

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

# An Update on the Development of Newborn Screening Guidance in Canada

July 11, 2024



# Agenda

- Background, scope, and how you can be involved (10 minutes)
- An inside look at the advisory panel's proposal (30 minutes)
- Next steps, Q&A (15 minutes)

# Background and Scope of Work

# National Strategy for Drugs for Rare Diseases



The Government of Canada announced a total investment of up to \$1.5 billion over 3 years in support of a National Strategy for Drugs for Rare Diseases in March 2023.

A key approach is **supporting improvements in screening and diagnostics** so patients with a rare disease have a better chance of getting access to effective treatments at the right time.

## What Is Newborn Screening?

- In Canada, newborn screening programs are established and funded by provinces and territories.
- Newborn screening refers to the tests that are done shortly after birth to check for serious but treatable diseases.
- It helps to identify certain conditions as early as possible to prevent serious health problems.
- Early identification of rare diseases through newborn screening may lead to more timely diagnosis and appropriate access to treatments and supports early in life.



# Newborn Screening Opportunity

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Provide jurisdictions with the best available information to support decision-making by:



Building on existing work to map current newborn screening coverage across the country



Facilitating engagement with interested parties to ensure the right voices are included



Convening experts to provide advice and guidance

## Benefits

- Leverage lessons learned **and** address common challenges
- Identify solutions to meet the needs of health systems

# Building on Existing Work on Newborn Screening in Canada



## Recommendations from:

- The Intergovernmental Newborn Screening Working Group presented a report to the ministers of health (January 2016)
- Immunity Canada and the Network of Rare Blood Disorder Organizations prepared a report on a national newborn screening program for rare disorders (January 2023)

# The Advisory Panel

- **Comprised of 2 co-chairs and 11 members\***
- Persons from across Canada with diverse representation (gender, race, culture) and perspectives
- Range of expertise and experience, including:
  - health care providers (medicine, midwifery, molecular genetics)
  - persons representing those with lived and living experience
  - persons working with Indigenous and other communities often made vulnerable through a combination of social and economic policy
  - individuals with backgrounds in law, bioethics, health policy, and research



\* Visit our [website](#) for advisory panel details.

# Overview of Project



Panel Meetings  
1 to 4



Engagement



Panel Meetings  
5 and 6



Engagement



Final Report

January to June

July to September

Fall and Winter

Winter

March 2025

**Note:** Interested parties will have an opportunity to provide input via written submission to Canada's Drug Agency from July 11 to September 11, 2024.

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# How You Can Be Involved

# Why Your Involvement Matters

## Your Input is Both Needed and Highly Valuable

- Used to inform the final report that will be submitted to Health Canada
- Shared with provincial and territorial governments
- Made publicly available

## Transparency

- By submitting written feedback, you agree to full disclosure of the information
- Submissions received will be posted on our website
- We will not publish any personal information information

## Summary of What We Heard

- Presented in a webinar in winter 2025

## How to Be Involved

1. Attend today's session
2. Download the **discussion paper** from [https://www.cadth.ca/sites/default/files/hta-he/HC0079\\_NBS\\_Discussion\\_Paper.pdf](https://www.cadth.ca/sites/default/files/hta-he/HC0079_NBS_Discussion_Paper.pdf)
  - Summary of panel's work
  - Questions included in online questionnaire
3. Review questions and **submit response**
  - **Online questionnaire** available at: <https://www.cadth.ca/pan-canadian-guidance-newborn-screening-building-foundations-early-detection-and-diagnosis>
  - Provide feedback via free-text response fields

**Online feedback deadline:** September 11, 2024

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# Advisory Panel's Draft Guidance

# Advisory Panel's Draft Guidance

1. Proposed guiding principles anchoring the advisory panel's approach

2. Exploring a future coordinated model for newborn screening in Canada

3. Opportunities to enhance newborn screening processes for adding, removing, or reassessing conditions

4. Developing criteria for adding and removing conditions to a proposed pan-Canadian newborn screening list

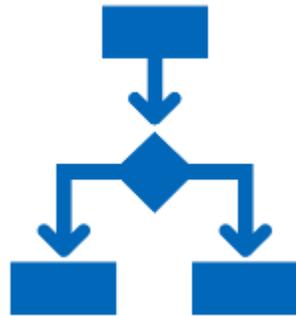
5. Proposed pan-Canadian newborn screening list

6. Anticipating emerging conditions

# 1. Proposed Guiding Principles Anchoring the Advisory Panel's Approach

# Proposed Guiding Principles

## Purpose



Inform and Shape  
Decision-Making



Aspirational  
Values

# Proposed Guiding Principles

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Health Rights of  
the Newborn



Equity



Effectiveness,  
Safety, and Quality



Transparency



Collaboration



Sustainability



Do you agree with the proposed guiding principles and definitions?

