Centres of Excellence: A Framework for Healthcare and Rare Diseases

Introduction

Centres of Excellence (CoE) in the healthcare context are specialised programmes within healthcare institutions that address the complex needs of specific medical fields. For rare diseases, which are often underdiagnosed, underreported and undertreated, CoE can serve as pivotal hubs for diagnosis, care, research, and innovation. Many rare diseases have a genetic basis, necessitating relevant expertise in genomic medicine, genetic testing, interpretation and counselling. Most rare conditions impact multiple organ systems, requiring coordinated, multidisciplinary clinical care and psychosocial support. CoE offer an interdisciplinary approach, integrating specialists, resources, care and management to achieve the best patient outcomes. While there is no single standardised definition of a CoE globally, there are widely accepted concepts that guide their establishment, and understanding these is essential as we work towards integrating CoE into South Africa's rare disease framework.

The aim of this document is to provide an overview of CoE, their potential role in addressing the unique needs of rare diseases in South Africa and recommendations for a way forward.

Rare Diseases in South Africa

Rare diseases are defined as conditions that impact ≤1 in 2,000 people, and with over 6000 rare diseases characterised to date globally, this diverse group of conditions represents a significant health burden (1, 2). In South Africa rare diseases are estimated to collectively impact >4 million South Africans, yet they are often neglected due to limited awareness, funding, and resources (1). The majority of rare diseases do not have an approved treatment. These conditions, sometimes called orphan diseases, demand specialised care frameworks integrating expertise, innovative treatments, and supportive policies.

What Problems Do Centres of Excellence Address?

Healthcare delivery for rare diseases poses numerous challenges. These include:

Challenges in Rare Disease Care

1. **Diagnostic Delays**: Patients with rare diseases often wait >5 years (3) for a diagnosis due to limited awareness, capacity and expertise. Many rare diseases are misdiagnosed and/or remain undiagnosed due to a lack of access to relevant clinical expertise and associated testing infrastructure, support, and counselling services.

2. **Fragmented Care**: The multidisciplinary nature of most rare diseases means that treatment often requires repeat visits to multiple specialists, creating logistical and financial burdens, particularly for affected families.

3. **Limited Access to Treatments**: Some rare disease therapies are expensive ("high-cost") and often remain inaccessible to patients. Other conditions can be managed by strict dietary controls and low-cost treatments, which may also have limited accessibility.

4. **Geographic Barriers**: Patients in remote areas outside major urban academic settings struggle to access relevant specialists and advanced care facilities.

5. **Insufficient Research**: Rare diseases lack the empiric data and funding needed to drive meaningful research and innovation, particularly in South Africa where communicable diseases have historically taken precedence.

How do CoE address these challenges?

1. **Centralised Expertise**: CoE bring together multidisciplinary teams to offer comprehensive care under one roof, facilitating referrals and easing the logistical and financial burden for the patient. While this provides the highest level of care it will also service a small proportion of South Africans. The way forward is a compromise between care and coverage and requires innovative approaches, including hybrid/virtual CoE for optimal, affordable coverage.

2. **Integrated Care Coordination**: Patients can benefit from seamless collaboration among healthcare providers, saving time and resources.

6. **Facilitation of Research**: CoE are hubs for clinical trials, data collection, innovation for diagnostics and therapeutics. Establishing biobanks, genomic databases and patient registries will enhance research opportunities and improve understanding of rare disorders in South African populations.

3. **Improved Access**: Economies of scale and centralised procurement reduce costs and improve access to medications, infrastructure and equipment.

4. **Support for Patients and Families**: CoE offer comprehensive psychosocial care including genetic counselling, education, and patient support networks, reducing the isolation many rare diseases patients and their families experience.

5. **Strengthening public-private partnerships:** To leverage private sector expertise and resources to improve access, efficiency, and innovation in a sector often underserved by public resources, addressing challenges in diagnosis, treatment, and research.

What Are Centres of Excellence?

Centres of Excellence (CoE) are defined as integrated practice units that combine specialised care, interdisciplinary teams, and advanced resources to deliver superior patient outcomes. They are particularly valuable in rare disease management due to their ability to centralise expertise and provide comprehensive, patient-centred care (4).

In South Africa the National Research Foundation (NRF) and Department of Science and Innovation (DSI) funds a specific type of CoE. While CoE in healthcare primarily enhance clinical care and treatment in specialised medical fields, the NRF/DSI funded CoE are focused on advancing research and development across various scientific disciplines. Both play crucial roles in their respective areas but are distinct in their focus and objectives – meaning NRF/DSI funded CoE are not an appropriate vehicle for rare diseases in the country.

Within this general context of CoE, there are many examples of different types of CoE for rare diseases globally, which vary in scope and function, from which key common elements can be identified.

