Canada's Drug Agency
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Drugs. Health Technologies and Systems. Médicaments, technologies de la santé et systèmes.

Building Toward a Potential Pan-Canadian Coordinated Approach for Newborn Screening

Summary of the Advisory Panel's Work and Engagement With Interested Parties

March 4, 2025

Overview

1. Work of the Advisory Panel: Context and Scope
2. Engaging With Interested Parties: Who We Heard From
3. What We Heard: Input Into the Advisory Panel's Discussion
4. Next Steps and Concluding Message

National Strategy for Drugs for Rare Diseases

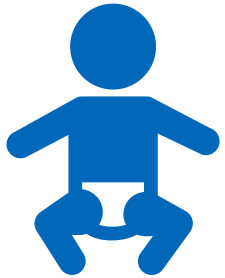
The Government of Canada announced a total investment of up to $1.5 billion over 3 years in support of a National Strategy for Drugs for Rare Diseases in March 2023.

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.

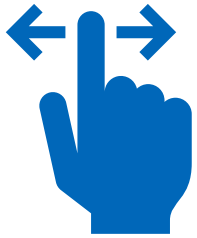
Pillar 3: Collect and Use Evidence

DRD Pipeline Repository

Newborn Screening



Customized Reviews



Disease-Based Registries and Real-World Evidence

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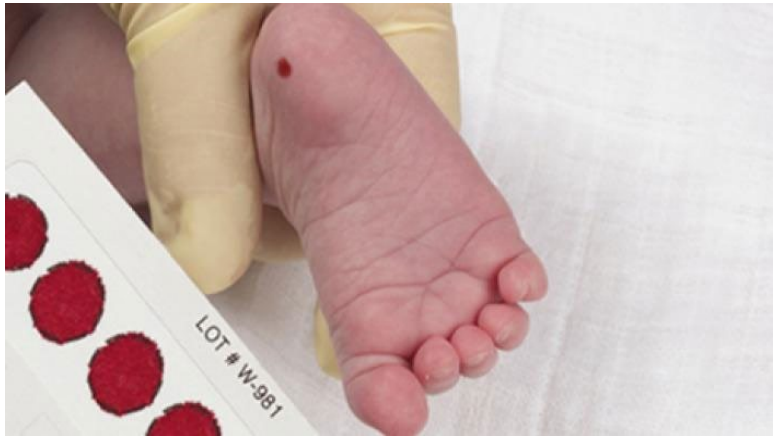
Work of the Advisory Panel: Context and Scope

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 disorders.
- Screening helps to identify certain conditions as early as possible to prevent serious health problems.
- Early identification of rare diseases through newborn screening allows for timely diagnosis and appropriate access to treatments and supports early in life.



Newborn Screening Opportunity

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

Building on existing work to map out the conditions that are being screened in newborns across the country

