

The impact of COVID-19 on patients affected by rare diseases and congenital disorders in South Africa: A scoping review

M C M Gomes,¹ MSc (Med), BSc Hons ; H L Malherbe,^{2,3} MSc, PhD 

¹ Department of Clinical Medicine, Nelson R Mandela School of Medicine, College of Health Sciences, University of KwaZulu-Natal, Durban, South Africa

² Centre for Human Metabolomics, North-West University, Potchefstroom, South Africa

³ Rare Diseases South Africa, Bryanston, South Africa

Corresponding author: M C M Gomes (mariannecm.gomes@gmail.com)

Rare diseases (RDs) are individually rare but collectively common, affecting an estimated 1 in 15 individuals in South Africa (SA). Patients with an RD often face a long diagnostic odyssey (>5 years on average) and many obstacles in accessing healthcare. A scoping review was conducted to assess the impact of the COVID-19 pandemic on the SA RD community. Fourteen studies met the inclusion criteria and were explored using thematic analysis, which showed that RD patients were further marginalised during the pandemic, particularly in access to healthcare. Increased inclusivity in policy creation and integrated community-based healthcare are recommended to ensure that RD patients are not an afterthought in future crises.

Keywords: COVID-19, rare diseases, congenital disorders, disability, healthcare access, marginalisation, discrimination.

S Afr Med J 2024;114(9):e1795. <https://doi.org/10.7196/SAMJ.2024.v114i9.1795>

Rare diseases (RDs) affect between 263 and 446 million individuals globally.^[1] Recent global consensus has defined an RD as 'a medical condition with a specific pattern of clinical signs, symptoms, and findings that affect fewer than, or equal to, 1 in 2000 persons living in any World Health Organization-defined region of the world'.^[2] Most RDs have a genetic basis and are also considered congenital disorders (CDs), since they are present from birth.^[3] Patients with an RD face numerous challenges, including a prolonged diagnostic journey (>5 years on average) marked by multiple visits to healthcare professionals (HCPs) and frequent misdiagnoses, with up to 50% remaining undiagnosed.^[4] Many patients are reliant on ongoing research for potential diagnoses and treatment, and where treatments are available, they are often inaccessible. RD patients also require supportive health services, educational support, palliative care and psychosocial assistance.^[4]

Low- and middle-income countries (LMICs), especially in sub-Saharan Africa, face even greater challenges in addressing RDs than high-income countries. Diagnosis in LMICs is often delayed or non-existent owing to limited access to testing, insufficient infrastructure, inadequate genetic services, and restricted access to care and treatment.^[5,6]

In South Africa (SA), an estimated 1 in 15 individuals, or 4.2 million people, are affected by RDs (Helen Malherbe, Rare Diseases South Africa, personal communication). These patients face further hurdles owing to competing health priorities in the country. The HIV/AIDS and tuberculosis epidemics of the 1990s significantly hampered progress in reducing child mortality and increasing life expectancy at birth.^[7] The diversion of funding to these health crises resulted in neglect of the genetic services developed to address the burden of RDs/CDs.^[8] The dual health system in SA (<16% private, 84% state)^[9] continues to face the parallel burden of communicable

and non-communicable diseases, including RDs/CDs. However, resources remain largely directed towards infectious diseases.^[7,10]

The COVID-19 pandemic

The COVID-19 pandemic began in 2020 and spread rapidly across the globe, with nearly 7 million deaths reported over 3 years.^[11] The global pandemic response shifted the focus from essential healthcare services to emergency services, primarily related to COVID-19.^[12] Patients with RDs encountered sudden, unexpected barriers to essential healthcare.^[4,13,14] Funding for research, crucial for developing new RD treatments,^[13] was also redirected, with many clinical trials delayed or terminated.

In SA, the first COVID-19 patient was confirmed on 5 March 2020, and 10 days later, the government declared a national State of Disaster^[15] and established the National Coronavirus Command Centre.^[16] A strict national lockdown was implemented that severely restricted movement, allowing individuals to leave their homes only for essential work or to purchase essential items. Schools, public leisure activities, and international and provincial borders were closed, and gatherings were restricted, with stringent physical distancing rules, mandatory mask-wearing, and a curfew from 20h00 to 05h00. Public transport capacity and operating hours were limited and the sale of certain goods, including clothing, tobacco and alcohol, was prohibited. Working from home was implemented, unless the work was considered essential, with others requiring permits to leave their homes.^[17] As the pandemic persisted, the country moved through different lockdown levels, ranging from level 5 (most severe) to level 1.^[18]

