

Governing population screening in an age of expansion: The case of newborn screening

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ABSTRACT

Newborn bloodspot screening is one of the most enduring and successful population screening initiatives. Yet technological innovation to permit simultaneous measurement of multiple biomarkers – and potentially, entire genomes – has spurred expansion and debate. Through a cross-jurisdictional comparison, we describe the varied roles and reach of screening-related governance structures in the United States, the United Kingdom, New Zealand and Canada, and highlight the distinct values and resources brought to bear by the genetics, public health and maternal-child health communities in adjudicating the benefits and burdens of expanded newborn screening. We call for the expansion of formal governance structures that are balanced in resources and perspective and mandated to ensure that the organization and delivery of newborn screening achieves optimal quality.

KEY WORDS: Mass screening; public health administration; public health; preventive medicine; health services administration; infant; newborn

La traduction du résumé se trouve à la fin de l'article.

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Newborn bloodspot screening (NBS) programs have expanded markedly in recent years, motivated by technological developments that have increased the number of conditions detectable and an accompanying press of parent and professional advocacy.¹ Heated debate has accompanied recent developments, with the salutary consequence that these once almost-unnoticed child public health initiatives now attract serious attention. Yet while debate on the ethics and outcomes of expanded screening is now commonplace,² discussion of the governance arrangements for airing and fairly resolving such debates has been largely absent. This is ironic because NBS expansion has been accompanied by a corollary expansion of governance arrangements to increase oversight and accountability – an expansion that remains incomplete.

In this commentary, we provide brief overviews of the evolution of governance arrangements for NBS in four different jurisdictions, to illuminate how different interest coalitions bring distinct values and resources to bear. Our analysis identifies the governance improvements that are underway and those needed to support fair, effective and accountable NBS programs: programs that can accommodate expansion potential while delivering the health gains that are the promise of, and justification for, these important public health interventions.

Newborn screening governance: Three interest coalitions, four jurisdictions

Formal governance arrangements for population screening are relatively new in most jurisdictions, and not uniformly present internationally.³ Their development reflects the state's need to enroll actors and expertise from outside government in the development of population screening policy, notably medical practitioners and medical science. It also reflects the need to utilize mechanisms of control beyond state sanction in program

implementation, such as professionalism. Their composition reflects the variable involvement and authority of different interest coalitions, each of which advances distinctive values and brings diverse resources to bear.

Three Interest Coalitions

Historically, governments have relied almost exclusively on actors and institutions from the 'genetics' world for the governance of NBS. Members of this interest coalition, including scientists, clinicians, patient advocates, industry, and sympathetic state actors, have advanced a commitment to rare genetic disease and have strongly valued the generation of diagnostic and reproductive risk information as ends in themselves. As well, they bring capacity to develop and conduct laboratory-based screening tests and a permissive approach to technological expansion in advance of robust evidence.⁴

Two other interest coalitions also have a potential role to play in NBS governance, though their value commitments and institutional resources have been more variably drawn upon. 'Public health' draws on preventive medicine and the science of epidemiology to bring attention to the benefits and burdens of screening across the full population of tested individuals, and across the full pathway from screening through confirmatory testing and effective early intervention. Further, it demands high-quality evidence to justify and sustain population-wide

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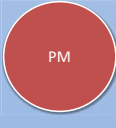
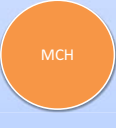
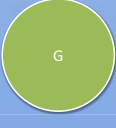
Interest coalitions	Core values	Key resources	Governance focus
Public health ^{5,16} 	<ul style="list-style-type: none"> •Focus on balance of benefits and burdens across population •Attention to screening as “pathway,” with ultimate clinical benefits as goal •Requirement of high quality evidence to justify intervention 	<ul style="list-style-type: none"> •Expertise in epidemiology and evidence-based medicine 	Evidence
Primary maternal & child health ^{17,18} 	<ul style="list-style-type: none"> •Focus on children and families •Attention to family wellness and patient engagement •Primary care (first contact, coordination, comprehensiveness, continuity); variably community-based 	<ul style="list-style-type: none"> •Infrastructure for sample collection and follow-up, education/consent for the full population •Infrastructure for clinical case finding as complement or alternative to screening 	System of care delivery
Genetics ^{8,19} 	<ul style="list-style-type: none"> •Focus on rare disease •Valuing information, including reproductive risk information, as an end in itself •Permissive approach to technological expansion in advance of robust evidence⁴ 	<ul style="list-style-type: none"> •Expertise in specialized laboratory testing •Expertise in treatment of rare genetic disease 	Testing proficiency

