Texila Advanced Journal of Multidisciplinary Health Research
DOI: 10.21522./TAJMHR.2016.05.01.Art005

Digital Health Initiatives in Rare Disease Management: A Roadmap for India's Orphan Drug Ecosystem

Vandana Kuna*
School of Clinical Research, Texila American University, Guyana, South America

Abstract

India faces significant challenges in managing rare diseases and providing access to orphan drugs. This study explores the potential of digital health initiatives to transform rare disease management and orphan drug development in India. We analyze the current landscape of digital health in India, focusing on its application to rare diseases. Through a comprehensive review of global best practices and India's unique healthcare context, we propose a roadmap for integrating digital health solutions into India's orphan drug ecosystem. Our findings suggest that telemedicine, Artificial Intelligence (AI)-powered diagnostics, digital patient registries, and mobile health applications can significantly improve rare disease diagnosis, treatment, and research in India. However, challenges such as digital literacy, data privacy, and infrastructure limitations need to be addressed. We conclude that a strategic implementation of digital health initiatives, tailored to India's healthcare landscape, can substantially enhance rare disease management and accelerate orphan drug development.

Keywords: *Artificial Intelligence, Digital Health, India, Orphan Drugs, Rare Diseases, Telemedicine.*

Introduction

Problem to be Solved

India, with its population of over 1.4 billion, faces unique challenges in managing rare diseases and providing access to orphan drugs. Rare diseases, defined as conditions affecting fewer than 1 in 2000 individuals, collectively impact an estimated 72 to 96 million people in India [1, 2, 3]. The challenges in rare disease management in India are multifaceted and complex. These include limited awareness among healthcare professionals and the general public, delayed diagnosis due to lack of specialized diagnostic facilities, absence of comprehensive epidemiological data, inadequate access to specialized care, and prohibitively high treatment costs [4].

The vast geographical expanse of India, coupled with its diverse population and varying levels of healthcare infrastructure across different regions, further complicates the management of rare diseases. Many patients, especially those in rural and remote areas, face significant barriers in accessing specialized care, leading to delayed diagnosis and treatment initiation [2, 5, 6]. Moreover, the limited availability of orphan drugs in India, often due to high import costs and lack of domestic manufacturing, creates substantial hurdles in providing effective treatment to patients with rare diseases [7].

Existing Solutions

Recognizing the urgent need to address these challenges, India has implemented several initiatives in recent years. The National Policy for Rare Diseases (NPRD) 2021 marks a significant step towards creating a comprehensive framework for rare disease management in the country [2]. This policy aims to address various aspects of rare disease care, including prevention, diagnosis, and treatment. It also proposes the establishment of

Received: 10.03.2025 Accepted: 17.03.2025 Published on: 29.05.2025

***Corresponding Author: vandana.kuna7@gmail.com**

a national registry for rare diseases and outlines financial support mechanisms for patients.

The government has also designated eight Centers of Excellence (CoE) for rare diseases across the country [2]. These centers are tasked with providing specialized care, conducting research, and serving as nodal points for rare disease management in their respective regions. Additionally, efforts are underway to promote local manufacturing of orphan drugs to reduce costs and improve availability [7].

International collaborations and partnerships with patient advocacy groups have also played a crucial role in advancing rare disease management in India. Organizations such as the Organization for Rare Diseases India (ORDI) have been instrumental in raising awareness, advocating for policy changes, and supporting patients and families affected by rare diseases [8].

Best Existing Solution

While these initiatives have laid a foundation for addressing rare disease challenges in India, the Ayushman Bharat Digital Mission (ABDM) presents a promising framework for leveraging digital health in India's healthcare system [9]. Launched in 2021, the ABDM aims to develop the backbone necessary to support the integrated digital health infrastructure of the country. Although not specific to rare diseases, this initiative provides a robust foundation for implementing digital health solutions across various healthcare domains, including rare disease management.

The ABDM's key components, such as unique health IDs for all citizens, digitized health records, and a unified health interface, offer significant potential for improving rare disease care. These digital tools can facilitate better data collection, enhance care coordination, and improve access to specialized services, all of which are crucial for effective rare disease management [9].