Lockdown, while essential to control the spread of the virus, had severe consequences, including significant income loss for many, and limited access to an already under-resourced healthcare system.^[12,19] Non-

essential services were discontinued, and essential services were redirected towards critical care, to the detriment of acute care. Telemedicine was introduced to manage chronic patients, home-based services were limited or stopped altogether, and elective surgical procedures were cancelled.^[20]

While these restrictions affected all patients accessing the healthcare system, owing to the implicit additional challenges and inequity faced by RD/CD patients, the impact upon this community and those living with disability was of a much greater magnitude of scale.^[21]

Prior to the COVID-19 pandemic, RD/CD patients in SA already faced substantial inequalities in accessing relevant healthcare services.^[22] For RDs with available treatments (<5%), these are often limited and unaffordable owing to their high cost.^[22,23] The lack of appropriate capacity to recognise, test, diagnose and appropriately refer RD patients exacerbates the challenges faced by this community. The SA National Health Act 61 of 2003 provides for medical genetic services for the 'health needs of vulnerable groups, including children and people with disabilities.'^[24] However, since 2001, medical genetic services have declined as a result of competing health needs, leaving RD patients without access to care and management.^[24] The 2021 Clinical Guidelines for Genetics Services in SA suggest a renewed commitment to CDs and RDs as part of National Health Insurance (NHI), SA's funding approach to universal healthcare.

The objectives of this scoping review are twofold: (i) to review the available literature documenting the impact of the COVID-19 pandemic on the SA RD/CD community; and (ii) to propose recommendations to mitigate against future disruptions to healthcare for RD/CD patients.

Methods

This review follows the scoping review framework as outlined by Arksey and O'Malley^[25] and the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) guidelines.^[26]

The research question

The research question was developed: 'What was the impact of the COVID-19 pandemic on individuals with an RD residing in South Africa?'

Identification of relevant studies

A PubMed search was performed to identify peer-reviewed journal articles published between 2020 and 2022. PubMed was used as the largest freely available biomedical database. A Boolean string was created using key search terms/word combinations relating to synonyms of RD, CD and Mesh terms across the life course, COVID-19 and SA (Table 1). Peer-reviewed and grey literature were included. All articles and reports were downloaded and imported into the reference manager tool Mendeley.

Study selection

Inclusion criteria for the peer-reviewed and grey literature were: (i) articles in English; (ii) full text available; and (iii) focused on the impact of the COVID-19 pandemic on RD/CD patients in SA. Systematic reviews, book reviews, case reports and case studies, publicly available theses and dissertations were excluded.

The term 'rare disease' was taken to refer to any impairment, whether physical or intellectual, and 'congenital disorders' were defined as 'abnormalities in structure or function present from birth'.^[3,27] Duplicates were removed, and initial screening based on article title and abstract was followed by full-text review undertaken by both authors.

Charting the data

Quantitative analysis of the identified literature was undertaken by author MCMG using a data form to extract the relevant data from each article, including author/s, study title and study aim. Full-text articles were imported into NVivo software, version 12 (QRS International, USA),^[28] where they were read and coded by both authors.

Collating, summarising and reporting results

The coded data were developed into themes and a thematic/qualitative framework was developed.

Ethical approval to conduct this research was granted by the Biomedical Research Ethics Committee, University of KwaZulu-Natal (ref. no. BREC/00006584/2023).

