Saving babies? The consequences of newborn genetic screening (Review)

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Medical genetics is currently a routine part of health policies, alongside and in relation to other forms of knowledge. Today, more than 4 million newborns are screened every year in the United States for more than 50 genetic conditions, for the most part metabolic. Some of these conditions are exceedingly rare and little information is available about their etiology, natural history or clinical relevance. In this context, *Saving Babies? The Consequences of Newborn Genetic Screening* is based on ethnographic research in a centre specializing in follow-up care for children with metabolic diseases in California. Taking a pragmatist approach, it focuses more specifically on examining the intended and unintended practical consequences and daily effects of expanded newborn screening for clinicians and for families, beyond the official discourse of prevention and life saving.

The authors begin by retracing the history of the expansion of newborn screening in the United States, showing in particular the role played by patient advocacy groups in this process. They then go on to revisit the situation of screened children who are not completely normal, biologically speaking, but not completely sick either, and who therefore find themselves in the limbo of diagnostic uncertainty, when the pathological nature of the anomaly detected remains uncertain. The clinicians' cautious attitude and the parents'

persistent anxiety show how, in the lives of the families concerned, this uncertainty takes on the shape of a real illness. More generally, and this will come as no surprise to sociologists of health, the results of these screening programmes alter the definition – in other words the ontological and epistemological status – of conditions that were previously detected through symptoms. The issue of normalization or difference based on objectivizing developmental and physical criteria is therefore raised for both clinicians and, in a more pressing fashion, parents: is the baby normal? (in cases of uncertainty) or how normal will the baby be? (in serious cases). Furthermore, by resolving anomalies and gradually changing the definition of the disease, geneticists avoid the onset of a crisis of credibility about the feasibility of newborn screening. In other words, they seek to close the gap between the promise of technologies and the realities of their implementation.

This brings us to the last part of the book, which looks at the limitations of prevention and the effects of these screening programmes in terms of public health. The authors underline, and rightly so in my opinion, the fact that screening is carried out in the United States without any request for parental consent (with a few exceptions) and that this reflects an extraordinary belief in the power of screening to save children's lives. However, regarding children with trajectories of serious illnesses, they ask: "What does it mean to say that patients who face significant developmental delays, frequent hospitalizations and serious risks of mortality have been 'saved' by early intervention?" Overall, it seems that screening can only be linked to more or less extensive beneficial effects for 10 of the 50 conditions concerned. Nonetheless, the authors specify that screening allows diagnostic odysseys to be avoided in cases where children are symptomatic and that parents are strongly supportive of the prevention aims of screening. Geneticists, for their part, adopt a reflexive stance and admit the lack of consensus regarding whether or not screening affords benefits to the most seriously affected patients.

The book concludes cautiously that it is too early to draw any conclusions about the future of newborn screening.

The work includes a particularly interesting section, especially for readers unfamiliar with the health care system in the United States, that focuses on the structural constraints and interpersonal dynamics that come to bear upon the follow-up of screened children. The authors argue that the social inequalities in the US can impede the opportunity to save lives despite the universal nature of newborn screening, something that is rare in the country. Problems linked to lack of health insurance cover, to which must be added the linguistic difficulties of non-English speakers and problems of family organization, accentuate the gap between the generalized funded of screening and the partial funding of follow-up care and therefore highlight what the authors – and they are completely right of course – call the myopic take on what is meant by public health genetics.

Overall, the book is clear, informative and easy to read. Description of the study results and bibliographical analysis are combined seamlessly and the authors provide extremely relevant thoughts in terms of public health. It is perhaps regrettable that certain aspects are not analyzed further, such as the anthropological meaning of these screening programmes, the role of early intervention or the light that can be shed on the vast area of contemporary anxiety that health has come to represent. Similarly, certain possible directions for reflection could perhaps warrant further substantiation, particularly concerning the moral dimension of normalization and the child's future, or the role of politics in decision-making about screening. Beyond this, it would be interesting to explore what exactly these screening programmes can tell us about the moral and political landscapes of wealthy countries and about notions that are taken to be self-evident (the power of screening, etc.). Nevertheless,

this book offers an excellent introduction for readers with an interest in newborn screening and the sociological transformations of biomedicine. And without a doubt, it encourages greater dialogue between research on these questions in the United States and in Europe.

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