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Newborn Screening Expansion Using Whole-Genome Sequencing: Benefits, Harms, and Public Health Decision Frameworks, Current Evidence and Gaps

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ABSTRACT

The expansion of newborn screening through whole-genome sequencing (WGS) represents a transformative shift from targeted biochemical testing toward comprehensive genomic population screening. This paper examines the potential benefits, harms, and public-health decision frameworks guiding the adoption of WGS in newborn screening programs, synthesizing current evidence and identifying key knowledge gaps. WGS offers the capacity to detect a broader spectrum of genetic disorders at or before symptom onset, improve diagnostic yield, and enable earlier clinical intervention and lifelong risk management. However, its implementation introduces substantial challenges, including incidental findings, uncertain variant interpretation, consent complexities, data-privacy concerns, inequities in access, and increased follow-up burdens on families and health systems. Public-health decision frameworks, such as cost-effectiveness analysis, value-of-information approaches, and phased pilot implementation, provide structured tools to balance potential benefits against risks while ensuring transparency and accountability. Despite growing international pilot programs, evidence on long-term clinical utility, population-level outcomes, economic sustainability, and ethical governance remains limited. Addressing these uncertainties requires coordinated research, robust infrastructure, and inclusive policy development. Overall, while WGS-based newborn screening holds significant promise for improving early detection and preventive health strategies, careful evaluation of its clinical, ethical, and societal implications is essential before large-scale adoption.

Keywords: Newborn screening**,** Whole-genome sequencing**,** Public health policy**,** Genetic testing ethics, and Population health implementation.

INTRODUCTION

Newborn screening for serious health conditions has achieved broad international adoption, traditionally relying on limited marker panels assayed soon after birth [1]. Recent technological advances enable laboratories to sequence entire genomes from dried-blood-spots collected at the same time. Countries around the world are beginning to explore whole-genome sequencing (WGS) as a universal approach for screening newborns [2]. It offers the promise of simultaneously screening for hundreds of conditions, far beyond the capacity of traditional, multiple, separately administered tests. Countries including Australia, Canada, Germany, the Netherlands, Sweden, Switzerland, and the U.K. have launched or prepared pilot programs [3]. Newborn screening refers to testing for a set of serious health conditions soon after birth [4]. These conditions are individually rare, but population screening enables early detection of sufficient cases to deliver statistically measurable preventative benefits [4]. Whole-genome sequencing refers to determining the complete DNA sequence of an organism's entire haploid genome. WGS on blood-spots is a laboratory procedure established for clinical diagnosis [1]. WGS newborn-screening entails sequencing an infant's genome from a blood-spot sample, applying a genetic-access-screening strategy to indicate the likelihood of developing select conditions later in life, and responding with further clinical follow-up focused on any conditions flagged as high-probability[2]. Benefits, harms, and decision frameworks supply the framing and integration needed to bring historical context to the present moment comprehensively [5]. Any advance in public health technology carries the potential for complications as well as

opportunities; even a clearly beneficial intervention may not receive support from others. The first-generation newborn-screening expansions implemented in the early 2000s, the current effort is sometimes called a second generation; established benchmarks for evaluation, and these apply as much to WGS as they did to tandem-mass-spectroscopy metabolite-analysis; the phrase "merely new" thus retains relevance. Certain larger frameworks remain in active discussion, reflecting ongoing public debate over the use of the technology in prominent health-care contexts, such as the clinical, reproductive, and clinical-reproductive interfaces [6]. Current domestic evaluations typically utilize screening panels rather than population-wide approaches. Early WGS programs elsewhere explicitly combine both broad genome-wide parallelism and access indicator screening within a single delay-conscious implementation perspective. Each global precedent thus possesses the ability to inform approaches tailored to diverse demographic realities [4]. These assumptions define the inquiry while limiting its primary focus to the public-health-policy rationale, which concentrates on benefits and harms [7]. Frameworks that overlap yet do not fully correspond, such as ethical, economic, and social assessments, lie firmly outside of scope [8]. A large body of work speaks to these development- and deployment-related themes, and addressing them within the present framework would merely distract from the benefits-harms comparison and deflect attention from the sustained-policy-discussion situation [9].

