The Plight of Rare Diseases in Southern Africa: Health and Social Services Policy Recommendations

Aneth David[1], Mohamed Zahir Alimohamed[2], Grantina Modern[3], Sharifa Mbarak[4], Husna Ahmad, Bianca Özcan[5], Nomsa Mtshali[4], Hlumela Tshijila[4], Kelly du Plessis[4], Eda Selebatso[6], Matshediso Letsholo[6], Nthabeleng Ramoeli, Trudy Nayakabangwe, Reon Van Der Merwe, Vivian Joseph, Farhan Yusuf, Maximilian Godwin Kilipamwambu, Kilaza Samson Mwaikono[2], Siana Nkya[2]

1 University of Dar es Salaam
2 Muhimbili University of Health and Allied Sciences
3 Nelson Mandela African Institution of Science and Technology
4 Rare Disease South Africa
5 Multiple Sclerosis Namibia
6 Botswana Organisation for Rare Diseases (BORDIS)

Funding: No specific funding was received for this work.

Potential competing interests: No potential competing interests to declare.

Abstract

Rare diseases (RDs) affect a small proportion of the population when compared to infectious and non-communicable diseases (NCDs) and thus receive limited attention. However, there are more than 10,000 known RDs affecting about 300 million people worldwide. The majority of rare diseases are hereditary and affect children. RDs are understudied and are difficult to diagnose, manage, and treat. The situation is worse in sub-Saharan Africa (SSA), where the public health infrastructure is weak, leaving a high health and cost burden on patients and caretakers. SSA is yet to adopt a common definition of what constitutes a rare disease as other regions, let alone establish the demographics of the kinds of RDs afflicting the population and the actual burden. Furthermore, policy frameworks and instruments to guide the management of RDs are non-existent. This gap contributes to a slow pace in the realization of universal healthcare and the sustainable development goals in this region.

We propose the adoption of a unified approach to tackle the burden of RDs across the SADC region by implementing common policies and measures aimed at improving the health and livelihood of people living with RDs. The recommendations include adopting a common definition, centralizing healthcare services, adopting preventive approaches, fostering collaborative research, and capacity building for healthcare workers. In addition, we propose shared cost-bearing models and special health insurance coverage that will ensure the provision of needed health and social services that improve the lives of people living with rare diseases (PLRD).

This position statement has been developed by a coalition of a professional human genetics organization, rare disease patient advocacy groups from six African countries, and the health youth cluster of the Southern African Development Community (SADC). This paper aims to inform policy issues on healthcare and social services that are necessary for improving the lives of PLRD. Harmonization of healthcare and social services policies and regulations across the SADC region will improve access to services and the livelihood of PLRD by allowing the effective utilization of resources and knowledge generated from common research goals.

Aneth Bella David[1,2], **Mohamed Zahir Alimohamed**[1,3,4,5], **Grantina Modern**[1,6], **Sharifa Mbarak**[7], **Husna Ahmad**[7], **Bianca Özcan**[8], **Nomsa Mtshali**[9], **Hlumela Tshijila**[9], **Kelly du Plessis**[9], **Eda Selebatso**[10], **Matshediso Letsholo**[10], **Nthabeleng Ramoeli**[11], **Trudy Nayakabangwe**[12], **Reon Van Der Merwe**[13], **Vivian Joseph**[13], **Farhan Yusuf**[13], **Maximilian Godwin Kilipamwambu**[13], **Kilaza Samson Mwaikono**[1,14], **Siana Nkya**[1,4,15]

[1] *Tanzania Human Genetics Organisation (THGO), Dar es Salaam, Tanzania*

[2] *Department of Molecular Biology and Biotechnology, University of Dar es Salaam, Dar es Salaam, Tanzania*

[3] *Department of Haematology and Blood Transfusion, Muhimbili University of Health and Allied Sciences, Dar es Salaam, Tanzania*

[4] *Department of Biochemistry, Muhimbili University of Health and Allied Sciences, Dar es Salaam, Tanzania*

[5] *Department of Genetics, University Medical Center Groningen, Groningen, The Netherlands*