## **2. Exploring a Future Coordinated Model for Newborn Screening in Canada**

# Exploring a Future Coordinated Model

A future coordinated model may:



Enhance  
Equity



Enable  
Efficiency in  
Process and  
Resources



Mitigate Risks

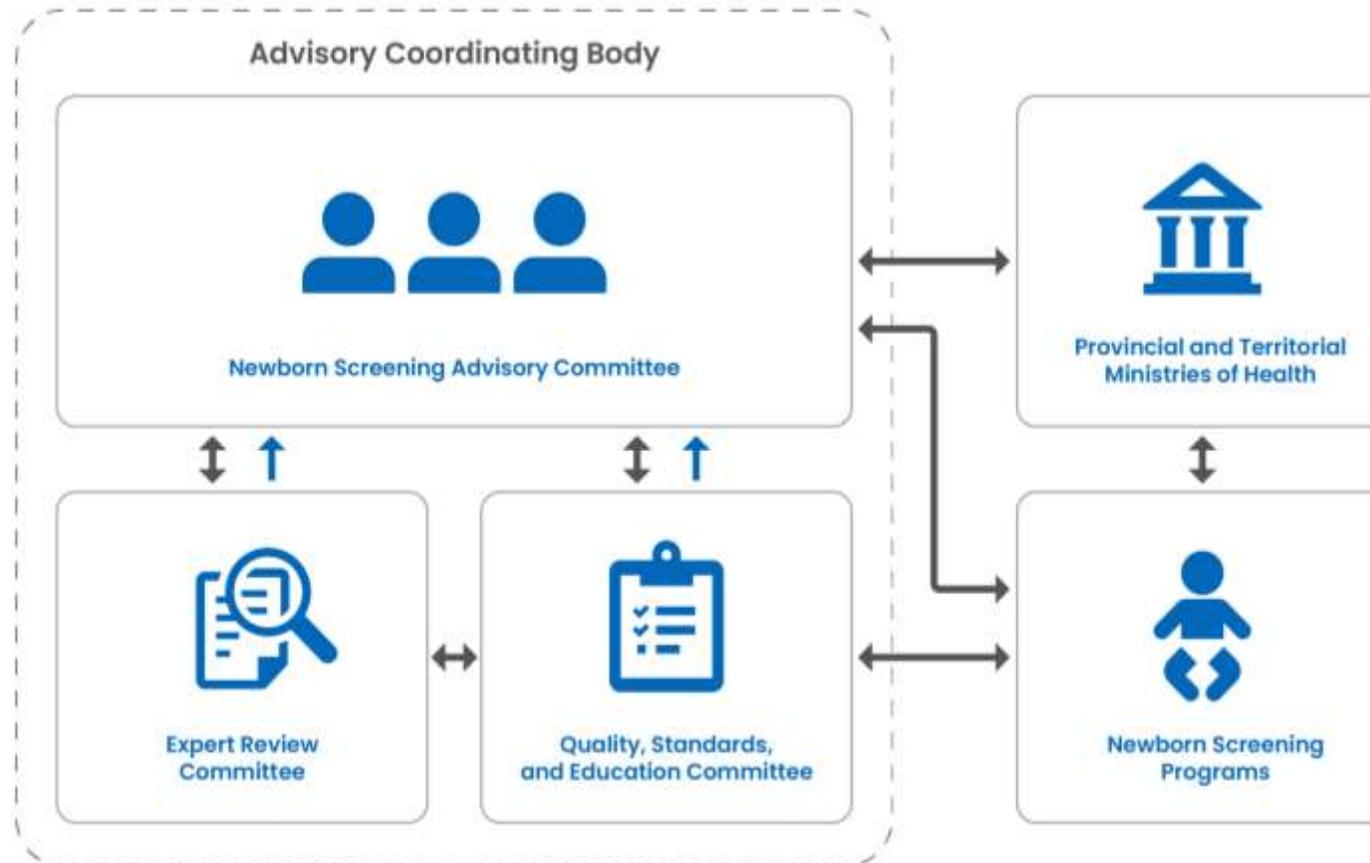


Facilitate  
Quality  
Improvement



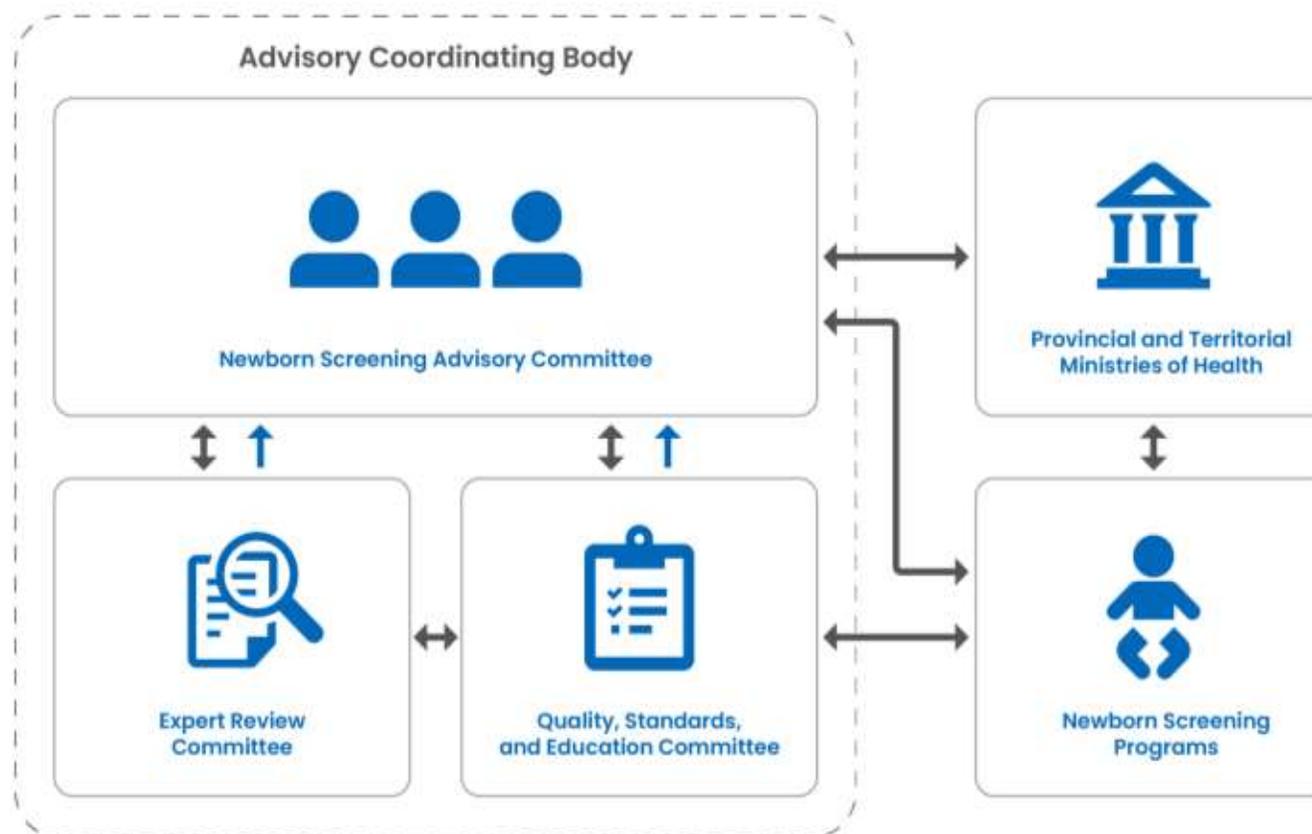
Support  
Anticipating and  
Responding to  
New Challenges