Background on Newborn Screening and Whole-Genome Sequencing

Newborn screening refers to a variety of techniques used to assist in the early detection of different medical conditions [10]. These techniques can help identify congenital disorders, inborn errors of metabolism, hearing impairments, infectious diseases, and critical congenital heart defects. While early detection may increase the chance of successful treatment, newborn screening is not a population surveillance tool. The screening of newborns can also be conducted at a later [11]. Children who are overdue and children born out of the planned schedule can also be screened. Whole Genome Sequencing (WGS) is a genetic analysis that tries to map out and know the whole genetic code of an organism in a very fast and affordable manner [3]. When screening newborns in genomics, understanding the parental genome is not a necessary part of the screening process. Parallel screening of the parents, on the other hand, can help improve the understanding of how damaging certain genetic variants can be and their association with the offspring [12]. If the WGS is done on the child, identifying pathogenic mutations becomes very difficult as the same comes from the parents [3]. In a clinical environment for genomics, WGS analysis has become more common, and it emphasizes the many different workflows one can adopt. WGS on the parents and offspring does increase the knowledge of the pathogenic nature of undiscovered genetic variants [13].

Benefits of Expanding Newborn Screening with Whole-Genome Sequencing

Expanding screening with whole-genome sequencing offers four benefits: [1] detecting genetic conditions before symptoms appear, [2] providing timely follow-up tests that clarify uncertain results, [14] enabling lifetime health planning for conditions unlikely to be diagnosed later, and [4] generating gains across the population through systematic record linkage, addressing non-communicable diseases and actionable risk factors such as alcohol dependence[2]. Widespread deployment of sequence-based screening has improved the diagnostic yield, timeframe to intervention, and clinical outcomes achieved in pilot studies [15]. Gains depend on prior screening systems, broader implementation challenges, data composition, and the unscreened fraction of the newborn cohort. Simulations suggest that expanding screening on a foundational system yields greater benefits than implementing a stand-alone service, even at reduced coverage [16]. The integrated model supports improved efficiency by updating the likelihood of pathogenicity using non-coding variants appearing after other conditions; aligning with the infectious disease or other facets of newborn care to streamline programmatic procedures; and cascading genome-sequence analysis from observed conditions during screening based on population-based prior probabilities [2].

Potential Harms and Ethical Considerations

The proposed expansion of newborn screening using whole-genome sequencing (WGS) raises potential harms and associated ethical considerations [5]. Incidental findings of importance unrelated to the infant's health, misinterpretation of readouts at the population level, gathering of excessive information, overmedicalization of normal variation, and overly burdensome follow-up can all occur. Some families may be disappointed by the absence of clinically actionable results [17]. There is a risk that the additional information gleaned from WGS is unequally distributed within and between communities already marginalized by the current newborn-screening paradigm [1]. Pertinent ethical considerations include the adequacy of consent, parental autonomy, and the dilemma of parental choice under uncertainty 3. Specifying which results will be disclosed, indicating the limitations of the method, and clarifying whether and to what extent data can be shared with other agencies present challenges for meaningful consent [18]. The nature and timing of communication about incidental findings and the distinction between clinically actionable and non-actionable results introduce further ethical complexity. Justice implications concern disparities not only in access to WGS itself but also in informed consent,

data interpretation, and the generation of follow-up recommendations [2]. Models of governance, regulation, and policy planning to mitigate these risks exist alongside the anticipated health advantages provided by WGS [4]. The broader concern is whether the acquisition of genomic information could reinforce existing inequities across a spectrum of health determinants ranging from health status and risk factors to treatment and follow-up that remain to be examined clinically or otherwise addressed [19]. Disconnections between information expected and information provided can lead to disappointment. Whether WGS can yield information beyond current practices remains open to investigation [1].

