

## ARTICLE

# The complex promise of newborn screening

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### Abstract

*Newborn screening has been practised as a form of preventive medicine since the 1960s, and has attracted increased attention in recent years as technological capacities expand. Like other emerging economies, India faces pressure to expand infant screening, though developments have been halting. The promise of newborn screening is the reduction of infant mortality and morbidity from a host of rare, typically genetic, disorders. Deciding what priority should be placed on the realisation of this promise, together with the practical challenge of coordinating the screening enterprise, requires the use of decision making frameworks that address both clinical criteria and values conflicts. Frameworks for public health ethics can aid sound policy development in India, and help to inform the larger international debate about the expansion and benefits of NBS.*

### Introduction

Population screening of newborns as a form of preventive medicine is of increasing interest internationally. Indeed, despite the fact that these programmes can make only a minor contribution to reducing the global burden of infant morbidity and mortality - focused as they are on rare, primarily genetic, diseases (eg, phenylketonuria) - they continue to make inroads in emerging and even depressed economies (1-3). Proponents argue that the contribution of genetic and congenital conditions to neonatal ill health is becoming increasingly relevant to the developing world as more immediate threats to neonatal health and survival are addressed and the "epidemiologic transition" advances (4-6). They note also that the growth of the middle classes in emerging economies increases the demand for these technologically sophisticated services (7). The argument for newborn screening (NBS) in the developing world is suggested as a matter of justice - that it is unfair to withhold the benefits of first world medicine from infants in under-privileged communities (8). Ultimately, however, developments in NBS are propelled by a range of more prosaic interests: private firms seeking commercial opportunity from the sale of screening tests, specialist professionals determined to advance the technological and clinical sophistication of their fields in their respective jurisdictions, and parent and consumer advocacy groups with powerful - often deeply personal - attachments to the clinical potential of early detection (9).

Like other emerging economies, India faces pressure to expand

NBS. To date, however, developments have been halting. Families with the financial means can purchase screening through public or private laboratories and clinics, particularly in urban centres. Yet access for the wider population is more limited. Meanwhile, obstacles to realising the health benefits of NBS are enormous in the Indian context. Population-wide benefits can only be achieved where sophisticated health systems exist to track screened infants and manage challenging and resource-intensive health needs. Further, even where these systems are well managed, additional harms are possible. These include the potential harms of false positive screening results, and that genetic diagnoses and reproductive risks may be misinterpreted as blameworthy.

In addition to these practical challenges, debates about the development and expansion of NBS concern its goals - what NBS is intended to achieve. Traditionally, the hoped-for benefits of NBS have primarily concerned clinical outcomes such as reduced mortality and morbidity for the screened infant. More recently, commentators have argued for an expanded interpretation of benefit, to include non-clinical improvements in the lives of infants' families (10). These contests over the ends of screening are especially glaring in emerging economies, where the potential to deliver expansively defined social benefits exists alongside basic, yet unfulfilled, clinical needs. Yet despite the intensity of these values conflicts, decision-making frameworks to guide NBS policy are focused on clinical considerations, and the role of values in decision-making remains opaque. Meanwhile, ethical reflection on these issues is both limited in quantity and narrowly concerned with the issue of informed consent. This is an important issue to be sure, but one that draws inspiration from clinical bioethics, which can offer limited guidance for public health interventions such as NBS.

Though generally thought of as a luxury of the developed world, NBS has always had an international orientation. Robert Guthrie, who first developed a viable screening assay in the early 1960s, took his campaign for NBS to emerging economies, relying on appeals to compassion as well as evidence. Early screening initiatives included parts of the developing world, and agencies that have advanced the cause of NBS have long worked with emerging economies (3,11). Yet as the relevance of NBS for the developing world comes into focus once again, the complex nature of its promise is increasingly salient.

