CHAPTER 9

Decision-Making About Newborn Screening Panels in Canada: Risk Management and Public Participation

Marisa Beck, Brendan Frank, Sara Minaeian, and Stuart G. Nicholls

Newborn Bloodspot Screening: An Under-Studied Risk Issue

The World Health Organization defines screening as "the presumptive identification of unrecognized disease in an apparently healthy, asymptomatic population by means of tests, examinations or other procedures that can be applied rapidly and easily to the target population" (Wilson & Junger, 1968). Population-based screening programs exist for different stages of life, from prenatal screening of the developing fetus, through newborn screening, to screening of adults.

M. Beck (✉) · B. Frank
Ottawa, ON, Canada
e-mail: mpenslar@gmail.com

S. Minaeian
University of Oxford, Oxford, United Kingdom

M. Gattinger (ed.), *Democratizing Risk Governance*,
https://doi.org/10.1007/978-3-031-24271-7_9

Initiated in the 1960s, and with programs now existing in all continents of the world (Therrell et al., 2015), newborn bloodspot screening (NBS) is the largest and longest running example of a population screening program internationally (Nicholls et al., 2014). However, programs vary in size and scope across jurisdictions.

NBS detects rare diseases in asymptomatic neonates. The process begins with taking a small blood sample 24–72 hours after birth, usually through a heel prick or heel lance. The sample is then screened for a range of biomarker targets that indicate elevated risk for a number of conditions. Newborns who screen positive for a condition undergo further testing to either confirm or rule out a diagnosis. If a diagnosis is confirmed, patients receive treatment from specialized healthcare providers. In Canada, screening is offered to all children as standard of care (Nicholls et al., 2014), predicated on early identification and early intervention, to ameliorate or prevent disease symptoms (Canadian Agency for Drugs and Technologies in Health, 2011). While provided as standard of care, parents can opt out of the screening program, in which case their child would not be tested for any of the conditions.

Over the past two decades, new and disruptive technologies have made it possible to include an increasing number of targets in screening panels at relatively low cost. In particular, the advent of tandem mass spectrometry technologies marked a step change for NBS, allowing for simultaneous detection of biomarkers for multiple disorders at minimal incremental costs (Levy, 1998). Progress in whole genome screening technologies could similarly trigger a sudden and substantive expansion of screening panels (Botkin & Rothwell, 2016; Bailey et al., 2021; Watson et al., 2022). But absent such technological breakthroughs, technical capacity for screening is only one factor of many in decisions about whether to add or remove conditions from screening panels.

S. G. Nicholls
Clinical Epidemiology Program, Ottawa Hospital Research Institute, Ottawa, ON, Canada
e-mail: snicholls@ohri.ca

The Expansion of Newborn Screening: An Exercise in Risk Governance

The universality of screening and the growing number of targets have sparked a discussion regarding accepted principles that underpin decision-making. For example, while decisions regarding the addition of targets have focused on the benefits to the individual child, there is a debate in the literature regarding what constitutes a benefit (Cornel et al., 2020). In some instances, there may be better health outcomes for the child as a result of early treatment of diagnosed conditions, but families may benefit in a number of ways following the diagnosis of a rare condition as well, such as the psychological comfort of avoiding the "diagnostic odyssey" and better knowledge to inform future reproductive decision-making (Bailey et al., 2006; Buchbinder & Timmermans, 2011; Potter et al., 2009; Bombard & Miller, 2012). Nevertheless, the US body responsible for NBS stopped considering benefit to the family in the nomination and review process for adding conditions to the panel (Watson et al., 2022). Early diagnosis and treatment of rare diseases can also significantly reduce long-term costs to the healthcare system (Sims et al., 2007; Shih et al., 2021). As such, NBS may provide benefits to both the individual and the healthcare system and, ultimately, society as a whole.

From a broader health system perspective, decisions about adding conditions to NBS panels must weigh several additional factors that may present challenges. Decisions to expand the list of conditions for newborn screening are decisions about benefits, but also risks. We use the term risk broadly to refer to the consequences—whether intended or unintended—of an event or activity for something that people value, including health, property, nature, beliefs, social institutions, and cultural practices (Renn, 2008; Beck, 1992; Stern & Fineberg, 1996). Risk is determined by two essential parameters: (1) the likelihood or probability that a consequence occurs and (2) the severity of the consequence for human health, well-being, or the natural environment.

In the context of NBS, the associated risks may accrue at different levels to different stakeholders. For example, screening tests commonly require decisions around thresholds. Setting this threshold requires a fine balance; too low and there may be many 'false positive' results—children that test positive but do not have the condition—but too high and there may be too many 'false negative' results—children who have the condition but screen negative. Including a condition where testing is insufficiently accurate can create personal and social costs or risks. There are also ethical

risks related to privacy and sample storage and short-term economic risks related to budget constraints. Indeed, any decision made within the context of a finite envelope of funds also involves the opportunity costs of alternate services that do not receive those funds (UK National Screening Committee, 2000; Ulph et al., 2017; Rogowski et al., 2014). Decisions about NBS panel expansions affect at least four distinct groups, two of which are less obvious: healthcare professionals who deliver the screening, babies and their families likely to benefit from any expansion, people who receive screening but who are unlikely to benefit from the expansion, and people who might lose access to healthcare resources that are now directed to newborn screening but could have been allocated elsewhere.

We thus contend that the decision-making process regarding the addition of a target to newborn screening panels is one of risk governance (Renn, 2008), where the goal is to reduce or prevent risks but do so "while taking into account social, cultural, ethical, political, and legal considerations" (Presidential/Congressional Commission on Risk Assessment Risk Management, 1997, 8).