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

Convening experts to provide advice and guidance

Benefits

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

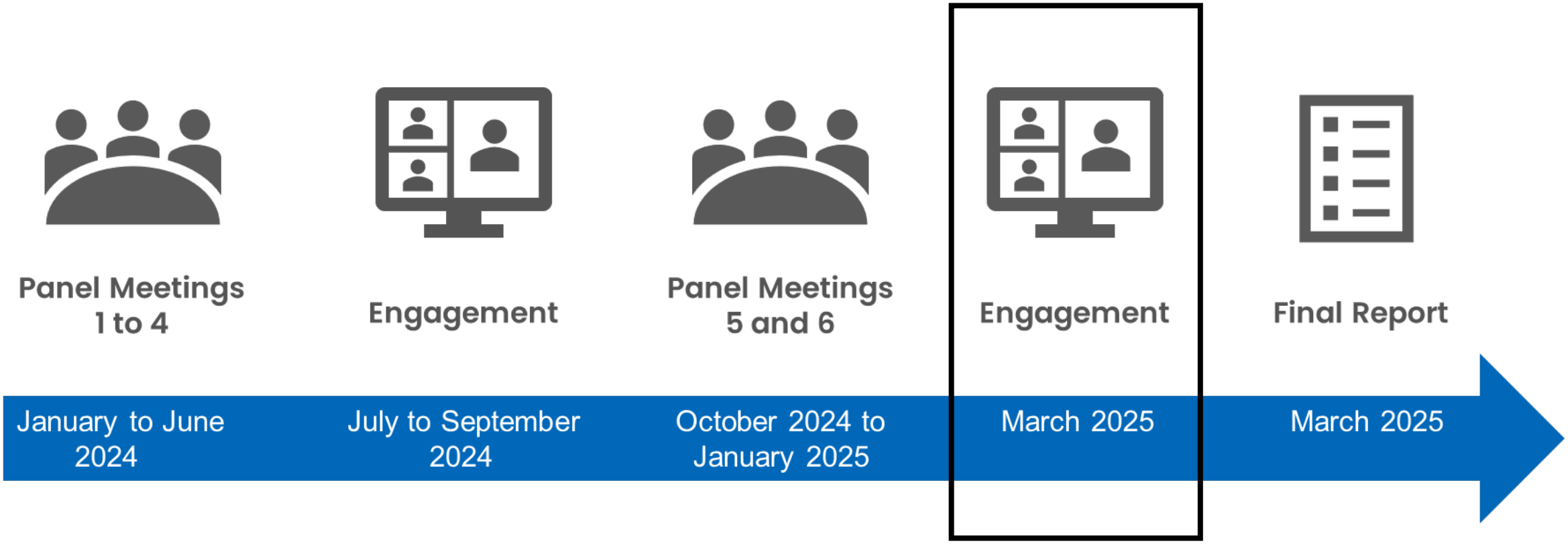
The Advisory Panel

Comprised of 2 co-chairs and 11 members*

- Persons from across Canada with diverse representation (gender, race, culture) and perspectives
- Ranges of expertise and experience, including health care providers (e.g., clinicians, program administrators, researchers), persons with lived and living experience, and individuals with backgrounds in bioethics, law, and health administration

* Visit our website for advisory panel details.

Project Timeline



Advisory Panel's Discussion Paper

1. Proposed guiding principles
2. Exploring a future coordinated model
3. Proposed processes to support adding or reassessing conditions
4. Proposed criteria for adding conditions and proposed considerations for reassessing a condition
5. Proposed pan-Canadian newborn screening list
6. Anticipating emerging conditions

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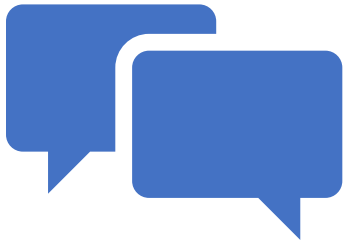
Engaging With Interested Parties: Who We Heard From

