



Arsht Research on Ethics and Community Grant

Assessing Parental Perspectives Regarding Genome Sequencing In Newborn Screening

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Abstract

Often coined the nation's most successful public health initiative, newborn screening (NBS) is also the largest genetic screening program in the United States. Controversial since inception, NBS consistently maintains ethical acceptability because of the undeniable benefits of reducing neonatal morbidity and mortality. With falling costs and increasing efficiency, genome sequencing (GS) technology is heralded to be the next major disruptor to the NBS program. With pilot projects incorporating GS into NBS, legislators may soon be faced with incorporating this technology into their state's NBS program. Ethical and practical concerns regarding GS stem from the complexity and volume of information generated, which makes balancing the risks and benefits significantly more subjective than with standard NBS. For example, should risk for adult onset conditions be reported, and, if so, what are the criteria? Will data, and hence benefits, be limited to the newborn period or be available throughout the life span? If data is available for a lifetime, who will have custody of the data and what protections will be taken to ensure privacy and prevent discrimination? These are just some of many questions that need to be answered. As policies emerge, the perspectives of all parents should be considered. Although studies have investigated the public perspectives on incorporating GS into NBS, no study has assessed parents of children with positive NBS screens and parents of children with conditions that could be screened by incorporating GS. Research shows parents of children with serious medical conditions have presented alternative views on other NBS related issues. The purpose of this study is to assess the awareness, attitudes, and concerns among these unique parental populations to aid in providing perspectives from all families. This study will use validated survey questions to assess these targeted parental populations with respect to awareness,

attitudes, and concerns on if and how to best incorporate GS into newborn screening. The four target populations will be parents of children with a (i) false positive NBS result, (ii) true positive NBS result, (iii) non-metabolic, Mendelian genetic condition that would be screened by incorporating GS, and (iv) negative NBS result and no known genetic conditions to represent the majority of parents. In addition, this study will measure how education regarding NBS and GS influences awareness and attitudes. Data will be analyzed using descriptive and comparative statistics and disseminated through submission to a conference presentation, and ultimately a peer-reviewed journal.

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