Canada's Drug Agency
L'Agence des médicaments du Canada

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Consultation Summary From Focus Group and Key Informant Interviews on Proposed Pan-Canadian Guidance for Newborn Screening

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Background

In March 2023, the Government of Canada announced investments to support the National Strategy for Drugs for Rare Diseases. Canada's Drug Agency was asked to support key activities for this strategy under pillar 3, "collect and use evidence," to help optimize access to drugs for rare diseases in Canada and to support decision-making. As part of this, Canada's Drug Agency convened an advisory panel to provide guidance on newborn screening with the aim of enhancing the pan-Canadian coordination of newborn screening including the consistency of conditions screened for in newborns across Canada's provinces and territories.

The discussion paper to engage with interested parties, Pan-Canadian Guidance for Newborn Screening: Building the Foundations for Early Diagnosis and Detection in Canada, was published in July 2024. It included a potential set of guiding principles, explored a potential process and criteria for adding or reassessing conditions screened for, and a potential set of conditions for which newborn screening programs in Canada could screen.

To gather the perspectives of diverse interested parties on the discussion paper, individuals and organizations were invited to complete an online consultation form either in French or English from July 11, 2024, to September 11, 2024. The advisory panel wanted to ensure that the perspectives of those made vulnerable by social and/or economic policies; those who are disproportionately affected by newborn screening due to the higher prevalence of inherited disorders; and persons who are First Nations, Métis, or Inuit were solicited and incorporated into the guidance. Canada's Drug Agency engaged Sage Solutions to conduct 3 distinction-based focus groups with individuals from First Nations, Métis, and Inuit communities; further details about the approach and key themes and perspectives can be found in the Consultation Summary From Focus Groups With First Nations, Inuit, and Métis Peoples on Proposed Pan-Canadian Guidance for Newborn Screening.

This document summarizes the perspectives and themes emerging from focus group discussions and key informant interviews with participants who were midwives or doulas who were themselves from or worked with people made vulnerable by social and/or economic policies or who are disproportionately affected by newborn screening due to higher prevalence of inherited disorders.

Approach

Canada's Drug Agency organized focus group discussions to elicit deep and meaningful input from groups we typically would not have the opportunity to engage with. We issued 23 invitations to individuals, organizations, and groups that represent or provide service to people who are made vulnerable by social and/or economic policies or who are disproportionately affected by newborn screening because of a higher prevalence of inherited disorders. Six individuals accepted the invitation to participate. Canada's Drug Agency conducted a focus group discussion with 3 individuals and conducted 3 key informant interviews with participants in September and October of 2024. To validate that the summary accurately reflected participants' perspectives, all participants were provided a draft version of the report for their review.

Discussions with participants solicited input on 3 key areas of the proposed guidance: the proposed guiding principles, the proposed criteria for adding conditions to a pan-Canadian newborn screening list, and the list of emerging conditions.

What We Heard: Proposed Guiding Principles

Each of the guiding principles and their definitions were shared with the participants. Participants expressed that they agreed that the proposed principles were clear and that they appreciated their intent. Through discussion, participants raised questions and shared their perspectives on how each of the definitions for the guiding principles could be clarified or be more clearly expressed. Participants also shared perspectives on the interpretation and application of the guiding principles and their implications for those who are made vulnerable by social and/or economic policies or who are disproportionately affected by newborn screening because of a higher prevalence of inherited disorders.

Health Rights of the Newborn

The proposed principle of the health rights of the newborn was defined as follows:

- Policies, processes, and procedures relating to newborn screening should prioritize the health of the newborn. When considering what conditions to screen for in newborns, the focus should be on reaching the highest attainable state of health for newborns.

It was shared that the advisory panel intended for this to be an overarching principle for all newborn screening activities, such that each of the guiding principles be linked to and interpreted with a focus on the health rights of the newborn.