Implications of Different Value Judgments: Why Public Participation Is Key to Effective Risk Governance

While conventional risk analysis quantifies all possible outcomes and multiplies them by their respective probabilities to arrive at a single indicator of risk, there are several challenges posed with this approach. First, people may value consequences differently. As such, risk assessment varies depending on whose perspectives are included (Zikmund-Fisher et al., 2007). Second, while the terminology of 'risk' assumes that we have sufficiently certain knowledge of potential outcomes and/or their associated probabilities, this knowledge is incomplete in many decision-making situations (Stirling, 2007). Indeed, in the context of rare diseases—the focus of newborn bloodspot screening—the scientific evidence may be limited (Watson et al., 2022). When available analysis or scientific knowledge is unable to reliably identify outcomes and/or probabilities, subjective judgments play an important role in risk assessment. In this context, highlighting the need to examine whose views and judgments are included in risk governance becomes even more crucial (Stirling, 2007).

Over the last two decades, scholars in risk governance have drawn attention to the crucial ways in which the opinions of scientific experts may differ from other stakeholders, such as those affected by the decisions

made, and have emphasized the need for public involvement in public policy decision-making (Renn, 2008; Jardine et al., 2009; Webler & Tuler, 2018). Public involvement generally refers to the engagement of multiple, diverse social groups in the formation of public policymaking or regulatory decision-making to address societal issues. While some authors such as Fiorino (1990), have argued that public participation is imperative for moral reasons (because it is the right thing to do), the engagement of groups affected by the decision may also have instrumental effects such as driving more publicly acceptable outcomes relative to decisions based on expert knowledge alone.

The inclusion of public(s) in policy decision-making challenges traditional notions about science and politics that underlie models of evidence-based decision-making. First, it problematizes the notion that science and politics—or facts and values—are separate and need to stay separate. Second, it undercuts the position that effective decision-making about risk should rely on scientific and expert knowledge alone. In reality, the two are intricately linked: not only does scientific evidence inform and shape political discourse but science itself is infused with politics and values—and legitimately so (see Douglas in this edited volume): involving citizens in risk governance may expose implicit value-judgments embedded in expert assessments (Nicholls et al., 2016; Kuzma, 2016). For example, empirical research shows that risk perceptions of experts frequently differ from those of the general population (Krewski et al., 2012) and that people's risk perception is strongly driven by their value commitment and cultural identity (Kahan, 2012). As a consequence, other forms of knowledge, including people's life experiences, 'local' and cultural knowledge are legitimate and valuable in risk governance and decision-making (Corburn, 2005).

The Need to Better Understand Decision-Making for Newborn Bloodspot Screening

Despite the acknowledged benefits and risks to the expansion of newborn screening panels and documented variation between programs internationally (Jansen et al., 2016), very little work has explored how decisions are made regarding the inclusion or exclusion of targets within newborn screening programs. To date, descriptions regarding the structures that support the decision-making process in NBS are essentially non-existent. Furthermore, despite established principles for population screening,

there is little if any examination of *how* criteria are applied (Jansen, 2017). A recent exception to this has been work by Jansen et al. (2016) who provide an overview of the decision-making process in the Netherlands (Jansen et al., 2021) as well as a brief description by Shone (2019) regarding the process in North Carolina.

This lack of data is problematic for several reasons. First, it precludes examination of the process and whether the decisions are fair or equitable; justifying the choice of diseases in an NBS program requires balancing the costs and benefits for society. This requires consideration of the broader population who, as taxpayers and recipients of healthcare services, are affected by decisions concerning funding and distribution of these services (van der Burg & Oerlemens, 2018). Second, it offers fewer opportunities to learn and understand the constraints placed on these decision processes as well as ways to improve them. Finally, it obfuscates the reasons for differences between provinces and territories, which may depend as much on value judgments and resource availability as they do on evidence (Nicholls et al., 2016).

This chapter reports on research findings about the process to expand NBS panels in Canada. Specifically, it focuses on how decision-making processes for NBS panel additions address risks, including how the public is involved in the process. To answer these questions, we draw on document analysis and interviews with key informants.

This chapter proceeds as follows. The next section, "Analytic Frameworks and Methods" introduces the analytical frameworks and methodology used in this study. The following section, "Who Decides How About NBS Panel Additions in Canada?" describes empirical results, and the Section, "Economic and Advisory Risk Management Tools Dominate" discusses these findings in the context risk management. The final section, "Where from Here? Avenues Forward for Decision-Makers and Scholars" offers concluding thoughts and identifies fruitful avenues for future research.

Analytic Frameworks and Methods

In this chapter, we examine the decision processes for NBS panel additions through a risk governance lens, with specific focus on the work of scientific advisory bodies. We consider these decision processes to address—if largely implicitly—the risks associated with NBS, including economic risks, health risks, and ethical risks identified above. In our

discussion, we draw on the typology of risk management tools identified in the REACT framework (Krewski et al., 2007, 2014) presented previously in Chapters 5 and 7. This framework provides an organizing structure for risk management tools that public authorities (governments and regulators) may choose to apply, and includes regulatory, economic, advisory, technological, and community interventions.

Given our focus on the democratization of decision-making, and public involvement specifically, we also apply the public participation spectrum developed by the International Association for Public Participation (IAP2) as a framework when determining the quality of public participation in decision-making. The IAP2 framework sets out levels of engagement that gradually transfer increasing amounts of agency to the public: (1) inform the public about the problems, alternatives, and solutions; (2) consult the public and ask feedback on assessments and alternative solutions; (3) involve the public to effectively incorporate perspectives and concerns; (4) collaborate with the public on every aspect of decision-making, (5) empower the public to have final decision-making authority (Fig. 9.1).

