Sequenziamento genomico neonatale: quali interessi considerare nella definizione del pannello di geni?

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Neonatal Genomic Sequencing: which Interests to Consider in Defining the Gene Panel?

Abstract: Newborn screening is a publicly funded test aimed at identifying genetic diseases in healthy infants where early diagnosis can lead to timely and effective clinical intervention. Recently, there has been growing interest in applying genomic sequencing, in particular Whole Genome Sequencing and Whole Exome Sequencing, to this screening, significantly increasing the number of identifiable conditions. Considering the promises of this approach and the specificity of genomic data, some have suggested that newborn sequencing could serve the interests of not only screened newborns but also their parents, relatives, and even society. Determining which interests should legitimately be considered in genomic newborn sequencing is crucial for defining which genes to include in the gene panel of neonatal genomic screening. Stemming from a bioethical perspective, this paper discusses which interests should be considered in newborn sequencing, what the implications are for gene panel definition, and what conflicts between interests might emerge. Specifically, the newborn clinical interest, the reproductive interest, the family clinical interest, personal utility, and social utility will be considered.

Keywords: Newborn screening, Genomic sequencing, Gene panel definition, Bioethics.

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