[6] *Decca College of Health and allied sciences, Dodoma, Tanzania*

[7] *Ali Kimara Rare Diseases Foundation (AKRDF), Dar es Salaam, Tanzania*

[8] *Multiple Sclerosis Namibia, Namibia*

[9] *Rare Disease South Africa, South Africa*

[10] *Botswana Organisation for Rare Diseases, Botswana*

[11] *Rare Disease Lesotho Association, Lesotho*

[12] *Child and Youth Care Zimbabwe, Zimbabwe*

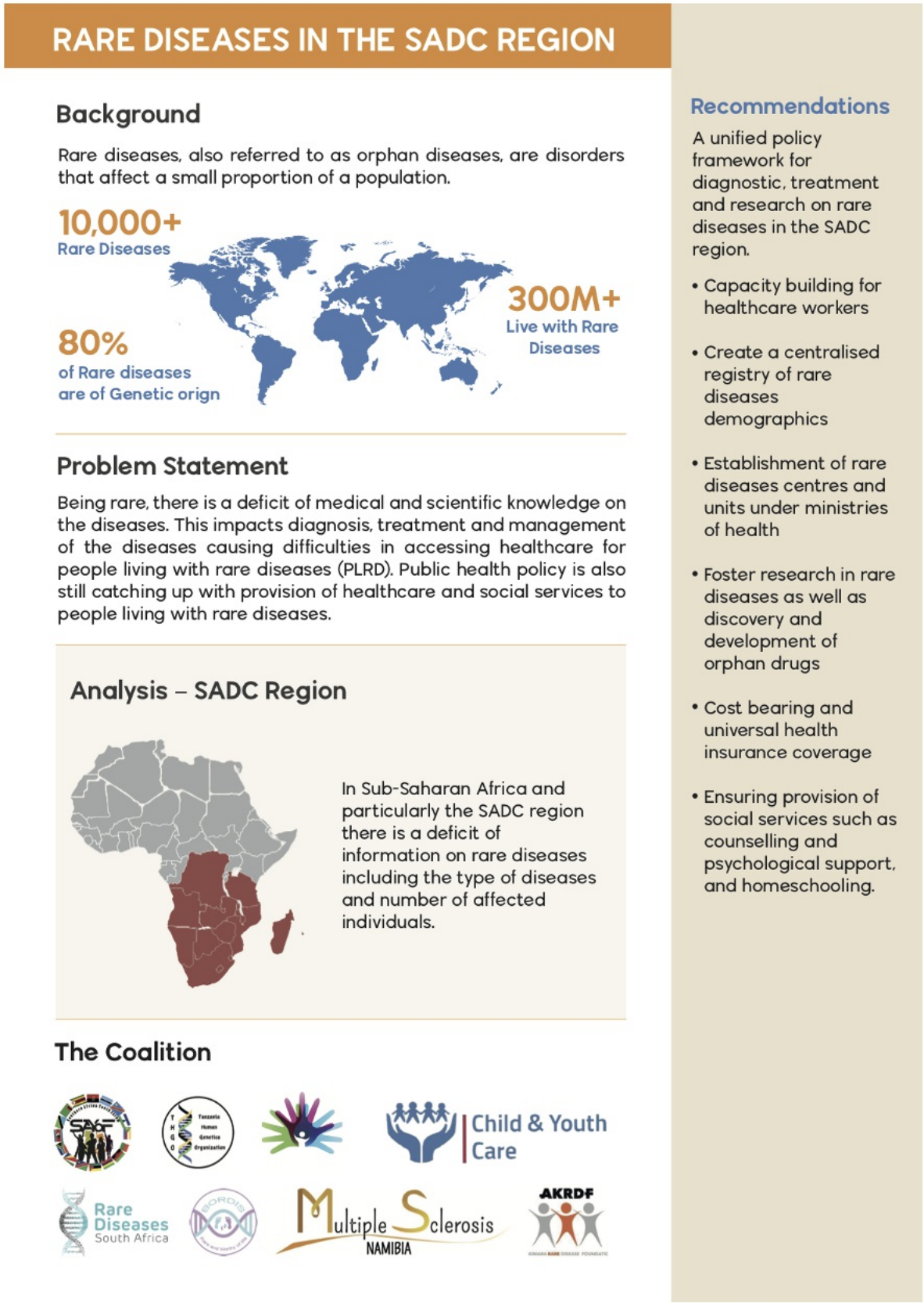
[13] *SADC Youth Forum, South Africa*

[14] *Department of Science and Laboratory Technology, Dar es Salaam Institute of Technology, Dar es Salaam, Tanzania*

[15] *Sickle Cell Program, Muhimbili University of Health and Allied Sciences, Dar es Salaam, Tanzania*

Keywords: Rare diseases, Southern African Development Community (SADC), Rare Diseases Africa, Rare diseases policy.

Graphical abstract



Background

Rare diseases, also known as orphan diseases, are disorders that affect a small portion of the population. The definition of a

rare disease varies among different populations and continents, but the most common definition states that a rare disease is a condition that affects 1 person per 2,000 (ORPHANET, 2012). There are over 10,000 known rare diseases (GARD, n.d.; Haendel et al., 2020; ORPHANET, 2012), most of which have a genetic origin (EURORDIS, n.d.; ORPHANET, 2012). Worldwide, it is estimated that 300 million people, which is about 3.5 - 5.9% of the global population, live with rare diseases (EURORDIS, n.d.; Nguengang Wakap et al., 2020). In Africa, the current understanding of rare diseases is hindered by a lack of reliable prevalence data. However, the prevalence is estimated to be 50 million patients (Lumaka et al., 2022), while some countries like South Africa indicate that there are approximately 3.6 million people with a rare disease, which translates to one in 15 people (RDAI, 2021; RDSA, n.d.). Adachi et al. (2023) suggested that a higher prevalence of rare diseases in South Africa compared to other African and neighboring countries may be due to a more developed tracking system. Therefore, despite being called rare, these diseases collectively impact a significant number of people.

Due to their rarity, there is a significant lack of medical and scientific knowledge about rare diseases. This deficiency has a substantial impact on the diagnosis, treatment, and management of these diseases, making it difficult for people living with rare diseases (PLRD) to access adequate healthcare. Additionally, public health policy is still in the process of catching up with the provision of healthcare and social services to PLRD (Krajnovic, 2012).