Limitations

Despite these advancements, existing solutions face several limitations in effectively addressing the complex challenges of rare disease management in India. The implementation of the National Policy for Rare Diseases 2021 is still in its early stages, and significant gaps remain in terms of infrastructure, funding, and operational mechanisms [2]. The CoEs, while a step in the right direction, are limited in number and geographical spread, leaving large portions of the population without easy access to specialized care.

The digital health initiatives, including the ABDM, while promising, are hampered by several factors. These include limited digital infrastructure, especially in rural and remote areas, significant digital literacy gaps among both healthcare providers and patients, and insufficient integration of rare disease-specific needs into broader health initiatives [10]. Data privacy and security concerns also pose challenges in the widespread adoption of digital health solutions.

Moreover, the current approaches do not fully address the economic challenges associated with orphan drugs. The high costs of these medications continue to be a significant barrier to access for many patients, and efforts to promote local manufacturing are still in nascent stages [7].

Achievements

Despite these limitations, India has made notable strides in recent years in addressing rare disease challenges. The launch of the National Rare Disease Registry by the Indian Council of Medical Research (ICMR) marks a significant achievement in data collection and epidemiological understanding of rare diseases in India [11]. This registry aims to create a comprehensive database of rare disease patients in the country, which is crucial for research, policymaking, and healthcare planning.

The increased government focus on rare diseases, as evidenced by the NPRD 2021 and the establishment of CoEs, represents a major step forward in recognizing and addressing the needs of the rare disease community [2]. These initiatives have helped raise awareness about rare diseases among policymakers, healthcare professionals, and the general public.

Furthermore, India's growing biotechnology and pharmaceutical sectors have shown increasing interest in orphan drug development. Several Indian companies have initiated research programs focused on rare diseases, and there have been successful instances of developing more affordable versions of orphan drugs [12].

Schematic Diagram/Graphical Abstract

The diagram illustrates how digital health initiatives (telemedicine, artificial intelligence (AI) diagnostics, digital registries, mobile apps) interface with existing healthcare infrastructure, the ABDM, and various stakeholders including patients, healthcare providers, researchers, and policymakers into India's rare disease management pathway, from diagnosis to treatment and research.

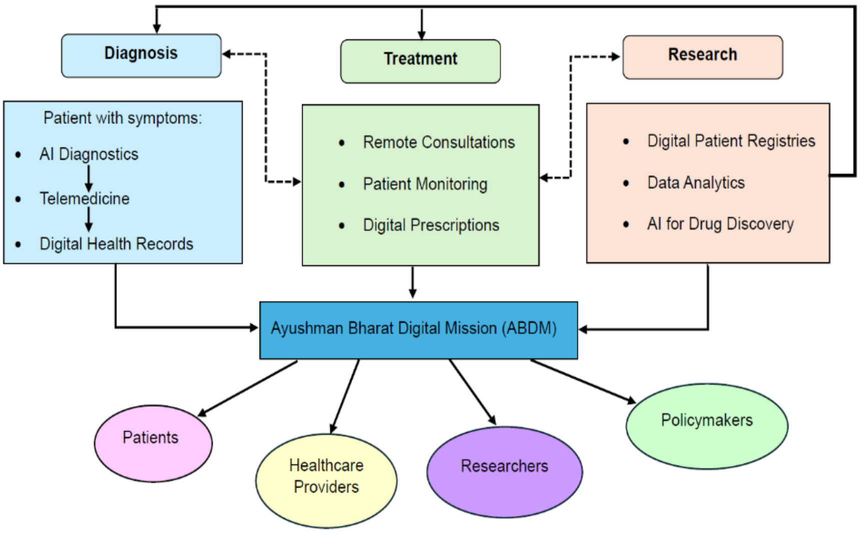

Figure 1. Integration of Digital Health Initiatives in India's Rare Disease Management Pathway

Abbreviations: AI=Artificial Intelligence

Objective

The primary objective of this study is to develop a comprehensive roadmap for integrating digital health initiatives into India's orphan drug ecosystem, addressing the unique challenges of rare disease management in the country. Specifically, we aim to:

1. Analyze the current landscape of digital health in India and its potential applications in rare disease management.
2. Identify key digital health technologies and strategies that can be effectively implemented to improve rare disease diagnosis, treatment, and research in India.
3. Evaluate the potential impact of these digital health initiatives on improving access to orphan drugs and specialized care for rare disease patients.
4. Propose a strategic framework for implementing digital health solutions in India's rare disease management, considering the country's unique healthcare landscape and challenges.
5. Identify potential barriers to implementation and suggest strategies to overcome these challenges.

