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Rare diseases: who pays which bill?

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The debate on rare diseases involving public health experts, politicians, and economists often focuses on the budget impact from the public health systems' and private health plans' incorporation of so-called orphan drugs. Since such drugs are limited to a reduced target public and require high investment in technological research for their development, they are scarcely profitable and bear little or no interest to pharmaceutical companies. This is certainly an important issue, but there are other dimensions to the economic and social impact of rare diseases that require attention.

The article by Pinto et al. [1], in this edition of CSP, makes important headway in this debate, tracing the profile of children and adolescents with three of these rare conditions, plus their caregivers, and more importantly, evidencing the financial and social impact of these diseases on the family. What we find are families that not only cope with the suffering of a sick child, but become indebted due to loss of income and increased expenditures. The loss of income is due to the inability to continue working, and the increased expenditures are due to the high costs of transportation (often involving long trips from home to hospital) and the need to hire caregivers for the sick child or for the family's other children, among others. It is not surprising to find that the principal burden falls on the mothers. These losses and their consequences are not resolved by government financial incentives alone.

Despite the particular context in which the data were collected (a pediatric referral hospital in the city of Rio de Janeiro) and the selection of a clientele mostly belonging to a representative patients' association, the authors expand the discussion with examples of the same phenomenon related to other diseases in different settings and populations.

The authors' initiative of estimating the costs of the disease from the patient's and family's perspective is praiseworthy. Economic assessments in health frequently overlook this aspect, since such studies are labor-intensive and costly and require informed consent from the interviewees, rather focusing exclusively on the funder's perspective. It was beyond the study's scope to assess quality of life for these children and their caregivers. However, with the data presented, it is not difficult to imagine the impact of the disease on this item, although not assessed in the article.

Some 250 new rare diseases are described every year due to the refinement of knowledge on pathophysiology and genomics. Thus, rare diseases are not all that rare, especially considering the total number of persons affected by these various diseases. Technological development in recent decades has created the possibility of treatment for many rare diseases. Associations of patients living with these diseases and health activists have propelled research for the development of new drugs. However, access to the drug is not enough, as clearly shown by Pinto et al. It is necessary to guarantee that the best care is offered. This requires more than monetary incentives. It is necessary to support and enable the caregivers' intense journey of dedication.

The United Nations' 2030 Agenda for Sustainable Development [2] proposes "to leave no one behind". Gender, social, racial, and religious inequities must be overcome. The article by Pinto et al. gives us food for thought on many of these inequities. Only a debate involving academic institutions, decision-makers, regulatory agencies, the pharmaceutical industry, and especially civil society through individual empowerment and advocacy associations [3,4,5] can answer the remaining questions: Who in fact is the orphan – the drug, the disease, or the affected family? Who should pay the bill? Which costs should be presented – in addition to the financial costs?

Additional information

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