Governance of state-mandated newborn screening programs: Engaging communities to inform the use of novel genome sequencing technologies.

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Nearly every child born in the United States is screened for several lethal or debilitating inherited disorders at birth through mandatory state-run newborn screening programs (NBS). The clear benefit and inclusivity has justified a public health approach built on universal screening, even in the absence of full transparency and active parental participation.

Recently, NBS programs have come under intense public scrutiny due to concerns about research use of newborn blood spots maintained by the states. While the NBS program holds great promise for improving the health of at-risk newborns as well as facilitating population-based research with leftover blood samples, public trust is at risk. Few people are fully aware of clinical NBS programs; fewer still understand when and how samples are banked for research purposes. This lack of awareness has led to escalating civic unease, prompting lawsuits in Minnesota and Texas, resulting in court-mandated destruction of over 5 million bloodspots.

While research use is one issue facing NBS programs, rapid technological growth in genomics is poised to further expand capabilities, calling into question the scope and breadth of NBS. The President's Council on Bioethics expressed concern about the rapid, technologically driven, increase in the number of conditions screened for since the introduction of tandem mass spectrometry. The advent of low-cost sequencing technologies capable of quickly and accurately screening a newborn's entire genome will further exacerbate these issues. Should NBS remain a public health approach, focused on population health? Or will it become the first step in personalized medicine for each newborn, a clinical test that brings with it myriad ethical concerns, including: the scope of results to return, unintended findings unrelated to treatable disorders that manifest in childhood, and the considerable uncertainty of genomic data?

Currently, we know little about the public's attitude towards whole genome analysis (WGA) in NBS. In the face of growing public concern and intense pressure for expansion based on technological innovation, the need for authentic, community guidance of NBS programs has reached a critical point. The UCSF/Kaiser Permanente "Center of Excellence" in translational genomics and ethics hosts several projects, including 1) a focus group study examining stakeholders' (pregnant women, clinicians, and parents of children with a genetic disorder). perspectives on WGA in NBS and 2) a two-part deliberative community engagement (DCE) with diverse California citizens, focused on policy challenges and governance of the California NBS program. DCE is rooted in a deliberative democracy framework and offers one approach to obtaining informed community feedback and assuring civic guidance.

We will present results from our focus groups and provide an overview of the theoretical foundations of our DCE as well as the design, and potential impact on governance and policy decision-making within the NBS program. Our approach expands beyond examining a single policy decision and instead focuses on multiple current and future issues. Only by examining the

issues holistically are recommendations likely to be adaptive to the changing landscape of genomic research and technological innovation.

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