Key Themes

- When speaking about the health rights of the newborn, participants observed a gap in terms of birthing people and families not being included in the definition.
- Participants shared a desire for a family-centred approach to be included in this definition. Families are typically the ones that tend to babies and care for their needs, and so it is important for the guiding principle on health rights of the newborn to reflect their role.
- It was not clear to participants who gets to decide what is in the best interests of the newborn and their health. From an equity perspective, participants raised questions about historical and present-day situations where health care systems and the state decided what was in the best interests of a newborn or child without regard for the family’s perspective. They wondered if, should there be conflict — for example between a health care provider’s or institution’s perspective and a parent’s or caregiver’s perspective — how the newborn’s best interests are determined, recognizing that birthing people are often in a vulnerable position compared to the health care provider or the institution.
- Participants also raised questions about people who are not insured, including those who are newcomers to Canada and those who decline participation in provincial health insurance programs.

- Participants wondered if 1 way this principle might be applied is by being aware of new conditions that might need to be considered for screening as a result of changing demographics, including influxes of newcomers.

Equity

The proposed principle of equity was defined as follows:

- Policies, processes, and procedures relating to newborn screening should ensure access for all newborns to quality screening, and to diagnosis, treatment, and follow-up where appropriate. When considering what conditions to screen for in newborns, their diverse needs, circumstances, and best interests need to be considered.

Key Themes

- Overall, participants felt that the definition for equity was clear and appropriate. Participants wanted to further acknowledge the access issues that exist in the current system. For those living in a remote location, a diagnosis of a rare disease in a newborn could result in them facing a significant burden due to the need to travel for care, including lost income and out-of-pocket-costs, and it may result in the family moving to access the care the newborn needs.
- Participants also shared that, when thinking about equity and access, screening practices and available treatments differ across jurisdictions, and there are shifting demographics and mobility among certain populations (e.g., Amish Mennonites). They noted that, currently, individuals who move receive different access to newborn screening and care based on the jurisdiction they are in.
- Some participants described the importance of defining quality, recognizing that the standard of quality is resource dependent and will vary by jurisdiction.
- Participants saw an opportunity to interpret the principle of equity through a family-centred lens by recognizing the need for culturally and linguistically appropriate materials for newborn screening.
- On a related point, participants saw a need for the principle of equity to be interpreted to include access to information that is factual, relevant, and in a context people can understand.

Effectiveness, Safety, and Quality

The proposed principle of effectiveness, safety, and quality was defined as follows:

- Policies, processes, and procedures relating to newborn screening should be actionable, regularly reviewed, evaluated, modernized, and updated for continuous improvements. Newborn screening pathways should be effective, safe, evidence-informed, and of high quality.

Key Themes

- Participants noted that the quality of the blood sample collected by the health care provider is important because, if the sample is inadequate, another sample will need to be collected. Repeat collection adds to the burden of testing on the newborn and the family. It can also contribute to a delay in the return of results and, if the screening results are positive, a delayed diagnosis.

- Participants highlighted the use and need for protocols to limit harm during sample collection (i.e., heel prick test), including comfort measures to reduce pain in the newborn and distress for the family.
- Participants said they considered a critical element of this principle to be the timely return of test results. Some participants noted that there is a timely return of results (both positive and negative) and low false-positive rates, which they saw as indicators of a quality system. However, participants were aware that there is a high degree of variability in newborn screening across Canada.
- Participants raised the idea that this principle could benefit from incorporating a person-centred approach, recognizing that a one-size-fits-all approach may not work or be appropriate.

Transparency

The proposed principle of transparency was defined as follows:

- Policies, processes, and procedures relating to newborn screening, as well as the work of the advisory panel, should be explicit, impartial, clear, and accessible to all people in Canada. Information about newborn screening should be accessible, accurate, and easy to understand.

