Rare diseases – progress on the path to improved health services

Photo credit: Shutterstock.com

Incremental improvements in the South African rare disease landscape have occurred over recent years. The Rare Disease Access Initiative (RDAI), a coalition of key stakeholders that includes participants from health industries and patient advocacy groups, have been the primary driving force behind raising awareness and promoting an environment which is favourable for people living with a rare disease (PLWRD).

The RDAI recently hosted their second Stakeholder Symposium, an open dialogue regarding actionable solutions to the challenges of access to healthcare that are experienced by approximately 3.7 million PLWRD in South Africa. This forum was chaired by Dr Helen Malherbe, director of Research at Rare Diseases South Africa (RDSA).

Barriers to equitable healthcare access

Kelly du Plessis, CEO and lead patient advocate of the RDSA, described fundamental challenges that present barriers to diagnosis, treatment, and health services for PLWRD, of which the cumulative effect limits universal and equitable access to quality health services. Implicit to a dual healthcare system are the differences in service offerings between private and public sectors and across provinces.

Ms du Plessis asserted "a lack of

knowledge and awareness of rare diseases among healthcare professionals creates clinical challenges, and a lack of public awareness creates stigma and societal challenges for those who live with a rare disease".

A lack of data quantifying the extent of rare diseases in South Africa poses numerous challenges. Due to many competing health priorities, improved allocation of funding is more likely when substantiated with a quantified evidence base. The National Department of Health currently does not undertake diagnostic surveillance of rare diseases. Also, South Africa does not have an Orphan Drug Policy, posing challenges regarding research and registration for drug development.

Strengthening of health systems to address the physical and mental health needs of PLWRD and implementing appropriate national measures to ensure equal access to benefits and services can only be achieved through collaboration between patients, healthcare providers and government. To this end, the RDAI has made a call for the inclusion and prioritisation of rare diseases in the national health agenda.

Conquering milestones

An essential step to the development of a National Rare Disease Policy is a consensus understanding of which conditions, specifically in the South African setting, are to be considered as a rare disease. Dr Noluthando Nematswerani, Discovery Health, is of the view that the lack of a formal definition is of universal concern, giving rise to different standards of access across the health industry.

The RDAI has previously proposed a definition for a rare disease as 'a condition affecting less than one in 2000 people', and this has been widely adopted by relevant stakeholders. The RDAI acknowledges that this definition serves as a guideline and is not intended to be exclusionary, as all cases need to be considered individually.

RDSA has designed a patient-led registry that can be utilised by all stakeholders to improve surveillance and data collection. Because technical registries are usually underutilised, the rare diseases registry has been designed to remove the reliance on data being generated by the clinician or the hospital.

Advancing into action

A perspective from Dr Nematswerani is that rare diseases pose unique challenges to the funding industry. Important considerations include a lack of capacity for diagnosis, treatment and follow-up, that clinical guidelines are not always available to facilitate decision-making regarding treatment and funding, and that delayed diagnosis and treatment may be costly for medical schemes. These barriers can be addressed through the creation and strengthening of Centres of Excellence.

Another confounding factor is that only 5% of rare diseases are treated with medicines approved for such diseases, and affordability of some high-cost and ultra-high-cost rare disease treatments must be weighed against the opportunity cost for maintaining equity of access to health services for all medical scheme members.

A lack of data quantifying the extent of rare diseases in South Africa poses numerous challenges

Of PLWRD, 95% will receive supportive care only due to the dearth of registered treatments. These barriers may be addressed through central procurement initiatives inclusive of the public and private healthcare sectors, and by application for Section 36 exemptions.

Dr Nematswerani concluded: "Setting up registries for local data collection is key in improving care. Without an understanding of the scope of rare diseases in South Africa, it is difficult to innovate around access to care, standardisation of care, and improved clinical outcomes."

Representing the Innovative Pharmaceutical Association of South Africa (IPASA), Gavin Bauer of Sanofi reiterated that rare disease patients deserve access to appropriate treatments in a timely, sustainable manner which enables improved health outcomes.

It is the view of IPASA that, in South Africa, PLWRD have limited opportunities for diagnosis and treatment, the diagnostic journey is complex and lengthy, and patients lack certainty regarding funding and their prognosis. IPASA is in accord with the need to co-create a National Rare Disease Policy and are committed to a coordinated plan of action to implementation.