Figure 1. Three interest coalitions with roles in the governance of newborn screening

interventions, and possesses the methodological wherewithal to generate and evaluate such evidence.⁵ Finally, a third interest coalition advances the values and resources of primary maternal and child health care. Variably composed in different jurisdictions, with a more medical (e.g., US, Canada) or community-based (e.g., UK, New Zealand) orientation, this ‘primary maternal-child’ interest coalition advances primary care’s family and wellness centred values,⁶ and provides the front-line infrastructure for delivering essential NBS services, including parent engagement (education, consent), sample collection, and follow up (e.g., referral). In its case-finding capacity, it also serves as a complement (i.e., for false negatives) or alternate to screening-based diagnosis (Figure 1).

Four Jurisdictions

The pace and scope of expansion in NBS has varied markedly internationally, as have developments in governance arrangements. The US has attracted particular attention, with the controversial American College of Medical Genetics (ACMG) report in 2005 that recommended an expanded uniform national panel (originally 29 primary + 25 secondary targets; currently 31 and 26⁷)* accounting for the technological opportunity offered by tandem mass spectrometry and the variability in conditions screened across independent, state-run programs.⁸ Despite criticism, the advisory committee with responsibility for national oversight consistently promoted this report, though it has recently enhanced its engagement with

public health, and reformed and expanded its evidence review process.⁹ The committee continues to advise on additions to the uniform panel and to recommend improvements in NBS programs (e.g., for implementation, evaluation, education). However, in the context of the fragmented system of US health care financing and delivery, structural governance is distinctly absent, as the committee lacks the mandate to oversee screening programs, including the delivery of follow-up testing or access to treatment.

The UK presents a notable contrast to the US with its recent expansion to only nine conditions on the blood spot screening panel[†] and a history of steadily tightening central control over local programs, including the cancellation of screening initiatives that did not meet criteria, and the institution of clear standards for implementation, quality assurance and management.¹⁰ NBS is governed through the UK National Screening Committee (NSC), which advises on all population screening initiatives across the four UK countries. Within England, the NSC also supports the implementation of eight non-cancer screening programs, including those falling under the auspices of the Fetal, Maternal and Child Health Coordinating Group (FMCH). With a dominant role for public health and a strong connection to primary and community based maternal and child health care, NBS is provided in association with a single-payer national health service, with most medically necessary physician, hospital and outpatient services free at point of care.

*This includes point of care screening programs, such as infant hearing screening and pulse oximetry.

†This does not include point of care screening programs, notably infant hearing screening and the newborn and infant physical exam.