# Illustrative Example of a Potential Coordinated Newborn Screening System



Note: ↔ = Communication and information sharing, → = Accountability

# A Potential Coordinated Newborn Screening System



Note: ↔ = Communication and information sharing, → = Accountability

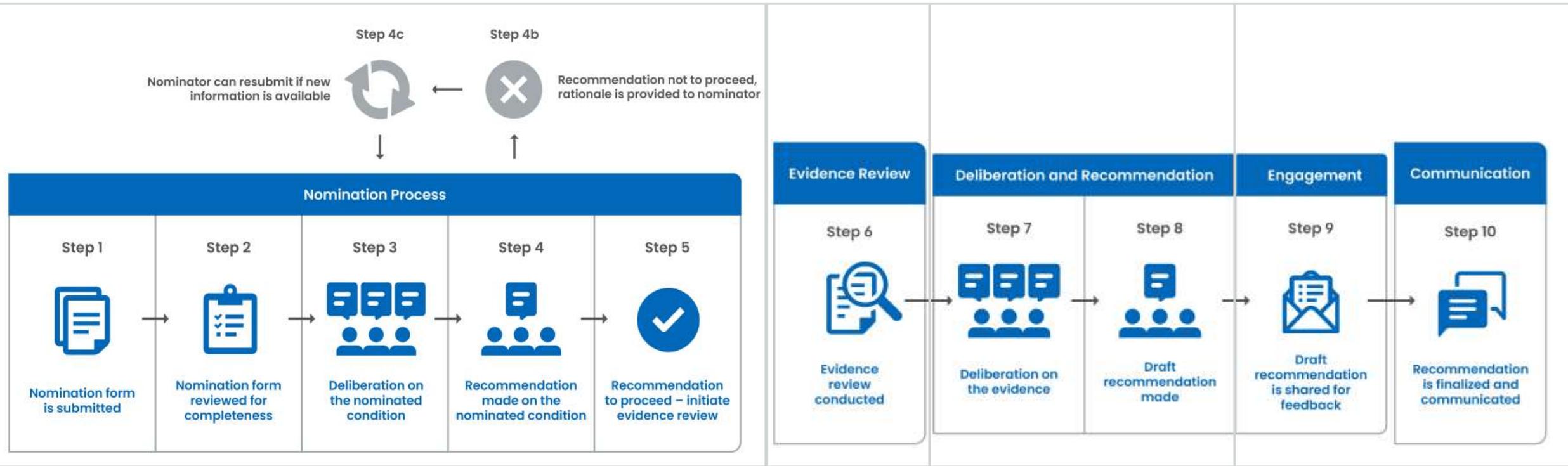


Do you have any specific changes that you would recommend for the proposed approach to the potential coordinated structures for newborn screening?