Public Health Decision Frameworks and Policy Instruments

Public health decision frameworks and policy instruments play a critical role in the evaluation and implementation of candidate programmes for newborn screening [20]. Such frameworks consider the separate decision-making process, the nature of the policy question, and the prioritisation of empirical and evidence-based questions, while policy instruments focus on how the advantages and disadvantages of screening can be implemented, monitored, and evaluated. The conspectus presented here provides an overview of public health decision frameworks and policy instruments at the moment being discussed in the context of expanding newborn screening with whole-genome sequencing (WGS) [21]. These frameworks have distinct, although nuanced, terms of reference, with the decision frameworks especially adaptable to the challenges posed by screening for heritable genomic conditions and integrating WGS in newborn screening at national and international levels[3]. Public health decision frameworks encompass different approaches to addressing the benefits and downsides of newborn screening, spanning classic cost-effectiveness and cost-utility analyses, value-of-information analyses, and precautionary, or safe-to-fail, principles. Similar frameworks supporting decision-making beyond the public health domain are being established internationally and are of potential relevance [22]. Efforts to strengthen the funding and governance of WGS for newborn screening would benefit from engaging with such proposals. A range of policy instruments constitutes mechanisms for implementing, monitoring, and evaluating the expansion of newborn screening with WGS [4]. Proposals from official bodies and practising clinicians, including international guidelines, robust reimbursement structures, pilot programmes spatially and temporally isolated from routine screening, and phased implementation schemes accompanied by comprehensive evaluation, would correspondingly support governance initiatives [4]. Policy instruments and public health decision frameworks promote transparency and a clearer understanding of the implications of widening newborn screening with WGS [23].

Evidence Base: Current Data, Gaps, and Methodological Challenges

Expanding newborn screening programs to include whole-genome sequencing entails substantial changes to existing screening practices, with implications for public health policy and frameworks [5]. There is considerable interest in the prospective benefits of expanding screening programs and generating population-wide genetic information, yet concern about the potential harms and uncertainties remains widespread. Limited data are available on the diagnostic yield, clinical utility, and broader health-system impact of whole-genome sequencing in this context [2]. Existing studies have examined early adoption in single jurisdictions, predictive evidence, and knowledge of screening programs in the specific cohort of deceased infants that was studied; however, current estimates of long-term outcomes and concomitant uncertainties are imprecise [1]. Questions regarding genotype-phenotype mapping still hamper routine analysis of many genetic variations, especially at the genome-wide scale. Systematic evaluations that characterize screening programs, patient cohorts, and health-system effects to better inform prospective jurisdictions and eliminate bottlenecks hindering broader adoption are thus essential[2]. Diagnostic yield, clinical actionability, and long-term population outcomes are critical determinants of the anticipated effects of introducing genome sequencing into neonatal screening. Whole-genome sequencing is the vital next step to clarify the consequences of introducing such a powerful diagnostic technique, particularly one directly targeting diagnosis and prognosis [24]. Uncertainties about screening type and procedure, therefore, figure prominently in discussions of high-level decision frameworks. Research that rigorously examines these aspects, moreover, has the potential to galvanize movement toward population-wide sequencing, which early evidence suggests may have pivotal implications for implementation in the pursuit of equity [3]. National surveys, modelling studies, and studies from several demonstrator sites have been employed by various groups to characterize the anticipated influence of genomic data in routine practice and on a population basis. However, these analyses are not entirely reflective of the anticipated and sought-after sequencing activities [1]. The population of interest is perinatal (within the first 28 days), but data on screening deaths refer primarily to stillbirths and infants who die soon after birth [2]. Estimating the long-term impact of testing on population health is inherently complex and may not even be feasible [4]. As all genome-sequence variants cannot be readily and universally interpreted at the present time, time-variant estimates of actionable rates are also significant, but favour the assurance that, especially with existing preliminary population genetic data, far fewer and closer-to-default settings (fixed action versus no action or similar) can be expected for the few variants that can efficiently be

recognized or degeneracy still is prominent; for genome-wide non-targeted screening, convention, expertise, literature, and provenance remain germane even in the absence of overreach on the inverse or otherwise[25].