In this paper, I consider the potential of NBS in the Indian

context, where the types of benefits that might arise are contested, and the challenges facing the realisation of these benefits are profound. I first examine what is known about the patchwork of developments in NBS in the country and then review the practical challenges that might arise in achieving population benefits from these programmes. I next consider the debates that have been aired about the goals of NBS, and the particular intensity of these values conflicts for emerging economies. I then turn to ethical frameworks for evaluating public health interventions, and consider how the framework proposed by Nancy Kass can guide values-explicit policy for NBS in India (12). I close by suggesting that consideration of the Indian case throws the complex promise of NBS into sharp relief, and provides guidance for wider debates about policy for infant screening.

### **Newborn screening in India**

In India today, access to NBS is growing but remains partial and incomplete. Access is primarily available at cost as private laboratories within India and from abroad introduce relevant testing technologies (13-16). Some NGOs work to provide access to those with fewer financial means. For example, the Spastic Society of Tamil Nadu (SPASTN, Chennai) operates NBS clinics as part of its preventive mandate (17,18). In addition, some medical colleges and hospitals provide access through free or subsidised pilot studies or local initiatives (19,20). Other initiatives provide screening under the auspices of research. Indeed, the Indian Council of Medical Research has worked since 2005 to initiate pilot projects in five cities, each screening 500,000 newborns free of charge, to establish the prevalence of certain genetic defects in the population (21); a 2008 report suggests that the programme is being launched on a reduced scale, screening 20,000 newborns in each of five cities for two conditions (22). Yet interest among states and union territories is increasing. In January 2007, the Union Territory (UT) of Chandigarh was declared the first state or UT in India to fund mass genetic screening (NBS and prenatal diagnosis), with a reduced fee for all and free access for the poor (23). In February the following year, Goa was said to be the first state to introduce mandatory NBS (24). Finally, in February 2009, the union cabinet was reported to have approved a proposal to establish an institute in Kalyani (West Bengal) to launch a large-scale programme for NBS (25).

Despite these developments, the relevance of NBS for India is hotly debated. Many commentators suggest that NBS is increasingly important, as the epidemiologic transition increases the significance of genetic disorders, and because of the particularly high burden of some genetic diseases in the population (eg, glucose-6-phosphate dehydrogenase deficiency, and haematological disorders) and high rates of consanguinity, especially in the southern states (4-6, 26-29). Yet others caution about the lack of data on the incidence of various genetic diseases, their natural history and the effectiveness of treatment in local conditions - data that are essential to the appropriate use of NBS technology (30,31). This is especially challenging given the large number of separate

endogamous communities in India, leading to distinct patterns of disease susceptibility in each sub-population (4). Further, the small contribution of genetic disease to overall infant mortality reduces the relevance of NBS for India (1). And the pressing weight of more basic healthcare needs calls into question the justice of allocating resources to screening services (31,32). Still, the advancement of genetic medicine in India is defended with the argument that the fruits of genomic science should not remain a luxury available only to the residents of developed nations (8). Indeed, as NBS becomes more readily available, though in patchy and uneven ways, calls for state action to introduce mandatory programmes are on the rise (33,34).

### **The complex promise of newborn screening: challenges in practice**

By definition, population screening is a broadly-based intervention involving pre-screening access to appropriate individuals, and post-screening care for those with positive results. Indeed, NBS is commonly discussed as a process with six key elements. In addition to the screening process itself (ie, properly timed specimen collection and transportation, laboratory analysis and reporting), the screening intervention includes the education of health professionals and consumers, early follow-up (including confirmatory testing), definitive diagnosis, clinical management (including counselling, immediate treatment and long term follow-up), and system evaluation (35,36). In short, the early detection of rare and treatable disorders can only result in reduced morbidity and mortality if screened infants are traced and managed. This requires sophisticated recruitment and follow-up mechanisms to ensure that all eligible infants are tested, and results communicated to families and their healthcare providers. It also requires well-developed health professional capacity, with primary and secondary care providers equipped to understand the significance of rare diagnoses and to prevent or manage disease in the infant. Finally, it requires systems that can ensure access to, and facilitate ongoing utilisation of, expensive dietary regimens or therapies.