Engaging With Interested Parties

Online consultation form



Focus group discussions and key informant interviews

Online Consultation

The form included 11 questions

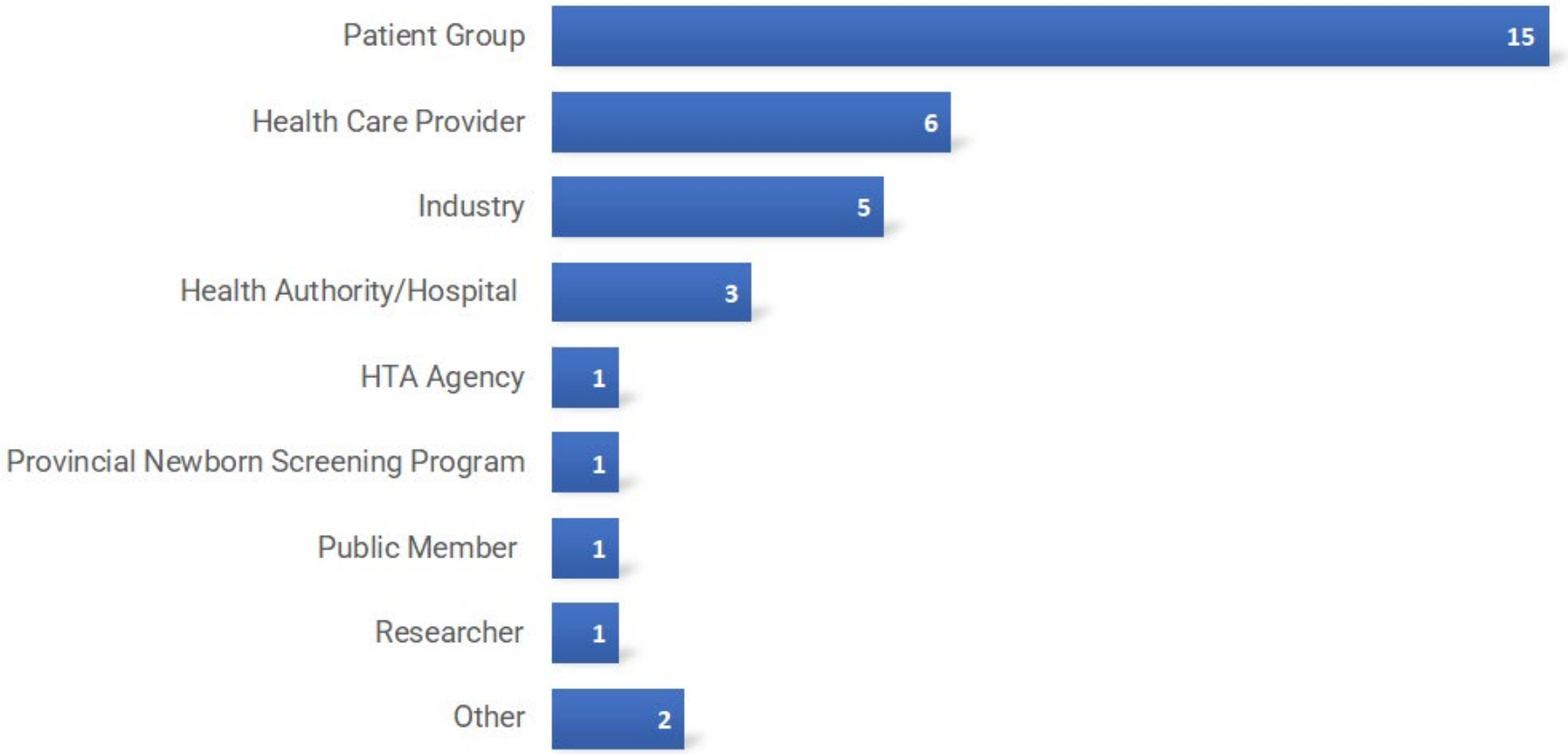
Submissions were received between July 11 and September 11

The form was available in French and English

Full submissions will be published online

Online Consultation: Who We Heard From

Online Consultations Received by Type of Respondent (N = 35)



Focus Groups and Key Informant Interviews

The panel felt it was critical to solicit and incorporate feedback from different perspectives

Canada's Drug Agency engaged Sage Solutions to conduct online focus groups with First Nations, Métis, and Inuit communities

Canada's Drug Agency extended invitations to participate in focus group discussions to individuals and organizations who provide community birthing care and who are from or work with underrepresented and underserved populations.

Focus group sessions and key informant interviews focused on the guiding principles, criteria for adding conditions, and emerging conditions

Focus Groups and Key Informant Interviews: Who We Heard From

Sage Solutions hosted 3 focus groups with people from First Nations, Métis, and Inuit communities (21 participants)

Canada's Drug Agency hosted 1 focus group and 3 key informant interviews with individuals who provide community birthing care and who are from or work with underrepresented and underserved populations (6 participants)

Process for Reviewing Input

- Discussed input over 2 meetings
- Carefully reviewed and reflected on input, discussed key themes, and incorporated suggestions into the final report
- Some out-of-scope topics were identified as important for considerations in future work

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What We Heard: Input Into the Advisory Panel's Discussions