Table 1. Boolean string used for literature search

Search #	PubMed Boolean string
#1	"Undiagnosed Disease"[Mesh] OR "Disease, Undiagnosed"[Mesh] OR "Orphan Disease"[Mesh] OR "Disease, Orphan"[Mesh] OR "Orphan Diseases"[Mesh] OR "Rare Disease"[Mesh] OR "Disease, Rare"[Mesh] OR "rare diseases"[Mesh] OR "rare disease*" [tiab] OR "orphan disease*" [tiab] OR "undiagnosed disease*" [tiab] OR "congenital defect*" [tiab] OR "congenital disorder*" [tiab] OR "congenital anomal*" [tiab] OR "congenital abnormalit*" [tiab] OR "congenital deformit*" [tiab] OR "congenital malformation*" [tiab] OR "congenital syndrome*" [tiab] OR "congenital dysmorphism*" [tiab] OR "congenital disease*" [tiab] OR "birth defect*" [tiab] OR "birth disorder*" [tiab] OR "birth anomal*" [tiab] OR "birth abnormalit*" [tiab] OR "birth deformit*" [tiab] OR "birth malformation*" [tiab] OR "birth syndrome*" [tiab] OR "birth dysmorphism*" [tiab] OR "birth disease*" [tiab] OR "genetic defect*" [tiab] OR "genetic disorder*" [tiab] OR "genetic anomal*" [tiab] OR "genetic abnormalit*" [tiab] OR "genetic deformit*" [tiab] OR "genetic malformation*" [tiab] OR "genetic syndrome*" [tiab] OR "genetic dysmorphism*" [tiab] OR "genetic disease*" [tiab] OR "fetal defect*" [tiab] OR "fetal disorder*" [tiab] OR "fetal anomal*" [tiab] OR "fetal abnormalit*" [tiab] OR "fetal deformit*" [tiab] OR "fetal malformation*" [tiab] OR "fetal syndrome*" [tiab] OR "fetal dysmorphism*" [tiab] OR "fetal disease*" [tiab] OR "foetal defect*" [tiab] OR "foetal disorder*" [tiab] OR "foetal anomal*" [tiab] OR "foetal abnormalit*" [tiab] OR "foetal deformit*" [tiab] OR "foetal malformation*" [tiab] OR "foetal syndrome*" [tiab] OR "foetal dysmorphism*" [tiab] OR "foetal disease*" [tiab] OR "single gene disorder*" [tiab] OR "disabilit*" [tiab] OR "inherited disease*" [tiab] OR "inherited condition*" [tiab] OR "inherited disorder*" [tiab]
#2	"Covid-19[Mesh]" OR "Covid-19[tiab]" OR "Covid19[tiab]" OR "SARS-CoV-2[tiab]" OR "SARS CoV 2"[tiab]
#3	"Republic of South Africa"[Mesh] OR "Union of South Africa"[Mesh] OR "South Africa"[Mesh] OR "south africa"[tiab]
#4	#1 AND #2 AND #3

Results

In total, 14 papers were included in the review: eight peer-reviewed/published articles and six grey literature (Fig. 1 and Table 2). The first authors of the majority of published articles (7/8, 88%) were from SA academic institutions: Stellenbosch University ($n=3/7$), the University of the Western Cape ($n=2/7$), and one each from the University of Johannesburg and North-West University.

A range of methodologies was found in the included articles (Table 3), with the majority using qualitative methodologies ($n=3$) or surveys ($n=6$). Of the three qualitative studies included, two used in-depth interview tools^[29,30] and the third used a reflective phenomenology study approach.^[31] The two mixed-method studies used a combination of interviews and survey instruments.^[32,33]

Discussion

The aim of this article was to review the impact of the COVID-19 pandemic on the SA RD/CD patient community and to recommend actions to mitigate against similar disruptions to healthcare in the future. The review included individuals with an RD/CD, and owing to the degree of disability associated with these conditions, also included those living with disability, although this was not included in the Boolean search terms. The literature indicated that these patients were further marginalised during the pandemic, particularly in terms of access to healthcare such as rehabilitation services, medication and mental health support, and access to information, education, basic needs and family support.

Of the articles included, almost half ($n=6/14$) were grey literature reports authored by international RD/CD or disability-related organisations. Of the peer-reviewed literature ($n=8/14$), the majority of primary authors were from SA academic institutions (two included international contributors) and most were published by academic institutions in Western Cape Province. There were no publications included from two of the largest academic institutions in SA, namely the University of the Witwatersrand and the University of KwaZulu-Natal. The relative contribution of grey literature, coupled with the lack of published studies on RDs/CDs in SA, reflects the lack of prioritisation of these issues in academic centres. Many articles were specifically related to persons living with disabilities ($n=10/14$; 71%) and not primarily RDs/CDs, although a proportion of congenital or acquired disability is associated with these conditions.