Yet many aspects of India's healthcare system give cause for concern that these conditions can be met. Access to healthcare for the poor, particularly in rural areas, is deeply challenging, and even the urban middle classes may experience serious problems in making good use of NBS test results (37). Genetic services are not widely available in India, nor are adequate laboratory facilities to diagnose inherited metabolic disorders - the most common targets of NBS tests (38-40). Further, little education in genetics is provided in medical colleges, leaving primary and secondary care clinicians poorly equipped to understand and manage genetic disease (39,42). In addition, neonatal care units, especially at the district level, are limited in their availability, with most neonatal care available through specialised tertiary units in urban areas (43). Finally, the special diets that are the mainstay of treatment for many conditions targeted by NBS (eg, phenylketonuria) are not available in India (though they can be imported) and are very expensive and generally unaffordable by most Indian families (31).

Alongside these practical challenges to realising its benefits, NBS has the potential to cause significant harm. The initial assessment of newborns involves a screening test that yields a high proportion of false positives for most screened conditions, requiring follow-up testing to distinguish infants who are truly affected from those who are not (31). While evidence is lacking from developing nations, evidence from developed countries indicates that false positive screening results can have serious negative consequences for some families (44), creating parental stress, lingering concerns about the infant's health, and substantial over-medicalisation (45-48), including the "impoverishment risk" for families who must pay for the associated medical costs (49). Further, prevalent "misunderstandings" of screening results may prove resistant to typical educational interventions, as interpretations of disease and disease risks are open to cultural influence. Duanna Fulwiley has documented, for example, the pervasive belief that the heterozygous (ie, carrier or "trait") state for sickle cell disorders - viewed as clinically benign by biomedical authorities - is clinically consequential in Senegal and perhaps in other jurisdictions (50). This is cause for concern in India, because NBS for the sickle cell disorders reliably identifies virtually all carriers (at rates of 17-100 carriers per affected child detected, depending on the ethnic composition of the screened population) and is recommended in countries such as India because of the relative prevalence of the disorders (51).

In addition, NBS for genetic disease (the vast majority of screened conditions) often leads to the assessment of reproductive risks in parents. Indeed, the opportunity to alert parents to the reproductive risks that they may face is a commonly identified benefit of NBS. Yet, the assessment of reproductive risks in parents and extended family members carries with it the potential for significant harm. Misattributed paternity may be detected through these assessments, with the consequent harms for the child, the family, and particularly the woman (51-53). Further, while data on genetic counselling in varied cultural contexts are limited, potential harms exist for families, and especially women, arising from culturally-specific interpretations of risk and responsibility for disease (54-56).

### **The complex promise of newborn screening: challenges in principle**

Achieving the benefits of NBS is a complex financial, organisational, and cultural task. Yet as has become increasingly apparent in recent years, the issue is not just how to achieve the benefits of NBS, but what benefits to achieve. As Pollitt has argued, the diversity of screening panels across international jurisdictions is not explained by variations in the incidence of disease in different populations, or differences in the organisation and capacity of health systems (7). Rather, different value judgments have been made about what NBS can and should accomplish (57). Further, the frameworks relied upon to support the development of NBS policy emphasise clinical measures of benefit and harm (including clinical measures of psychological impact). Thus, despite the significance of values and values conflicts in NBS policy, robust

consideration of values in decision making is poorly supported and accompanying ethical debates are impoverished.

To date, much NBS policy relies on the 1968 WHO framework of Wilson and Jungner which identifies criteria for evaluating the effectiveness of screening programmes (7). The ten "principles" of screening require that there be adequate knowledge of the condition to be screened, the screening test and the treatment (that the natural history of the disease be known, that a suitable test be available, that accepted treatment and treatment facilities be available, etc) and that the costs be balanced (58). While broadly informative, these criteria have been criticised as too vague to inform clear decisions, and not fully relevant to NBS (7,57). Even in the UK, where these principles have been carefully revised to provide a more complete set of criteria, distinctions between "adequate" and "not quite good" enough remain qualitative and thus ultimately subjective (7,57,59).