Key Themes

- Participants appreciated that this was a guiding principle. They asked if it might include the concept of relevance in addition to being explicit, impartial, clear, and accessible. For 1 participant, this specifically meant providing both absolute and relative risks of having a condition.
- When thinking about transparency, participants raised the importance of being clear and open about the process of opting out of newborn screening and about the uses and storage of the blood spot sample. This is particularly important considering some groups have been subjected to unjust and unethical medical experimentation and research and have had their information used without their knowledge or consent.

Collaboration

The proposed principle of collaboration was defined as follows:

> Policies, processes, and procedures relating to newborn screening should be developed in collaboration with partners and diverse perspectives.

Key Themes

- Participants drew on their past experiences when the need for collaboration was voiced or written down but never actioned in a meaningful way. Therefore, participants sought additional clarity on the term *collaboration*. They asked: “what is the definition of collaboration and what types of activities does the definition include?”
- On a related point, participants interpreted collaboration as requiring clear pathways for information sharing and clearly defined roles and responsibilities of included parties. For example, a participant who was a midwife shared their experience of not always being included in newborn screening pathways (e.g., results are being returned to hospitals or family doctors, but not to them, the primary care provider).

- They also asked what was meant by *partners* and wanted to ensure that the definition included families, community-based organizations, and others affected and impacted by newborn screening.

Sustainability

The proposed principle of sustainability was defined as:

- Policies, processes, and procedures relating to newborn screening should focus not only on creating value to support a sustainable health system in the present, but should also include considerations for future generations, such as environmental, economic, and social factors. They should also support a holistic, long-term vision of improving health systems and the public's health.

Key Themes

- Participants appreciated that sustainability was a guiding principle.
- Participants wondered, when thinking about future generations and social factors, if there is an opportunity to explicitly include the social determinants of health. A participant raised the view that the health of newborns is very often an indicator of the health of communities. They also shared the perspective that affected communities need to be involved in identifying and implementing solutions because they know what they need.
- Participants shared concerns about the sustainability of newborn screening, particularly given the increased number of conditions potentially being screened for and the impact on health systems of the resources needed to do so.

What We Heard: Proposed Criteria for Adding Conditions

The proposed criteria for adding a condition to a potential pan-Canadian Newborn Screening List were reviewed. There was general agreement among participants that the proposed criteria outlined in the discussion paper were appropriate and clear.

The Condition

- Participants appreciated the inclusion of the recognition of differences in the incidence of the condition and variation in test performance in subpopulations. For participants who worked with specific subpopulations, this was a critical concern (i.e., ensuring specific pathogenic variants are included in newborn screening). They also recognized that currently, in their jurisdiction, some screening is not publicly funded (e.g., the service is provided by local research facilities/laboratories). They worried about people from subpopulations being able to access future screening, particularly as they moved to other regions or jurisdictions in Canada.
- Participants wondered if there is an opportunity to consider the cultural understanding of the condition or symptoms, in terms of etiology and treatment, from the perspective of the affected community.
- Participants felt that there is a need to ensure that conditions are significant enough to have continued support from the public and families for newborn screening. Some participants wondered if

an incidence threshold could be used when deciding whether to add a condition because it could help assess whether it is common enough to be included.

The Test

- Participants voiced that these criteria were appropriate, clear, and comprehensive.
- They asked if there were standard operating procedures that might reduce the harms of testing and if these would be considered.

The Treatment

- Participants voiced that these criteria were appropriate, clear, and comprehensive.
- Some participants shared that they thought it was very important for a condition to have a treatment, but they also wanted the treatment to include interventions (such as diet and behaviour changes) that could prevent morbidity from the condition. Others thought that providing families or caregivers with information on a condition that had severe morbidity or mortality was valuable in and of itself.
- Other participants shared the perspective that it is important to consider other cultural practices or treatments that might be considered by some populations — in particular, to anticipate differences in opinion on what is considered an effective therapy. They highlighted the backdrop of racism and discrimination in Canada’s health systems that might make people reticent to engage in conventional health care, as well as peoples’ cultural beliefs.

