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

Michèle Ramsay[1]

1 University of Witwatersrand

Potential competing interests: No potential competing interests to declare.

It is very encouraging to read about regional initiatives in Africa to address diagnosis, treatment and policy around rare diseases. Rare diseases have not received sufficient attention in Southern Africa (SA) at many different levels, and this has led to great suffering for patients and their families. In some cases a definitive diagnosis could lead to lifesaving interventions and treatments. I commend the efforts of my colleagues and hope that they will continue the discussions and the pursuit of mechanisms and approaches to make a meaningful differences in the lives of affected families.

There are a few things that require further consideration and that would help clarify objectives and perhaps guide a more practical approach. Much still need to be done to lobby governments for support and to ensure that they invest in these activities.

Definition of rare disease in the context of SA: It is suggested that if it affects 1 person (or fewer) per 2000 it should be considered rare. We do not have accurate prevalence for most diseases in SA so perhaps we need to think of another way of defining ‘rare’.

The current list of rare diseases includes a mixture of monogenic, multifactorial and environmentally triggered diseases and a careful delineation of the different types of diseases and potential approaches to diagnosis and treatment should be explored. This will also feed into approaches to health improvement for people with rare diseases.

Some activities can be encouraged and implemented across multiple countries within a region, but others require country level interventions. The sphere of influence should be assessed and addressed at the appropriate level.

Developing databases, capacity strengthening through specialised training courses, fostering collaborative research, developing guidelines and best practices, and establishing regional centers of excellent are ideal for implementation across multiple countries in a region. This will help cross-fertilisation of ideas and brainstorming of approaches that can then be applied in different setting.

However, initiatives like introducing rare diseases in training curricula, cost sharing and health insurance coverage, access to social services, and establishing national rare diseases centers of excellence in multiple countries are important but need support and funding. These activities therefore, and most importantly, require political buy-in within a country. They cannot be actioned at regional level, unless there are agreements between individual countries to jointly support these initiatives. This could be very challenging as each country sets its own health priorities.

Consideration could also be given to priority setting. What could be done first and how could other initiatives then follow. It is challenging when there are few resources, and not many people involved, to tackle a problem for all sides simultaneously.