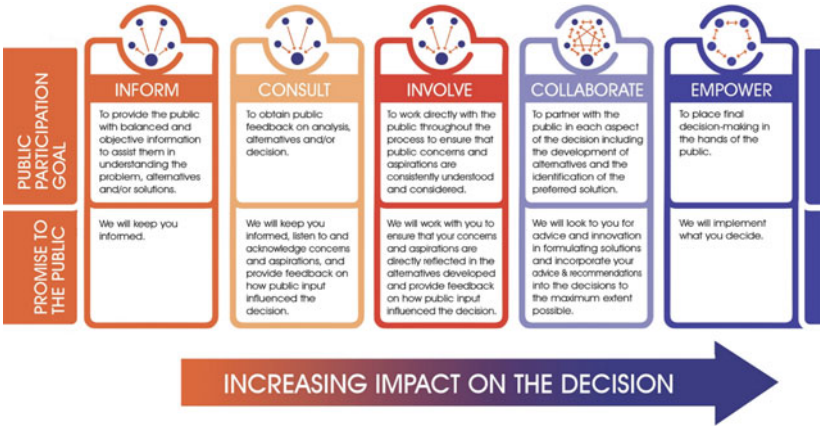

Fig. 9.1 The International Association for Public Participation (IAP2) Spectrum of Public Participation (© International Association for Public Participation www.iap2.org, retrieved from https://www.iap2canada.ca/foundations/ 22 September 2022. Reproduced with permission)

Choosing and designing appropriate means for public participation is highly dependent on local contexts and resource availability (Webler & Tuler, 2018). Involving the public constructively in risk decision-making can be expensive; it requires effort, skill, and learning.

Regarding the democratic quality of public involvement in risk governance, the introduction of this edited volume identified four principles:

> ***Transparency*** concerns the ease with which stakeholders can access information about risk-related decision processes and outcomes.
> ***Inclusiveness and representativeness*** focus on whether those who are impacted or concerned by risk issues have formal opportunities to make their voices heard in decision-making about these risks (inclusiveness). This principle also refers to whether the range of stakeholders involved, including marginalized social groups, is representative of potentially affected or concerned populations (representativeness).
> ***Deliberative quality*** refers to the 'how' of public engagement: is there a genuine opportunity for members of the public to engage in dialogue and exchange? Are their voices heard and seriously considered in the deliberations?
> ***Accountability of decision-makers*** focuses on the accountability of public authorities involved in risk-related decision-making toward citizens (through elected officials) or non-elected officials (e.g., bureaucrats, expert committees).

The combination of the IAP2 spectrum and the four principles of the democratic quality of public involvement in risk governance is original to this chapter. We use these frameworks as analytic structures to analyze: publicly available information about the process of test addition, the scholarly literature, and interviews with individuals who have direct experience at the scientific advisory juncture of the decision-making processes within Canadian newborn screening programs. Interviews with final decision-making authorities within provincial and territorial governments were out of scope for this research.

We conducted documentary analysis (of websites, publicly available materials) as well as semi-structured interviews with eight participants involved in five different NBS programs across Canada. We made great efforts to speak with participants from all Canadian screening programs,

but only representatives from Saskatchewan, Alberta, British Columbia, the Maritimes (Nova Scotia, Prince Edward Island, and New Brunswick), and Quebec accepted our invitation to participate in this study.[1] Some of our interviewees have experience with both the medical and administrative components of NBS but are not involved in the final decision. Interviews were conducted over the phone and focused on the governance structures and decision-making processes concerning additions to NBS panels.

Who Decides How About NBS Panel Additions in Canada?

Canadian NBS Programs: Great Variety, Little Transparency

In Canada, jurisdiction over NBS programs and screening panel composition lies with provinces and territories and there is no central organizing body. While all babies born in Canada today have access to screening, the number of conditions included in the screening panels differs between jurisdictions (Potter et al., 2008) and not all provinces/territories have their own screening facilities (Table 9.1). Prince Edward Island and New Brunswick share a regional facility with Nova Scotia; Yukon sends its samples to British Columbia; the Northwest Territories and Nunavut share facilities with Alberta (Kitimeot) and Ontario (Baffin). Provinces and territories that use the same facility share the same screening panels. The number of conditions screened for in Canada ranges from 11 in Québec to 40 in Manitoba.

The lack of standardization across Canada not only means that populations in different regions have unequal access to testing but also that decisions regarding the composition of screening panels likely differ across the country. At the same time, and consistent with the broader newborn screening literature, publicly available information about these processes is scarce and uneven across programs. In most provinces, it is close to impossible for members of the public to learn about the evidence that decisions are based on and the mechanisms that operate when decisions are made. For example, while all NBS programs have websites, the available resources about NBS are targeted toward expectant parents and

[1] Since British Columbia and Alberta provide testing services to Nunavut, the Northwest Territories, and the Yukon, our participants are effectively involved in NBS programs in 10 provinces/territories across Canada.

healthcare professionals and provide little information about the decision-making process itself. Ontario's NBS program is an exception; its website provides information on the test addition process and the factors that are considered by the Advisory Council when considering addition of a

Table 9.1 NBS programs and testing facilities in Canada

Province/territory	*NBS program, testing facility, website*	*# of conditions included*[2]
British Columbia Yukon	BC Newborn Screening Program http://www.perinatalservicesbc.ca/our-services/screening-programs/newborn-screening-program	24
Alberta Nunavut (Kitimeot) Northwest Territories	Alberta Health Services, https://www.albertahealthservices.ca/info/page9014.aspx	22
Saskatchewan	Roy Romanow Provincial Laboratory https://www.saskhealthauthority.ca/facilities-locations/roy-romanov-provincial-laboratory/screening-and-reference-services	over 30
Manitoba	Cadham Provincial Laboratory https://www.gov.mb.ca/health/publichealth/cpl/baby.html#:~:text=For%20more%20information%20about%20newborn,at%20204%2D945%2D7458	around 40
Ontario Nunavut (Baffin)	Newborn Screening Ontario, Children's Hospital of Eastern Ontario https://www.newbornscreening.on.ca/	28
Québec	Québec Neonatal Blood and Urine Screening Program https://www.quebec.ca/en/health/advice-and-prevention/screening-and-carrier-testing-offer/blood-and-urine-screening-in-newborns	11 (by blood)

(continued)

[2] Information retrieved from the programs' websites on September 20th, 2022.