Proposed Guiding Principles

Highlights of What We Proposed

Health Rights of the Newborn

Equity

Effectiveness, Safety, and Quality

Transparency

Collaboration

Sustainability

General agreement

Proposed Guiding Principles

Highlights of What We Heard

- Considerations are needed for the family or caregivers and communities
- A focus on the well-being of the newborn and holistic care should be incorporated
- Further explanation of terms and incorporating additional concepts
- A need to address additional barriers and challenges to access
- Acknowledging the tension between the guiding principles
- Clarifying linkages between the guiding principles

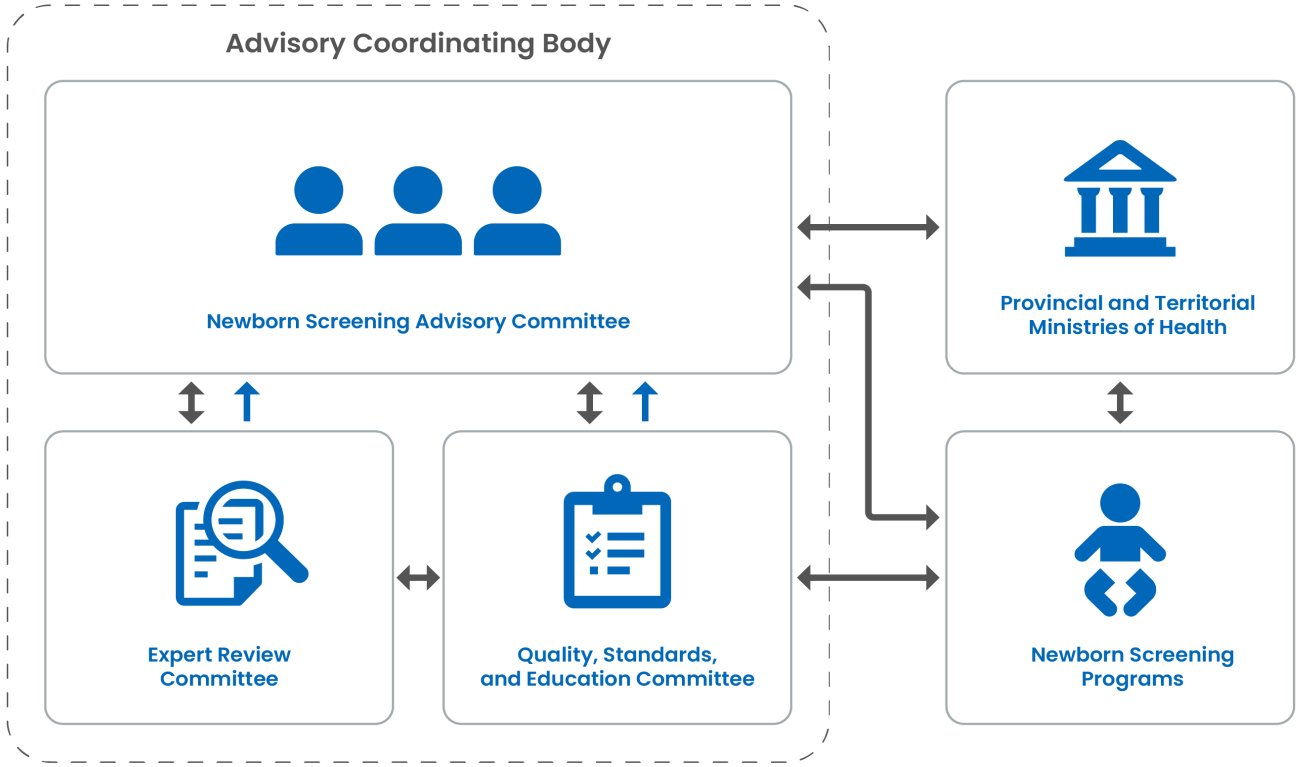
Proposed Guiding Principles

Highlights of the Advisory Panel's Discussion

- Additional explanatory text will be included, terms will be clarified, and additional concepts will be incorporated into the final report
- Changes to the equity definition will be made
  - Examples of barriers and challenges to access will be included
- The purpose and interplay of the guiding principles will be clarified