In the US, the basic approach of the Wilson and Jungner criteria has been dispensed with in favour of a more complex heuristic. At the behest of the US Health Resources and Services Administration, the American College of Medical Genetics convened an expert panel, solicited input from diverse specialist and consumer experts, and commissioned literature reviews to recommend a "uniform" screening panel to US states (60). The committee's approach strayed far beyond traditional principles of screening in emphasising non-clinical benefits for the families of screened infants, and positively valuing the capacity of multiplex technology (notably tandem mass spectrometry) to screen for multiple conditions at once. The set of recommendations developed in consequence has been criticised for its inadequate methods, and the biased membership that advanced them (47,61). Further, though the relevance of values is more apparent in this set of recommendations, the role of values in reaching final decisions remains opaque.

As is apparent from the US recommendations, the debate about what benefits NBS should achieve is growing. There are calls to change the "dogma" that NBS should emphasise the provision of clinical benefits such as reduced mortality and morbidity for the screened infant (10). Such commentators have argued for an expanded interpretation of benefit, to include non-clinical improvements in the lives of infants' families, in particular, the opportunity to inform life planning and reproductive decision making, and to circumvent the sometimes prolonged process of reaching a clinical diagnosis of a sick child's condition ie, the "diagnostic odyssey" (7,10,62). These commentators argue that the identification of untreatable conditions can provide benefit to families by increasing knowledge about the natural history of rare conditions, and providing opportunity to advance research into disease treatments (10). Yet though widened in one respect, in encompassing the affected infant's family in its sights, the emphasis of this vision remains strangely limited, ignoring as it does all unaffected infants and families whose lives are touched, sometimes profoundly, by the unintended effects of NBS.

Alongside an increasingly strident debate about the

appropriate goals of NBS is growing discussion of the ethics of NBS. Commentators suggest that, as NBS ceases to consistently provide definitive clinical benefits to screened infants, its justification shifts from addressing a “public health emergency” to providing a “public health service” (63). While an “emergency” justification may provide warrant for mandatory NBS - as is the case in most US states - there is an increased need for parental consent when screening meets a “service” function (63,64). Yet while the growing need for informed consent in NBS is an important consideration, the focus on autonomy as the primary ethical issue reflects the biases of traditional bioethics and is surely insufficient (65).

The values conflict that is increasingly evident in the published - primarily Euro-American - literature is intensified in emerging economies. In countries such as India, the values of a growing middle class with respect to the benefits of information and the relevance of informed consent exist alongside basic and unfulfilled human needs. Indeed, demands for the most technologically sophisticated medicine exist alongside the quest for basic medical and community care to address entirely preventable causes of maternal, infant and child mortality. The existence of these contrasting needs and values is not surprising, but their close coexistence renders the conflict more vexing. These contrasting values and imperatives exist cheek by jowl within single political jurisdictions for which coherent and consistent public health policy must be crafted and on behalf of which health services for diverse populations must be organised and delivered. Decision making within these jurisdictions cannot rely on frameworks that draw exclusively on clinical considerations, or that cloak value judgments within qualitative assessments of a “good enough” outcome.

**Public health ethics: considering the complex promise of newborn screening**

There is no ready resolution to the conflict of values apparent in the development and expansion of NBS, and no framework

can provide a definite calculus for policy decision making. Yet there is a clear need for more sophisticated and values-explicit frameworks that reflect the status of NBS as a public health intervention, and extend moral considerations beyond the confines of autonomy-focused bioethics. In recent years, scholarly interest in public health ethics has blossomed, and several frameworks have been proposed that identify principles to justify public health interventions such as NBS (12,66,67). Though none are expressly applicable to NBS, I apply the six-step framework developed by Nancy Kass to consideration of this case (12). (Table 1)

The first issue to consider is the goal of the public health programme. Kass argues that the end benefits of public health interventions must be health related - involving “an ultimate reduction in morbidity or mortality”. She does not question the value of other more proximate or process goals, such as wider familial or social benefits; indeed, she argues that these should be given “strong consideration”. Thus, this framework suggests that current debates about the benefits to be achieved by NBS are less about which benefits to pursue, and more about what priority to place on all possible benefits that may arise. Applying Kass’ approach, the ultimate goal of reducing mortality and morbidity remains, with wider social goals as secondary targets of screening.