Table 9.1 (continued)

Province/territory	*NBS program, testing facility, website*	*# of conditions included*
New Brunswick Nova Scotia Prince Edward Island	Maritime Newborn Screening Program, IWK Health Centre https://www.iwk.nshealth.ca/newbornscreening	22
Newfoundland and Labrador	Provincial Medical Genetics Program, Health Sciences Centre St. Johns http://www.nlma.nl.ca/FileManager/Notices_and_Advisories/docs/2015/What_is_newborn_screening_brochure.pdf	19

Source Authors' own source

condition (Newborn Screening Ontario, n.d.). The lack of transparency also precludes any assessments of which decision processes engage patients and/or the public and consider their perspectives.

Governance Structures for Newborn Screening

For the most part, processes for adding new conditions to newborn screening panels are not guided by official government policies or regulations. But across all programs, governments have final authority over the decision to expand screening panels.

Most NBS programs have established standing Advisory Committees tasked with providing evidence-based scientific advice to governments and recommending changes to NBS panels. However, they possess no executive authority to make these changes to the panels themselves. In Ontario, for example, the Newborn Screening Ontario Advisory Council (NSO-AC) exists as a standing advisory committee that has within its mandate development of the process and review of proposals for potential new screening targets. In the Maritimes (i.e., New Brunswick, Nova Scotia, and Prince Edward Island), there is also a Diagnosis Committee that reviews submissions from medical professionals proposing additional conditions to the panel. Some Advisory Committees also strike specialized ad hoc working groups when evaluating whether or not to add new conditions to NBS screening panels.

These standing Advisory Committees consist largely of physicians with various specializations (pediatrics, public health, genetics, neonatology, endocrinology, etc.), laboratory staff, and occasionally economists (Québec) or government representatives (British Columbia, Ontario). Some Advisory Committees also require geographic representation among their members. For instance, Ontario's Advisory Committee includes members from across the province, and the Committee in the Maritimes includes representatives from all three participating provinces.

Most of the roles, responsibilities, and procedures of these Committees have evolved organically over time. In some provinces, according to some interviewees, issues as simple as meeting intervals are not formalised or do not proceed at a regular schedule. Some use terms of reference or similar guiding documents, but there are few external rules or pressures guiding their activities. One participant described their dissatisfaction with the informality of these arrangements:

> It is frustrating for clinicians and for the program and for the public, especially parents with children who are afflicted with these conditions not to have a clear or consistent process.

Decision Processes

With regard to decision-making processes for adding new conditions to NBS panels in Canadian programs, they tend to fall under two broad categories.

First, there are processes that are bound by legislation, regulation, directive, or other types of formalized guidance from a health agency or similar government body. Very few of the Committees' operations are covered by legislation. In most cases, the legislation mandates that an NBS program exists but is silent on their operations and decision-making processes for adding or removing conditions from the panel.

The exceptions are Saskatchewan and Alberta. Saskatchewan introduced *The Newborn Screening Regulations*, Chapter P-37.1 Reg 15, which formalized newborn screening procedures in 2014, including program administration, sample collection, testing and follow-up, disclosures, and adoption of guidelines. The regulations are, however, silent on the topics of risk governance and additions to the panel, but afford the minister of health tremendous latitude and flexibility on public engagement.

> The minister shall: (a) cause the [Newborn Screening Guidelines] to be made available to the public in any form or manner that the minister considers appropriate; and (b) take any steps that the minister considers appropriate to bring the guidelines, and the manner and form in which the guidelines are available, to the attention of the public.

Similarly, Alberta established a province-wide NBS program in 2009, alongside the formation of Alberta Health Services. This amalgamated and harmonized NBS program had previously been managed and administered by regional health authorities. However, both Saskatchewan and Alberta's regulations do not refer to the addition of new conditions to NBS panels.

Second, there are processes that are not bound by legal instruments or official policy. Participants within the interviews indicated that the decisions involving additions to NBS panels overwhelmingly fall into this category. While these activities are often guided by terms of reference or other internally developed procedures, there are no regulations guiding the development of terms of reference or their contents. Activities that fall into this category include internal deliberations by Advisory Committees, working groups and the government decision-makers who ultimately have the discretion to act or not act upon their advice. Within the Advisory Committees, working groups and other advisory bodies, the process of deciding if and when to make a recommendation is largely consensus-based. As one participant described it:

> No legislation, no. The Advisory Committee has terms of reference. It is done on a consensus basis… In terms of composition and structure of the committee, that was done by the [centre] when the program expanded. They consulted with the other provinces, tried to see what is done in other provinces and then established who should be on theirs.

Most participants were not aware of codified processes to guide or inform the decision-making of the Advisory Committees and similar bodies. In the absence of such guidance, Advisory Committees have developed processes and procedures internally as needed to provide recommendations to decision-makers. This includes decisions regarding adding new conditions to the NBS panel as well as the screening procedures themselves.

Factors Affecting Decisions About Panel Expansion

Advisory Committees draw on multiple information sources when looking for new conditions to add to the NBS panels. In addition to the scientific literature, participants across provinces also noted that the Advisory Committees' activities are heavily informed by the work of their counterparts across Canada, particularly Ontario, as well as the United States, the European Union, and the World Health Organization:

> Ontario does great research, and we just use their studies…quite often we rely on what other provinces do. That's how it works.

This is largely due to budgetary constraints, as Advisory Committees simply do not have the resources to exhaustively scan new scientific literature. This forces them to draw on knowledge generated and mobilized in other jurisdictions. Indeed, despite the provincial mandate of the programs, interviewees consistently referred to inter-provincial discussion and collegiality, albeit often unofficial.

Participants said that the final decision takes a range of factors into account, including scientific, economic, and political considerations. As noted earlier, occasionally, separate groups are struck to assess these considerations separately. For example, in British Columbia, a specialized working group reviews every new condition under consideration and iterates with the NBS Advisory Committee. One participant described the unique considerations required for adding a specific condition:

> We develop a working group that is specific to the condition that is being reviewed. So, for example, we just finished our review of spinal muscular atrophy. And so we invited a couple of neurologists with more expertise in neurogenetics to participate in that review.