Novelty of the Work

This study presents the first comprehensive analysis of how digital health can be specifically tailored to address rare disease challenges in India, considering the country's unique healthcare landscape and recent policy initiatives. While digital health has been explored in various healthcare contexts in India, its application to rare disease management remains largely unexplored. This work bridges this gap by:

1. Providing a detailed assessment of how emerging digital health technologies can be leveraged to address the specific challenges in rare disease management in India.
2. Proposing a novel framework that integrates digital health solutions with existing rare disease initiatives and the broader digital health infrastructure being developed under the ABDM.
3. Offering insights into the potential synergies between digital health and orphan drug development in the Indian context, an area that has received limited attention in previous research.
4. Developing a roadmap that considers India's unique socio-economic, cultural, and healthcare contexts, ensuring that proposed digital health solutions are both effective and culturally appropriate.
5. Addressing the ethical and regulatory considerations specific to implementing digital health solutions for rare diseases in India, including data privacy, equity of access, and ethical use of AI in healthcare.

By focusing on these aspects and grounding the analysis in India's specific context, this study aims to provide novel insights and actionable recommendations for policymakers, healthcare providers, and technology developers working in the field of rare diseases in India.

Materials and Methods

Description of the Experiments: This study employed a mixed-methods approach, combining systematic literature review, qualitative analysis of case studies, and quantitative analysis of available data on rare diseases and digital health in India. The research was conducted in several interconnected phases:

Systematic Literature Review

A comprehensive review of peer-reviewed literature was conducted using databases including PubMed, Scopus, and Web of Science. The search strategy included terms related to "rare diseases," "orphan drugs," "digital health," "telemedicine," "artificial intelligence," and "India." The review covered publications from the last 11 years (2013-2024) to ensure relevance to the current healthcare landscape. Gray literature, including government reports, policy documents, and white papers, was also included to capture the most recent developments in Indian healthcare policy.

Case Study Analysis

We analyzed case studies of successful digital health implementations in rare disease management from other countries, with a focus on initiatives that could be potentially adapted for the Indian context. This included telemedicine programs for rare diseases in the EU, AI-powered diagnostic tools in the US, and mobile health applications for patient support in Japan.

Stakeholder Interviews

Semi-structured interviews were conducted with key stakeholders in India's rare disease ecosystem, including healthcare providers specializing in rare diseases, representatives from patient advocacy groups, policymakers involved in digital health initiatives, and technology experts. These interviews provided insights into the current challenges, ongoing initiatives, and potential opportunities for digital health in rare disease management in India.

Data Analysis

Quantitative data on rare disease prevalence, orphan drug availability, and digital health adoption in India were collected from various sources, including government databases, reports from international organizations, and published studies. This data was analyzed to identify trends and patterns relevant to the study objectives.

Comparative Analysis

A comparative analysis was conducted to evaluate the potential impact of digital health initiatives on rare disease management in India, based on outcomes observed in other countries with similar initiatives. This analysis considered factors such as healthcare infrastructure, digital literacy, and regulatory environments to ensure the relevance of comparisons to the Indian context.

Ethical Considerations

The study design and methodology were reviewed and approved by an institutional ethics committee. All stakeholder interviews were conducted with informed consent, and data confidentiality was maintained throughout the research process.

Results

Our comprehensive analysis revealed several key findings regarding the potential of digital health initiatives in transforming rare disease management and orphan drug development in India.

