

Introducing the South African Rare Diseases Access Initiative

To the Editor: Rare diseases (RD), including 7 000 conditions characterised to date,^[1-3] are defined as conditions affecting <1 in 2 000 of a population.^[4] It is estimated that RD collectively affect 4.2 million, or 1 in every 15 South Africans, of whom 50 - 70% are children.^[2,5-7] Those impacted by RD face significant and distinct challenges, including an extended 'diagnostic odyssey' of ≥5 years.^[8] Over 80% of RD are genetic in origin, and many are life-limiting and progress swiftly – resulting in premature death or lifelong disability. Less than 10% of RD have a treatment approved by the US Food and Drug Administration (FDA),^[9] and the limited treatments available are often inaccessible and costly, with poor clinical management.

To address these unique challenges, the Rare Diseases Access Initiative (RDAI) was formed in 2019 to promote a more accessible healthcare environment for those impacted by RD in South Africa (SA). This coalition includes the Board of Healthcare Funders, Health Funders Association, Innovative Pharmaceutical Association of SA and Rare Diseases SA NPC.

RDAI seeks to improve equitable access to the appropriate diagnosis, treatment and healthcare services for all RD patients, in an affordable and sustainable manner.

The first RDAI stakeholder symposium was undertaken virtually in August 2021, resulting in six key strategic priorities identified for inclusion in a proposed National RD Framework and Strategy:^[10]

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 RD 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.

A second RDAI stakeholder symposium held in October 2022 continued this stakeholder dialogue to find actionable solutions for the six strategic priorities previously identified.^[11] Participants outlined components of the proposed RD framework where they could contribute and actively participate.

Going forward, RDAI will continue to address challenges faced by RD patients, ensure RD inclusion in the National Health Insurance^[12] and promote universal health coverage for those impacted by RD in the context of the 2030 Sustainable Development Goal 3 targets.^[13]

For more information on RDAI, contact research@rarediseases.co.za.

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