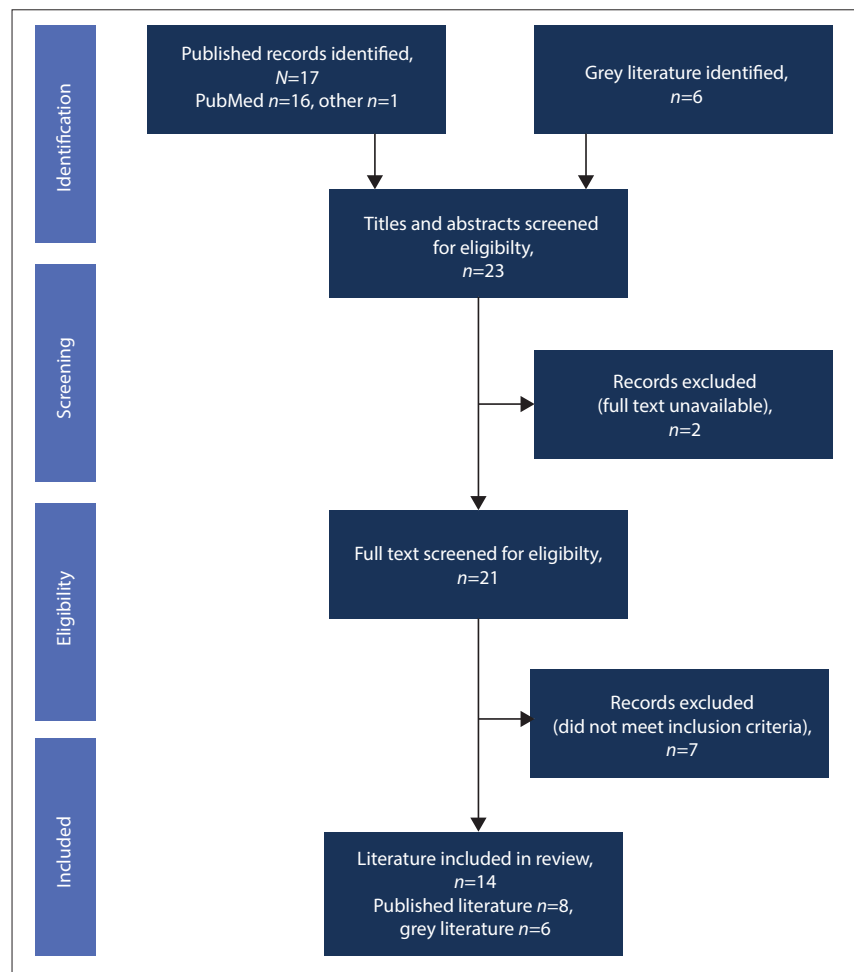


Fig. 1. Preferred Reporting Items for Systematic reviews and Meta-Analysis (PRISMA) flow diagram of scoping review.

The qualitative analysis undertaken provides a framework of key challenges, opportunities and recommendations identified in this scoping review (Fig. 2). A predominant and recurring theme was the increased marginalisation of RD/CD patients during the pandemic. Pre-existing barriers to care, whether to critical care, normal care or rehabilitation services, deteriorated further^[34] (Fig. 2). The themes identified are discussed in detail here, following the qualitative framework (Figs 2 and 3).

Challenges

While many of the challenges identified in this study were shared by other patient groups during the pandemic, there were also challenges unique to the RD/CD community. Collectively, these are:

Access challenges

Non-emergency and chronic care/management. For many RD/CD patients, timeous medical interventions are required to prevent secondary complications.^[33] However, health-

care services that are considered essential to RD/CD patients, such as rehabilitation services, were not classified as 'essential' during lockdown.^[31,33,35,36] Defining 'non-essential' appeared to be subjective and hospital specific,^[37] resulting in extended interruptions in therapies and postponed treatment. The closure of schools prevented children living with disabilities from accessing rehabilitative and therapeutic services that they received at school. Most parents/caregivers had limited resources to address this treatment deficit via other (private) means.^[34] Non-essential surgeries (e.g. paediatric surgery for cleft lip and/or palate that must be performed timeously for optimal outcomes) were also cancelled or postponed, and it is estimated that the backlog will take 2 years to catch up.^[38]

Mental health services. Patients with RDs/CDs lived in constant fear of contracting the virus and were concerned about accessing healthcare if they did contract it.^[31,33] Those in residential institutions were not allowed to be visited by their families for fear of contracting