Governance history	Structural characteristics of oversight	Other organizational features	Role of interest group
<p>USA *</p> <ul style="list-style-type: none"> Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Established 2003 Advocated for adoption of ACMG Uniform Panel Has since reformed the evidence review process to consider additions 	<ul style="list-style-type: none"> Segregated: Remit limited to newborn screening; focus on blood spot screening, but growing oversight of point of care testing, including hearing screening Fragmented: No oversight and no responsibility for system of care (structurally disconnected from service delivery and financing) 	<ul style="list-style-type: none"> Evaluative criteria specific to newborn screening (NBS); explicit concessions for rare disease No process for reconsideration/deletion of conditions on the panel NBS is mandatory in most US states 	<p>Extent of authority vs Extent of involvement. MCH (orange) is low on both. PM (red) is medium. G (green) is high on both.</p>
<p>Ontario, Canada †</p> <ul style="list-style-type: none"> Provincial Council for Maternal, Child Health, with Advisory Committee for Maternal Child Screening, and Newborn Screening Subcommittee Established 2010 Inherited ACMG Uniform Panel, which was adopted from 2005 through 2008 Has since reformed the evidence review and governance process 	<ul style="list-style-type: none"> Coordinated: Remit for population screening in the maternal-child context; focus on newborn bloodspot screening and antenatal screening Integrated: Partial oversight of screening pathway, with shared responsibility (with provincial health insurance system) for full system of care 	<ul style="list-style-type: none"> Evaluative criteria specific to NBS; no explicit concessions for rare disease Ability to consider additions and deletions to panel NBS is not mandatory in Ontario but there is no requirement of express consent; consent is often implied 	<p>Extent of authority vs Extent of involvement. MCH (orange) is low. PM (red) is medium. G (green) is high on both.</p>
<p>New Zealand ‡</p> <ul style="list-style-type: none"> National Screening Unit, with Newborn Metabolic Screening Program Governance Team Established 2002 Adopted expanded Australasian NBS panel Has since reformed the governance process 	<ul style="list-style-type: none"> Coordinated: Remit for all population screening Integrated: Oversight of screening pathway, with shared responsibility (with national health service) for full system of care 	<ul style="list-style-type: none"> Evaluative criteria specific to NBS; no explicit concessions for rare disease Ability to consider additions and deletions to panel NBS, as all other population screening programs, requires informed consent 	<p>Extent of authority vs Extent of involvement. MCH (orange) is medium. PM (red) is low. G (green) is high on both.</p>
<p>England, UK §</p> <ul style="list-style-type: none"> UK National Screening Committee, with Fetal, Maternal and Child Health Coordinating Group, and Blood spot program advisory committee Established in 1996 Adopted uniform 9-condition panel across England 	<ul style="list-style-type: none"> Coordinated: Remit for all population screening Integrated: Oversight of screening pathway, with shared responsibility (with national health service) for full system of care 	<ul style="list-style-type: none"> General evaluative criteria – not specific to NBS Ability to consider additions and deletions to panel NBS, as all other population screening programs, requires informed consent 	<p>Extent of authority vs Extent of involvement. MCH (orange) is low. PM (red) is high. G (green) is low.</p>

Notes:

*Because new authorizing legislation was not passed to prolong its mandate in 2013, the Secretary used the Act's discretionary powers in April of 2013 to re-establish the Committee as the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC). For convenience, the old name and acronym are used here.

See:

Authorizing Legislation: Title 42-PHSA-IX-Part A 300b SACHDNC Legislation (PDF - 55.9 KB).

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†Canada is a decentralized federation, and the delivery of most health care and public health services is coordinated at the provincial level. There is currently no national framework for the governance of newborn or other population screening services. Ontario is Canada's largest province, with a population of approximately 14,000,000. Like most provinces, Ontario funds a universal system of medical insurance providing free at point of care access to all medically necessary physician and hospital services, as well as partial coverage for other services (e.g., drugs, technologies); medical foods for persons diagnosed through NBS are covered for life in Ontario.

See:

Ontario Newsroom. Ontario becomes national leader in newborn screening. 2005. [January 15, 2013]. Available from:

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‡See:

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http://www.nsu.govt.nz/files/ANNB/Newborn_Metabolic_Screening_Programme_Policy_Framework_June_2011.pdf.

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United Kingdom National Screening Committee. History of the United Kingdom National Screening Committee. 2013. [May 4, 2014]. Available from:

<http://www.screening.nhs.uk/history>; United Kingdom National Screening Committee. United Kingdom National Screening Committee Cross Programme Work. 2013.

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Embargoed 00.01, Friday 9 May, 2014, "New screening will protect babies from death and disability" <http://www.screening.nhs.uk/meetings>, accessed May 13, 2014.