Situational analysis of rare diseases in the SADC region

In Sub-Saharan Africa (SSA), particularly in the Southern African Development Community (SADC) region, there is a lack of information regarding rare diseases, including their types, prevalence, and distribution (Kaywanga et al., 2022; Owings, 2021). Several diseases have been reported to affect people in the region, including Pompe disease, lupus, Ehlers-Danlos syndrome, mucopolysaccharidosis type (MPS), Gaucher disease, epilepsy, myasthenia gravis, multiple sclerosis, Guillain-Barré syndrome, hemophilia, neuromyelitis optica spectrum disorders (NMOSD), muscular dystrophy (MD), juvenile idiopathic arthritis, neurofibromatosis, trisomy 18, and Wiskott-Aldrich syndrome (Personal communication, Owings, 2021; Sevittz et al., 2022). Some diseases have been reported in case reports, such as Takenouchi–Kosaki syndrome (Flynn et al., 2021) and hereditary spastic paraplegia type 11 (Landouré et al., 2020). However, these represent only a few cases, as the majority of rare diseases remain undiagnosed or misdiagnosed due to limited information about their associated symptoms and a lack of guidelines for capturing their information within the healthcare systems of the SADC region.

On the other hand, there are no clear guidelines on the treatment and management of rare diseases, leaving the burden on patients and their families. This lack of information and delayed and/or misdiagnosis can result in a significant expenditure of resources and an increased burden on healthcare systems, patients, and their families. The diverse nature of rare diseases requires a multi-stakeholder approach to address the challenges, as patients often require care from multiple specialized departments. While the number of patients with a specific rare disease is low and spread across a large geographical area, collectively rare diseases affect a significant portion of the population. Furthermore, most rare diseases manifest at an early age, leading to a diminished quality of life (Gissen et al., 2021) and reduced life expectancy (Gorini et al., 2021). Affected patients require healthcare services throughout their lives, resulting in considerable costs. In the absence of comprehensive health insurance coverage, these expenses are borne by patients and their families (Adachi et al., 2023). Moreover, misdiagnosis, which is common with rare diseases, further exacerbates these costs and resource wastage. Timely and

accurate diagnosis, as well as appropriate management, could enable patients to lead normal lives and make contributions to society. However, research, diagnosis, genetic testing, and treatment for these diseases are limited due to constraints in human and financial resources.

Policies, laws, legislations, national plans, and strategies for guiding the provision of health and social services to PLRD in the SADC region are few, vary between countries, and are fragmented. A review of national plans, policies, and government actions for rare diseases in 23 countries across all continents by Khosla and Valdez (2018) found no published literature about existing or planned national plans, laws, or strategies related to rare diseases in Africa. Similarly, Adachi et al. (2023), in their review of the evidence, policies, and challenges to enhancing equitable access to rare disease diagnosis and treatment, noted the same gap in policies concerning rare diseases in Africa compared to other regions such as Europe, holding back progress for the diagnosis, treatment, and research of the conditions. The Africa health strategy of 2016 – 2030 mentions several health challenges facing the continent, including infectious diseases like AIDS, infant mortality, and even non-communicable diseases, but no mention of rare diseases (AU, n.d.).

However, efforts and progress are being made in individual countries. Regarding the provision of healthcare to PLRD, Tanzania has food, drugs, and cosmetics orphan medicines regulations put in place in 2018 to guide all regulatory matters related to registration, importation, and monitoring of the quality and safety of human and veterinary medicines designated as orphan medicines in Tanzania Mainland (URT, 2018). Furthermore, earlier this year, during the commemoration of the Global Rare Diseases Day, the President of Zanzibar, Dr. Hussein Mwinyi, ordered state institutions to incorporate rare disease treatment in the health policy to allow children with rare diseases to receive specialized care (Lamtey, 2023), although the translation into actionable policies may take longer. South Africa is reported to have begun developing a National Policy Framework and Strategy for Rare Diseases in 2019, with further stakeholder engagements in 2021 (Owings, 2021; RDAI, 2021). The framework was developed to help address imbalances in access to healthcare by PLRDs. It is not clear whether the framework has been adopted. Regionally, there are sparse initiatives that aim to collectively address rare diseases in Africa, such as the African Rare Diseases Alliance (African Alliance for Rare Diseases, n.d.) and the African Summit on Rare Diseases (EJPRD, 2021; FIN, n.d.; RDI, n.d.). Furthermore, information on different initiatives is notoriously difficult to obtain. Most initiatives are country-wise, such as those making up this coalition (Appendix 1).

Concerning research on rare diseases, in South Africa, the Centre for Metabolomics (CHM) is working towards establishing a rare disease biobank with the main focus on collecting samples and information on congenital rare diseases. The biobank lays a foundation for future similar initiatives and will go a long way in addressing the research gaps that exist on the continent as far as rare diseases are concerned (Adachi et al., 2023; Conradie et al., 2021). Other similar initiatives include the Rare Disease Working Group of the Human Heredity and Health in Africa Consortium (H3Africa), a collaborative partnership that focuses on addressing challenges associated with rare genetic diseases in Africa (Lumaka et al., 2022).