# **3. Opportunities to Enhance Newborn Screening Processes for Adding (or Removing) Conditions**

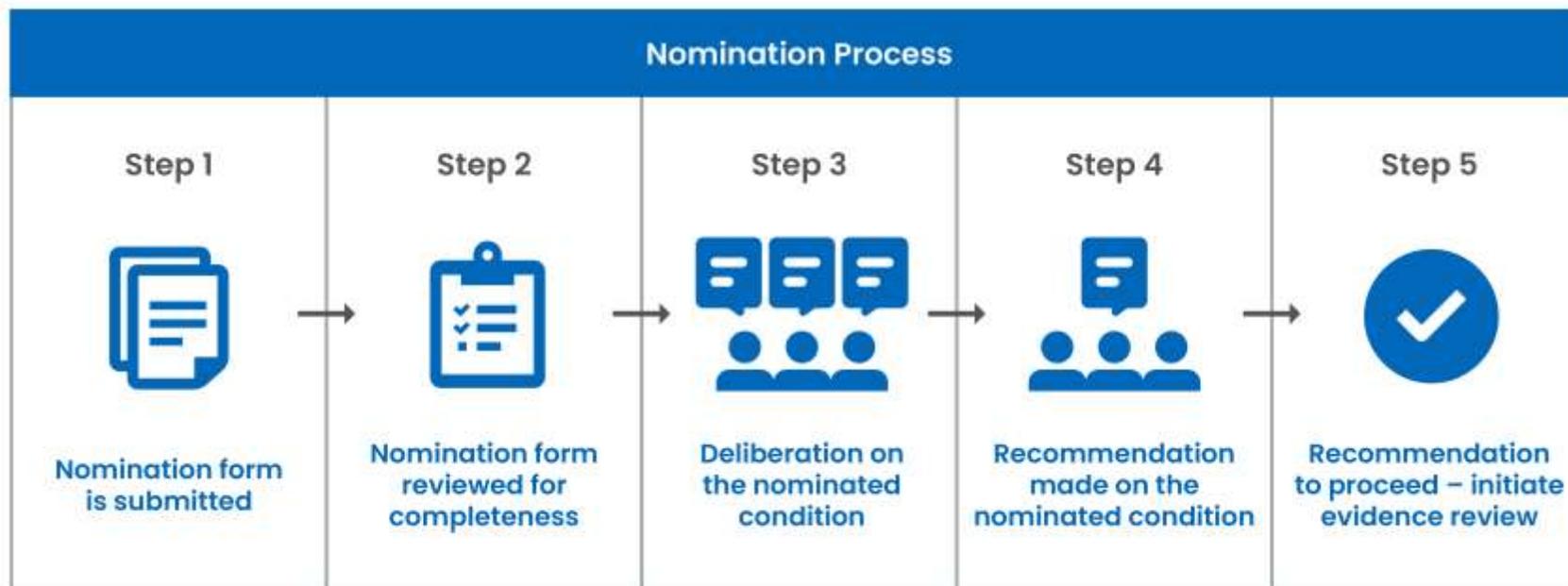
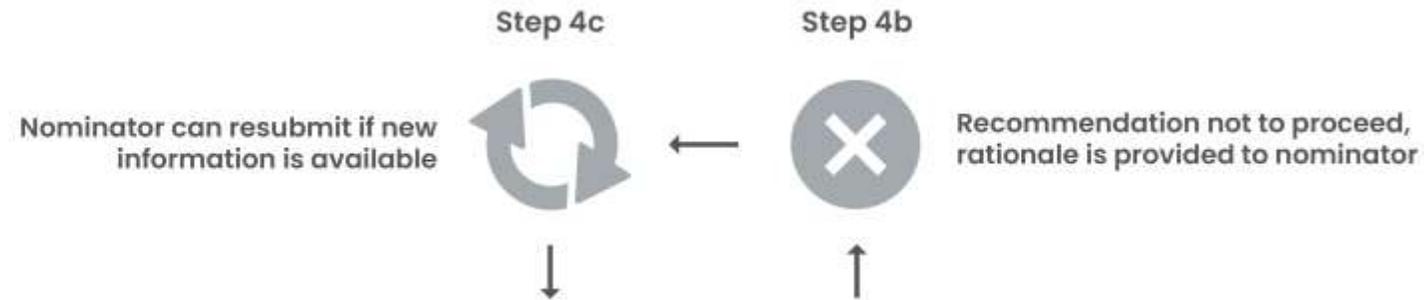
# Proposed Processes to Support Adding (or Removing) Conditions

