SMA (January 2020), it has been adopted in Alberta, Manitoba, Ontario, Saskatchewan, and Nunavut/Northwest Territories.(Groulx-Boivin et al. 2023) It is planned in Quebec and Nova Scotia. The absence of or delay in state financing for its inclusion was the most often cited barrier, followed by a lack of human and financial resources. (**N**iri et al. 2023) Ontario has no false positive cases, whereas no sufficient studies have been done regarding this in other provinces.(Groulx-Boivin et al. 2023)

The majority of Latin American nations have national and/or regional newborn screening programmes; however, not all of them now test for SMA. In Brazil, legislation was approved in June 2021, requiring all states to start newborn screening programmes by June 2022 and gradually add spinal muscular atrophy. However, current difficulties could prevent the

accomplishment within the allotted period.(Giugliani et al. 2022) SMA is planned for the fifth stage of the expansion in Brazil.(Brinckmann et al. 2023)

To conclude, we can say that Canada is far ahead of Latin America in the successful implementation of SMA in the NBS program. Although Latin America has an implementation of neonatal screening for various pathologies, SMA is not included in it yet.

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