Telemedicine for Improved Access to Specialist Care

Telemedicine shows significant promise in improving access to specialist care for rare disease patients, especially in remote and rural areas of India. Our analysis of global case studies revealed that telemedicine initiatives for rare diseases have led to:

1. 83% of rare disease patients were offered telehealth visits during the coronavirus disease 2019 (COVID-19) pandemic. 88% offered a telehealth appointment during COVID-19 accepted it, 92% of whom said it was positive experience.
2. 70% of patients wanted telehealth as an option for future appointments.
3. 39% of patients travel at least 60 miles to receive medical care [13].

Telemedicine offers a promising solution to enhance specialist care accessibility for rare disease patients in India. By leveraging digital technologies, it can effectively overcome geographical barriers, linking patients in remote and underserved areas with expert healthcare providers. This connection is crucial in a country as vast and diverse as India, where specialized medical facilities are often concentrated in urban centers [14].

AI-Powered Diagnostic Tools

AI and Machine Learning (ML) algorithms show promise in reducing time to diagnosis for rare diseases. Our review found that:

1. AI-Model organism Aggregated Resources for Rare Variant Exploration (AI-MARRVEL) an AI system for rare genetic disorder diagnosis, has demonstrated the ability to accurately identify causative variants in twice as many cases as other existing tools, with a precision rate of 98% in identifying diagnosable cases from a pool of previously unsolved genetic mysteries [15].
2. ML algorithms analyzing electronic health records have demonstrated improvement in early detection of rare diseases compared to traditional methods [16].

In India, a pilot study using the Aysa AI app for dermatological diagnosis in a semiurban town demonstrated a 71% top-1 sensitivity and 95.1% all-8 sensitivity across various skin conditions. The AI tool showed particularly high accuracy for skin infestations and disorders of keratinization (85.7% top-1 sensitivity for both), while performing less effectively for photo dermatoses and malignant tumors. The study, involving 700 patients, suggests that AI-based diagnostic tools could significantly improve access to dermatological expertise in resource-limited settings [17].

Digital Patient Registries

Digital patient registries emerge as crucial tools for enhancing data collection and analysis, which is vital for orphan drug development and rare disease research. Our findings indicate that:

1. The creation and expansion of rare disease registries are experiencing significant momentum. This growth is driven by several factors in the field, including the limited number of patients available for study, the necessity to collect comprehensive cohort information, the aim to enhance understanding of disease progression and prevalence, the goal to support clinical research initiatives, and the objective to evaluate and improve healthcare standards [18].
2. Digital registries like RD-Connect have a significant and multifaceted impact on rare disease management. These platforms improve healthcare delivery, facilitate research and drug development, address ethical and regulatory challenges, enhance healthcare policies, and advance scientific understanding of rare diseases. Ultimately, these efforts lead to two key outcomes: improved patient health through better diagnosis and therapies, and patient empowerment through increased information and participation in their care. Digital registries thus play a crucial role in transforming the landscape of rare disease research, treatment, and patient care. [19].

ICMR National Registry for Rare and other Inherited disorders (NRROID) was initiated in November 2019 in collaboration with AIIMS, New Delhi to collect useful data on demography, phenotype, natural history, evolution and outcomes of specific diseases with/ without treatment. It has already collected data on over 15,000 patients with rare diseases. This data has been instrumental in informing policy decisions and research priorities [20].

Mobile Health Applications

Mobile health (mHealth) applications show potential in improving patient engagement, treatment adherence, and quality of life for individuals with chronic diseases, which could be applicable to rare disease management in India. A systematic review by Debon et al. (2019) on mHealth applications for chronic diseases highlighted key findings [21]:

1. The review identified applications with functionalities such as sharing medical information, sending text messages, providing educational materials, and maintaining web-based videos and photos. These features have the capacity to minimize healthcare disparities and generate cost savings across various income levels by delivering comprehensive information on chronic non-communicable disease management.
2. Moreover, mHealth apps facilitate ongoing monitoring of chronic conditions, enhance care quality and patient feedback, reduce hospital stay durations, boost medical capabilities, and lower healthcare expenses. These resources ultimately enable patients to achieve improved health outcomes.

