Facilitating a framework for managing rare diseases in South Africa: Comparative insights from the UK and Italy

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Managing and treating rare diseases is a global concern, largely because of their rarity and the complexities associated with their care. Robust ethico-legal frameworks governing rare diseases are essential as they establish coordinated guidelines that ensure patients receive the care and support which they require. This article examines and compares the existing frameworks for rare diseases in the United Kingdom and Italy, with those in South Africa (SA), highlighting the differences and similarities in how they operate, towards facilitating a national framework that suits our context. The recommendations offer proposed solutions to add to the ongoing dialogue regarding the current lack of governance for rare diseases in SA. The article further calls for a coherent national framework to better address and represent the needs and rights of individuals affected by rare diseases. SA requires a systematic approach, with clear guidelines for diagnosing, treating and funding rare diseases. This approach would establish a basis for efficient resource allocation and equitable access to essential treatments.

Keywords. Rare diseases prescribed minimum benefits, orphan drugs, medical law, healthcare, medicine, national health insurance, human rights, access to healthcare.

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In the context of global healthcare, rare diseases are fast becoming a complex issue for communities to manage, both internationally and in South Africa (SA).[1] By definition, rare diseases affect a small percentage of the population, adding complexity to their diagnosis and treatment. South Africa has adopted the European Organisation for Rare Diseases' (EURORDIS) definition of a rare disease, which serves as a widely recognised reference for rare diseases definitions, classifying a condition as rare when it individually affects 1 in 2 000 people.[2] The World Health Organization (WHO) recognises an estimated 5 000 - 8 000 known rare diseases affecting as many as 400 million people worldwide, which amounts to approximately 3.5 - 5.9% of the world's population.[3] In SA, an estimated 1 in 15 people will likely experience a rare disease at some point during their lifetime,[4] with 50% to 70% of those affected being children.[5] Many people who have rare diseases often face their challenges alone without adequate support. As the number of successfully diagnosed rare diseases remains low, very little attention is devoted to them, further limiting funding for research, and delaying the development of new drugs.[6]

Rare diseases are seldom considered in healthcare planning and policy-making in SA, as policymakers prioritise more prevalent health issues that affect a larger proportion of the population, such as HIV/AIDS and diabetes.[7] In addition, the reimbursement of rare diseases is complex and involves both private and public healthcare stakeholders. Only 18% of the SA population has access to private healthcare, while the majority 82% who cannot afford to pay for private healthcare, fall within the public health sector.[8] While rare diseases should be covered in full under the prescribed minimum benefits (PMBs), this often does not materialise. Matters are worse in the public sector as resources are limited despite the government's responsibility, under Section 27 of the Constitution of the Republic of SA 1996 (the Constitution), to take reasonable legislative and other measures, within its available resources, to achieve the progressive realisation of the right to have access to healthcare services. Owing to a small number of rare diseases being diagnosed, medication for these rare diseases in the public health sector is not always accessible, and rare diseases are often completely disregarded, resulting in an unequal allocation of resources in the healthcare system.[7]

The purpose of this article is to explore possible policy solutions to increase access to orphan drugs (drugs that are not developed by pharmaceutical companies because of economic factors, even though these drugs meet public health needs)[9] and treatments, in accordance with the healthcare rights guaranteed by the Constitution and the WHO. As such, the UK and Italy serve as useful comparisons for the management of rare diseases in SA, given their well-developed policies, tailored methods for reimbursing rare disease treatments, their commitment to promoting patients' rights to access these treatments as well as their distinctive healthcare systems. The comparison between these countries can generate realistic strategies for SA to adopt, by prioritising affordability and equitable access to rare disease treatment.

This article provides a comparative analysis of academic literature, national plans, legislation and policies from SA, the UK and Italy,

compiling the similarities and differences between their varying approaches to rare diseases. The article further examines the inadequate provisions for rare diseases in SA's governance framework and aims to generate recommendations for a more equitable system, amid the newly enacted National Health Insurance Act 20 of 2023 (NHIA).

Comparative analysis of governance frameworks for rare diseases in SA, the United Kingdom and Italy

Access to healthcare in SA is shaped by the country's dual healthcare system and encompasses both public and private sectors. The Constitution's Bill of Rights declares access to healthcare services a fundamental right in sections 27(1) and (2) and directs the government to take the necessary steps within its resources to realise this right. In support of these provisions, the National Health Act 61 of 2003 (NHA), and the Patients' Rights Charter[10] aim to progressively achieve this right by building a unified health system for the nation.