## Four Sequential and Dependent Processes



# Proposed Processes to Support Adding (or Removing) Conditions

## 1. Nomination Process



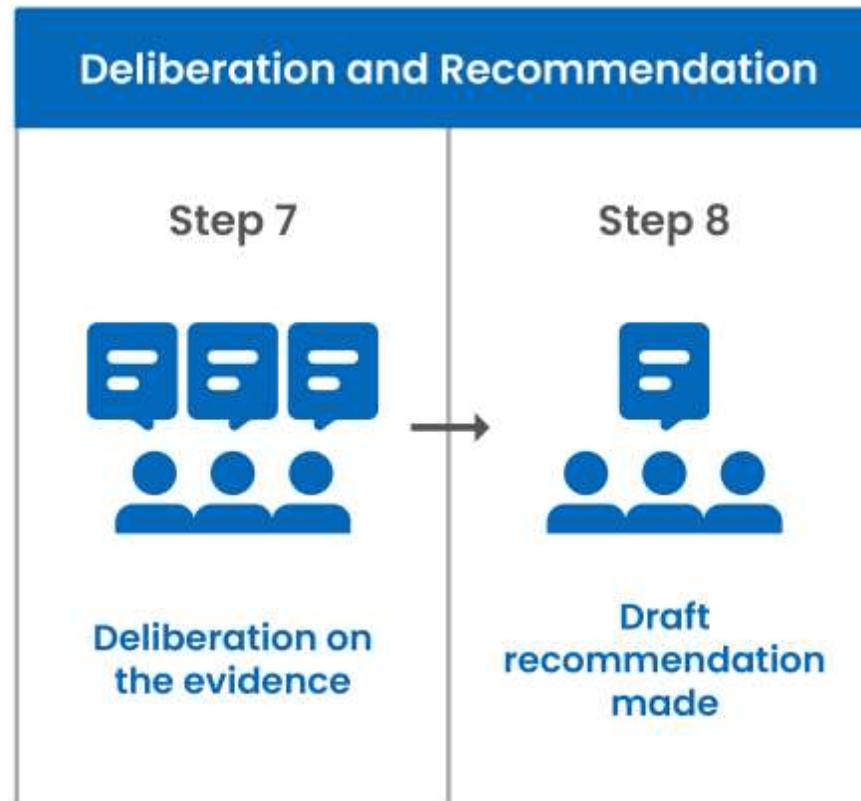
# Proposed Processes to Support Adding (or Removing) Conditions

## 2. Evidence Review



# Proposed Processes to Support Adding (or Removing) Conditions

## 3. Deliberation and Recommendation



# Proposed Processes to Support Adding (or Removing) Conditions

## 4. Engagement and Communication



## Proposed Processes – Questions for Feedback

Do you agree with the proposed process for nominating a condition?

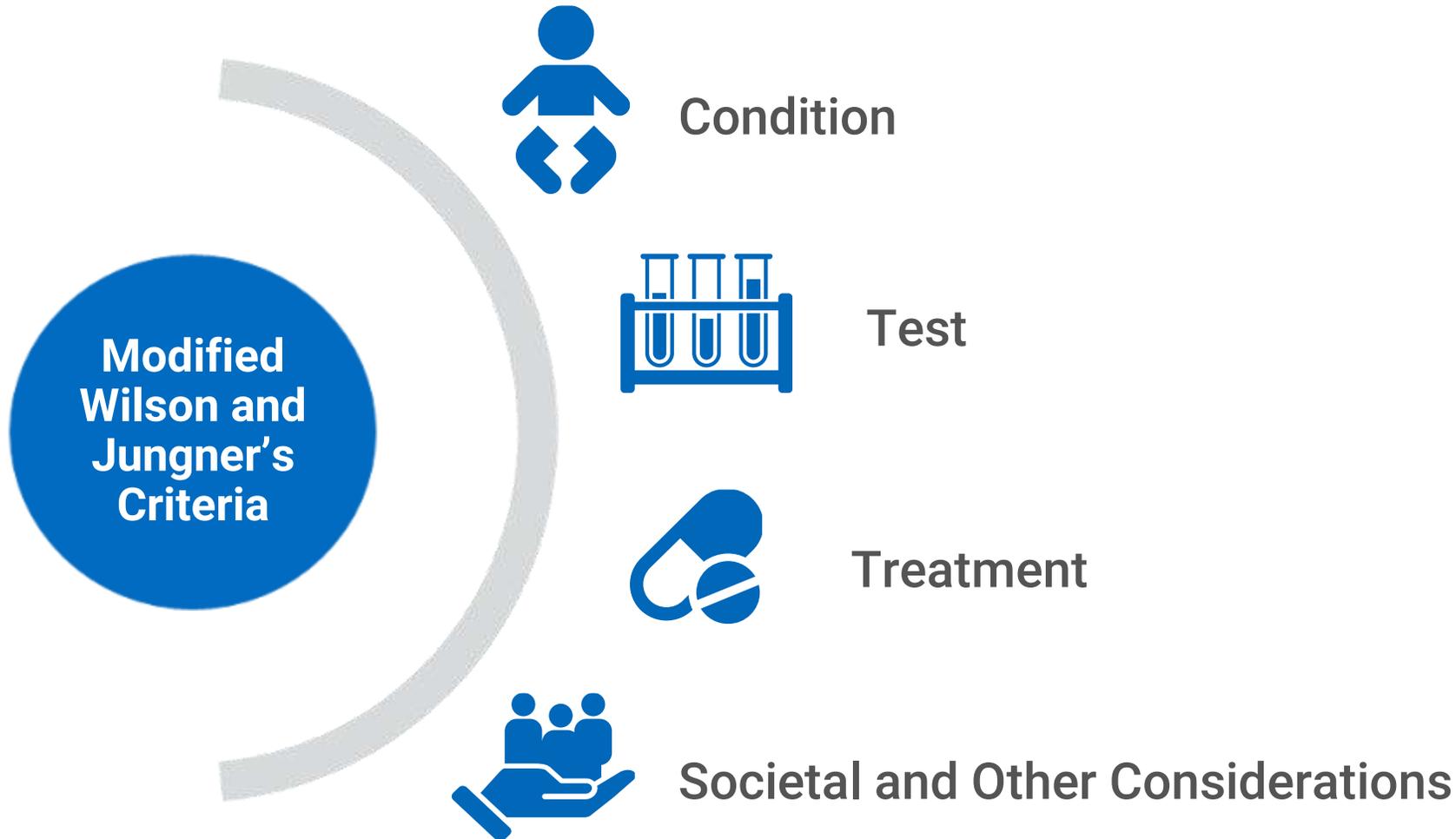
Do you agree with the proposed process for evidence review?