Global Examples of CoE

1. Collaborative Global Network for Rare Diseases

The Collaborative Global Network for Rare Diseases (GNRD) is a Rare Disease International (RDI) initiative that began in 2020 in collaboration with the World Health Organization (WHO) (https://www.rarediseasesinternational.org/collaborative-global-network/). The GNRD aims to engage and support the rare disease community to develop a person-centred, global network of care and expertise for all rare disease patients.

The GNRD vision is an evolved, multidisciplinary model of care to address rare diseases. This "networked care" model proposes that expertise can be accessed from across a national, regional and global network of experts to inform care locally, (similar approach to the European Reference Network https://www.ern-rnd.eu/). Developing such an international network will harness digital technologies and advances to drive peer-to-peer learning and the sharing of knowledge, resources and capacities to strengthen health systems for rare diseases (5).

Following wide stakeholder consultation across all WHO regions to identify the global population needs, a framework has been developed for a GCRD. Components of the proposed Network include:

- **National Hubs** – nationally endorsed centres connecting under a national hub-and-spoke model.
- **Regional Hubs** – virtual multi-centre collaborative hubs of expert centres and patient organisations

- **Global Network** – a global learning system and knowledge-sharing network, informing global public health action and leveraging technology to extend care for rare diseases around the world.

While the development of the GCRD is ongoing, it offers an opportunity for South Africa to address these issues internally to designate and develop relevant geographical and specific disease CoE to ensure readiness for alignment with the GNRD.

2. ***EURORDIS Centres of Expertise (Europe)***

EURORDIS CoE are designed to provide specialised care and research for rare diseases. (https://www.eurordis.org/) They focus on:

- Multidisciplinary collaboration and cross-border partnerships.
- Disease registries to monitor outcomes and guide research.
- Integration of social and paramedical services into patient care.

3. **NORD Rare Disease Centres of Excellence (USA)**

The National Organisation for Rare Disorders (NORD) program (https://rarediseases.org/) focuses on:

- Reducing diagnostic delays.
- Expanding multisite clinical trials.
- Training new specialists in rare diseases.

4. **Murdoch Children's Research Institute (Australia)**

This institute (https://www.mcri.edu.au/) emphasises:

- Accelerated diagnosis and care.
- Discovery of new treatments and diagnostic methods.
- Equitable access to services.

5. **Cystic Fibrosis Centre of Excellence (USA)**

Cystic Fibrosis (CF) is a complicated, multi-organ disease with a plethora of functional defects, although most of the morbidity and mortality associated with CF results from the progressive loss of lung function. The mission of the Centre for Cystic Fibrosis and Airways Disease Research (CF-AIR) at Children's Healthcare of Atlanta, USA (https://pedsresearch.org/centers/cf-air/cf-center-of-excellence/cf-center-of-excellence-mission) is:

- To build a comprehensive programme that will lead to new breakthroughs in CF research that will advance the quality and longevity of life for patients.
- Through collaboration, seeks to become the best comprehensive CF programme in the country, excellent in research, clinical care, and education.

Examples of Rare Disease Centres of Excellence in South Africa

In South Africa, outside of the NRF/DSI CoE context there are several centres focusing on rare diseases that may be considered as CoE. One example is the **Red Cross War**

Memorial Children's Hospital (RCWMCH) in Cape Town, which exemplifies a paediatric healthcare CoE, particularly in the management of rare diseases. As one of only a few dedicated child health institutions in South Africa, RCWMCH offers a comprehensive range of specialist paediatric services and serves as a pivotal training ground for specialist child health professionals.

Key Features of RCWMCH as a CoE:

- ***Specialised Services:*** RCWMCH provides multidisciplinary care for complex and rare paediatric conditions, including neurometabolic, neurodegenerative, and neuromuscular disorders. The hospital's neurology department manages large cohorts of children with conditions such as epilepsy and neurocutaneous conditions.
- ***Allied Healthcare:*** The hospital offers a comprehensive range of allied healthcare services relevant for rare disease patients, including genetic counselling and testing, physiotherapy, occupational therapy, speech and feeding therapy, social intervention, and educational placement.
- ***Training and Education:*** Affiliated with the University of Cape Town, RCWMCH is a premier centre for postgraduate specialist paediatric medical and surgical training in Africa. It offers extensive training programmes for healthcare professionals across the continent, ensuring the sustainability of specialised paediatric care.
- ***Research and Innovation:*** RCWMCH engages in groundbreaking research on childhood illnesses, contributing to global medical knowledge and the development of innovative treatments. This research focus enhances the quality of care provided to patients with rare diseases.
- ***Community Outreach:*** RCWMCH is active in paediatric outreach and support programmes across South Africa, extending its expertise to underserved areas and improving access to specialised care for rare diseases.

By integrating specialised clinical and rehabilitative services, comprehensive training, research initiatives and community outreach, RCWMCH addresses many of the multifaceted challenges associated with rare diseases.