Regarding the provision of social services, in 2022, Tanzania approved the formalization of homeschooling for children living with rare diseases in response to the efforts of patient advocacy and professional groups (URT, 2021). However, such initiatives are still lacking in other SADC region countries.

These examples of policies, initiatives, and activities related to rare diseases demonstrate that progress is being made,

although it is slow and fragmented. The harmonization of healthcare and social services, along with their corresponding policies and regulations across a geographic region, enhances access to healthcare and improves the well-being of people living with rare diseases. This can be observed in the case of the European Union (EU) and other regional blocs, where it facilitates the effective utilization of resources and the sharing of knowledge to achieve common goals. Several similar proposals have been introduced, such as the national call to action policy framework in Tanzania (Kaywanga et al., 2022) and the global call for action on rare diseases in Africa, developed during the 11th International Conference on Rare Diseases and Orphan Drugs (ICORD) in South Africa (Baynam et al., 2020).

Policy recommendations for the diagnosis, treatment, and research of rare diseases in the SADC region

We propose a unified policy framework to guide the management and service delivery of RDs across the SADC region aimed at improving the health and livelihood of PLRDs. We propose the establishment of policy frameworks that will guide the healthcare systems in providing timely diagnosis, access to care, treatments and drugs, social support for patients with rare diseases in the SADC region, as well as fostering research aimed at gathering relevant RDs evidence.

The recommendations are summarized in Figure 1.

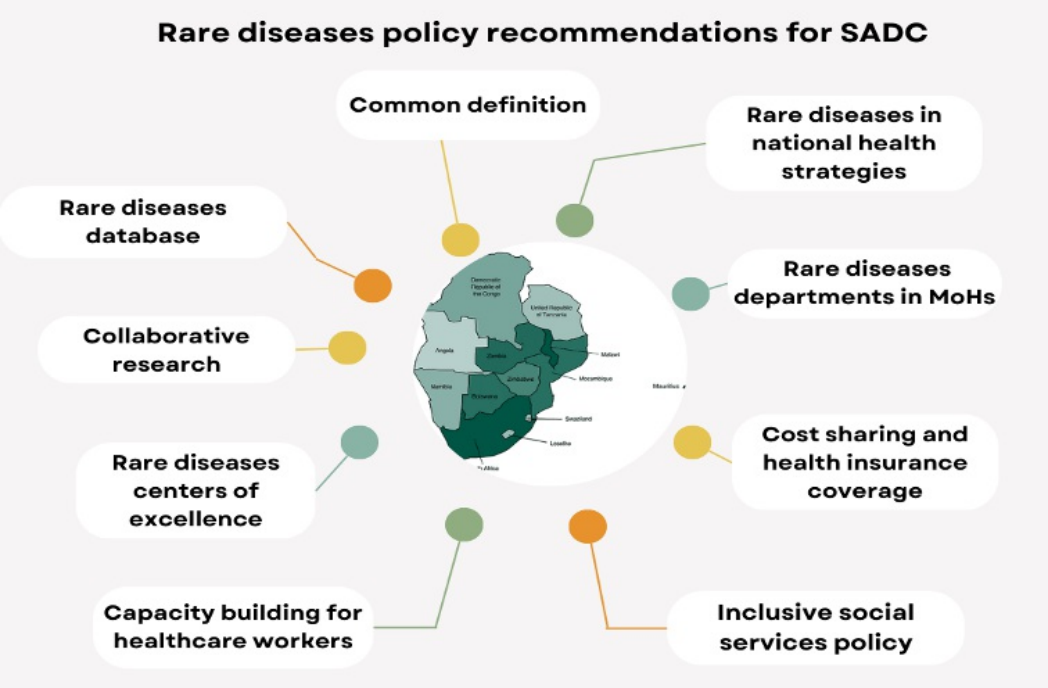


Figure 1. A summary of policy recommendations for rare diseases in the SADC region

Adoption of a common definition of a rare disease

The creation and adoption of a common definition for the entire SADC region will increase awareness and recognition of what constitutes a rare disease. This, in turn, will facilitate research into diagnosis and treatment options and improve access

to treatment. We propose using the most widely accepted definition of a rare disease, i.e., a condition that affects 1 person per 2,000.

Rare diseases database

We propose the establishment of a centralized registry of rare diseases and their demographics, which will help increase knowledge on the epidemiology of rare diseases in the region. The registry will identify and classify rare diseases in member states, their prognosis, diagnosis, and treatment regimens. This data is critical to establishing the actual needs and determining the type, level, and costs of the support needed. The database will also assist in research and appropriate policy development.

Incorporate rare diseases in national health strategic plans for NCDs and allocate budget

The NCDs' strategic plans should include various aspects of rare diseases, not just diagnosis and treatment, but also preventive services such as genetic counseling, prenatal and newborn screening, and genetic testing. This will ensure that rare diseases are given due consideration and are allocated the resources needed for diagnosis, treatment, and research, similar to other NCDs and infectious diseases such as cancer, TB, and AIDS. Furthermore, the inclusion of rare diseases in national strategic plans with clear budget commitments will overall lower the burden of rare diseases through preventive services and improve the livelihood of PLRDs.

