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COMMENT

Taking seriousness seriously in genomic health

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The 'seriousness' of a genetic condition has important implications for genetic health care. Indeed, this notion is both widely used in clinical practice and frequently referred to in policy. The seriousness (or severity) of a genetic condition is most often raised in a reproductive setting (both screening and diagnosis), although it may be relevant to other forms of clinical care and population screening, as well as debates over emerging technologies such as genome editing.

Yet for a concept so central to genomic health care and especially to prenatal genomics, it is surprising that a detailed account of 'seriousness' has received so little attention. It has been observed that determining whether a condition is deemed to be 'serious' has significant implications [1] and that the concept is complex and uncertain in many ways [2], but it has not really been pinned down. This is especially true when it comes to research that digs into what 'serious' might mean at a conceptual or philosophical level, or which draws on the lived experience of patients.

In this issue, Boardman and Clark provide a welcome contribution to addressing this gap [3]. Their introduction shows just how symptom-focused and data-driven the existing definitions of 'serious' are, and how they tend to reflect the views of health professionals [4, 5], or healthy people who have had testing [6]. This overlooks the importance of hearing from people who can tell us about what it is actually like to live with a genetic condition.

Boardman and Clark address these gaps by bringing together interview and survey data obtained from people with a range of 'serious' genetic conditions. Most of their participants report good quality of life, even though they also experience suffering (another concept that needs further unpacking) and may not always be in good health. Interestingly, those with lifelong conditions view it as inherent to their identity—it cannot be separated from who they are. Some participants who might appear, to an outsider, to live with a serious disability report good health. They distinguish their disability from their health.

Genetic conditions with onset later in life have tended to be more controversial to prevent prenatally. This is because emphasis is usually placed on years lived without symptoms. However, participants in Boardman and Clark's study who had SMA4, the latest-onset and mildest form of this disease, evaluated their health as being the worst. They combined their disability with their health rather than their identity. This raises interesting questions as to whether our assumptions about prenatal prevention of late-onset genetic conditions are appropriate—but such a question must be left to a different commentary.

We wish to focus here on the idea that we need to do more to take 'seriousness' seriously. Any notion of 'serious' needs a variety of approaches, including ethical and conceptual analysis, the viewpoints of those who live with conditions and commentary from health care professionals. Descriptions should not be limited to medical indicators, nor to creating buckets to categorise these, but should incorporate social, cultural and environmental factors too.

The concept of 'serious' also needs to reflect the variety of ways (positive, neutral and detrimental) that a genetic condition can impact a person's life. The fact that the experience of a condition will likely change or vary over time (including as treatments are developed) and between people is also relevant, as is Boardman and Clark's observation that positive adaptation—by incorporating one's condition into one's identity—regularly occurs. Crucially, if we are to use 'serious' well, we should strive not to disembody the concept from the person: a genetic condition cannot exist without a person to have it. We need to hear and account for these factors.

Taking seriousness seriously will also involve thinking about when and how it should be used. Should it, for example, be used in a universal way, with 'one size fits all'? Or should it be applied more contextually? Or both? It may be that different notions of 'serious' are needed for different uses, such as designing a population policy as compared to supporting a couple in clinical practice.

Ultimately, this may mean that taking seriousness seriously becomes a question of procedure as well as substance. The substance of severity is subjective, contextual, uncertain and changeable. Rather than strive to settle on a single definition, we could aim to develop a framework to operationalise particular meanings of 'serious' in different contexts. This could assist in a range of activities, from designing screening panels to supporting individuals in clinic. The framework could be representative of diversity and transparent in application. It could incorporate ideas such as adaptation and the importance of social and environmental support. It could draw attention to the importance of health communication and decision support.

If we do not take seriousness seriously, then the concept will lose its meaning and become lost in a series of checklists, taxonomies and algorithms. These technically robust approaches will fail to genuinely improve our understandings of the lived experiences of serious genetic conditions, or even more importantly, help a person or couple faced with an important and life-changing decision.

The answer to the question 'what is a serious condition?'—and even the processes we should follow to help individuals, couples, health professionals and policy-makers arrive at an answer to it—remain elusive. While their research does not quite answer the question they pose, Boardman and Clark certainly point us to some key aspects concerning that important question. There is now a need for further work, to consider in greater detail the role

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severity plays in genomic health care, what this concept actually means in varying contexts (drawing on a wide range of perspectives) and how we can best use it to provide equitable and ethically defensible care to individuals and populations.

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COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

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