Rare Diseases and Difficulty Accessing Orphan Drugs: A Topic That Needs Discussion

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The difficulty in accessing orphan drugs by rare disease patients has raised questions in our clinical practice regarding our relationship with our patients and with the national authority.

These problems, which are widely discussed outside of Portugal,[1-5] will be more difficult for us to manage due to financial difficulties.

The number of rare diseases (diseases affecting fewer than 5 in 10,000 individuals, according to the definition adopted in the European Union) has been steadily increasing. Currently, there are more than 7,000 and about two-thirds have genetic causes. Most orphan drugs, which are used for the treatment of these diseases, are intended for the pediatric population.[6]

Firstly, it is important to emphasize that the scientific knowledge generated by the demand for orphan drugs has an important intrinsic value and is independent of any consideration of individual cases. In fact, a therapeutic innovation has sometimes reached far beyond the results obtained in the patients in which it was used, allowing the same therapy to be applied to other diseases.

Patient associations have been playing a decisive role in the growth of orphan drug research. Likewise, the Orphan Drug Act of 1983 in the United States and the Orphan Regulation of 1999 in the European Union promoted this research.[3] Incentives, such as market exclusivity, led to what was called the "gold rush" of pharmaceutical companies. The price of orphan drugs has been high from the beginning, as they are intended for a smaller target population and the research costs must be covered.

Questions about the price of these medicines quickly arose to defend the need to safeguard the interests of non-rare disease patients, as funders through their taxes, especially in countries where care is provided through a public health system.

Some of the raised questions are:

- Should individuals who are severely affected by a given disease have a proportionately greater allocation of resources?
- Does this not constitute a limitation to the health care of others?
- Should people who are in current need of expensive medical care for a serious and life-threatening illness be deprived of such care in the name of the present or future well-being of others?

The concept of need was articulated with the concept of the ability to benefit. Is unusually expensive treatment appropriate for someone seriously ill with limited ability to benefit? By definition, the need does not exist if there is no capacity to benefit.

This utilitarian interpretation of ethics considers that moral rules express a set of commitments through which we reconcile our personal interest with the interest of others in order to obtain the best possible result for as many people as possible.

The deontological approach to ethics, on the other hand, considers that an action for the benefit of another, even if it may bring us a personal disadvantage will be morally correct if arising from a duty and does not need any additional justification. The Code of Medical Deontology reflects this deontological view of ethics.[4-5]

This code affirms the principles of autonomy and beneficence.

The principle of autonomy presupposes respect for the opinions of the patient about how their diagnosis and treatment process should be guided. The current reality of patient "emancipation" in relation to the old paternalistic model of exercising medicine is rather evident in the field of rare diseases. Patients and their families play an increasingly larger role in therapeutic decisions.

If a treatment is available and the patient is informed and agrees, we must arrange for it to be administered in accordance with the principle of beneficence.

We conclude that, when there is a treatment that can bring significant benefits, and in general greater benefits with earlier administration, the delay or non-administration not only increases the cost/benefit ratio but is also ethically unjustifiable.

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It is not a question of defending the individual interests of our patients as opposed to the interests of the majority, but rather of upholding a general principle applicable to all in similar circumstances. In our view, this principle is imperative and we will globally – society in general, market authorization holders, policymakers, and health authorities – have to assume the search for a solution to the question of the price and accessibility of orphan medicines.

Keywords: Rare Diseases/therapy; Orphan Drug Production

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