The majority of the population is served by the public sector, which is under-resourced and encounters challenges such as long waiting times, staff shortages and outdated equipment.[11] On the other hand, the private sector offers high-quality care with advanced facilities to those who can afford health insurance. The Medical Schemes Act No. 131 of 1998 (MSA) seeks to protect the interests of medical scheme beneficiaries and makes specific minimum healthcare services available to them, listed as PMBs and are accessible with full reimbursement, and no co-payment or deductions, regardless of their chosen healthcare plan.[12] Although the purpose of PMBs is to provide continuous care that improves patients' health and well-being while making healthcare more affordable,[12] patients still undergo delays in treatment or are often left without access to treatment when their claims are disregarded by healthcare funders, even for treatments that should be covered under PMBs.[13]

Limited government focus creates barriers in regulating rare diseases, impairing access to healthcare and treatment opportunities. Currently, the treatment of rare diseases is determined on a case-by-case basis, with the MSA directing which costs and healthcare services are included as PMBs.

The NHIA came into effect on 15 May 2024 to promote universal access to healthcare across SA. Despite government efforts to implement the National Health Insurance (NHI) in accordance with the NHIA's objectives, the healthcare system is still struggling with inequality, inefficiency, corruption and the burden of other communicable diseases such as HIV/AIDS and non-communicable diseases such as diabetes.[14] Over recent years, rare diseases have gained increased recognition which has prompted efforts by government and non-governmental organisations to boost awareness, research and policy development. Several experts believe that the NHI could achieve these objectives in the future;[8] however, attention should be given to the prevailing gaps in the healthcare system.

The integration of rare disease management into the broader healthcare system is still evolving, with prominent regional inequalities across provinces, influencing the availability and quality of care. Financial shortcomings in the healthcare system also impact which conditions are prioritised. Considerable budget cuts, nearing a billion Rand, have forced the National Department of Health,(NDoH) to evaluate which conditions should be included in the NHI treatment plan.[15]

A proposal – the Rare Diseases Framework document (RDF document) – has been presented to government to facilitate the availability and affordability of treatments for rare disease patients through the collaborative efforts of the non-profit organisation, Rare Diseases SA (RDSA), and the Rare Diseases Access Initiative (RDAI).[16] The RDF document advocates for collaboration among the government and main organs of state to develop and implement a policy framework for rare diseases. According to RDSA, RDAI seeks to cultivate a fair and equitable environment for patients that incorporates both the private and public sectors and that aligns SA with other approaches like the UK, EU and BRICS.[17]

By working together, the NDoH, pharmaceutical organisations, and other relevant entities can develop practical and affordable strategies for rare diseases without straining the healthcare budget.

United Kingdom

In the United Kingdom (UK), rare diseases are managed under the National Health Service (NHS), which provides access to healthcare for all persons in the UK. The NHS' strong funding mechanism creates a reliable system for the reimbursement of rare diseases.[18]

Healthcare in the UK has been partially decentralised since the late 1990s, with England, Wales, Scotland and Northern Ireland establishing individual NHS administrations, enabling them to set their own healthcare policies and goals.[19] Accompanying the NHS is the National Institute for Health and Care Excellence (NICE), an independent body responsible for defining national guidelines and standards for healthcare services in the UK.[20] NICE evaluates the best quality of healthcare services, treatments and interventions, and ensures that they are cost-effective and safe; as such, the NHS often relies on its recommendations to determine whether particular treatments, drugs or healthcare services should be publicly funded or included in medical insurance coverage. Empowered by the Health and Social Care Act, 2012,[21] NICE can evaluate high-cost rare disease therapies independently to establish a 'robust, independent and transparent' system for procuring orphan drugs.[22]

As a party to the International Covenant on Economic, Social and Cultural Rights (ICESCR),[23] the UK is obliged to progressively promote the right to health, alongside its domestic human rights law. The Human Rights Act incorporates the rights from the European Convention on Human Rights (ECHR) into UK law, outlining the essential rights and freedoms available to everyone in the country.[24] As such, the UK government identified the need to address the challenges that rare disease patients encounter, together with their families, and consequently established a national strategy in 2013, the UK Strategy for Rare Diseases, prioritising their wellbeing.[25] This strategy facilitates access to the latest treatments and promotes strong coordination throughout the healthcare system. It focuses on identifying and preventing rare diseases, providing early diagnosis and intervention, establishing specialised treatment centres and executing controlled processes for the development and reimbursement of orphan drugs.