Do you agree with the proposed processes for deliberating and developing recommendations?

Do you agree with the proposed process for engagement and communication?

# 4. Proposed Criteria for Adding (or Removing) Conditions

# Proposed Criteria to Add a Condition



# Proposed Criteria to Add a Condition

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Condition



Test



Treatment



Other  
Considerations

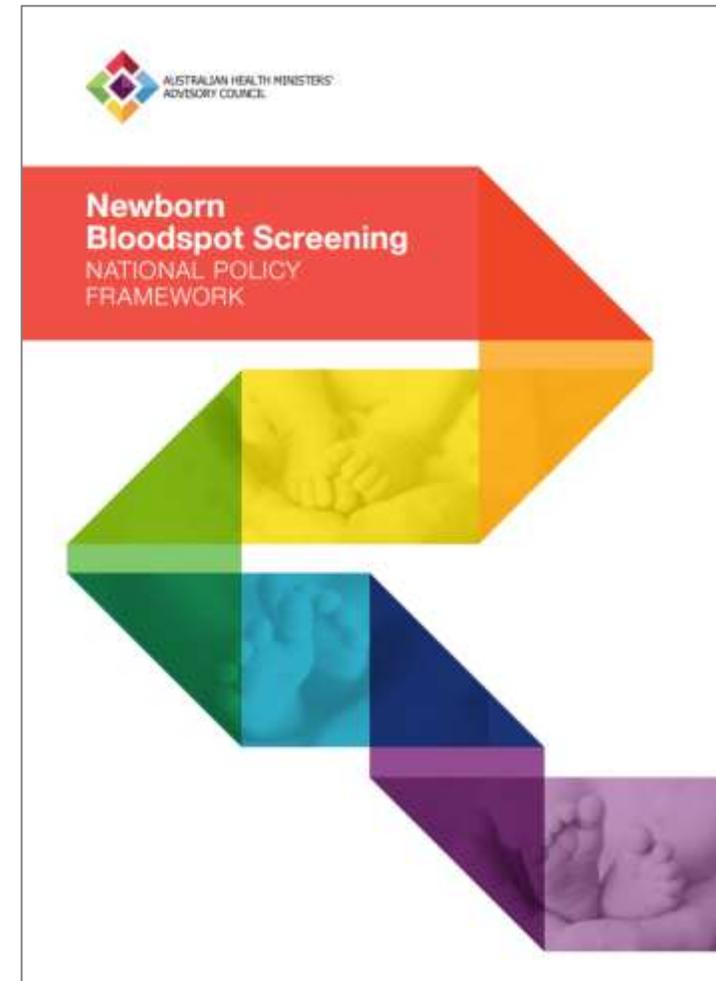


Do you agree with the proposed criteria for adding conditions from a proposed pan-Canadian Newborn Screening List?