Exploring a Future Coordinated Model

Highlights of What We Proposed



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

Exploring a Future Coordinated Model

Highlights of What We Heard

- Opportunities for existing organization(s) to support the proposed coordinated model
- Opportunity for federal, provincial, and territorial ministries of health to participate in the proposed committees
- Keep committee structure streamlined
- Opportunities to build linkages with community-based organizations and patient groups

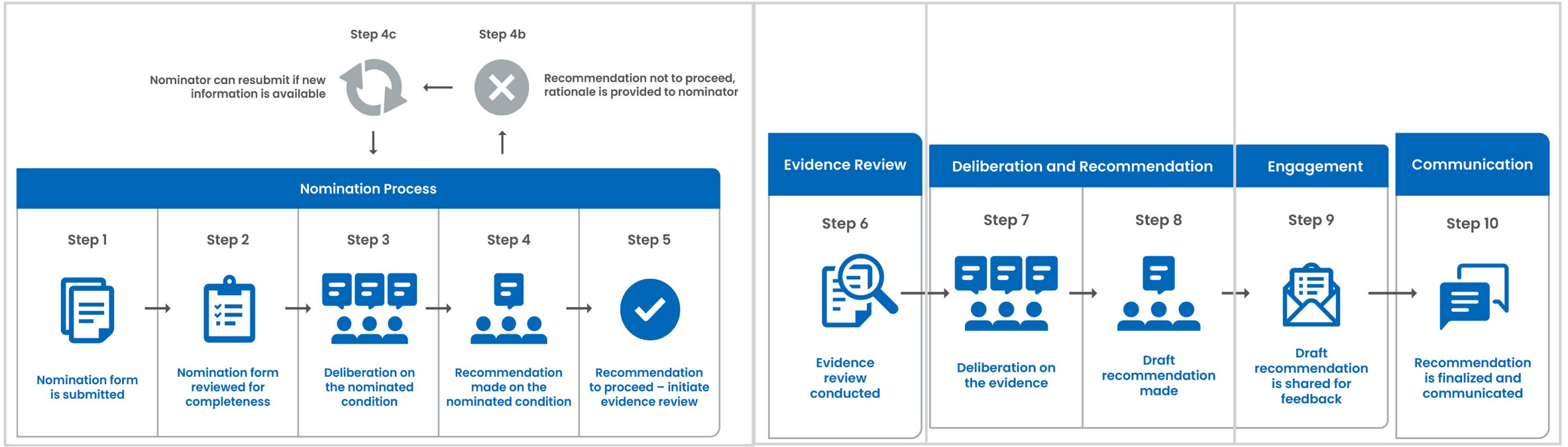
Exploring a Future Coordinated Model

Highlights of Advisory Panel's Discussion

- Acknowledged opportunities to implement the coordinated model by working with and building from existing organization(s)
- Revised committees' compositions and functions to highlight opportunities to engage representatives from federal, provincial, and territorial ministries of health as observers
- Clarified that governance model should seek to ensure efficiency and streamlined processes whenever possible
- Recognized the importance of engaging with the public and patients and articulated opportunities for linkages in committees' composition and function