The UK has taken an active approach to creating awareness of rare diseases and has implemented a series of national guidelines and policies for their management. In 2021, the UK Rare Diseases Framework (the Framework) was introduced, building on the duties outlined in the UK Strategy. The Framework was followed by the release of the Rare Diseases Action Plan in 2024, marking England's third action plan addressing rare diseases.[26]

Despite ongoing challenges, the UK continues to focus on supporting individuals affected by rare diseases by formulating clear and organised processes for access to essential healthcare and by fostering collaboration among healthcare professionals, researchers and patient advocacy groups.

Italy

Considering Italy's membership of the European Union (EU), it is necessary to outline the European approach to provide context to its role in the management of rare diseases. The EU has assumed a unified approach to managing rare diseases, with all member states adopting a common definition of rare disease, (less than 5 cases per 10 000 individuals), based on the proportion of cases in their populations. Their efforts are guided by international legislation, starting with the Orphan Medicinal Product Regulation (EC) No. 141/2000 (1999), which promotes research on rare diseases and supports the development of orphan drugs, followed by EC Regulation 847/2000, which sets out the criteria that classify a medicinal product as an orphan medicinal product. Another EU development in rare diseases is the European Project for Rare Diseases National Plan (EUROPLAN), designed to facilitate the development of national plans across EU member states.

Italy predominantly operates under a public healthcare system and is managed by the National Health Service (Servizio Sanitario Nazionale, SSN), affording universal healthcare services to all citizens and legal residents.[27] Only an estimated 15% of Italians use private healthcare insurance for specialised private hospital services such as minor surgical procedures and obstetrics.[28]

Italy's decentralised healthcare system is organised into national, regional and local tiers.[29] Within this structure, the Agenzia Italiana del Farmaco (AIFA) oversees the regulation of drugs in the Italian market and rare disease policies. Each region maintains its autonomy, with national guidelines and policies integrated regionally, meaning that access to treatments for rare diseases and implementation of policies vary depending on the region's resources and healthcare infrastructure.

The legal framework for rare diseases in Italy is outlined in Ministerial Decree No. 279 of May 2001, introducing regulations for the formation of a national network for rare diseases, furnishing guidelines for exemptions from costs associated with healthcare services. On 27 November 2021, Italy published the Consolidated Law, Law No. 175/2021, to protect the health rights of persons with rare diseases in the country. This law supports the SSN's objective for access to universal healthcare services and orphan drugs among all the regions and is dedicated to strengthening the national network for disease surveillance, diagnosis and treatment.[30] Early access to orphan drugs is possible with this law, securing funding for treatments, including those offered internationally but not yet available in Italy. Furthermore, the Italian legal framework has consistently supported the development and testing of orphan drugs and introduced the Balduzzi Law,[31] to speed up their availability at the regional level.

Aligned with EUROPLAN's objectives, Italy established different national plans as part of its public health strategy, detailing its priorities, actions, budget and timeline for implementation. For instance, the National Prevention Plan (NPP) was introduced to provide a structured approach for passing health policies that focus on health promotion, disease prevention, and evidence-based interventions that achieve superior health standards.[27]

The National Health Plan (NHP) is the foundation for Italy's national health strategy. It is reviewed every three years, and each region adapts it into its regional prevention plan, with the NPP being a vital component of public health strategy. Additionally, Italy adopted its first National Plan for Rare Diseases from 2013 to 2016,[32] followed by a new National Rare Disease Plan (NRDP), for 2023 - 2026, reinforcing its leadership within the EU in rare disease regulation.[33]

The Italian NRDP supports individuals living with rare diseases and their families, by promoting equitable healthcare through advanced diagnosis, prevention, treatment and rehabilitation. Various nationwide complementary initiatives have also followed from this plan to address their needs, such as the National Centre for Rare Diseases, the Rare Diseases Unit and the Italian Undiagnosed Rare Disease Network.

Table 1 summarises the status of the legal and policy frameworks and implementation plans within SA, the UK and Italy, identifying common practices and differences.

Discussion

The above comparative analysis reveals that despite RDSA and RDAI's efforts for policy advancements and increased access to care, SA is still in the stages of infancy with respect to developing a rare disease framework as it struggles with resource limitations and systemic challenges. As it stands, without a clear framework, patients with rare disease have little to no prospect of success in accessing coordinated care, and are in some cases forced to turn to the courts for a determination[34] – a time consuming and costly exercise.