Establishment of special rare diseases departments

Alongside the incorporation of rare diseases in national strategic plans, we also propose that SADC member states establish rare diseases departments within Ministries of Health to coordinate activities related to rare diseases prevention, treatment, and research within the ministries.

Foster collaborative research in rare diseases

A large percentage of rare diseases have a genetic origin; thus, understanding human genetics is a key part of tackling them. However, there is currently little genetic data on Africans in global databases, especially for people with rare diseases. Furthermore, research into rare disease diagnosis and treatment is not attractive to commercial companies due to the small customer base. We propose policies that will foster collaborative research to understand genetic factors and heredity patterns of rare diseases, the development of diagnostic tools, and the discovery and development of orphan drugs by:

a. Allocating funding for research on rare diseases diagnosis, heredity, and treatment
b. Creating a harmonious research environment in the region with shared resources, protocols, databases, and discoveries.
c. Harmonization of the regulatory environment for research, clinical trials, and drug development for rare diseases.
d. Incentivizing and encouraging the development of orphan drugs by fast-tracking the approval of protocols, waiving fees associated with the discovery and production of orphan drugs, and centralizing the processes and protocols for drug development. We also propose tax incentives for drug development companies, such as a certain level of tax credits and

tax reduction for preclinical research, as well as market exclusivity for a number of years.

Establishment of rare diseases centers of excellence

Since rare disease treatment and management are often complex and costly, requiring services from multiple departments, it can be difficult and costly to arrange at primary healthcare facilities. The establishment of centers of excellence for rare diseases will allow for the effective utilization of resources for diagnosis, treatment, and research, as well as make it easier for patients to access the services affordably. Centers of excellence will also promote the sharing of information and expertise among healthcare providers and between them and researchers, thus hastening the development of treatment options and accessibility for PLRDs.

Cost sharing and health insurance coverage

Partly because they affect few people, approved treatments are currently available for only 2.5-5% of rare diseases, many of which are prohibitively priced and often not registered for use in SADC countries. This means that patients in the region may not have access to treatments even if they are available in other parts of the world. Cost sharing will alleviate the burden of healthcare services for patients or healthcare providers and reduce barriers to accessing necessary healthcare services, particularly for lower-income individuals and those with chronic conditions. Universal health insurance coverage will ensure that everyone has access to necessary healthcare services without financial barriers. Cost sharing and universal health coverage may include subsidizing expensive and life-saving drugs for patients with serious rare medical conditions and establishing special national health insurance coverage for rare disease patients who face unique challenges in accessing necessary healthcare services.

Capacity building for healthcare workers

There is a need to increase the ability of healthcare workers to diagnose, treat, and manage rare diseases, as well as provide resources to patients. This may include the development of specialized training programs for healthcare providers and providing financial and other support. Other ways may include establishing referral networks to help healthcare providers connect with specialists who have expertise in the diagnosis and treatment of rare diseases, while promoting collaboration between providers and researchers to allow access to the latest diagnostic and treatment options. Furthermore, increasing access to up-to-date information on rare diseases and encouraging exchange between providers may improve their ability to diagnose and treat these conditions effectively.

Establish inclusive policies for access to social services

Living with a rare disease can be particularly challenging for children, as they may not be able to access social services such as education in normal settings due to significant physical, cognitive, and social impacts. We propose the adoption of policies to provide an environment for the social inclusion of rare disease patients in wider society. This can include institutional homeschooling and working from home as aspects of education and employment, which can offer tailored education, individualized instruction, flexible schedules, and remote work opportunities that can help rare disease patients

manage their health needs while also achieving independence, purpose, and financial stability.

Conclusion

Rare diseases receive limited attention in the SADC region, resulting in significant costs not only to healthcare systems but also to individuals living with these conditions. With no clear guidelines, the diseases are often misdiagnosed or completely undiagnosed, exacerbating the situation. We propose a unified policy framework for health and social services to address the gaps in rare disease diagnosis, treatment, and management in the SADC region. This approach has been shown to improve health outcomes for PLRDs by collectively addressing the burden and ensuring the effective utilization of resources, rather than tackling individual diseases in small geographical areas.

List of abbreviations

CHM	Centre for Metabolomics
NMOSD	Neuromyelitis Optica Spectrum Disorders
MPS	Mucopolysaccharidosis type
MD	Muscular Dystrophy
NCDs	Non-Communicable Diseases
PLRD	People Living with Rare Diseases
SADC	Southern Africa Development Cooperation
SSA	Sub-Saharan Africa

Declarations

Ethics approval and consent to participate: Not applicable

Consent for publication: Not applicable

Availability of data and materials: Not applicable

Competing interests: None

Funding: None

Authors' contributions: All authors participated in brainstorming ideas. ABD initiated the first draft, and all authors improved and polished it to produce the final version. All authors have approved the manuscript for submission.

Acknowledgements

We are grateful to the communities of rare disease patients who have shared their experiences of dealing with rare diseases in the Southern Africa region.