Practical steps to improve rare disease diagnosis, treatment and outcomes include bringing internationally available medicines to South African patients through an accelerated registration process, to co-create and innovate sustainable access such as employing alternative reimbursement models, and to advocate for access for all rare disease patients.

Dr Chris Vorster, head of the Centre for Human Metabolomics at the North-West University, discussed crucial challenges in the diagnosis of rare diseases in South Africa. He maintains that public sector tertiary hospitals are currently better equipped than the private sector for diagnosing the 'out-of-the-ordinary'. Diagnostic costs are moderate to very high and, consequently, many patients cannot afford to get a rare disease diagnosis.

Dr Vorster calls for the establishment of an advisory board that will oversee sustainable development of virtual Centres of Excellence that cater to the diversity of rare diseases, the establishment of diagnostic and treatment guidelines, as well as education and training activities; "If ever there is a time for this, it would be now." He also considers the development of a local registry and Biobank as critical because research is key to advancing rare disease care.

Matthew Bolz-Johnson, representing the Global Network for Rare Diseases, is inspired by what existing pockets of expertise in South Africa are achieving. He considers the country to be leaders in solving challenges for PLWRD, and thereby laying the groundwork for other nations in Africa. Possible pilot activities that the Global Network can assist South Africa to implement include the provision of virtual consultation by digital platform, capacity building of expertise for a disease area or thematic area, and to improve diagnostic access.

Recent victories - improved access for people living with hereditary angioedema or cystic fibrosis

For patients with hereditary forms of bradykinin-mediated angioedema, characterised by episodes of severe swelling that is potentially life-threatening, diagnosis is a primary challenge because the condition is not easily distinguishable among the many other non-genetic causes of histaminergic angioedema. The danger of a missed diagnosis is that the usual treatments will not work in these cases.

According to Prof Jonathan Peter of the ACARE Centre of Excellence, University of Cape Town, this illustrates other important issues in the journey of PLWRD

beyond access to expensive medications. Other key challenges are difficulties in accessing appropriate care, a lack of knowledge regarding the danger signs of an angioedema episode on the part of patients, educators and doctors, and that patients do not carry targeted therapies with them to counteract an episode as it occurs.

Recently launched, the Angioedema Hotline aims to support patients in remote areas and to improve diagnosis and the use of targeted therapies. Research to identify a hereditary angioedema biomarker in the African population is ongoing.

Regarding targeted therapies, the Global Access programme is assisting public and private sector patients and the price of certain therapies have been substantially reduced. A virtual Centre of Excellence is currently being established, for which Prof Peter has pioneering hopes.

Virtual centres of excellence will allow access not just for our patients in South Africa, but hopefully also for other groups around the continent that have the same problems with unrecognised and undiagnosed forms of angioedema, he said.

Dr Marco Zampoli, chair of the South African Cystic Fibrosis Medical and Scientific Advisory Committee, shared milestones achieved in improving cystic fibrosis (CF) care. CF has been well acknowledged, having established Centres of Excellence in the private and public sectors of some provinces, and Dr Zampoli credits the efforts of patient organisations in supporting clinicians regarding CF advocacy and awareness.

Rare Diseases South Africa

About the RDAI

The RDAI was formed as a coalition of interested groups, with the aim to promote an environment which is favourable for those with rare diseases. Participants in the RDAI and supporters of the objectives include the Board of Healthcare Funders, the Health Funders Association, the Innovative Pharmaceutical Association South Africa (IPASA), and RDSA.

RDAI RARE DISEASES ACCESS INITIATIVE

The South African CF registry was established in 2018 as a public–private initiative, and is a valuable resource for data, research and advocacy. Funding for part-time data collectors has proved to be a successful model for quantifying the CF landscape in South Africa.

The diagnosis of CF remains a challenge in many provinces even though early diagnosis improves outcomes, prevents early onset complications, and reduces the cost of treatment over a lifetime. Diagnosis is expensive and requires special equipment and expertise, and newborn screening is not routinely performed.

Importantly, Dr Zampoli maintains that complete genotyping is no longer a nice to have, but a must have, as it is necessary prior to the use of new treatments.

The Advisory Board has published comprehensive consensus guidelines on all aspects of CF care in the South African context, are actively engaged with the CF community, and have ongoing engagement with the pharmaceutical industry to access new medicines.

