



## Introducing the South African Rare Diseases Access Initiative (RDAI)

16 February 2024  
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# The Rare Diseases Access Initiative



**A growing organisation with big ambitions**

**Seeking fair treatment for those affected by rare diseases**

## Formed in 2019

The RDAI was formed in 2019 to promote a more favourable environment for those impacted by rare diseases in South Africa.

## Collaboration

The RDAI works in collaboration with Rare Diseases South Africa (RDSA) whose mission is to ensure a better tomorrow for all those impacted by rare diseases.

## Equitable Access

The RDAI seeks to improve equitable access for Rare Diseases patients regardless of their socioeconomic status or healthcare sector they access.

RDAI is led by a steering committee made up of representatives drawn from various leading organisations



# Rare Diseases in Context

Rare diseases are often:

- life threatening, life limiting or chronically debilitating;
- complex, often affecting multiple body systems and requiring specialised and coordinated care that comes at considerable cost to families and the health system;
- incurable, many with no effective treatment and symptoms often worsen over time.

Demographics of Rare diseases:

- Rare diseases (RD), including 7 000 conditions characterised to date, are defined as conditions affecting <1 in 2 000 of a population.<sup>1-4</sup>
- Effect an estimated 4.2 million, or 1 in every 15 South Africans, of whom 50 - 70% are children.<sup>2,5-7</sup>
- 80% of rare diseases are genetic, and therefore not readily preventable.



1. Orphanet. Prevalence and incidence of rare diseases: Bibliographic data. Diseases listed by decreasing prevalence, incidence or number of published cases. Paris: Orphanet, 2022

2. European Organisation on Rare Diseases. Rare diseases: Understanding this public health priority. Paris: EURORDIS, 2005. [https://www.eurordis.org/wp-content/uploads/2009/12/princeps\\_document-EN.pdf](https://www.eurordis.org/wp-content/uploads/2009/12/princeps_document-EN.pdf) (accessed 23 April 2023).

3. Haendel M, Vasilevsky N, Unni D, et al. How many rare diseases are there? Nat Rev Drug Discov 2020;19(2):77-78. <https://doi.org/10.1038/d41573-019-00180-y>

4. Rare Diseases International. Operational Description of Rare Diseases. A Reference to Improve the Recognition and Visibility of Rare Diseases. Paris: RDI, 2022. <https://www.rarediseasesinternational.org/description-for-rd/> (accessed 30 March 2022).

5. Statistics South Africa. Statistical Release P0302. Mid-year population estimates 2022. Pretoria: StatsSA, 2022. <https://www.statssa.gov.za/publications/P0305/P03052021.pdf> (accessed 20 February 2023).

6. Council of the European Union. Council recommendation on action in the field of rare diseases. J Eur Comm 2009;C151/7-C151/10. <https://eur-lex.europa.eu/legal-content/EN/TXT/?uri=celex%3A32009H0703%2802%29> (accessed 23 April 2023).

7. Wakap SN, Lambert DM, Olry A, et al. Estimating cumulative point prevalence of rare diseases: Analysis of the Orphanet database. Eur J Hum Genet 2020;28(2):165-173. <https://doi.org/10.1038/s41431-019-0508-0>

# Key Challenges for Rare Diseases in South Africa

- The SA health system is not equipped to respond to the needs of those living with rare diseases, being overwhelmingly geared towards supporting diseases that affect large numbers of people.
- The burden of rare diseases remains unacceptably high for patients, families, communities and the health system.



# RDAI Key Strategic Priorities to Address Challenges

Six key strategic priorities were identified for inclusion in a proposed National Rare Diseases Framework and Strategy:<sup>8</sup>

1. **Diagnosis:** Timely, accurate RD diagnosis saves lives, prevents disease progression and mitigates disability.
2. **Access to treatments:** RD patients require equitable and timely access to clinically appropriate treatments in a manner that is financially sustainable to the healthcare system.
3. **Data collection and management:** Empirical data are required to quantify the RD burden of disease to enable an appropriate healthcare response to be mounted via independent, comprehensive and transparent health technology assessment.
4. **Co-ordinated care:** Many RDs are complex, long-term, and affect multiple organ systems. Co-ordinated, multidisciplinary care is needed to both improve patient outcomes and optimise allocation and implementation of available resources.
5. **Access to services:** Quality health services should be equitably provided, accessible and affordable to RD patients.
6. **Collaborative research:** Nationally co-ordinated research, including active participation by patients/carers and advocacy groups, should be integrated into national plans and clinical care for RD.

8. Rare Diseases Access Initiative. Call for a National Rare Disease Framework. Johannesburg: Rare Diseases South Africa, 2020.

# How RDAI Plan to Achieve our Goals

## POLICY DEVELOPMENT STEPS

HOW CAN THIS BE ACHIEVED? —>—

- Establish a RD advisory committee
- Map gaps & opportunities
- Establish system requirements
- Create a roadmap
- Plan a phased implementation
- Determine an implementation timeline



### A NATIONAL RARE DISEASES FRAMEWORK What are the goals?

- Rare Disease Guidelines
- Co-ordinated care
- Centres of Excellence
- Improved patient outcomes
- Access to services
- Health needs assessment
- Collaborative Research

Quantify disease burden

01

Address & build capacity

02

Strengthen the capacity of health systems to address RDs

03

04

05

06

Define standards of care

Facilitate appropriate diagnosis, treatment & continuity of care

Research & measure: Data, outcomes and monitoring



# RDAI Activities and Outputs

## Symposiums and Outcomes

2021 RDAI Symposium

Outcome:  
Rare Diseases framework document was adopted

2022 RDAI Stakeholder Workshop

Outcome:  
Stakeholder proposals on implementation of the Rare Diseases framework

## Documents and Publications

Rare Diseases framework document, 2019

Recognising Rare Diseases: Access and action:  
Specialist Forum, September 2021

RDAI call to action, white paper, 2022

Call for inclusion and prioritisation of Rare Diseases in the national health agenda. October 2023

Rare-Diseases – progress on the path to improved health services; Specialist Forum, January 2023

SAMJ publication, 2023

## Key Stakeholders Engagement

Council for Medical Schemes

National Department of Health

- Chief Director, NDoH
- Deputy Director General NHI
- Child, Youth, and School Health Chief Directorate

# Conclusion

- Recognizing rare diseases as part of public health is not just an ethical imperative but a strategic necessity.
- By addressing rare diseases within the framework of public health, we acknowledge the shared responsibility to ensure timely and accurate diagnosis, equitable access to treatment, and comprehensive support for affected individuals and their families.
- In doing so, we champion inclusivity, foster medical innovation, and contribute to a healthier society overall.
- Going forward, RDAI will continue to address challenges faced by RD patients, ensure RD inclusion in the National Health Insurance<sup>9</sup> and promote universal health coverage for those impacted by RD in the context of the 2030 Sustainable Development Goal 3 targets.<sup>10</sup>

**"No country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases."**

- Helen Clark, United Nations Development Programme  
RareX/ICORD 2016, Cape Town, South Africa



# THANK YOU



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