Appendices

Appendix 1: The SADC rare diseases coalition

We are a group of patient advocacy groups, human genetics researchers and regional youth health activists made up of the following organizations:

SADC Youth Forum (SAYoF)

The Southern Africa Youth Forum (SAYoF) is a regional development platform for youth in the SADC region. SAYoF works with the SADC Secretariat, among other regional organizations, to empower young people and ensure sustainable inclusion. SAYoF is the official convener of the SADC Youth Forum and SADC Youth Parliament, which are the largest youth gatherings in Southern Africa, aimed at co-creating solutions for youth and regional development. SAYoF was founded based on the increasing demand by young people to be important players and stakeholders in regional and national processes, provide solutions at national and regional levels, and participate in sustainable transformation. Through its Health Cluster, SAYoF is committed to ensuring disability support, access to information, and social support for persons living with rare diseases. SAYoF is also dedicated to supporting the empowerment of rare disease advocates through knowledge exchanges, awareness raising, policy promotion, networking, international collaboration, and joint action.

Type of organization	Regional Organisation (SADC)
Years in operation	4 years
Contact person	Misheck Gondo
E-mail: info@sayof.org	Mobile: +263 77 534 9603

Tanzania Human Genetics Organisation (THGO) (Tanzania)

Tanzania Human Genetics Organization (THGO) is a dedicated Tanzanian human genetics advocacy organization with the mission of coordinating and enhancing human genetics research, services, and related activities in the community. Its goal is to generate knowledge and recommendations for the prevention, diagnosis, and treatment of genetic diseases and the promotion of health. THGO was conceptualized in 2017, inaugurated in 2019, and officially registered on 24 March 2021 as a non-profit organization. THGO strongly believes that if the underlying root causes of violence against children and adolescents are identified and addressed early, violence against children can be entirely preventable.

Type of organization	Professional Society
Years in operation	4 years
Contact person	Dr. Siana Nkya
E-mail: humangeneticstz@gmail.com	Mobile: +255 784 193 349

Ali Kimara Rare Disease Foundation (AKRDF) (Tanzania)

The Ali Kimara Rare Disease Foundation (AKRDF) is a registered charitable foundation in Tanzania whose purpose is to advocate for the inclusion of children living with rare diseases in health and education policies in Tanzania. The foundation is named after Ali Kimara, a 12-year-old boy born in Zanzibar who has been battling a rare disease since he was 2 years old. Ali lost his sister, Nasreen, to a rare disease at the age of 3. The painful experiences and exposure to the reality and challenges of living with a rare disease were the reasons behind the establishment of the foundation. AKRDF is a voice for children affected by rare diseases in Tanzania.

Type of organization	Patient Advocacy
Years in operation	7 years
Contact person	Sharifa Mbarak
E-mail: sharifambarak1@gmail.com	Mobile: +255 712 644 570

Rare Disease Lesotho Association (RDLA) (Lesotho)

The Rare Diseases Lesotho Association is a non-profit organization registered in October 2017 in accordance with the Societies Act of 1966 of Lesotho. The organization assists all patients affected by rare diseases in accessing treatment and support care to improve their health and quality of life. The intention is to facilitate and support advocacy and engagement among those with the ability to prevent, intervene, treat, and provide supportive care for patients and families affected by chronic and rare diseases in Lesotho.

Type of organization	Patient Advocacy
Years in operation	4 years
Contact person	Nthabeleng Ramoeli
E-mail: mramoeli@gmail.com	Mobile: +266 5388 5912

Child and Youth Care (CYC) (Zimbabwe)

Child and Youth Care (CYC) is a Zimbabwean-based Private Voluntary Organization (PVO) that empowers vulnerable and neglected children who are impacted by rare diseases. CYC endeavors to defend, champion, and advance the interests of people living with rare diseases in Zimbabwe (RDZ) and their caregivers in Zimbabwe to improve their quality of life. CYC is able to provide the much-needed intervention in supporting children living with RDZ by setting up different support groups and by making early diagnosis, treatment, and care more accessible with the help of local and international stakeholders.

Type of organization	Civil Society Organization (CSO)
Years in operation	7 years
Contact person	Trudy Nyakambangwe
E-mail: trudynyakambangwe@gmail.com	Mobile: +263 77 540 7510

Rare Diseases SA (RDSA) (South Africa)

Rare Diseases South Africa is a registered non-profit organization advocating for a better tomorrow for the 1 in 15 South Africans impacted by rare diseases and congenital disorders. This includes seeking greater recognition, support, improved health services, and an overall better quality of life. By connecting patients, families, and patient groups, bringing together stakeholders, and mobilizing the South African rare disease and genetics community, Rare Diseases South Africa strengthens the patient voice and shapes research, policies, and patient services.

Type of organization	Patient Advocacy
Years in operation	9 years
Contact person	Nomsa Dlamini
E-mail patients@rarediseases.co.za	Mobile: +27 72 476 7552

Botswana Organisation for Rare Diseases (BORDIS) (Botswana)

BORDIS is an NGO that was formed by a family having children with rare diseases. The challenges they went through while trying to access diagnosis and treatment inspired them to raise their voice so that other families going through the same can receive support. The mandate of BORDIS is to create an environment where rare disease patients are able to thrive. This is accomplished by building collaborations and networks in Botswana and globally in order to harness the necessary resources for accessing diagnosis, treatment, and management of rare disease patients in Botswana.