For instance, if British Columbia's Advisory Committee endorses adding a condition to the panel, another specialized group develops a business case and cost-benefit analysis for presentation to the Ministry of Health for final decision. Interviewees indicated that government decision-makers are forced to weigh a number of considerations; as a result, the decision about the inclusion of a target or condition is not exclusively based on medical and scientific evidence.

> It took two years for the government to mandate the conditions be added to our panel. So there was quite a gap and during that time there were a lot of things happening. A change in government usually means a change in priorities and direction. […]. There are lots of things to consider.

Indeed, all interviewees indicated that budgetary considerations are a key factor in the government's decision-making about panel additions. One participant stated that adding a single condition to the panel requires approximately $500,000 per year per condition but noted that adding a condition generates a "return on investment" of roughly 20 to one in medical costs. However, these savings accrue over decades and do not fit comfortably within budgeting or political cycles. The Advisory Committees appear to be well aware of these hurdles, and interviewees reported that they often make recommendations strategically with political considerations in mind. If budgetary constraints prevent the addition of a new condition to the panel, participants noted that Committees will often submit a rejected condition for reconsideration the following year, aware that the rejection may not have been made due to a lack of scientific merit.

As noted above, governments have the final say on whether or not a condition is ultimately added to an NBS panel, either directly or indirectly through budgeting decisions. It was not clear, however, from the interviews, how many steps removed the Advisory Committees are from these final decision-makers, be they the relevant minister or another senior official, or how their advice is weighed against other factors. The frequency and nature of interactions between the Advisory Committees and the decision-makers who take their advice was also unclear.

Public Involvement in the Screening Decision Process

Across all provinces examined, the Advisory Committees and the working groups consist of specialists and experts who are trained to evaluate the medical and technical rationales for adding new conditions to the NBS panel. There are no formal or structured opportunities for the public to become involved in this decision-making (although in Ontario members of the public can nominate conditions for review). The exception is in Nova Scotia, which at the time of our study was in the process of recruiting two parents to the panel, one from a city and another from a rural region of the province. In this case, the interviewee indicated

that this was an initiative of the Advisory Committee, not a result of a government directive.

Although some participants noted that inclusion of parents is considered or planned, accessing the Advisory Committees generally requires parents to take initiative. Some participants noted resistance to parents and citizens sitting on expert panels, for a variety of reasons:

> It is difficult to find any one parent to speak for or represent the vast majority of parents. I am reluctant to have public representation on our advisory committee for this reason. Yet we must be in step with the wishes, values, and concerns of parents with respect to newborn screening. A more comprehensive and democratic way of doing this is through structured well-designed surveys and or group interviews. This can be resource intensive for any one province but could be coordinated at a national level.

Participants indicated that lack of expertise is the key obstacle to including the public in their decision-making. Some also mentioned a perceived lack of interest by the public, as demonstrated by the dearth of organized advocacy groups in this space. Advocacy groups that *are* organized and well-funded (one participant mentioned cystic fibrosis) are already included in the screening criteria, so there is no additional or incremental work that they can undertake with respect to the screening panel. Since new additions to the screening panel will generally be rare, public awareness may be low, and consequently advocacy may be limited.

Interviewees indicated that, in most provinces, there is minimal interaction between parents and citizens and the Advisory Committees and decision-makers who determine the details and composition of the NBS panels. Parents are largely passive participants in the screening process, their involvement limited to reading, conversations with medical professionals who are collecting samples, and providing the information necessary for informed consent. Some provinces do, however, seek post hoc patient feedback to improve the patient experience. For instance, in British Columbia, parent feedback is solicited after the fact to improve the overall NBS screening process.

Despite the noted reluctance by some to involve parents, other interviewees indicated a general openness toward greater public involvement, but none suggested it was a priority or an explicit part of the mandate. Participants noted that information on the NBS screening process, including new additions, is publicly accessible. However, interviewees did

not suggest that promoting public awareness of NBS panels is a priority. Changes to the NBS screening panels are typically communicated to the public via websites, newsletters, ministerial press releases, and updates to the medical brochures and literature that medical professionals provide to new parents prior to the procedure.

Economic and Advisory Risk Management Tools Dominate

Decision-making about the composition of NBS panels is—whether implicitly or explicitly—an act of risk governance. Based on our documentary analysis and interviews and applying the REACT typology as a conceptual framework, we analyze the approaches taken to address the various risks related to NBS (Table 9.2).

Our findings indicate a relatively light use of **regulatory risk management approaches**. Certainly, governments are the final authority in decisions about panel composition, but the decision-making process and the composition and procedures of the advisory bodies are almost entirely unregulated in the jurisdictions examined (with the exceptions of Alberta

Table 9.2 Risk management in NBS panel decision-making in Canada

Risk management approach (REACT framework)	*Application in NBS panel decision-making*
Regulatory interventions	Governments have final decision authority, but the decision process itself is lightly regulated
Economic interventions	Cost-benefit considerations importantly drive governments' and advisory bodies' decisions
Advisory interventions	Advisory bodies assess potential panel additions and provide recommendations to governments
Community interventions	Overall, limited formalized opportunities for public involvement in decision making. Even information about the decision process is not publicly available in most programs
Technological interventions	Technological capability is a necessary condition for panel expansion, but limited insights from this study about how technology is being used to address risks associated with expansions

Source for REACT framework: Krewski et al. (2007) and Krewski et al. (2014). Authors' own source for application to NBS panel decision-making

and Saskatchewan). Instead, the structures have evolved organically and occasionally function on an ad hoc basis.

In contrast, **economic approaches to risk management** seem paramount in the considerations of the advisory bodies and ultimately, governments' decisions about panel additions. Our findings indicate that governments use formal and informal economic analysis to measure and examine the societal impacts of adding conditions to the screening panels. What remains unclear are the parameters upon which these assessments are made: how are short-term budgetary burdens weighed against long-term health benefits and savings for the entire healthcare system?