In its three decades of democracy, SA has made substantial progress in promoting its population's healthcare needs; however, it continues to encounter various challenges that burden the healthcare system, particularly the public sector. These challenges include the burden of diseases such as HIV, tuberculosis, diabetes and cancer, along with inadequate record keeping, the shortage of human and

Table 1. Rare disease legal and policy frameworks and implementation plans in SA, the UK and Italy

Category	SA	UK	Italy
Legal frameworks	Fragmented, emerging regulations	Centralised, NICE, national guidelines	Decentralised, AIFA, regional differences
Policy frameworks	Emerging integration, regional disparities across provinces	Integrated within NHS, specific pathways	Regional integration, variability in access
Implementation plans	Limited resources, evolving support structures	Strong national funding, established pathways	Variable regional resources and access

NICE = National Institute for Health and Care Excellence; AIFA = Agenzia Italiana del Farmaco; NHS = National Health Service.

medical resources, and poor service delivery,[14] all of which impede the progressive realisation of the right to healthcare.

On the other hand, the UK and Italy displayed more progressive legislative approaches to rare diseases. For instance, both the UK and Italy have developed national plans, laws, frameworks and strategies for these disorders. The UK's healthcare system is supported by the NHS and includes dedicated rare disease services, and a well-defined strategy for their treatment by way of its national plan and framework.

Italy's forward thinking in keeping up with its healthcare demands and in creating regulations and policies for rare diseases places it among the forefront member states in the EU.[35] Its framework for rare diseases is based on multiple measures and national and regional collaborative networks to deliver high-quality treatments. Ministerial Decree No. 279 and the Italian National Rare Diseases Plan are pivotal in driving policy improvements for rare disease care.

Consequently, each country's framework is showcased within its specific healthcare context, suggesting that the most successful rare disease care strategies may emerge from integrating national leadership with regional flexibility.

Recommendations

It is submitted that SA can overcome its challenges in the lack of governance for rare diseases, by adopting the following recommendations:

1. A national policy focused on rare diseases should be developed to achieve universal healthcare for patients with rare diseases. RDAI's RDF document is a starting point; however, it needs to be revised and brought in line with international models such as the UK's and Italy's rare disease plans and frameworks. Government's approval and support is crucial to finalise and implement this framework that would streamline diagnosis, treatment and funding, thereby improving patient access to healthcare and ensuring equitable care.
2. Securing funding for the treatment and management of rare diseases can pave the way for access to better treatments and promote scientific and medical innovations that give rise to speedier and more accurate diagnoses.
3. To gain a better understanding of rare diseases, a rare disease registry should be created to collect and evaluate patient data, monitor health outcomes and ascertain the effectiveness of treatments.
4. Setting up designated centres of excellence would offer patients individual, specialised care and training to healthcare professionals, improving access to treatment and clinical research in SA.
5. Equitable access to orphan drugs should become a priority, ensuring that rare disease patients receive the same healthcare rights as those with common diseases.
6. Support structures like the RDSA rely on government's participation in making a difference in the lives of vulnerable groups in society. Increased support from government can empower patient advocacy groups to achieve their objectives and influence healthcare policies that reflect patients' needs.
7. SA stands to benefit from engaging in international collaboration by analysing how countries such as the UK and Italy handle rare diseases, to strengthen its own healthcare strategies for rare diseases.
8. Implementing Health Technology Assessment (HTA) for rare diseases could optimise the allocation of resources and lead to affordable treatments.
9. Training of healthcare professionals in managing rare diseases can facilitate earlier diagnosis and reduce the strain on the healthcare system.
10. Ethical overview by ethical regulatory bodies would reinforce fair access to healthcare founded on Constitutional values and principles.
11. Investing in local manufacturing and research allows resource-limited countries like SA to rely less on costly imported drugs.

Conclusion

For SA to cultivate a more inclusive healthcare setting for rare diseases, it is vital that the implementation of a policy framework dedicated to rare diseases is consistent with the legal principles and standards established by national laws in the country. Data from the comparative jurisdictions above suggest that the starting point to recognising the challenges faced with the reimbursement of medical expenses for rare diseases, is to establish sound legal and policy frameworks that address these challenges as well as the needs of those affected by them. Without a reliable funding system such as the UK's NHS and Italy's SSN, SA will continue to grapple with access to healthcare. Furthermore, a strong influence from ethical regulatory bodies and patient advocacy groups can offer support to rare disease patients through their unpredictable healthcare journeys and accelerate the need for improved access to healthcare.

It is anticipated that the phased rollout of the NHI will unlock the potential to incorporate rare diseases in its healthcare packages, which will in turn inspire a sense of optimism in future developments and reinforce its commitment to providing universal healthcare coverage for all South Africans – specifically in this context to people suffering from rare diseases. International collaboration can further advance the NHI's blueprint for rare diseases.

This article highlights the crucial need to implement a coherent framework that assists in strengthening the rights of healthcare users by promoting fair access to healthcare services, protecting their interests and building a more transparent healthcare system.

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