Type of organization	Patient Advocacy
Years in operation	7 years
Contact person	Eda Selebatso
E-mail: rareconnectbotswana@gmail.com	Mobile: +267 71 692 565

Multiple Sclerosis Namibia (MS NAMIBIA) (Namibia)

Multiple Sclerosis Namibia is a registered welfare organization founded by a Multiple Sclerosis (MS) sufferer in July 2007. MS Namibia operates under the auspices of the Ministry of Health and Social Services. Since its establishment, MS Namibia's main priorities have been to raise awareness of the disease, make contact with, and support Namibians suffering from Multiple Sclerosis. MS Namibia is actively involved in fundraising, awareness campaigns, patient outreach, and support, including providing wheelchairs, therapy, and eventually medication. Multiple Sclerosis Namibia aims to support each and every person with MS in Namibia by being the voice for each patient, offering comfort to the lonely and isolated,

disseminating information, and facilitating connections between healthcare institutions.

Type of organization	Patient Advocacy
Years in operation	15 years
Contact person	Bianca Olivia Özcan
E-mail: info@msnamibia.org	Mobile: +264 81 142 3543

References

- Adachi, T., El-Hattab, A.W., Jain, R., Nogales Crespo, K.A., Quirland Lazo, C.I., Scarpa, M., Summar, M., Wattanasirichaigoon, D. (2023). Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges. *International Journal of Environmental Research and Public Health*, 20(6), 4732. https://doi.org/10.3390/ijerph20064732
- African Alliance for Rare Diseases. (n.d.). *Terms of reference for African Alliance for Rare Diseases*. Retrieved from https://africa-rare.org/wp-content/uploads/2016/10/Terms-of-Reference-Africa-Rare.org_.pdf (accessed on June 1, 2023).
- AU - African Union. (n.d.). *Africa Health Strategy 2016–2030*. Retrieved from https://au.int/sites/default/files/documents/24098-au_ahs_strategy_clean.pdf (accessed on June 1, 2023).
- Baynam, G.S., Groft, S., Van Der Westhuizen, F.H., Gassman, S.D., Du Plessis, K., Coles, E.P., Selebatso, E., Selebatso, M., Gaobinelwe, B., Selebatso, T., Joel, D., Llera, V.A., Vorster, B.C., Wuebbels, B., Djoudalbaye, B., Austin, C.P., Kumuthini, J., Forman, J., Kaufmann, P., Chipeta, J., Gavhed, D., Larsson, A., Stojiljkovic, M., Nordgren, A., Roldan, E.J.A., Taruscio, D., Wong-Rieger, D., Nowak, K., Bilkey, G.A., Easteal, S., Bowdin, S., Reichardt, J.K.V., Beltran, S., Kosaki, K., Van Karnebeek, C.D.M., Gong, M., Shuyang, Z., Mehrian-Shai, R., Adams, D.R., Puri, R.D., Zhang, F., Pachter, N., Muenke, M., Nellaker, C., Gahl, W.A., Cederroth, H., Broley, S., Schoonen, M., Boycott, K.M., Posada, M. (2020). A call for global action for rare diseases in Africa. *Nature Genetics*, 52, 21–26. https://doi.org/10.1038/s41588-019-0552-2
- Conradie, E.H., Malherbe, H., Hendriksz, C.J., Dercksen, M., Vorster, B.C. (2021). An Overview of Benefits and Challenges of Rare Disease Biobanking in Africa, Focusing on South Africa. *Biopreservation and Biobanking*, 19(2), 143–150. https://doi.org/10.1089/bio.2020.0108
- EJPRD - European Joint Programme on Rare Diseases. (2021).*African Summit on Rare Diseases*. Retrieved from https://www.ejprarediseases.org/event/first-african-summit-on-rare-diseases-2021/#:~:text=The%20Summit%20will%20bring%20together,%20December%201st%20%E2%80%93%203rd%2C%202021 (accessed on June 1, 2023).
- EURORDIS. (n.d.). What is a rare disease? *Rare Diseases Europe (EURORDIS)*. Retrieved from https://www.eurordis.org/information-support/what-is-a-rare-disease/ (accessed on May 28, 2023).
- FIN - Fabry International Network. (n.d.). *The African Summit on Rare Diseases*. Retrieved from https://www.fabrynetwork.org/what-is-fabry/ (accessed on June 1, 2023).
- Flynn, K., Feben, C., Lamola, L., Carstens, N., Krause, A., Lombard, Z., for DDD-Africa as members of the H3Africa

Consortium (2021). Ending a diagnostic odyssey—The first case of Takenouchi–Kosaki syndrome in an African patient. *Clinical Case Reports*, 9, 2144–2148. https://doi.org/10.1002/ccr3.3966