Table 2. Thematic summary of articles included in scoping review

			Themes																					
			Challenges								Recommendations													
Author(s), year	Title	Literature type (study design)	Basic needs	Caregivers/residential care	Communication and information	Consumables, e.g. PPE	Cost/financial	COVID-19 testing and vaccination	Discrimination	Education and employment	Emergency access	Increased risk of infection	Medication and assistive devices	Mental health support	Non-emergency care (continuity)	Pre-existing inequality in care	Social-based protection	Transport	Education and awareness	Targeted research	Policy-making inclusivity	Financing mechanisms for NPOs	Integrated community-based healthcare	
Makuyana, 2022 ^[30]	Towards interventions on school dropouts for disabled learners amidst and post-COVID-19 pandemic	Peer reviewed (qualitative study: in-depth interviews and focus groups)	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
McKinney <i>et al.</i> , 2020 ^[36]	COVID-19, disability and the context of healthcare triage in South Africa: Notes in a time of pandemic	Rapid review	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
McKinney <i>et al.</i> , 2021 ^[34]	Access to healthcare for people with disabilities in South Africa: Bad at any time, worse during COVID-19?	Discussion paper	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Mupaku <i>et al.</i> , 2021 ^[29]	Transitioning to adulthood from residential childcare during COVID-19: Experiences of young people with intellectual disabilities and/or autism spectrum disorder in South Africa	Qualitative study of semi-structured interviews	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Roy <i>et al.</i> , 2022 ^[41]	Impact of COVID-19 on gender-based violence prevention and response services in Kenya, Uganda, Nigeria, and South Africa: A cross-sectional survey	Cross-sectional online survey	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Van Biljon and van Niekerk, 2022 ^[31]	Working in the time of COVID-19: Rehabilitation clinicians' reflections of working in Gauteng's public healthcare during the pandemic	Phenomenology study using critical reflection method	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
Ned <i>et al.</i> , 2021 ^[33]	Experiences of vulnerability of people with disabilities during COVID-19 in South Africa	Online survey and qualitative follow-up interviews	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
			...Continued																					

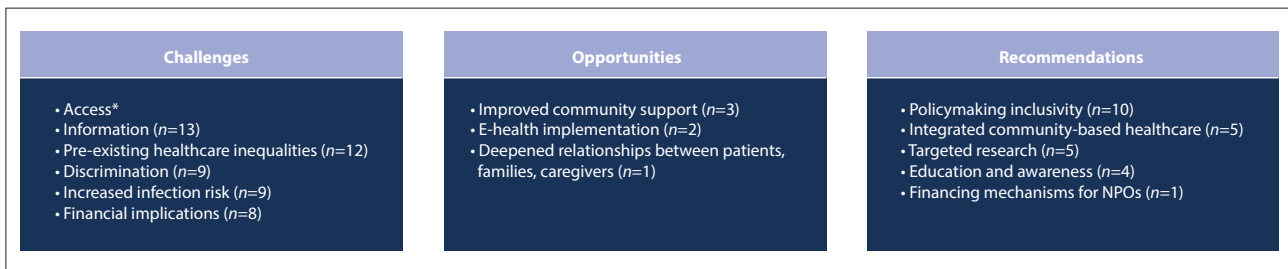


Fig. 2. Qualitative framework derived from thematic analysis of coding of included articles. (n = number of articles referring to the theme; NPO = non-profit organisation; *See Fig. 3 for access challenges.)

