Review of: "The Plight of Rare Diseases in Southern Africa: Health and Social Services Policy Recommendations"

Laura Magazzini[1]

1 Sant'Anna School of Advanced Studies

Potential competing interests: No potential competing interests to declare.

The Study offers an interesting perspective on rare diseases (RDs) in Southern Africa. Despite the adjective “rare”, these diseases affect globally a significant number of people. The Study reports 3.6 million people in South Africa (one in 15 people).

Around the world, RDs are increasingly perceived as a public health issue, and specific provision for fostering research on available therapies have been enacted in many countries. Available studies mostly present data and analysis related to USA and Europe. In these countries, healthcare policies directed to providing incentives for RDs are thought to be effective in fostering research targeted to those pathologies, and the number of available treatments is increasing over time (Yin, 2008; Braun *et al.*, 2010; Westermark *et al.*, 2011).

The Study proposes a multi-stakeholder approach that considers the different layers of the healthcare system from research, to caregivers and patients. This unified approach is aimed at improving the health and livelihood of people living with these (understudied) pathologies, making proposals directed to research, prevention, better diagnosis and treatment of RDs, as well as capacity building of healthcare workers. The proposal also makes suggestions to enhance the provision of health and social services including shared cost-bearing models and special health insurance coverage. The authors claim that in Sub-Saharan Africa (SSA) there is a deficit of information on RDs including their type and the number of affected individuals.

The proposal seems to be inspired by the policies designed in other countries. From these, some lessons may be learned that could be helpful in devising incentives and strategies in the context of SSA countries.

First, it is certainly very important to map the RDs that are mostly prevalent in SSA regions and characterizing their genetic characteristics and prevalence. Collaboration between different states and stakeholders can play an important role to improve management of RDs.

RDs are heterogenous in terms of prevalence, with some diseases affecting less than 1 individual in million and some that instead are closer to the threshold selected for being eligible to RDs incentives. Available studies show that orphan drug legislation has been effective in spurring research directed to RDs, with larger efforts directed towards the less rare among RDs (i.e., those diseases with prevalence closer to the threshold – see Yin, 1998, Gamba *et al.*, 2022).

In this respect, Gamba *et al.* (2022) show that push mechanisms (e.g. tax credit) and pull mechanisms (e.g., market

exclusivity) have a differential impact on RD research as a function of the disease prevalence. In particular, the effect of pull mechanisms seem to be stronger at higher prevalence, whereas the direction of the effect is ambiguos when the effect of push mechanisms is analyzed as a function of prevalence. Empirically, Gamba *et al.* (2022) find that, within the class of orphan diseases, the increase in R&D efforts is concentrated mainly on less rare diseases. In the European context, this tendency may have been exacerbated because European legislation almost exclusively relies on pull incentives.

The results have implications for policy. If reducing the number of diseases with no therapeutic option available is to be set as a priority, a proper balance of incentives between pull and push incentives should be devised. A more radical proposal is based on the idea of replacing the existing approach based on a single threshold with prevalence-dependent incentives.

References

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