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

Erik Tambuyzer

Potential competing interests: No potential competing interests to declare.

The article is a welcome paper on rare diseases in Southern Africa. It is well written and understandable, and testifies that the authors are knowledgable in the field. Hereby, I want to offer a few comments and possible edits that may be useful for the authors to consider. Please consider my comments hereunder as constructive with the aim to help.

- General
    - It is a great first step that a coalition such as this one has been set up.
    - It would be good to mention in the paper also what is specific in the field of rare diseases for the region compared to other geographical areas which some examples.
    - Astract: the proposal for a unified approach accross the area is to be applauded, as only a large population base, managed under common rules, and international collaboration will create enough knowledge and expertise to help alleviate (some of) the problems for patients suffering from rare diseases. International collaboration could already start in the area of diagnosis since no country will be able to diagnose all thousands of possible rare diseases. In this context, the creation of centers of excellence is important, but will nations agree that they will all not have centers of excellence for all rare diseases? And if so, will patients of different nations be able to travel to the center of excellence for their disease in another country?
    - Prevention should be mentioned in addition to diagnosis, management and treatment of rare diseases throughout the article.
    - Graphical abstract: “cost bearing and universal health insurance coverage”: is there a political will to collaborate on this issue between nations in the region, or if not yet existing, should it not be mentioned how such political collaboration will be achieved?
    - Rare disease database: It will be very important to create a register of rare diseases for the area. Such register will be useful for policy makers, but also for developers of orphan drugs. I would recommend to cover this point in more detail in the article especially how it can be set up and maintained.
    - Situational analysis: it is very important as mentioned that the situation of rare disease patients is discussed and managed in a multi-stakeholder approach, which has also been proven to work in the European Union and elsewhere.
    - in addition to the issues of which rare disease patients suffer, it would be good to mention, where possible and appropriate, and was done in some cases already; proposals of what can be done as first steps to alleviate some of these issues.

  - It may be important to also strive for the inclusion of the Democratic Republic of the Congo in the coaliton, since it is one of the largest countries in the area.
- Detail
  - Background, Adoption of a common definition: There is no existing definition of a "rare disease" or “rare diseases” in the USA of in the European Union. However, what exists is a definition of a product (orphan drug, orphan medicinal product/medicine) that is used to treat a rare disease. The definition for orphan drug in the USA is a product to treat a rare disease with a prevalence of less than 200,000 in the US population. The definition of an orphan medicinal product in the European Union is for a product to treat a rare disease with a prevalence of less than 5/10,000 in the EU population, for which there is no approved treatment in the EU or, if such treatment exists, that is of significant benefit to patients. In spite of the fact that such common definition does not yet exist, does not mean that it cannot be helpful, and can be strived for in the area. Please correct as necessary in the paper.
  - Background: In relation to that, the term “orphan diseases” does not exist, and is confusing as it may lead to believe that orphans have specific diseases which (of course) is not the case.. The term orphan diseases is derived from the term orphan drugs or orphan medicines/medicinal products. Rather the authors should use “rare diseases”.in the article.
  - Situational analysis: please note that MS (multiple sclerosis) in most regions will not be considered rare as the prevalence is mostly too high.