Dr Zampoli concluded that "the value of a multidisciplinary rare disease team lies in the support provided to each other, their patients and the families of patients.

The way forward

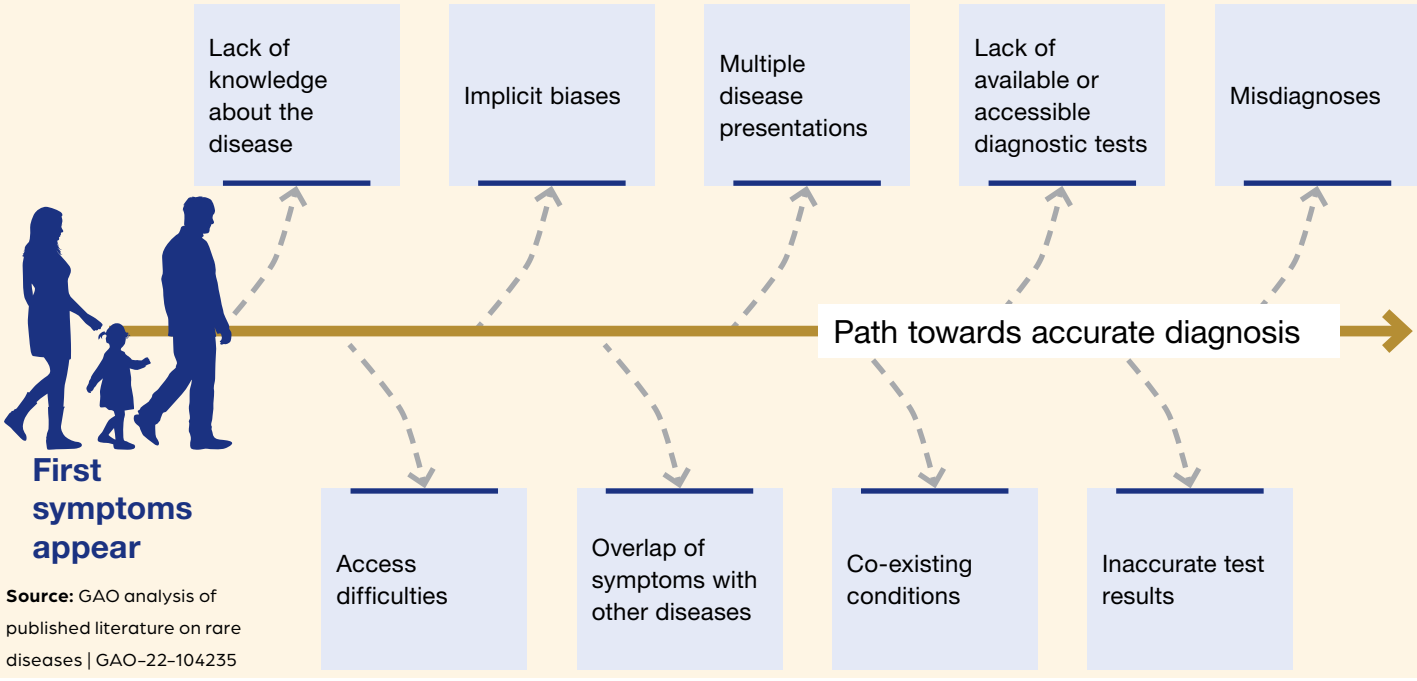
The RDAI intends to improve access by driving solutions whereby rare disease patients have some certainty. The development of a National Rare Disease Policy would quantify the disease burden, define the standards of care, address capacity among healthcare professionals and ensure that there is appropriate diagnosis and monitoring.

Foundational to such a policy are an improved clinical and public knowledge base, central procurement to ensure access to high-cost treatments, the setting up of data registries, and the establishment of advisory boards to develop guidelines for care.

Ms du Plessis had an important take-home message: The reality is that these challenges need to be addressed as a parallel process to the NHI. We have issues that need to be addressed today, we cannot keep saying to patients just wait for the NHI to come along.

YouTube link:
https://youtu.be/VIygZnRZRTI SF

Figure 1: Factors that may present a barrier to an accurate rare disease diagnosis



Source: GAO analysis of published literature on rare diseases | GAO-22-104235