- GARD. (n.d.). About GARD. *Genetic and Rare Diseases (GARD)*. Retrieved from https://rarediseases.info.nih.gov/about (accessed on May 28, 2023).
- Gissen, P., Specchio, N., Olaye, A., Jain, M., Butt, T., Ghosh, W., Ruban-Fell, B., Griffiths, A., Camp, C., Sisic, Z., Schwering, C., Wibbeler, E., Trivisano, M., Lee, L., Nickel, M., Mortensen, A., Schulz, A. (2021). Investigating health-related quality of life in rare diseases: a case study in utility value determination for patients with CLN2 disease (neuronal ceroid lipofuscinosis type 2). *Orphanet Journal of Rare Diseases*, 16, 217. https://doi.org/10.1186/s13023-021-01829-x
- Gorini, F., Coi, A., Mezzasalma, L., Baldacci, S., Pierini, A., Santoro, M. (2021). Survival of patients with rare diseases: a population-based study in Tuscany (Italy). *Orphanet Journal of Rare Diseases*, 16, 275. https://doi.org/10.1186/s13023-021-01907-0
- Haendel, M., Vasilevsky, N., Unni, D., Bologa, C., Harris, N., Rehm, H., Hamosh, A., Baynam, G., Groza, T., McMurry, J., Dawkins, H., Rath, A., Thaxon, C., Bocci, G., Joachimiak, M.P., Köhler, S., Robinson, P.N., Mungall, C., Oprea, T.I. (2020). How many rare diseases are there? *Nature Reviews Drug Discovery*, 19, 77–78. https://doi.org/10.1038/d41573-019-00180-y
- Kaywanga, F., Alimohamed, M.Z., David, A.B., Maeda, D., Mbarak, S., Mavura, T., Nkya, S., Ishengoma, D.S. (2022). Rare diseases in Tanzania: a National Call for Action to address policy and urgent needs of individuals with rare diseases. *Orphanet Journal of Rare Diseases*, 17, 343. https://doi.org/10.1186/s13023-022-02498-0
- Khosla, N., Valdez, R. (2018). A compilation of national plans, policies and government actions for rare diseases in 23 countries. *International Rare Diseases Research Consortium (IRDR)*, 7, 213–222. https://doi.org/10.5582/irdr.2018.01085
- Krajnovic, D. (2012). Ethical and social aspects on rare diseases. *Filozofija i društvo*, 23, 32–48. https://doi.org/10.2298/FID1204032K
- Lamtey, G. (2023). Mwinyi wants health systems to include care for rare diseases. *The Citizen*.
- Landouré, G., Dembélé, K., Diarra, S., Cissé, L., Samassékou, O., Bocoum, A., Yalcouyé, A., Traoré, M., Fischbeck, K.H., Guinto, C.O. (2020). A novel variant in the spatacsin gene causing SPG11 in a Malian family. *Journal of the Neurological Sciences*, 411, 116675. https://doi.org/10.1016/j.jns.2020.116675
- Lumaka, A., Carstens, N., Devriendt, K., Krause, A., Kulohoma, B., Kumuthini, J., Mubungu, G., Mukisa, J., Nel, M., Olanrewaju, T.O., Lombard, Z., Landouré, G. (2022). Increasing African genomic data generation and sharing to resolve rare and undiagnosed diseases in Africa: a call-to-action by the H3Africa rare diseases working group. *Orphanet Journal of Rare Diseases*, 17, 230. https://doi.org/10.1186/s13023-022-02391-w
- Nguengang Wakap, S., Lambert, D.M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y., Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics*, 28, 165–173. https://doi.org/10.1038/s41431-019-0508-0
- ORPHANET. (2012). About Rare Diseases. *ORPHANET: The portal for rare diseases and orphan drugs*. Retrieved from https://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN (accessed on March 14, 2022).
- Owings, L. (2021). In-depth: What happens to people in SA who have rare diseases? *Spotlight*. Retrieved from https://www.spotlightnsp.co.za/2021/09/06/in-depth-what-happens-to-people-in-sa-who-have-rare-diseases/ (accessed on June 1, 2023).

- RDAI. (2021). Recognising rare diseases access and action. *Medical Academic*. Retrieved from https://www.medicalacademic.co.za/uncategorised/recognising-rare-diseases-access-and-action/ (accessed on May 28, 2023).
- RDI. (n.d.). Program for the African summit on rare diseases. *Rare Diseases International*. Retrieved from https://www.rarediseasesinternational.org/wp-content/uploads/2021/10/Program-Outline-29-October-2021-for-circulation.pdf?utm_source=Africa+Rare+Disease+Summit&utm_medium=agenda (accessed on June 1, 2023).
- RDSA. (n.d.). About Rare Diseases South Africa (RDSA). *Rare Diseases South Africa (RDSA)*. Retrieved from https://www.rarediseases.co.za/our-purpose (accessed on May 27, 2023).
- Sevittz, H., Laher, F., Varughese, S.T., Nel, M., McMaster, A., Jacobson, B.F. (2022). Baseline characteristics of 32 patients with Gaucher disease who were treated with imiglucerase: South African data from the International Collaborative Gaucher Group (ICGG) Gaucher Registry. *South African Medical Journal*, 112, 13518.
- URT. (2021). Ministry of Education, Science and Technology: national strategy for inclusive education 2021/22 - 25/26.
- URT. (2018). The Tanzania food, drugs and cosmetics (orphan medicines) regulations. Retrieved from https://www.tmda.go.tz/uploads/publications/en1581793767-GN%20412%20TFDA%20(ORPHAN%20MEDICINES)%20REGULATIONS,%202018.pdf (accessed on June 1, 2023).