Implementation Considerations: Equity, Logistics, and Feasibility

The variety of logistical and technical challenges prevents nationwide implementation of whole-genome sequencing in newborn screening across the United States [3]. Key considerations include laboratory capacity and supply chains, turnaround times, quality assurance, data systems, consent workflows, and integration with existing newborn screening programs [26]. Ensuring widespread accessibility and equity remains a priority. Scalable, context-sensitive sequencing models can accommodate variable resources and support under-resourced communities [4]. Laboratory capacity, supply chains, and turnaround times for sequencing samples depend on the optimal number of samples processed per week and how many weeks a state can prioritize newborn screening before commercial contracts take precedence [5]. Developing a robust newborn supply chain with obvious benefits for preservation and consent increases safety, generates substantial interest, and identifies analytical pipelines. Quality assurance is particularly challenging in settings without an established pipeline [5]. Efforts to collect samples, provide results, and monitor ongoing availability motivate the exploration of a sequencer kit similar to a polymerase chain reaction (PCR) kit to support widespread exploration of possible sequencing for newborn screening [27]. Laboratory information management systems, data-analysis pipelines, and electronic laboratory requisition systems require substantial investment, and consent workflows generate concerns about clarity, simplicity, and accessibility [28]. A variety of web-based, app-based, and paper-based consent approaches exist as alternative methods for secure patient outreach, data collection, and communication feedback [3]. Newborn screening already occurs in many jurisdictions, and sequencing can be added to existing systems. Such efforts can highlight bottlenecks and, therefore, demonstrate rapid local capabilities while also supplying capture devices support for organizations targeting re-sequencing [29].

Economic Evaluation and Value Assessment

Newborn screening programs vary widely across the world [5]. The World Health Organisation (WHO) recommends a minimum of six conditions that should be included in any newborn screening programme. These conditions capture a large number of modifiable and transferable risk factors in a targeted group; geographical variations in the existence of these risk factors drive the recommendation to include only six. In Finland, screening with a targeted seventeen condition-in-laboratory panel at birth that contributes to rapid identification and precise follow-up remains the status quo [1]. Cost-effectiveness analysis of screening operations is well established [1]. The well-studied economics surrounding cost per event remain relevant to whole-genome sequencing when modelling the long-run impact on newborn screening [30]. The basic methodology revolves around modelling the costs and the probability of generating at least one actionable result over a predefined time period. Cost-effectiveness analysis is well studied, but datasets remain scarce, and a credible literal interpretation is unsuitable [5].

Stakeholder Perspectives and Communication Strategies

Newborn screening programs of the early twenty-first century carry visions inspired by their pioneering predecessors, yet they operate in a world of technological and epistemic diversity inconceivable in the 1960s[5]. The genome became widely available in the 1970s, but an embryonic conception of genomics in the early twenty-first century operated under the domination of other regulatory concerns [31]. More than a decade later, whole-genome sequencing (WGS) is under serious consideration for newborn screening in some jurisdictions, although official adoption worldwide remains distant [2]. Most extensive genome-based screening efforts to date focus on serious disorders with early-onset manifestations; many public health indices were formulated to foster well-functioning, equitable care systems from diagnostics through intervention, emphasizing habitually serious screening targets. A conceptual universe centres on the individual, less preoccupied with matters of equity, and has wider consequences [32]. The potential health gain is larger, but so are the knock-on uncertainties. Such expansion rests on four interrelated foundations: [1] uncertainties cluster around technological and computational transitions, supply-chain capacities, stakeholder involvement, and policy frameworks; [2] certain developments in regulatory and ethical debates during the first half of the twenty-first century nevertheless offer a wider perspective on the whole-genome issue; [33] genomic conditions would, even in the absence of expanded implementation, enter standard care routes nonetheless; and [4] comprehensive delivery platforms equip empirical, real-world assessments, collected prioritization, and wider stakeholder engagement.