The RCWMCH model demonstrates the critical role of academic institutions in establishing CoE, providing superior patient care, advancing medical research, ensuring the continuous education of healthcare providers and patient participation. This example underscores the potential for other institutions in South Africa to develop similar CoE that effectively tackle rare diseases, leveraging academic partnerships to enhance patient outcomes and foster innovation.

Why Academic Institutions for Rare Diseases in South Africa?

Academic institutions in South Africa are uniquely positioned to host CoE for rare diseases, offering a mutually beneficial arrangement for patients, healthcare providers, and institutions.

Advantages of Academic Institutions

Advantage	Description
Superior Patient Care	Access to specialists and advanced technologies improves outcomes for patients with rare diseases.
Research Opportunities	Academic environments foster the development of innovative treatments, expansion of genetic research, deepened disease understanding and patient-led research opportunities.
Training and Education	CoE serve as teaching grounds for future healthcare providers, ensuring sustainability of care. They also provide training opportunities for genetic counsellors, medical geneticists, and laboratory scientists to enhance genetic literacy among non-genetics healthcare professionals.
Specialist Retention	Institutions with CoE attract and retain top talent in medical specialties.
Economies of Scale	Centralised procurement lowers costs for infrastructure, equipment and treatments.

Key Characteristics of CoE for Rare Diseases

Feature	Description
Multidisciplinary Teams	Collaboration across medical, allied healthcare, social services and patient groups to address diverse patient needs.
Comprehensive Services	Offers diagnosis, including appropriate screening and testing, treatment, follow-up care and management, counselling services (genetic and psychosocial), rehabilitative care (physio, speech, and feeding therapy), palliative care and research participation.
Patient-Centred Approach	Tailors care to individual needs, ensuring equity, accessibility and compassion.
Continuous Improvement	Regular monitoring and evaluation of care quality and outcomes including patient feedback.

Detailed Criteria for CoE

Drawing from international frameworks and established best practice, CoE for rare diseases in South Africa should meet the following criteria:

Criterion	Description
Expert Teams	Skilled clinicians, genetic counsellors and allied health workers adhering to evidence-based guidelines.
Adequate Caseload	Sufficient patient volume to maintain expertise.
Integrated Care Coordination	Seamless scheduling and communication among specialists to optimise the patient experience.
Ongoing Research	Active participation in clinical trials, genetic studies, and biobank initiatives, and knowledge-sharing initiatives, involving the patient community.
Quality Monitoring	Continuous evaluation of care quality and outcomes from relevant stakeholder perspectives.
Patient Engagement	Ensure patients and families are involved in decision-making, have access to genetic counselling and testing and are referred to relevant patient and family support networks for psychosocial support.

Summary

To establish effective CoE for rare diseases in South Africa, the following actions are critical:

Action	Benefit
Develop Multidisciplinary Teams	Provides holistic care tailored to the complex needs of rare disease patients, including specialised genetic services.
Create Disease Registries	Enables tracking of disease prevalence and burden, outcomes, and care standards, including genetic data for better understanding of hereditary conditions.
Foster Research Collaboration	Advances understanding and treatment options for rare diseases, including the integration of genomic medicine and genetic research initiatives.

Action	Benefit
Adopt Global Standards	Aligns care delivery with international best practices.
Reduce Geographic Barriers	Facilitates outreach programmes and telemedicine to support remote populations.

Conclusion

CoE are pivotal in addressing the unmet needs of rare disease patients in the country. By leveraging the strengths of academic institutions, aligning with global and best standards and adapting them to the South African healthcare context, we can build a sustainable framework that enhances patient care, drives research, and ensures equitable access to resources.

A coordinated effort among government, academic institutions, genetic specialists and patient organisations is essential to realise this vision.

Recommendations

To ensure the effective establishment of CoE for rare diseases in South Africa, a situational analysis is required to understand what is currently available. An audit of existing centres/groups of expertise is recommended, including supporting capacity and infrastructure to identify strengths, gaps, and collaborative opportunities.

References

1. Wang C, Whiting A, Rath A, Anido R, Ardigò D, Baynam G, et al. Operational Description of Rare Diseases: A reference to improve the recognition and visibility of rare diseases (Forthcoming)*Orphanet J Rare Dis* 2024. Orphanet J of Rare Dis. 2024.
2. Wakap SN, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020;28(2):165-73. doi: https://doi.org/10.1038/s41431-019-0508-0.
3. The Lancet Global Health. The landscape for rare diseases in 2024. The Lancet Global Health. 2024;12.
4. Elrod JK, Fortenberry JL. Centers of excellence in healthcare institutions: what they are and how to assemble them. BMC health services research. 2017;17:15-24.
5. Rare Diseases International. Global Network for Rare Diseases 2023. Available from: https://www.rarediseasesinternational.org/collaborative-global-network/.

Version 21 May 2025 H. Malherbe