Figure 2. Governance of newborn screening in four jurisdictions

New Zealand and Ontario, Canada represent intermediate positions between these two poles, embodying more coordinated and integrated governance structures than the US but less than the UK. Like the UK, New Zealand oversees all population screening initiatives, including NBS, through a national governance structure. The National Screening Unit (NSU) was made responsible for NBS in 2005,¹¹ but the program's expansion at that time (to 28 conditions)[‡] adhered to the recommendations of the Joint Newborn Screening Committee of the Human Genetics Society of Australasia and the Royal Australian College of Physicians, and made no reference to national criteria for population screening. Nonetheless, the NSU has formal responsibility for oversight of quality along the full screening pathway, within a national health service that ensures access to necessary physician, hospital and relevant outpatient services.

The governance of NBS in Canada is a provincial responsibility, though recent initiatives support some national coordination.¹² In Ontario the influence of the US was early and notable, with an expansion in 2005 that effectively adopted the ACMG recommendations, and more recent additions paralleling those in the US. However, these developments also initiated experimentation with new governance arrangements. Building on reforms related to Ontario's maternal and child health system,¹³ the governance of NBS was restructured (2009–2010) to encompass population screening in the maternal, newborn and child health context, in association with a provincial data infrastructure to measure maternal child health status and outcomes to inform policy and health system improvements. The remit of these governance arrangements extends beyond the provision of advice on NBS disease targets, to encompass the screening pathway and relevant high quality care within a provincial health insurance system that ensures access to medically necessary hospital and physician services (Figure 2).¹⁴

Towards better governance

This review of NBS governance arrangements identifies several patterns and opportunities for change. First, while genetics has historically dominated the governance of NBS, across the four jurisdictions that have formalized their arrangements, the involvement of two other interest coalitions – public health, and primary maternal-child health – is notable and laudable. Second, there is increased reliance on formal evidence review processes, drawing on well-established public health principles, with consideration of both additions and deletions to screening panels. As well, there is increased attention to screening as a program or comprehensive pathway, which includes parent education and engagement and concern for short- and long-term outcomes. Notably, those jurisdictions with the most robust governance arrangements (UK, NZ) most strongly support informed consent – highlighting that voluntarism is no substitute for good governance. It is only with a continued emphasis on evidence-based screening targets, pathway of care thinking, and ongoing evaluation, that NBS quality care can be assured.

These developments also suggest ways in which the organization of governance arrangements influences function and mandate. Specifically, governance arrangements that address NBS alongside other population screening initiatives, notably other maternal-child screening initiatives (e.g., prenatal screening, hearing screening), appear to bring with them involvement from broader interest coalitions, which bring distinct but complementary expertise and values. Similarly, greater emphasis on systems of care delivery may enhance the role of the primary maternal-child health community. Bringing NBS out of its silo and into a multi-disciplinary governance framework-placing it as one among many population screening programs, especially those relevant to maternal-child health – is of salutary importance.

As NBS programs contemplate a next wave of exponential increase in screening panels, notably through genome sequencing,¹⁵ new governance arrangements that represent a broader set of interests, values and resources will be essential to ensure that technological opportunities serve as midwives to core goals: to enable early identification of conditions where effective treatments and high-quality and universally accessible screening programs can enhance health for all children and their families.

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RÉSUMÉ

Le dépistage des taches de sang chez les nouveau-nés est une des initiatives de dépistage de masse les plus durables et fructueuses. Pourtant, l'innovation technologique pour permettre la mesure simultanée de multiples marqueurs biologiques, et peut-être des génomes entiers, a suscité une expansion et un débat. Nous décrivons au moyen d'une comparaison inter-compétence la variété de rôles et la portée des structures de gouvernance du dépistage aux États-Unis, au Royaume-Uni, en Nouvelle-Zélande et au Canada, et soulignons les valeurs et ressources distinctes mises à profit par les communautés de la génétique, de la santé publique et de la santé maternelle et infantile en jugeant les bénéfices et le fardeau du dépistage élargi chez les nouveau-nés. Nous demandons une expansion des structures de gouvernance officielles qui soit équilibrée en termes de ressources et de perspective et ayant le mandat de s'assurer que l'organisation et la prestation du dépistage chez les nouveau-nés soient d'une qualité optimale.

MOTS CLÉS : dépistage de masse; administration de la santé publique; santé publique; médecine préventive; administration des services de santé; nourrisson; nouveau-né