Governance, Privacy, and Data Stewardship

Full genomic data availability raises important questions regarding stewardship, governance, and privacy at multiple levels [3]. Data ownership, access, sharing, and governance differ at levels including subnational jurisdictions, population-based screening initiatives, health system organizations, and research institutions. Genetic and genomic data have a distinctive life cycle, with periods of generation, primary and secondary use, retention, and erasure [34]. The extent of individual rights to genomic information, including data that reveal

traits, risks, ancestry, and family relationships, remains actively debated. Where the level of data sharing is exceptionally high, questions arise regarding stewardship of individual rights at a collective level, balancing respect for individuals against collective benefit [1]. Different governance arrangements are needed to align with the wide-ranging modes and objectives of potential genomic data use [4]. Stipulating acceptable data stewardship procedures can help reassure parents about retention periods and erasure protocols that maintain the privacy of genomic information once it is no longer considered relevant for screening. Periods of genetic data retention can vary, but most jurisdictions opt for longer eras than are applied for traditional newborn screening [4]. The pledges to ensure privacy and responsible handling of information must match the significance of the data being shared, mechanisms to assure compliance must be clear, and methods for addressing a breach must be defined. Data retention policies after newborn screening need to be informed by ethical and privacy research concerning both genomic and health information [35].

International Experiences and Comparative Analysis

Newborn screening (NBS) has evolved significantly since its introduction in the 1960s, evolving from a focus on single diseases to screening for many conditions simultaneously. Genome sequencing (GS) and whole-genome sequencing (WGS) have emerged as powerful new tools capable of identifying many conditions in a comprehensive fashion [36]. As of mid-2023, several countries are piloting WGS-based approaches to NBS. These developments provide an opportunity to learn about the integration of WGS into NBS in different contexts, and gain insights into the challenges, implementation strategies, and policies adopted in diverse regulatory environments and health systems [2]. A better international understanding of the potential benefits and associated challenges of WGS-based NBS can facilitate evidence-informed decisions [37]. Early pilots of WGS-based NBS are underway in France (NBS4C project), Norway, Japan, and Singapore. The selection of conditions for sequencing varies, but Neurodevelopmental Disorder is included in all programs. Deadly Congenital Metabolic Disorders are included in all but the French pilot. Reports on pilot implementation from several countries highlight pathways, lessons, and recommendations that could inform decisions elsewhere [1].

Research Agenda and Priority Topics

Significant evidence gaps hinder progress toward an informed decision on whether to expand newborn screening with whole-genome sequencing (WGS) [38]. To address this knowledge deficit, a targeted research agenda identifies high-impact, practical studies that can close specific evidence gaps, improve decision-making, and advance policy and implementation efforts [4]. Filling these priority evidence gaps requires appropriate study designs, including long-term monitoring of technology pilots and real-world utility assessments based on observational data. Additional near-term priorities include equity analyses and operational evaluations that assess progress in addressing disparities in access, infrastructure, and support capabilities and the effectiveness of related outreach efforts [39]. Accompanying consent-related research focuses on best practices for enrolment, consent, and communication materials that align with the ethical obligation to support informed parental decision-making while mitigating the material risk of misuse, misinterpretation, and overmedicalization through effective risk articulation [2]. Strategic interventions to enhance the available evidence base include harmonizing shared data infrastructures and establishing collaboration networks to coordinate initiatives across multiple jurisdictions. Broader tractability can be pursued through leveraging policy-relevant funding mechanisms and aligning with existing national and regional collaboration initiatives, such as national genomics strategies, biobanking projects, and frameworks for responsible GBS deployment [40-45].

CONCLUSION

Whole-genome sequencing has the potential to redefine newborn screening by enabling comprehensive early detection of genetic conditions and supporting more proactive, personalized healthcare across the lifespan. Nevertheless, its integration into routine screening programs must be approached cautiously. The technology introduces ethical, logistical, economic, and interpretive challenges that extend beyond those associated with traditional screening panels, including managing uncertain findings, ensuring informed parental consent, safeguarding genomic data, and preventing inequitable access to benefits. Effective public-health decision-making should therefore rely on transparent evaluation frameworks, phased pilot programs, and continuous monitoring of clinical utility, cost-effectiveness, and social impact. Strengthening laboratory infrastructure, developing standardized interpretation protocols, and investing in stakeholder communication will be critical to successful implementation. Future research should prioritize long-term outcome studies, equity-focused analyses, and governance models that balance individual rights with collective health benefits.

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