Call for the inclusion and prioritisation of rare diseases in the National Health agenda

To address the barriers to healthcare experienced by people with rare diseases, this call for open dialogue is intended as the first steps towards the development of a National Rare Disease Policy for South Africa. The intent is to secure equality of care for those who need, but who do not currently have access to the appropriate diagnosis, treatment, and healthcare services.

1. Introduction

The Rare Diseases Access Initiative (RDAI) in South Africa was formed as a coalition of interested groups, with the aim to promote an environment which is favourable for those with rare diseases. In particular, the participants seek equitable access for those who need, but who do not currently have, the appropriate diagnosis, treatment, and healthcare services, regardless of healthcare sector.

The development of a National Rare Disease Policy would quantify the disease burden, define the standards of care, address capacity amongst healthcare professionals, and ensure that there is appropriate diagnosis and monitoring.

1.2 Participants in the Rare Diseases Access Initiative

Board of Healthcare Funders (BHF)
Discovery Health
Government Employees Medical Scheme (GEMS)
Health Funders Association (HFA)
Innovative Pharmaceutical Association South Africa (IPASA)
Medihelp
Medscheme
Momentum
Rare Diseases South Africa (RDSA)
The South African Medical Technology Industry Association (SAMED)
Observer: Council for Medical Schemes

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2. The Problem

2.1 Lack of data

There is no universal definition for rare diseases. Rare diseases are serious chronic diseases that may be life-threatening, most of them with a genetic basis. There are estimated to be between 5,000 and 8,000 rare diseases worldwide and one person out of every 15 could be affected by a rare disease.[1]

The thresholds used to define rare diseases range widely across countries. The WHO and the EU use a threshold of not more than 5 people per 100,000 population. Disease prevalence seems to be the preferred way by which all jurisdictions define rare diseases.[2] South Africa lacks data on rare disease prevalence.

2.2 Impact on patient lives

RDs are life threatening, life limiting or chronically debilitating diseases, often of genetic origin. RDs are complex, often affecting multiple body systems and requiring specialised and coordinated care that comes at considerable cost to families and the health system.

Centres of excellence, dealing with certain rare diseases, exist in various centres in South Africa. These institutions set an example of what can be achieved but are needed on a much broader scale. This presents an opportunity for collaboration to improve the South African health system and health outcomes for people with rare diseases.

3. Global Trends – Policy for Rare Diseases

In 2019, Rare Diseases International signed a Memorandum of Understanding with the World Health Organisation (WHO) with the key focus of shaping an international rare disease policy and to strengthen the capacity of health systems to address rare diseases.[3]

Further to this, in December 2021, the United Nations (UN) General Assembly formally adopted a Resolution recognising the over 300 million Persons Living with a Rare Disease (PLWRD) and their families worldwide. This Resolution was ground-breaking as it was the first UN document to formally acknowledge the challenges faced by PLWRD and their families. The Resolution represents a major shift in global policy which should see rare diseases integrated firmly into the agenda of the UN. Key actions asserted in the Resolution are as follows:[3]

"Recognizing the fundamental importance of equity, social justice and social protection mechanisms as well as the elimination of the root causes of discrimination and stigma in health-care settings to ensure universal and equitable access to quality health services without financial hardship for all people, particularly for those who are in vulnerable situations, including those living with a rare disease".

"To strengthen health systems, notably in terms of primary health care, in order to provide universal access to a wide range of healthcare services that are safe, of quality, accessible, available and affordable, timely, and clinically and financially integrated, which will help to empower persons living with a rare disease in addressing their physical and mental health needs to realize their human rights, including their right to the highest attainable standard."

"The implementation of appropriate, national measures to ensure that persons living with a rare disease are not left behind, recognizing that persons living with a rare disease are often disproportionally affected by poverty, discrimination and lack of decent work and employment, and that they may require assistance in order to enjoy equal access to benefits and services, notably in the fields of education, employment and health, and to promote their full, equal and meaningful participation in society, and to commit to working towards the social integration and physical and mental well-being of persons living with a rare disease and their families and caregivers without any discrimination".

3.1 Need for data and a registry to determine burden of disease.

In Europe, information on patients who are affected by rare diseases is fragmented across hundreds of registries, both nationally and regionally, making it difficult to quantify the burden of these diseases. To plan appropriately for services, being able to quantify the burden of disease is imperative. To address this problem, the European Union Rare Disease Platform (EU RD Platform) was established. This Platform aims to set standards for data collection and data exchange to ultimately act as a "knowledge generation centre" for researchers, healthcare providers, patients and policymakers. Maximising the information from each registry and making it more accessible should improve knowledge, diagnosis, and treatment of rare diseases.

4. Rare Diseases in South Africa

4.1 Demographics

In South Africa, it is estimated that 4.2 million people are living with a rare disease; however, there is no formal definition of rare diseases in the country, nor is there any record keeping of incidence, prevalence or treatment, making it difficult to quantify the burden of rare diseases nationally. In addition, the management of patients with rare diseases largely follows an individualistic clinical approach, with differences across provinces, and between the private and public healthcare sectors. Many rare diseases go undiagnosed, and the current healthcare system is not geared to responding to the various challenges that individuals with rare diseases face.[4]

4.2 Proposed definition of RDs for South Africa

Based on the available research and with its application to the South African population, RDAI proposes the adoption of the following definition for a Rare Disease:
"a condition affecting less than 1 in 2000 people (50 per 100,000 of population".[2]

5. Need for SA Policy on RDs

The proposed development of a national rare disease policy for South Africa is crucial to addressing these imbalances. A National Rare Disease Policy would:

- quantify the disease burden
- define the standards of care
- build capacity amongst health care professionals
- ensure that there is appropriate diagnosis and monitoring
- provide for continuity of care
- create the need for a digital platform to record data that will allow for health outcomes to be monitored
- result in cost benefits for rare disease patients and the health system

5.1 Way forward – Recommendation for a Think Tank

The RDAI recommends the establishment of a 'Think Tank' platform, bringing together the relevant stakeholders including:

- policy makers
- clinical experts
- researchers
- epidemiologists
- civil society
- funders both public and private
- regulators
- patient advocacy groups
- pharmaceutical and technology companies.

Working groups could then be established from this platform, tasked with addressing specific issues including, but not limited to:

- Quantifying the national incidence of each rare disease
- Defining the standards of care/treatment guidelines including eligibility and exit criteria for each treatment option.
- Developing a monitoring and evaluation framework that will track the health outcomes as per the defined indicators identified.

5.2 RDAI offers to partner in these efforts

The RDAI, as a coalition on interested parties including patients and healthcare players, is willing to participate in the process to achieve the goals set out herein.

6. Conclusion

The RDAI calls on the Honourable Minister of Health to take accountability for rare diseases and include and prioritise rare diseases in the National Health agenda, as an ethical imperative.

South Africans living with rare diseases have the potential to live improved, longer lives if they are diagnosed early and able to access appropriate and coordinated healthcare services timeously. The solutions identified should be based on the principles of financial sustainability, equitable coverage, and patient accessibility to treatment. These principles are in line with the objectives of the proposed National Health Insurance Framework.

In the absence of a policy on rare diseases in South Africa, the achievement of this goal will only be possible through collaboration between patients, healthcare providers and government, with government offering leadership in the establishment of a national policy on rare diseases in South Africa.

7. References

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