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Genetic conditions and the Social and Human Cciences of health: contributions to the debate

In the last decades publications around "rare diseases", within the Social and Human Sciences, have been gradually increasing. However, few works are focused on those people who are directly experiencing and interpreting their own condition. Care, which is centered on the family's actions or institutionalized in civil associations, tends to see the diagnosed as a needy person in need to help from others. The consequences of this perspective obscure the subjective, intersubjective and organizational dimension of those who daily live with the symptoms of the condition and resist the imperative of normality[1]. The result is the (re) production of a discourse impregnated by the charitable vision and the continuous suffering, which leaves the person with the diagnosis of disease incapable of fighting for their rights, which is put by necesity into the voice of another.

On the other hand, as we look at the field of rare diseases, we see that their linkage to genetic "diseases" encourages parents to search for a cure by repairing the genetic chain- a fact stimulated the dissemination of diagnostic technologies. This search generates a discrepancy within the field of rare diseases, where the priority of action ends up focusing on the 3% of the genetic conditions that possess drug treatment, covering/obscuring/overshadowing the daily needs of those who, even with diagnosis and symptoms, see themselves as active rather than ill people. This difference produces gaps that will only be filled from a multi and interdisciplinary view attentive to the relationship between the biomedical concept and the sociocultural conception of disease and their contexts. In this sense, the "disease" is determined by the gene that instantiates the 'proper' understanding of the health-disease process, diagnosis, extended and equitable care, as well as the historical, political and social constructions involved in these elements[2].

In this scenario, the present thematic number has contributions of different researchers on genetic conditions from a perspective of disability studies and/or qualitative research. The authors come from different areas of research, from both national institutions and from abroad. Their contributions are theoretical, methodological and empirical texts on the subject. The works are based on the perspective of the person diagnosed with a genetic condition and they intend to reflect on the understanding of "disease", its impact and the effects on the experiences and the organization of life itself. Also, they consider the perspective of those who experience and act (individually or collectively) in the face of there percussions of the "illness", whether on a personal, familial, professional, organizational, associative or institutional level, with different care modes.

Hence, this work may contribute to broadening debates about genetic conditions and to add to the rights and recognition of the person, not the medicine, as the epicenter of action in the field of "rare diseases".

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