Advisory risk management interventions are another key approach employed in the decision-making about NBS panel composition. All NBS programs have one or more advisory bodies, largely made up of healthcare professionals who are responsible for making recommendations regarding panel additions based on scientific evidence and their medical expertise. While these advisory bodies have no final decision authority, interviewees indicated that governments generally follow the committee's recommendations—if budgetary considerations allow. Some interviewees also indicated that advisory bodies sometimes anticipate such budgetary constraints when developing their recommendations.

Finally, before discussing community interventions, it should be noted that our results offer little insights into how **technological risk management tools** are applied in the decision-making process. Advisory committees for the screening programs involved in this study all include a representative from the testing laboratories, but we are not aware of more concrete mechanisms for considering technology as a risk mitigation tool. For example, one way of directly applying technological risk mitigation approaches would be to make an addition to the panel dependent on the use of a specific testing technology that reduced the risk of false positives. Our interviews do not provide evidence of such measures.

Great Potential to Increase Public Participation

According to our findings, **community-based risk management tools** currently receive limited attention. We see little evidence of formalized public involvement in decision-making processes. An exception is the opportunity for anyone in Ontario to suggest adding a condition to the screening panel. If engagement with parents takes place, it seems to be of an informal nature (for instance, advocacy with the government) or occur

mostly after a new condition was added to the panel (for instance, in the BC program).

Table 9.3 categorizes the public participation activities identified in this study along the IAP2's spectrum (the *what*) and also considers these activities in reference to the four principles of democratization (the *how*). Based on the evidence considered, we find great potential for improvements across all four democratization principles: transparency, inclusiveness and representativeness, deliberative quality of the interaction, and accountability.

With respect to the levels of involvement and engagement, all interviewees indicated that NBS programs do inform the public about panel composition and decision outcomes regarding additions. These types of community risk management interventions—information and post hoc consultation—sit on the lower end of the spectrum of public participation, as defined by the International Association for Public Participation (International Association for Public Participation, 2018). However, with regard to transparency, information about the decision process is largely lacking. The exception to this is Newborn Screening Ontario, which offers publicly available information about the process and the criteria that the advisory committee considers when developing recommendations. However, what is clearly lacking for all programs is easily accessible and comprehensive public information about the decision process itself—who makes decisions and based on what information.

Few programs illustrated more active involvement of the public; outreach appears to be limited and tends to focus on consultation about implementation of the program, as opposed to the proposed decision to add a condition. While in theory any member of the public could nominate a condition using the process developed in Ontario, engagement practices generally appeared to focus on patients. This is despite the earlier proposition that there may be multiple stakeholder groups (including the general public not affected by a condition on the screening panel) that may be affected by or interested in NBS panel additions and that may hold different perspectives on the issue. Consequently, we suggest that consultations, where they exist, are generally not inclusive and representative of populations affected by changes in the NBS panel composition. Indeed, even when there were indications of improved engagement (e.g., the Maritimes), they tended to focus on parents.

We did not find evidence of public(s) having decision-making power; indeed, the final decision-making power appeared to lie with elected

Table 9.3 Public participation in decision-making for NBS panel additions

Spectrum of public participation (IAP2) Principles of democratization	*Inform publics about NBS and panel expansions*	*Consult publics for feedback on process and/or outcome*	*Involve publics in process to hear and consider concerns*	*Collaborate with publics at each step of decision process*	*Empower publics by giving final decision power*
Transparency: What information is available?	Communication by NBS programs largely about decision outcomes, not process (exception: ON)	n/a	The actual impact is not transparent because involvements mostly informal	n/a	n/a
Inclusiveness and representativeness: Who is invited/involved/heard? Who are the involved publics? (e.g., parents/advocacy groups/general public)	Existing information about NBS is accessible to the general public through program websites. Much of the information is targeted at parents though	Post hoc feedback from the public (BC)	Interviews indicated that there is a proposal for including parents in the advisory body of the Maritime NBS program NS Ontario enables the general public to suggest a panel addition	n/a	n/a
Deliberative quality: How are publics engaged?	Activities constitute one-way communication but websites typically include contacts for questions	Inconsistent and generally requires parents to take initiative; limited outreach on part of decision-makers (exception: QC, post hoc)	Not clear	n/a	n/a

Spectrum of public participation (IAP2) Principles of democratization	*Inform publics about NBS and panel expansions*	*Consult publics for feedback on process and/or outcome*	*Involve publics in process to hear and consider concerns*	*Collaborate with publics at each step of decision process*	*Empower publics by giving final decision power*
Accountability: How can citizens hold decision-makers accountable?	Elected governments have final authority and are democratically accountable to citizens; possibly problematic for jurisdictions without their own programs				n/a

Sources Spectrum of public participation (International Association for Public Participation, 2018), principles of democratization (Chapter 1), authors' own source for application to NBS screening

government officials and the provincial public service. While governments are democratically accountable to their citizens, multiple jurisdictions in Canada do not have their own testing facilities, instead joining screening programs of other jurisdictions. In these jurisdictions, accountability relationships are weaker and the claim to government as a proxy for public involvement is reduced.

As we show in Table 9.1, Maritime provinces share a screening program and testing facilities. The Advisory Committee includes representatives from all three provinces, but final authority over panel additions lies with Nova Scotia government in Halifax, where the testing facility is physically located. For Nunavut and the Northwest Territories, which have joined Alberta's screening program and the Yukon, which has joined British Columbia, we have no indication from our interviews that representatives from the North are included in any of the involved bodies. As a result, there is a tension between democratic accountability and healthcare resourcing needs in the North.