The second question asks how effective the programme will be in achieving its goals. Thus, this criterion demands full consideration of the practical challenges facing a total NBS programme (including pre-screening and post-screening care), and some evidence to corroborate the assumption that these elaborate elements can be knit together in the Indian context. Kass suggests that the quality of the evidence required depends on the burdens that might be imposed. She argues also that the need for evidence exists even in the face of our most deeply held assumptions, and indeed, precisely because such assumptions about benefit are unlikely to be otherwise called into question.

**Table 1. An ethics framework for public health (12)**

Key questions	Considerations
What are the public health goals of the proposed programme?	<ul style="list-style-type: none"> <li>• Identify the programme goals</li> <li>• Attend to ultimate, not only proximate or process, goals</li> <li>• Public health benefits are the ultimate goal of public health interventions, though other social goals may be valued</li> </ul>
How effective is the programme in achieving its stated goals?	<ul style="list-style-type: none"> <li>• Identify assumptions about how the programme will work</li> <li>• Ensure that data exist to substantiate these assumptions</li> <li>• The quality of data required should correspond with the burden imposed</li> </ul>
What are the known or potential burdens of the programme?	<ul style="list-style-type: none"> <li>• Identify burdens or harms that might occur</li> <li>• Three broad categories of moral harm exist: risks to privacy and confidentiality; risks to liberty and self-determination; risks to justice</li> </ul>
Can the burdens be minimised? Are there alternative approaches?	<ul style="list-style-type: none"> <li>• Determine potential modifications to minimise burden without greatly reducing programme’s efficacy</li> </ul>
Is the programme implemented fairly?	<ul style="list-style-type: none"> <li>• Consider whether the benefits and burdens are distributed equitably</li> </ul>
How can the benefits and burdens of a programme be fairly balanced?	<ul style="list-style-type: none"> <li>• Make decisions about whether the expected benefits justify the identified burdens</li> <li>• Fair process required to achieve resolution of inevitable values conflicts</li> </ul>

The third issue involves the need to consider the burdens or harms of the programme, both clinical and moral. This criterion asks first for identification of the full spectrum of potential harms, attention to which should be at least as expansive as is consideration of the potential benefits to be pursued. Thus, if the benefits to be achieved by NBS include relief of the "diagnostic odyssey" for families in which an untreatable disease is identified in a screened infant then we not only need evidence of this accomplishment, but corollary attention to the range of diagnostic harms possible for unaffected infants.

Nancy Kass suggests that the moral infringements likely from public health interventions are threefold: risks to privacy and confidentiality, risks to liberty and self-determination, and risks to justice. Yet these risks have particular contours in the context of NBS. The risks to privacy and confidentiality that arise with any programme that collects data about members of the public are exacerbated in the case of NBS because the information collected concerns the infant, and not her parents. Concerns for the infant's privacy and confidentiality are counterbalanced by potential clinical benefits where the information generated is relevant for medical management. Yet these infringements lack mitigating benefits when the information is incidental and non-health-serving (eg, information about benign variants or carrier status).

Risks to liberty and self-determination arise in several ways through NBS. As noted previously, much recent commentary has focused on the growing need for informed consent in NBS. Kass has pointed out for public health interventions generally, that, given evidence that the adoption of explicit consent for NBS is not particularly difficult, that it does not result in significantly reduced uptake, and that when well managed, it can generate benefits in the form of better educated parents (68,69), there is no warrant in public health ethics to pursue compulsory approaches. Yet risks to liberty and self-determination may remain, even where NBS is offered rather than required.