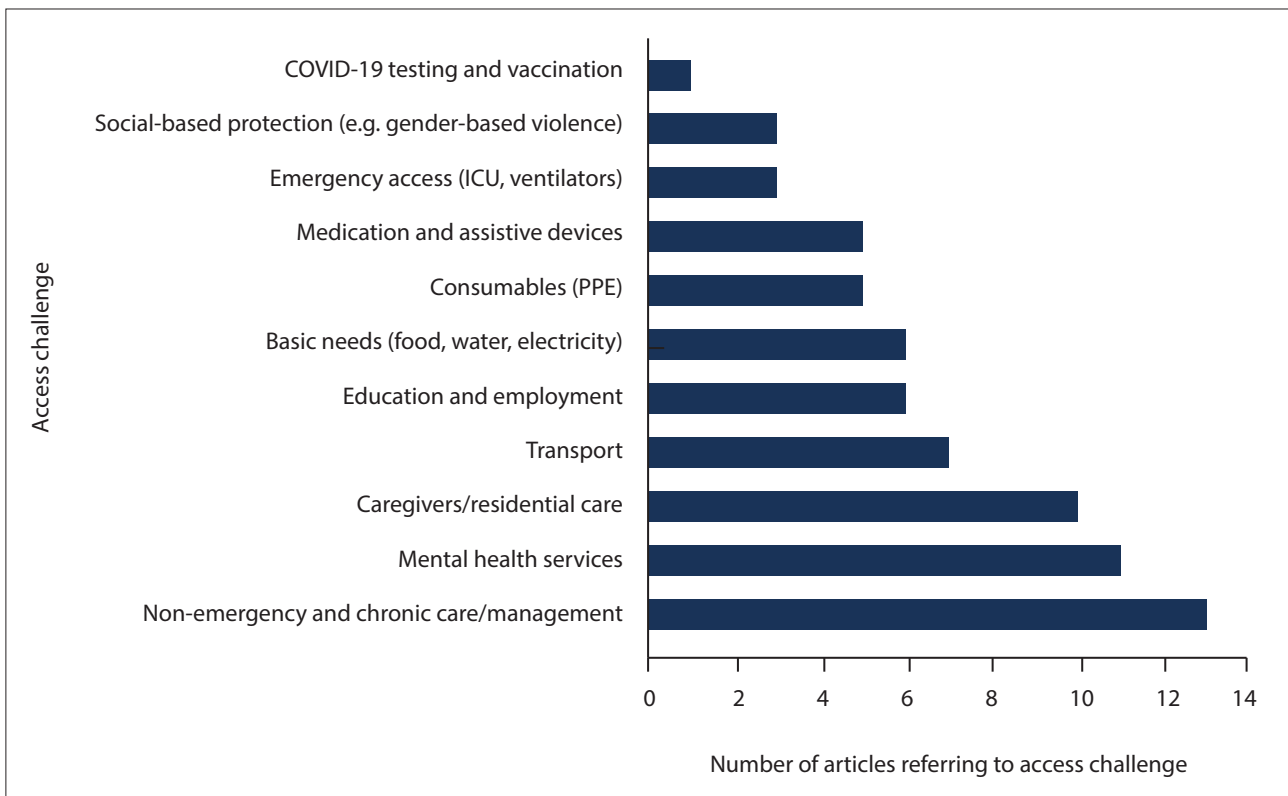


Fig. 3. Breakdown and ranking of access challenges identified through thematic analysis of articles. (ICU = intensive care unit; PPE = personal protective equipment.)

Table 3. Methodologies used in included articles

Type of study	n
Survey	6
Qualitative	3
Mixed methods	2
Review	2
Discussion paper	1
Total	14

the virus,^[36] leading to isolation and loneliness.^[16,21] Adolescents with intellectual disabilities transitioning towards adulthood experienced additional mental health challenges. The imposed lockdown kept them isolated, preventing them from developing independence.^[29] Those scheduled for transition from care homes and facilities to home-based care suffered from anxiety and depression as a result of continued postponement.^[16,29,34] Mental health challenges may also have contributed to the increased incidence of school dropout of children living with disabilities during the pandemic.^[30] Despite the

increased burden of mental health challenges, the number of clinics providing mental health services declined during the pandemic,^[34,35] resulting in widening of the pre-existing gap in access to mental health services for RD/CD patients.^[33]

Caregivers and residential care. Many RD/CD patients are highly dependent upon their caregivers. In SA, many caregivers are not provided, or financially supported, by the government and are instead paid out of pocket by the families.^[34] The risk of a caregiver contracting COVID-19 and having to quarantine, with no provision for a replacement caregiver, was a daily concern.^[34] Some care homes were forced to close, leaving RD/CD patients at home solely in the care of family members who may not have had the skills needed to care for them effectively.^[36] Caregivers are often the sole voice for RD/CD patients with communication challenges.^[16] For deaf and hard-of-hearing patients, when access to their caregivers was obstructed and sign language interpretation services were unavailable,^[36] communication with HCPs unable to use sign language was challenging.^[34] This situation was exacerbated by the mandatory wearing of medical masks, preventing deaf patients from lip-reading.

Transport. Transport challenges were exacerbated by the pandemic.^[33,34] Public train services were suspended, limited buses were in operation, and restrictions were placed on taxi occupancy capacities.^[34] These transport limitations severely impacted on the ability of non-live-in caregivers to travel. There was also a sense of fear associated with using public transport, with many caregivers and patients apprehensive about contracting the virus by travelling in a confined space.^[34] Discrimination by transport operators, as well as other passengers, often resulting in refusal to transport those with visible conditions or using assistive devices, further contributed to access challenges faced by RD patients.^[16,21,34]

Education and employment. As lockdown persisted, employers (including organisations serving those with disabilities) were forced to make changes, requiring employees to take unpaid leave, reduced pay and retrenchment.^[21,32] The lack of availability of sign language interpreters in schools impacted on children with hearing disabilities.^[35] Heads of mainstream schools were unwilling to enrol children living with disabilities, as they lacked the skills and capacity to support them.^[30]

Basic needs. The SA government provided a stimulus package including a grant and food parcel earmarked for citizens who earned a combined monthly household income of <ZAR3 600 or existing recipients of South African Social Security Agency grants, including disability, child welfare and pensioner grants. However, many people with disabilities did not receive the food parcel or were unable to access it.^[16,35] The gap in access to food for those with an RD/CD or living with a disability was addressed to some extent by non-profit organisations (NPOs).^[32]

Consumables. The fear of infection for RD/CD patients was compounded by lack of access to personal protective equipment (PPE) for themselves, their caregivers and HCPs.^[31-33,35] Some state hospitals in Gauteng Province ran out of mandatory PPE,^[31] and organisations supporting persons with disabilities became reliant on PPE donations for their staff.^[32]