Finally, while multiple interviewees identified a lack of expertise in engaging with the public, there was interest from some interviewees in greater public involvement. At the same time, others perceived lack of knowledge and interest on the side of the public. These comments are in line with what has become known as the 'knowledge-deficit model' of public engagement; the public is viewed as uninformed and thus unable to grasp the science upon which the decisions are based. While we have no evidence to indicate that public input would not be considered, the suggestion that a lack of understanding about NBS would preclude public involvement fails to consider the other societal risks outlined earlier, especially opportunity costs brought about by decisions made to fund certain healthcare interventions at the expense of others. This may indicate a lack of openness among some of the expert advisors involved in the process toward genuine democratization of decision-making or a perception that broader considerations are beyond their remit.

Where from Here? Avenues Forward for Decision-Makers and Scholars

This chapter discussed decisions about NBS panel additions as exercises of risk governance. While this study is exploratory in nature, we can identify multiple avenues forward for practitioners, including governments and NBS program leads, and future research directions for scholars.

A key takeaway from this study is that transparency about NBS panel decisions in Canada is generally low. While public information is available about decision outcomes (i.e., the list of conditions included in screening panels), even basic process information remains mostly hidden from the public, including who is involved in decision-making about panel expansions (e.g., the composition of the Advisory Committees) and how the decisions were made (e.g., explanations of why certain conditions were added or not). To improve transparency and accountability of decision-makers toward the public, this information should be as publicly accessible as possible.

Future research could examine options for diversifying the risk governance approaches used in decision-making about NBS panel composition. Such research should aim to better understand public perception of risks and benefits associated with NBS and help to examine the various publics affected by NBS—families, advocacy groups, and the public at large. The perceptions of risks and benefits associated with NBS may differ significantly across these groups. In particular for NBS programs serving diverse populations in multiple jurisdictions, such research may also identify how cultural differences may affect risk perceptions and preferences. Regarding the choice and design of mechanisms for public participation, the risk governance literature indicates that there is no one-size-fits-all approach. Therefore, research investigating risk perceptions and preferences for risk governance at a local level is crucial for strengthening the democratic character of decision-making across Canadian jurisdictions.

Resource limitations are at the heart of decision-making about NBS in Canada. Our work identified funding considerations as a key driver of decisions regarding panel additions. Moreover, they are likely a key driver in the design of the decision process itself. Specifically, community-based risk management approaches—currently used scarcely in Canada—can be resource-intensive in terms of time, expertise, and funding. Absent a champion for public involvement and engagement from within government, there will likely be little if any non-expert input into the process. Future research can help to inform arguments about why public engagement is worth the investment, as it can produce robust and acceptable decision outcomes and thus be worthy of investment.

There remains a great opportunity, and potential costs, in developing more transparent and engaging approaches to decision-making regarding the expansion of newborn screening panels. Indeed, a key question—much like that for newborn screening programs themselves—is whether

such changes are worth the cost; a question that science cannot answer on its own.

Acknowledgements The authors thank Alexandra Guay-Charette for her excellent research assistance.

References

(1997). *Presidential/Congressional Commission on risk assessment and risk management. Risk assessment and risk management in regulatory decision-making* (Final Report. Vol. 2). United States Environmental Protection Agency Washington: https://cfpub.epa.gov/ncea/risk/recordisplay.cfm?deid=55006.

Bailey, D.B. Jr, Ackerman Porter, K., Andrews, S.M., Raspa, M., Gwaltney, A.Y., & Peay, H.L. (2021). Expert evaluation of strategies to modernize newborn screening in the United States. *JAMA Network Open*, 4(12), Article e2140998. https://doi.org/10.1001/jamanetworkopen.2021.40998.

Bailey, D.B. Jr, Beskow, L.M., Davis, A.M., & Skinner, D. (2006). Changing perspectives on the benefits of newborn screening. *Mental Retardation and Developmental Disabilities Research Reviews*, 12(4), 270–279.

Beck, U. (1992). *Risk society. Towards a new modernity*. Sage.

Bombard, Y., &, Miller F.A. (2012). Reply to Ross' commentary: Reproductive benefit through newborn screening: Preferences, policy and ethics. *European Journal of Human Genetics*, 20, 486–489.

Botkin, J.R., & Rothwell, E. (2016). Whole genome sequencing and newborn screening. *Current Genetic Medicine Reports*, 4(1), 1–6.

Buchbinder, M., & Timmermans, S. (2011). Newborn screening and maternal diagnosis: Rethinking family benefit. *Social Science and Medicine*, 73(7), 1014–1018.

Canadian Agency for Drugs and Technologies in Health. (2011, August 26). *Newborn screenings for disorders and abnormalities in Canada*. Retrieved September 22, 2022, from https://www.cadth.ca/sites/default/files/pdf/Newborn_Screening_es-26_e.pdf.

Corburn, J. (2005). Street science: Community knowledge and environmental health justice. The MIT Press.

Cornel, M.C., Rigter, T., Jansen, M.E., & Henneman, L. (2020). Neonatal and carrier screening for rare diseases: How innovation challenges screening criteria worldwide. *Journal of Community Genetics*, 12(2), 257–265.

Fiorino, D.J. (1990). Citizen participation and environmental risk: A survey of institutional mechanisms. *Science, Technology, & Human Values*, 15(2), 226–243.

International Association for Public Participation. (2018). *Public participation spectrum*. Retrieved September 22, 2022, from https://iap2canada.ca/foundations.

Jansen, M.E. (2017). Reply to a Mackie. *European Journal of Human Genetics*, 25(7), 791–792.

Jansen, M.E., Klein, A.W., Buitenhuis, E.C., Rodenburg, W., & Cornel, M.C. (2021). Expanded neonatal bloodspot screening programmes: An evaluation framework to discuss new conditions with stakeholders. *Frontiers in Pediatrics*, 9, 635353.

Jansen, M.E., Metternick-Jones, S.C., & Lister, K.J. (2016). International differences in the evaluation of conditions for newborn bloodspot screening: A review of scientific literature and policy documents. *European Journal of Human Genetics*, 25(1), 10–16.

