

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

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Potential competing interests: No potential competing interests to declare.

Review report

The study is of interest. The overall structure is well designed. Below are few suggestions and recommendations to the authors.

Abstract:

1. "The situation is worse in sub-Saharan Africa, where the public health infrastructure is weak" to change it with "The situation is worse in sub-Saharan Africa, where the healthcare system and infrastructure are weak".
2. "Leaving a high health and cost burden on patients and caretakers": This sentence needs to be edited.

Background:

1. The first sentence needs a reference "Rare diseases, ... the population".
2. "This deficiency has a substantialadequate healthcare": This statement needs a reference of a prior study that justified this fact.

Situational analysis of rare diseases in the SADC region

1. The title of this paragraph could be modified into "Current situation of rare diseases in the SADC region".
2. From "On the other hand..... proportion of the population": References need to be added to support authors statements and opinions.
3. "Khosla and Valdes (2018) found no published literature about existing or planned national plans, laws..." to be edited into "Khosla and Valdes (2018) found no published literature about existing or national plans, laws...".
4. (AU, n.d.) The year needs to be added.
5. "This can be observed in the case of the European Union (EU).... common goals": Reference about facilitated cases in European countries with good outcomes to be cited.
6. Throughout this paragraph, the authors mentioned mostly URT and South Africa. What is the current situation in the other SADC countries? Why no other SADC country had been mentioned?

Policy recommendations for the diagnosis, treatment, and research of rare diseases in the SADC region

1. “We propose a unified policy framework to aimed at improving the health and livelihood of PLRDs” to be changed into “We propose a unified policy framework to aiming at improving the health and livelihood of PLRDs”.
2. “We propose the establishment of in providing timely diagnosis” to be changed into “We encourage the establishment of in providing real-time diagnosis”.

Adoption of a common definition of a rare disease

1. “This, in return, will facilitate research into diagnosis and treatment options and improve access to treatment” to edit into “In return, the adoption of a common definition will facilitate the diagnosis procedures and increase the treatment options and access”.

Rare diseases database

1. “We propose the establishment of a centralized registry for rare diseases and their demographics which ... in the region” to modified into “We suggest the establishment of a centralized registry for rare diseases and their geographical, socioeconomic, and demographic factors which will increase the epidemiological knowledge of rare diseases in the region”.

Foster collaborative research in rare diseases

1. Change first sentence into: “A large percentage of rare diseases have a genetic origin; thus, understanding human genetics is the necessary for tackling and declining their burden”.

Establishment of rare diseases centers of excellence

1. First sentence to be modified into: “Since rare diseases treatment and management are often complex, costly, and require services from different departments, difficulties can be approved financially in primary healthcare facilities”.
2. “Centers of excellence will also promote the sharing of information and expertise among healthcare providers and between them and researchers” edit it into: “Centers of excellence will also promote the availability of information and expertise among healthcare providers and researchers reciprocally”.

Cost sharing and health insurance coverage

1. As the first sentence provides a statistical information, authors should cite a reference.
2. “Universal health insurance coverage... without financial barriers” change it into “Universal health insurance coverage ... without financial, gender, and geographical barriers”.

Capacity building for healthcare workers

1. Check for grammatical mistakes in the paragraph. For example, “... healthcare providers and providing financial and other support” should be healthcare givers and providing financial and other supports”.

Establish inclusive policies for access to social services

1. Authors should add the establishment of laws to protect those suffering rare diseases, especially children in rural and urban areas.
2. The creation of specialized facilities and structures to decline the discrimination.

Conclusion

1. “We propose...” to change into “The proposed unified policy framework...”.

On the first pages of the manuscript, the authors referred to rare diseases as RD. However, this reference has changed instead for the employment of “rare diseases” all along towards the end. Authors should keep using RD throughout the manuscript to design rare diseases as the abbreviation was defined from the first use, and in the list of abbreviations.