Medication and assistive devices. Disability-specific interventions were not prioritised during lockdown, resulting in patients using wheelchairs and other assistive devices being unable to access device upgrades and repairs.^[33] RD/CD patients had limited access to medications, including several stockouts where specific medication was unavailable countrywide.^[33] In some instances, parents were arrested when they left the house to try to buy medicine, and other essential items, for their children with disabilities.^[35]

Emergency care. Existing healthcare capacity, infrastructure and services were redirected and reallocated to the COVID-19 pandemic emergency response.^[32,34] The triage policies implemented in facilities to assist HCPs in directing the scarce resources during the pandemic did not specifically address patients living with RD/CDs or disabilities, and largely excluded them based on their diagnosed condition without further evaluation.^[34,35] Triage policies in SA are linked to the Clinical Frailty Scale and directly exclude individuals with disabilities from accessing intensive care unit admission and ventilator access at times when resources are scarce.^[34,36] This exclusion raises a red flag in terms of how life and quality of life are evaluated, and is contrary to the Bill of Rights in the SA Constitution.^[39]

Services for gender-based violence (GBV) prevention and response. GBV services are mainly provided by NPOs, many of which experienced a decrease in funding during the pandemic.^[40] GBV services were not explicitly classified as 'essential' services. The lack of prioritisation of GBV services, together with the lockdown restricting individuals to their homes, contributed to a

well-documented increase in GBV against women in SA during the pandemic, and particularly those living with disabilities.^[33,41]

COVID-19 testing and vaccination. The roll-out of COVID-19 vaccination was not disability inclusive (no accessibility measures were undertaken) and did not prioritise vulnerable communities such as RD/CD patients. Instead, vaccination roll-out was phased according to an individual's occupation and age,^[42,43] despite RD/CD patients being more vulnerable and at greater risk of developing severe disease as a result of their underlying health conditions.^[43]

Communication and information

The information on COVID-19 provided by the SA government to the public was very generic, at times contradictory, and not tailored to people living with communication challenges and disabilities.^[33,43] Braille and audio versions of key information were often lacking for those with visual impairment. Similarly, for the deaf and hard of hearing, print versions were unavailable and sign language interpretation, subtitles and captioning were often not provided for pandemic-related television broadcasts.^[16,33] While the SA Department of Women, Youth and Persons with Disabilities is responsible for supporting individuals living with disabilities, their communication was unclear, particularly when stating which caregivers could request permission to travel during the lockdown.^[34]

Discrimination

Patients living with RDs/CDs and disabilities often face discrimination when accessing employment, education and healthcare.^[36] When policies are developed to address these issues, they are typically very broad, grouping all individuals living with disabilities into one category, despite the diversity of this community.^[35] This lack of acknowledgement is insensitive and ill-informed, and demonstrates ignorance about the specific and varying needs of individuals living with RDs/CDs and disabilities.^[35]

Increased risk of infection

Many patients living with RDs/CDs and associated disabilities are at an increased risk of COVID-19 infection due to pre-existing conditions, impaired immune systems and comorbidities, and because of where and how they live.^[32-34,43] Many live in communal residential facilities requiring close contact with caregivers, or live in poverty, with limited or no access to running water, sanitation, and hygiene facilities.^[36]

Financial challenges

COVID-19 brought additional and unexpected expenses, particularly for those living with RDs/CDs and disability.^[34] Relief measures tended to be based on the general population and not the more vulnerable, with specific needs.^[35] Individuals using assistive devices were encouraged to disinfect their devices regularly, but without financial support this was unrealistic for many living hand to mouth,^[36] demonstrating a lack of awareness of the financial circumstances of those living with disability in the country.

Opportunities

A limited number of opportunities were identified:

Improved community and NPO support

Many HCPs and NPOs were innovative in adjusting and adapting to the changing circumstances brought about by the lockdown, providing support and sharing information.^[32,43] Some NPOs provided financial assistance, while others stepped in to continue to

implement government programmes that had ceased to operate, such as therapy services for babies with disabilities.^[32]

E-health implementation

Increased implementation of virtual healthcare, such as telemedicine, enabled HCPs to continue to consult with patients remotely,^[32] and prescriptions were sent via email (EURORDIS Rare Diseases Europe, 'Rare barometer: How has COVID-19 impacted people with rare diseases? South Africa – November 2020', available from the authors on request). Online consultations also helped alleviate the financial burden associated with travelling to appointments, and should be considered for continued implementation post pandemic.