Jardine, C., Turtiak, M., & Driedger, S.M. (2009). Public participation and risk governance: Opportunities and barriers. *International Journal of Risk Assessment and Management*, 13(3/4), 260–275.

Kahan, D.M. (2012). Cultural cognition as a conception of the cultural theory of risk. In S. Roeser, R. Hillerbrand, & M. Peterson (Eds.), *Handbook of risk theory: Epistemology, decision theory, ethics, and social implications of risk* (pp. 725–759). Springer.

Krewski, D., Hogan, V., Turner, M.C., Zeman, P.L., McDowell, I., Edwards, N., & Losos, J. (2007). An integrated framework for risk management and population health. *Human and Ecological Risk Assessment: An International Journal*, 13(6), 1288–1312.

Krewski, D., Turner, M.C., Lemyre, L., & Lee, J.E.C. (2012). Expert vs. public perception of population health risks in Canada. *Journal of Risk Research*, 15(6), 601–625.

Krewski, D., Westphal, M., Andersen, M.E., Paoli, G.M., Chiu, W.A., Al-Zoughool, M., Croteau, M.C., Burgoon, L.D., & Cote, I. (2014). A framework for the next generation of risk science. *Environmental Health Perspectives*, 122(8), 796.

Kuzma, J. (2016). Policy: Reboot the debate on genetic engineering. *Nature*, 531, 165–167.

Levy, H.L. (1998). Newborn screening by tandem mass spectrometry: A new era. *Clinical Chemistry*, 44(12), 2401–2402.

Newborn Screening Ontario (n.d.). https://www.newbornscreening.on.ca.

Nicholls, S.G., Newson, A.J., & Ashcroft, R.E. (2016). The need for ethics as well as evidence in evidence-based medicine. *Journal of Clinical Epidemiology*, 77, 7–10.

Nicholls, S.G., Wilson, B.J., Etchegary, H., Brehaut, J.C., Potter, B.K., Hayeems, R., Chakraborty, P., Milburn, J., Pullman, D., Turner, L., & Carroll, J.C.

(2014). Benefits and burdens of newborn screening: Public understanding and decision-making. *Personalized Medicine*, 11(6), 593–607.

Potter, B.K., Avard, D., Entwistle, V., Kennedy, C., Chakraborty, P., McGuire, M., & Wilson, B.J. (2009). Ethical, legal, and social issues in health technology assessment for prenatal/preconceptional and newborn screening: A workshop report. *Public Health Genomics*, 12(1), 4–10.

Potter, B.K., Avard, D., & Wilson, B.J. (2008). Newborn blood spot screening in four countries: Stakeholder involvement. *Journal of Public Health Policy*, 29(1), 121–142.

Renn, O. (2008). White paper on risk governance: Toward an integrative framework. In O. Renn & K.D. Walker (Eds.), *Global risk governance: Concept and practice using the IRGC framework* (pp. 3–73). Springer Netherlands.

Rogowski, W.H., Grosse, S.D., Schmidtke, J., & Marckmann, G. (2014). Criteria for fairly allocating scarce health-care resources to genetic tests: Which matter most? *European Journal of Human Genetics*, 22(1), 25–31.

Shih, S.T.F., Farrar, M.A., Wiley, V., & Chambers, G. (2021). Newborn screening for spinal muscular atrophy with disease-modifying therapies: A cost-effectiveness analysis. *Journal of Neurology, Neurosurgery and Psychiatry*, 92, 1296–1304.

Shone, S.M. (2019). Newborn screening policy decisions: Adding conditions. *North Carolina Medical Journal*, 80(1), 42–44.

Sims, E.J., Mugford, M., Clark, A., Aitken, D., McCormick, J., Mehta, G., Mehta, A., & UK Cystic Fibrosis Database Steering Committee. (2007). Economic implications of newborn screening for cystic fibrosis: A cost of illness retrospective cohort study. *The Lancet (British Edition)*, 369(9568), 1187–1195. https://www.sciencedirect.com/science/article/abs/pii/S0140673607605650.

Stern, P.C., & Fineberg, H.V. (1996). *Understanding risk: Informing decisions in a democratic society*. Committee on Risk Characterization, National Research Council. https://doi.org/10.17226/5138.

Stirling, A. (2007). Risk, precaution and science: Towards a more constructive policy debate. Talking point on the precautionary principle. *EMBO Reports*, 8(4), 309–315.

Therrell, B.L., Padilla, C.D., Loeber, J.G., Kneisser, I., Saadallah, A., Borrajo, G.J.C., & Adams, J. (2015). Current status of newborn screening worldwide: 2015. *Seminars in Perinatology,* 39(3), 171–187.

UK National Screening Committee. (2000). *Second report of the UK national screening committee*. London, UK, Department of Health.

Ulph, F., Wright, S., Dharni, N., Payne, K., Bennett, R., Roberts, S., Walshe, K., & Lavender, T. (2017). Provision of information about newborn screening antenatally: A sequential exploratory mixed-methods project. *Health Technoogyl Assessment Reports*, 21(55), 1–240.

van der Burg, S., & Oerlemans, A. (2018). Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening. *Bioethics*, 32(3), 171–183.

Watson, M.S., Lloyd-Puryear, M.A., & Howell, R.R. (2022). The progress and future of US newborn screening. *International Journal of Neonatal Screening*, 8(3), 41–66.

Webler, T., & Tuler, S. (2018). Four decades of public participation in risk decision making. *Risk Analysis*, 41(3), 503–518.

Wilson, J.M.G., & Jungner, G. (1968). Principles and practice of screening for disease (No. 24). World Health Organization. Retrieved September 22, 2022, from https://apps.who.int/iris/handle/10665/37650.

Zikmund-Fisher, B.J., Smith, D.M., Ubel, P.A., & Fagerlin, A. (2007). Validation of the subjective numeracy scale: Effects of low numeracy on comprehension of risk communications and utility elicitations. *Medical Decision Making*, 27(5), 663–671.