# Proposed Considerations for Removing a Condition

Adapted from  
Australia's Framework

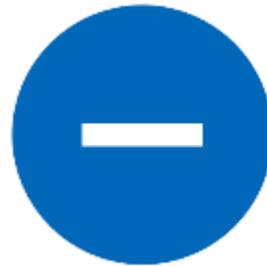
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Source: [Newborn Bloodspot Screening – National Policy Framework \(health.gov.au\)](http://health.gov.au)

# Proposed Considerations for Removing a Condition

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## Removing a Condition



Do you agree with the proposed considerations for removing conditions from a proposed pan-Canadian Newborn Screening List?

# 5. A Proposed Pan-Canadian Newborn Screening List

# A Proposed Pan-Canadian Newborn Screening List

## Building on:

- What is currently being screened for in Canada
- What was proposed in the past
  - 2016 Intergovernmental Working Group



# A Proposed Pan-Canadian Newborn Screening List

Classic phenylketonuria	Maple syrup urine disease	Tyrosinemia, type 1	Citrullinemia, type 1	Argininosuccinic acidemia	Medium-chain acyl-CoA dehydrogenase
Trifunctional protein deficiency	Long-chain 3-hydroxyacyl-CoA dehydrogenase	Very long-chain acyl-CoA dehydrogenase	Carnitine uptake defect	Severe combined immunodeficiencies	Isovaleric acidemia
Glutaric acidemia, type 1	Methylmalonic acidemia (methylmalonyl-CoA mutase deficiency)	Methylmalonic acidemia (cobalamin disorders)	Propionic acidemia	Primary congenital hypothyroidism	Congenital adrenal hyperplasia
Classic galactosemia	Biotinidase deficiency	Cystic fibrosis	S, S disease (sickle cell anemia)	S, C disease	S, beta-thalassemia

Spinal muscular atrophy

# A Proposed Pan-Canadian Newborn Screening List

## Further Evidence Review Required

<b>•Guanidinoacetate methyltransferase deficiency</b>	<b>Homocystinuria</b>	<b>HMG-CoA lyase deficiency</b>
<b>Carnitine palmitoyltransferase I deficiency</b>	<b>Carnitine palmitoyltransferase II deficiency</b>	<b>Carnitine acylcarnitine translocase deficiency</b>
<b>Mucopolysaccharidosis, type I</b>	<b>Congenital cytomegalovirus (hearing loss risk factor)</b>	<b>X-linked adrenoleukodystrophy</b>

# A Proposed Pan-Canadian Newborn Screening List

## Proposed Pan-Canadian Newborn Screening List

- 25 conditions

## Proposed for Further Evidence Review

- 9 conditions



Do you agree with the proposed pan-Canadian newborn screening list?

# 6. Anticipating Emerging Conditions

# Anticipating Emerging Conditions for Newborn Screening

## Definition of emerging:

- Not currently screened for in Canada
- Under review or adopted by a newborn screening program in another country
- Treatments in the pipeline



# Anticipating Emerging Conditions for Newborn

## Potential Emerging Conditions

- Pompe disease
- Fabry disease
- Mucopolysaccharidosis, type 2
- Mucopolysaccharidosis, type 3
- Batten's disease
- Niemann-Pick disease, type A and B
- Gaucher disease
- Duchenne muscular dystrophy
- Krabbe disease
- X-linked agammaglobulinemia
- Metachromatic leukodystrophy



Are there other emerging conditions that you would recommend the advisory panel consider in its final report?

# Next Steps

# How to Provide Feedback?

July

11

## Today

Release of the NBS Advisory Panel's discussion paper for consultation

- Leveraging existing partnerships and relationships to seek input
- Focus groups with patient and equity-deserving groups
- Online feedback forum supported by discussion document to guide and inform participants when answering questions

Sept

11

## Deadline for Feedback From Interested Parties

Feedback period for the discussion paper closes

# Questions for Feedback

## Feedback Requested

Questions asked will include topics, such as:

- draft guiding principles and definitions
- proposed approach for potential coordinated structures for newborn screening
- proposed processes (e.g., nomination, evidence review, deliberation, recommendation, engagement, and communication)
- proposed criteria for adding and removing conditions for newborn screening
- proposed newborn screening conditions that could potentially be added to a future pan-Canadian list
- emerging conditions not yet screened for in Canada



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# Questions

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