Proposed Processes to Support Adding or Reassessing Conditions

Highlights of What We Proposed



General agreement, some contention within the evidence review process

Proposed Processes to Support Adding or Reassessing Conditions

Highlights of What We Heard and the Advisory Panel's Discussion

Conflict of interest disclosures

During the nomination and engagement processes

Different perspectives

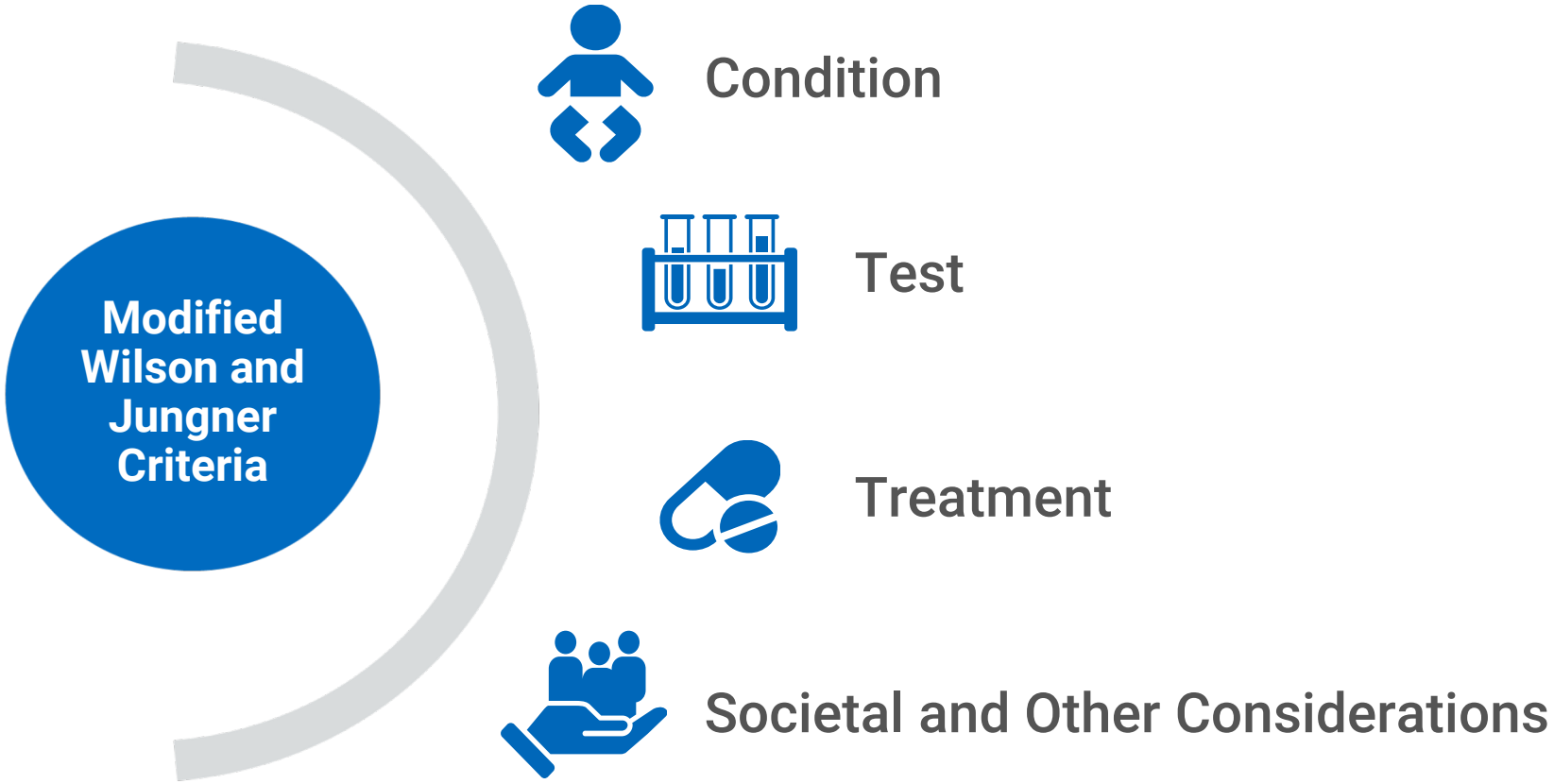
Include experts specific to the disease under review, those with lived and living experience, the public, and the jurisdictions

Short- and long-term implications

Consideration of short- and long-term implications for newborns, families, and health systems

Proposed Criteria for Adding Conditions

Highlights of What We Proposed



General agreement with some contention

Proposed Criteria for Adding Conditions

Highlights of What We Heard

- Some expressed a preference for the original Wilson and Jungner screening criteria
- Clarify how the criteria will be interpreted and operationalized highlighting the need to have deliberative methods
- Focus should be on conditions that arise early in life and are preventable and treatable
- Clarify that there needs to be evidence of benefit to screening, early diagnosis, and treatment for the newborn
- Clarify the concept of presymptomatic or latency period
- Clarify how the criteria will account for variability in populations

