Equity call for rare diseases healthcare

For those impacted by rare diseases, equitable access across the continuum of care will not only extend and improve their quality of life, it will also be of economic benefit to the individual and the health system.

Source: ©niyazz 123rf

Currently, regardless of the healthcare sector, the South African health system is not geared to respond to the particular challenges that are common to many different rare diseases.

A rare disease is defined as a disease that affects less than one in 2,000 people. Approximately 8,000 different rare diseases have been described to date and, cumulatively, there are 3.7 million South Africans living with a rare disease.

Often complex, they can affect multiple body systems, requiring expensive specialised and coordinated care.

Call for equity

A recent webinar, coordinated by the Rare Diseases Access Initiative (RDAI), in association with Rare Diseases South Africa (RDSA) has led to a call for a coordinated approach to diagnosis and care, and for increased equity in healthcare, for people living with rare diseases.

The webinar brought together an expert panel to discuss the challenges facing those affected by rare diseases in South

Africa and to explore possible solutions.

“South Africans living with rare diseases could have extended and improved lives if they are able to freely access appropriate and affordable healthcare services without barriers,” says Kelly du Plessis, on behalf of the RDAI.

“Our goal is to put South Africa on par with the EU, UK and BRICS, by having a National Rare Disease Policy.

“Without a Rare Disease framework and policy, South Africa is falling behind other countries and our chance of achieving universal healthcare is diminished,” she says.

Improving access

Keynote speaker, Professor Shahida Moosa, specialist consultant in medical genetics at Tygerberg Hospital and associate professor of medical genetics at Stellenbosch University, explained that for families with rare diseases, the journey to a diagnosis becomes an odyssey.

This journey is punctuated by multiple tests and investigations, some that are painful and invasive and many that are unnecessary, and that despite consultation with numerous specialists, many are still left without a diagnosis or find themselves on a misdiagnosis odyssey.

The transition from disjointed to coordinated care for those with rare diseases can be effectively managed by the creation of ‘centres of excellence’ that provide the comprehensive services of a multidisciplinary team with requisite support and collaboration between stakeholders.

NHI – rare diseases in context

Dr Crisp, deputy director general National Health Insurance, National Department of Health, and keynote speaker noted that all healthcare providers, public and private, should be accessible to everybody purely based on their need for healthcare.

Rare diseases are currently treated in the public sector although there are the obvious limitations of capping of care and rationing of services, which happens even in wealthy countries.

The National Health Insurance (NHI) fund will enable efficiencies that can redirect more money to pay for rare disease products, although there may be a point where it is not cost-effective for the individual or the nation.

Inclusion will be based on the cost-effectiveness of the health product rather than which health product is the least expensive.

Dr Crisp elaborates further advantages of the NHI fund as a single purchaser: “Over time, the artificial intelligence in the system makes it possible for us to understand where the products are being used, by whom, are there variations, and what is rarely used?

“This enables budget accommodations to be made, particularly in the case of rare diseases, so that the other people living in that area are not penalised for those few who are receiving very expensive care.”

Data collection, monitoring and evaluation

The National Public Health Institute of South Africa will have the functions of conducting research to inform policy and guideline formulation; monitor and evaluate the impact of policy and system implementation; strengthen epidemiology and surveillance; and strengthen advocacy, social mobilisation and partnerships.

Dr Natalie Mayet: deputy director at the National Institute for Communicable Diseases, also a keynote speaker, is of the

view that these functions can only be achieved through collaborative networks and partnerships with stakeholders.

Specific elements of multiple existing policy and regulatory frameworks for health information systems across South Africa can be leveraged to support a proposed data framework for a rare disease surveillance plan.

“This data framework is underpinned by healthcare professional capacity building and a robust communication strategy,” Mayet notes.

Collaborative research

Ideally, research should be embedded into clinic care and should be aimed at solving particular problems.

The generation of statistically robust evidence from the trial setting is challenging in rare disease research due to low patient numbers and the ethical implications of including children in such studies, and this limits the development of treatments