As Francis et al have argued, the consideration of ethics of public health calls for a more sophisticated understanding of autonomy, one that recognises our vulnerability and relatedness (70). The standard vision of autonomy emphasises "reasoned choice by a competent individual" with a clear idea of her values and preferences (70). In the context of NBS, however, the typical view of autonomy is falsified both because the decision maker is not the individual who is screened (an infant), and because the agent who must make decisions about screening (a parent) is profoundly vulnerable in her relationship to this infant (70). Even voluntary forms of NBS demand that parents, at a stressful and intensely emotional time, make reasoned judgments regarding an offer of screening from a public health authority promising benefit. A US video (<http://youtube.com/watch?v=yqQRio1-P6c>) promotes NBS as "a test that can save your baby's life". A fully autonomous agent - in the traditional vein - might be able to consider her preferences and make sound judgments in such a context. But the profound need to do the best for our children

renders parents intensely vulnerable to the seduction of such promises. Given this context, it may be that the goals of liberty and self-determination are best served by offers that can reasonably be expected to provide clear clinical benefits, rather than expecting that parents can rationally adjudicate between those services that will, and those services that might not, save the lives and improve the health of affected infants.

Risks to justice arise in NBS where the benefits and burdens fall inequitably on different populations. This issue is especially salient in the Indian context, where the benefits of identifying affected infants are most likely to be fully realised by more affluent populations. Similarly, the harms that arise through the identification of false positives or incidental findings are likely to be unevenly distributed, if only because the "impoverishment risk" of associated medical costs weighs more heavily on less affluent members of society.

The fourth question asks whether burdens might be minimised, and demands that we act to minimise these burdens if the programme can be modified without greatly reducing its efficacy. Here the question of alternative ways to achieve the same or similar goals comes into focus. In the Indian context, the potential benefits of NBS should be considered alongside other public health commitments to maternal, infant and child health. Can NBS build on and enhance these commitments, or will investment in NBS compete with and potentially reduce these other commitments? Will investments in these other commitments achieve more benefits, with a more equitable distribution of benefits and harms?

The fifth question asks if the programme can be implemented fairly. Relevant to the case of NBS is the potential for the benefits and burdens to fall unfairly on specific populations. This potential harm arises because genetic risks are often unevenly distributed across ethnic communities. Thus, even when NBS is not targeted at specific ethnic groups, it may have the practical effect of differentially implicating and impacting such groups.

The sixth and final question raises the challenge of governance - of collective decision making for collective goals. This question asks us to fairly balance the identified benefits and burdens of NBS in a specific context. Rather than a clear calculus of adjudication, Kass calls for a system of fair procedures to consider and weigh this complex balance.

As Kass points out in suggesting this set of six questions, different societies will reach different decisions even when applying the same ethical principles (12). Thus, the goal of the above exercise is not to reach a single decision about what should be done in India, or more accurately, by each of its states and union territories, but to highlight the relevance of such a values-explicit public health ethics framework for the development of sound NBS policy.

## Conclusion

The development and expansion of NBS offers a complex promise to the citizens of India. The practical challenges facing

the realisation of the benefits of screening are so egregious that it is tempting to focus on these to the exclusion of all other issues. Yet the conflict of values inherent in the development of this technologically sophisticated strategy for reducing infant mortality and morbidity is equally deserving of attention. Indeed, the Indian case throws these complex questions of practice and principle into stark relief. A full consideration of these complexities requires the use of decision making frameworks that explicitly address both clinical criteria and values conflicts. Emerging frameworks for public health ethics provide a useful starting point. Their use might aid sound policy development in India, and help to inform the larger international debate about the expansion and benefits of NBS.

**This paper was presented at the Indian Journal of Medical Ethics Second National Bioethics Conference, December 6-8, 2007.**

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