Proposed Criteria for Adding Conditions

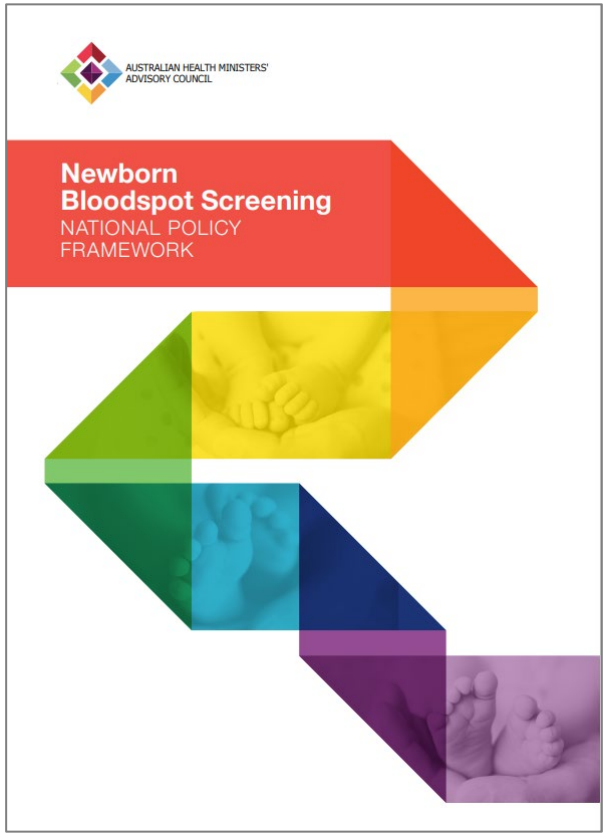
Highlights of Advisory Panel's Discussion

- The proposed modified criteria were revised to clarify the following:
  - the focus is on conditions that manifest in early life
  - the benefit of screening is for the newborn
  - the concept of the condition having a presymptomatic or latency period
- Further work on deliberative processes to support the use of the criteria was discussed

Proposed Considerations for Reassessing a Condition

Highlights of What We Proposed

Adapted from Australia's framework



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

General agreement

Proposed Considerations for Reassessing a Condition

Highlights of What We Heard

- The role of reassessment needs to be clarified

Highlights of Advisory Panel's Discussion

- Further development of the process for reassessment is needed

A Proposed Pan-Canadian Newborn Screening List

Highlights of What We Proposed

Proposed Pan-Canadian Newborn Screening List

- 25 conditions

Proposed for Further Evidence Review

- 9 conditions

General agreement with some contention

A Proposed Pan-Canadian Newborn Screening List

Highlights of What We Proposed

- Clarification on the positioning of the pan-Canadian list
- Addition of conditions to the proposed newborn screening list and the list of conditions requiring an evidence review
- Clarification of how the conditions will be defined

A Proposed Pan-Canadian Newborn Screening List

Highlights of Advisory Panel's Discussion

- The intent of the proposed pan-Canadian newborn screening list was clarified
- Why conditions are on the list of conditions that require an evidence review was clarified
- The need for case definitions was discussed

Anticipating Emerging Conditions

Highlights of What We Proposed and Heard

11

Conditions proposed in the discussion paper

19

Additional conditions proposed by respondents

Other Considerations

Highlights of What We Heard and the Advisory Panel's Discussion

- Other types of newborn screening testing
- Genomic sequencing
- Laboratory infrastructure
- Data sharing, privacy, and quality metrics
- Educational materials

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Next Steps and Concluding Message

Next Steps

Publication of the online consultation submissions and summaries of the focus group discussions and key informant interviews

The final report will be posted on the Canada's Drug Agency website at the end of March

Concluding Message

The advisory panel would like to thank the individuals who participated in our engagement activities. Your engagement and perspectives have been instrumental in supporting the advancement of the newborn screening guidance. We truly appreciate your time and contributions.

Thank you

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