Deepening of relationships

When caregivers and families were unable to leave their patients with intellectual disabilities due to the extended lockdown travel limitations, this led to a strengthening of their bond.^[29] In general, individuals felt safer going to public places during the pandemic because of the sanitising and social distancing protocols enforced.^[4]

Recommendations

A number of recommendations were identified in the included literature, mostly related to greater inclusivity of those with an RD/CD or disability.

Inclusivity in policymaking

The RD/CD and disability community should be included, at every stage of the process, in policy discussion, creation and implementation to ensure that their needs are understood and addressed. Triage policies need to be redesigned to be more inclusive.^[35] When formulating future disaster management policies, a child and their parent (and not solely mothers and neonates) should be viewed as a dyad rather than as separate individuals, so they remain together upon admission to hospital.^[44]

Greater emphasis on integrated community-based healthcare

The introduction or expansion of more community-based healthcare services and their integration into primary healthcare, that advocates for equitable access to care for all across all levels,^[45] may also assist in bridging the gap in access to healthcare services for RD/CD patients and those with a disability.^[33] These services should include expansion of currently limited mental health services, and rehabilitation services as a vital component of quality of life of many patients.^[33] Psychosocial support should be prioritised for individuals with intellectual disabilities and those unable to self-advocate, particularly when transitioning into adulthood and from paediatric to adult care.^[29]

Targeted research on the needs and experiences of RD/CD/disability communities

Increased awareness at all levels will be facilitated through expanding the evidence base of issues facing the RD/CD community. Research into RD/CD and disability, as well as patient experiences, should be prioritised, published and made available to policy planners. Comprehensive, quantifiable data on RD/CD and disability in SA, which are currently inadequate, are also essential to inform proper emergency scenario planning.^[33]

Education and awareness

In SA, the lockdown implemented did not consider the specific needs of the RD/CD patient and disability community. This failure led to

further marginalisation with many 'new' hurdles to overcome to access essential healthcare. To overcome these challenges, an active drive to increase education and awareness related to these conditions is required among key stakeholders and the lay public. Patients also need to be appropriately educated and empowered to optimise their participation and advocacy role during the creation of relevant policies.^[35]

Financing mechanisms for NPOs

Organisations that support RD/CD patients and those living with disability were significantly affected by the pandemic, with severely limited funding forcing some organisations to close. Accessible short-term financing is recommended to sustain these organisations through similar future disruptions, as measures such as the Solidarity Fund were restricted.^[32] These organisations should also be consulted when analysing the failures and pitfalls of the pandemic response, and should participate in monitoring the systems put in place to mitigate future disruptions.^[36]

Study limitations

While a comprehensive Boolean search string was implemented for this scoping review, disability and related terms were not included as they were not deemed relevant at the start of this study. However, review of the identified articles indicates that expanding the search term to include terms related to congenital and acquired disability could substantially increase the scope and content of relevant literature.

Few opportunities were identified in this study (Fig. 2). This may be attributed to the limited literature on RD/CD patients and those with associated disabilities published to date. Future research, as yet unavailable, may be identified in a subsequent and expanded review, and highlight additional issues of relevance. However, this review serves as a starting point for the future dialogue and research on these issues for inclusion in relevant policy planning.

Conclusion

This study examined peer-reviewed and grey literature published between 2020 and 2022 on the impact of the COVID-19 pandemic on RD/CD patients, and those with disabilities, in SA. The pandemic has shown that health systems were ill-prepared for COVID-19 and measures did not serve the needs of RD/CD patients, who have specific needs that require inclusion in healthcare-related policies. The expansion of community-based healthcare services would help address the lack of access during the pandemic. It is also recommended that RD/CD patients, and the organisations that support them, be included as active participants in policy creation and planning for future disruptions, and not just considered as end users. This is particularly important within the context of NHI and the journey towards universal healthcare in SA to ensure that no one is left behind.

Data availability. The datasets generated and analysed during the present study are available from the corresponding author (MCMG) on reasonable request.

Declaration. The research for this study was done in partial fulfilment of the requirements for MCMG's PhD (Medicine) degree at the University of KwaZulu-Natal.

Acknowledgements. This work was supported by Rare Diseases South Africa and the College of Health Sciences, University of KwaZulu-Natal.

Author contributions. MCMG: study conceptualisation and design, data

capture, data analysis, first draft preparation, manuscript revision. HLM: study conceptualisation, data analysis, critical expert review of draft and final manuscript.

Funding. None.

Conflicts of interest. None.

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Received 11 December 2023; accepted 19 May 2024.