Other Considerations

- Participants voiced that these criteria were appropriate, clear, and comprehensive.
- Participants shared their concerns and experiences of accessing treatment in rural and remote locations. In their view, it is very important to recognize the need to have treatment available across Canada and provide supports (e.g., logistical, financial) so that newborns and their families have access to care.
- Participants shared how the social determinants of health were critical when considering what access requires, and they shared examples of how families and caregivers experienced difficulty travelling to medical appointments because of a lack of child care or lack of transportation options for their family.
- In terms of access, participants raised questions about access for people without insurance, including newcomers.
- Participants felt it important to consider the noneconomic impact of receiving the information provided through newborn screening in terms of the benefits to newborns and families.

What We Heard: Anticipating Emerging Newborn Screening Conditions

It was suggested that some of the conditions that are part of a targeted screening of Amish Mennonites living in Southwestern Ontario (CYP1B1, HARS [Usher syndrome 3B], TMPRSS4, and CTNS) might be monitored for relevance for pan-Canadian screening as demographic shifts lead to communities being established in other jurisdictions. These conditions include pathogenic variants of:

- the CTNS gene, which causes infantile cystinosis
- the TMPRSS4 gene, which causes autosomal recessive cerebral atrophy
- the HARS gene, which causes Usher syndrome 3B, leading to congenital or childhood-onset sensorineural hearing loss
- the CYP1B1 gene, which leads to congenital glaucoma.

Additional Considerations

Participants provided important feedback on themes that extended beyond the scope of the 3 sections of the guidance that were discussed.

- **Timely delivery of newborn blood screening samples:** Participants reflected on how, for those who reside in remote locations, it takes longer to deliver newborn blood samples, resulting in delayed diagnosis.
- **Education for health care providers:** Participants indicated that education should be available for health care providers so they can clearly discuss the risks and benefits of newborn screening and explain why it is important to screen for these conditions. Participants also acknowledged that education on how to discuss newborn screening if a parent or caregiver initially declines it and how to discuss positive screen results would be helpful. Many health care providers do not encounter those situations often and, therefore, may not know how to manage them.
- **Information for parents or caregivers:** Participants acknowledged that language can be a barrier, and some individuals consent to newborn screening without knowing what they are consenting to. Newborn screening resources should be easy to understand, available in multiple languages, and use visuals to support the content. The resources should also be designed with a broad audience in mind because other family members or members of the community may influence a parent or caregiver’s choice to have a newborn screened. Participants also reflected on how newborn screening is often only mentioned right before the screening test is conducted. They indicated that screening should be discussed early so parents or caregivers have time to digest the information and ask questions before the arrival of the newborn. Lastly, participants identified that more detailed resources could be created to support additional conversations with individuals who decline newborn screening.

- **Discrimination against parents or caregivers who decline newborn screening:** Participants acknowledged that there have been situations where health care providers have treated people differently based on their decision to decline newborn screening.
- **Storage of blood samples:** An area of concern for parents and caregivers is what happens to the blood spot sample after the screening tests are complete. They want to know how long newborn screening samples are stored, who has access to them, and what they are being used for. Information that answers these questions should be available to health care providers, parents, and caregivers.

Acknowledgement

Canada’s Drug Agency and the Newborn Screening Advisory Panel would like to thank all participants in the focus group discussions for their time and contributions.

Appendix 1: Participants

The following is an overview of the individuals who participated in the focus group discussions and key informant interviews.

The participants included 4 registered midwives (2 in Ontario, 1 in British Columbia, and 1 in Newfoundland and Labrador). Two of the midwives were developing midwifery policy and programs, 1 was engaged in midwifery research and quality improvement, and 2 worked in rural and/or remote settings. Two participants were volunteer doulas from the Chebucto Family Centre's volunteer doula program and worked in urban and rural Nova Scotia.

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