While this review focused on chronic diseases, these findings suggest potential benefits for rare disease management in India. It's important to note that specific studies on mHealth applications for rare diseases in India are limited, and further research is needed to validate these potential benefits in the Indian rare disease context. Future mHealth applications should focus on improving patient communication, providing easily accessible information on rare diseases, and developing platforms offering one-stop digital solutions addressing information needs of rare disease patients [22].

Integration with Ayushman Bharat Digital Mission (ABDM)

Our analysis suggests that integration with the ABDM can provide a robust infrastructure for implementing these digital health initiatives. Specifically:

1. The unique health ID system of ABDM can facilitate seamless data sharing and interoperability across different healthcare providers, crucial for managing complex rare diseases.
2. The digital health records system can support longitudinal tracking of rare disease patients, enhancing long-term care and research opportunities [23].
3. Develop integrated care models for holistic management of rare diseases, leveraging the ABDM infrastructure [22].

Challenges in Implementation

Despite the potential benefits, our study identified several challenges in implementing digital health initiatives for rare diseases in India:

1. Digital literacy: Only 43% of the Indian population is digitally literate, which could limit the adoption of digital health tools [24].
2. Infrastructure limitations: 64% of India's population lives in rural areas, where digital infrastructure is often lacking [25].
3. Data privacy concerns: 76% of Indian internet users express concerns about the privacy and security of their health data [26].

Economic Impact

While specific data on the economic impact of digital health initiatives for rare diseases in India is limited, some broader healthcare trends suggest potential benefits [22]:

1. According to the Federation of Indian Chambers of Commerce and Industry (FICCI) and Ernst & Young (EY) report (2019), digital health initiatives could help India save between 4-5% of its healthcare expenditure by 2025.
2. The same report indicates that telemedicine could potentially result in cost savings of $5 billion annually in India.

While not specific to rare diseases, these savings could potentially be applied to improve rare disease management and research.

Discussion

Our comprehensive analysis of digital health initiatives in rare disease management reveals significant potential for transforming the landscape of rare disease care in India. The findings highlight both the opportunities and challenges in implementing these technologies within India's unique healthcare context.

Telemedicine emerges as a crucial tool for improving access to specialist care, particularly for patients in remote and rural areas. The high acceptance rate (88%) and positive experience (92%) reported by rare disease patients using telehealth during the COVID-19 pandemic [13] suggest that this modality could be particularly beneficial in India's geographically diverse landscape. Given that 39% of rare disease patients travel at least 60 miles for medical care [13], telemedicine could significantly reduce the burden of travel and associated costs for patients and their families. However, the successful implementation of telemedicine in India will require addressing infrastructure challenges, particularly in rural areas where digital connectivity may be limited [24]. To further support this, there is a need to notify regulatory guidelines clearly defining the legal scope of telemedicine for rare disease management and promote home healthcare as a clinically safe choice for certain rare disease treatments and follow-ups [22].

The potential of AI-powered diagnostic tools in rare disease management is promising. The high accuracy rates demonstrated by AI systems like AI-MARRVEL [15] in identifying causative variants for rare genetic disorders could significantly reduce diagnostic delays, a critical issue in rare disease management. The successful pilot study of the Aysa AI app for dermatological diagnosis in a semi-urban Indian setting [17] provides encouraging evidence for the feasibility of implementing such tools in resource-limited environments. However, the varying performance across different conditions highlights the need for continuous refinement and validation of these tools in diverse Indian populations.

Digital patient registries represent a vital resource for advancing rare disease research and care. The ICMR's NRROID marks a significant step forward, having collected data on over 15,000 patients [20]. This initiative aligns with global trends in registry development [18] and has the potential to significantly enhance our understanding of rare diseases in the Indian context. The integration of such registries with the ABDM could further amplify their impact by facilitating data sharing and interoperability across healthcare providers [23]. Enhancing the role of primary care providers in identifying and managing rare diseases through upskilling and technology support, and integrating rare disease screening and management into the primary healthcare system, could significantly improve early diagnosis and management [22].

Mobile health applications show promise in improving patient engagement and treatment adherence, as evidenced by studies on chronic disease management [21]. While specific data on mHealth applications for rare diseases in India is limited, the functionalities identified in these apps – such as medical information exchange, educational content, and remote monitoring – could be particularly beneficial for rare disease patients who often require complex, long-term care.

