

Rare diseases: Should they be included into Plan AUGE?

This case has been developed by Marcelo Muñoz (Office of Bioethics, Ministry of Health of Chile¹); with the support of PAHO's Regional Program on Bioethics; based on Bastías G, et al. "Policy Brief: Financing Options for the treatment of rare diseases in Chile." (EVIPNet-Chile, June 30, 2011) and the subsequent "Report on Policy Dialogue: Financing Options for Rare Diseases in Chile" (EVIPNet-Chile, October 2011) authored by Pedro Crocco and Cynthia Arguello.²

Background

A rare disease is a disease that occurs infrequently in the general population. In order to be considered "rare", each specific disease cannot affect more than a limited number of people out of the whole population. That number varies. In Europe it is defined as 5 in 10,000 inhabitants. In Chile it is defined as 0.18 in 10,000 citizens.³ This figure can also be expressed as 18 patients with rare diseases out of one million citizens. In a total population of 17 million citizens this could mean as many as 306 individuals for each rare disease. The number of patients with rare disease varies considerably from disease to disease. Some diseases affect only one in 100,000 people, and there are diseases that are even rarer and only affect a couple of dozen patients.

Most rare diseases are genetic, and many of them appear early in life. It is estimated that about a third of patients with rare diseases will die before reaching the age of five. Many rare diseases can only be treated with so-called "orphan drugs." Orphan drugs are pharmaceutical agents developed specifically to treat rare diseases. Ultra-orphan drugs are medicines used to treat exceptionally rare diseases. Due to the low number of patients, orphan drugs are as a rule non-profitable for pharmaceutical companies. These drugs are very expensive on a per-patient basis, so they are as a rule not cost-effective. Moreover, the cost of developing orphan drugs makes pharmaceutical companies less willing to invest in the research and development of these drugs, so many diseases do not have adequate treatments.

Case description

In Chile, there is no specific public health policy to deal with rare diseases, or a systematic, formal mechanism to determine funding of orphan drugs. Due to the progressive development of health technologies, the diagnosis of rare diseases is becoming more common in Chile. The cost of treatment for rare diseases is financially catastrophic because of the high cost of orphan drugs, and the fact that a significant part

¹ Disclaimer: This case study is solely an educational exercise and does not necessarily reflect the position of the Ministry of Health of Chile on this issue.

² EVIPNet is an initiative promoted by WHO and led in the Americas by Evelina Chapman (PAHO).

³ According to the Bill on Rare Diseases.

of the cost of drugs is paid in Chile out-of-pocket. These high costs hinder access to treatment or leads affected families to poverty or further impoverishes them. The vulnerability caused by diseases that are often life-threatening and severely debilitating, gets exacerbated by the difficulties –and sometimes inability— to access treatment.

Chile’s economic and social development in the last 30 years has walked hand-in-hand with the development of its democratic institutions. These developments have favored the conditions for citizens to voice their demands for the protection and promotion of their rights. Associations of patients with rare diseases and their families have become very vocal in the media: They raise awareness about the situation of patients with rare diseases in Chile and advocate for coverage for treatment. The right to healthcare has been invoked to put additional pressure on the government about coverage of rare diseases in many cases that are currently being reviewed in courts.

Plan AUGE (“Existing Guarantees and Universal Access”) currently includes 69 prioritized pathologies or health conditions for which treatment and coverage are guaranteed by law. The law specifies that conditions should be prioritized taking into account the health situation of the population, the effectiveness of the interventions, their contribution to the extension or quality of life and, if possible, cost-effectiveness. Some rare diseases (based on the definition of Orphanet ⁴) are included among these pathologies, such as cleft lip and palate, Tetralogy of Fallot and other congenital cardiopathies, myelomeningocele (most common type of spina bifida), some forms of cancers in persons younger than 15 years of age, hemophilia A and B, cystic fibrosis.

In addition to the AUGE Plan, two other mechanisms have been used to finance treatment of rare diseases. First, the system of public health insurance (FONASA), through its Special Program for High Cost Drugs, finances very high-cost orphan drugs to treat a few specific conditions such as dystonia, Guillain Barre, severe dwarfism, and Gaucher disease, and botulism. Due to limits in budget, patient coverage is occasionally limited. Moreover, financial projections suggest that these funding mechanisms are not sustainable. Second, “Extraordinary Aid Funds” have also been used to finance orphan drugs and thus alleviate patients’ demands.

Discussion

Influence of interest groups and legal action seem to have had bearing on the government’s decision to fund treatment of certain rare diseases. Yet it seems that a formal national policy on rare diseases, that specifies priority-setting criteria, is necessary. Should rare diseases that can be treated with orphan drugs be included as prioritized conditions in AUGE?

⁴ Orphanet. The portal for rare diseases and orphan drugs. <http://www.orpha.net/consor/cgi-bin/index.php>. Orphanet uses the European definition (prevalence of 500 in one million inhabitants), so it includes diseases that might not count as rare diseases according to the definition proposed in Chile (18 in a million) and/or according to the prevalence of the disease in Chile.

Since treatment of rare diseases is extremely expensive, it is obvious that funding such treatment would imply restricting funding for treatment of people with more common diseases, or for other public health interventions. What trade-offs are acceptable? How should the goal to maximize (health) benefits for society be balanced against the moral imperative to save the life of these individuals that were extremely unfortunate to be born with a rare disease?

What role does the moral and/or legal right to health care play in this discussion? Which obligations do health authorities have in regards to access, quality and financial protection of health services for patients with rare diseases and their families? If only some conditions can be funded, how should those conditions be determined?

Finally, how should the government ensure fairness in resource allocation given the differential power of different patient groups? What is the proper role for advocacy groups in such decision-making? Is there one?