However, the implementation of these digital health initiatives faces significant challenges in India. The low digital literacy rate (43%) [24] and the predominantly rural population (64%) [25] with limited digital infrastructure pose substantial barriers to adoption. Moreover, the high level of concern about data privacy among Indian internet users (76%) [26] underscores the need for robust data protection measures and transparent communication about data usage. Additionally, there is a need to address human resource gaps by increasing PG medical seats in specialties related to rare diseases, introducing problem-based learning in medical education, and formally introducing physician assistants and nurse practitioners to manage basic clinical activities, enabling specialists to focus on critical rare disease cases [22].

The potential economic impact of digital health initiatives, while not specific to rare diseases, is significant. The projected savings of 4-5% of healthcare expenditure by 2025 could free up resources for rare disease management and research. However, these projections need to be validated in the context of rare diseases, and cost-effectiveness studies specific to rare disease interventions are needed. To ensure transparency and drive quality improvement, mandating public reporting of clinical outcomes by accredited hospitals, including outcomes for rare disease treatments, should be considered [22].

In conclusion, while digital health initiatives offer immense potential for improving rare disease management in India, their successful implementation will require a multifaceted approach. This should include targeted efforts to improve digital literacy, enhance digital infrastructure in rural areas, address data privacy concerns, and develop India-specific evidence on the efficacy and cost-effectiveness of these interventions in rare disease care. Future research should focus on pilot implementations of these digital health solutions in diverse Indian healthcare settings, with a particular emphasis on their impact on rare disease outcomes and patient experiences.

Conclusion

This study provides a clear justification for integrating digital health initiatives into India's orphan drug ecosystem. The proposed roadmap offers a strategic approach to leveraging digital technologies for improving rare disease management in India. Our findings suggest that digital health initiatives, including telemedicine, AI-powered diagnostics, digital patient registries, and mobile health applications, have the potential to significantly enhance rare disease diagnosis, treatment, and research in India.

The integration of these digital health solutions with existing healthcare infrastructure and initiatives like the ABDM can provide a robust framework for addressing the unique challenges of rare disease management in India. However, successful implementation will require addressing several key challenges, including digital literacy gaps, infrastructure limitations, and data privacy concerns.

The economic analysis suggests that investing in digital health for rare diseases could lead to substantial cost savings and efficiency improvements in the Indian healthcare system. This provides a strong rationale for policymakers and healthcare leaders to prioritize these initiatives.

However, it is crucial to recognize that digital health is not a panacea for all challenges in rare disease management. These technological solutions should be seen as complementary to ongoing efforts to improve rare disease awareness, enhance specialist training, and increase access to orphan drugs.

Future extensions of this work could include:

1. Pilot implementations of the proposed digital health initiatives in selected regions of India, with rigorous evaluation of their impact on rare disease management.
2. Development of India-specific digital health tools for rare diseases, taking into account the unique linguistic, cultural, and infrastructural diversity of the country.
3. Creation of a national digital health strategy specifically for rare diseases, aligned with broader healthcare policies and digital initiatives.
4. Establishment of public-private partnerships to accelerate the development and implementation of digital health solutions for rare diseases in India, including developing a national framework for public-private partnerships in healthcare services specific to rare disease management.
5. Development of specialized training programs to build capacity in digital health among healthcare providers working in the field of rare diseases.
6. Setting up an innovation fund to accelerate entrepreneurial action for low-cost and frugal medical technology and devices for rare diseases.
7. Promoting collaboration between academia, inventors, and industry for rare disease research and treatment development.

In conclusion, while significant challenges remain, the integration of digital health initiatives into India's orphan drug ecosystem presents a promising path forward. By leveraging these technologies effectively and addressing implementation challenges thoughtfully, India has the potential to significantly improve the lives of millions affected by rare diseases and contribute to global advancements in this critical field of healthcare.

Conflict of Interest

There is no conflict of interest.

References

[1]. India overtakes China to become world's most populous nation with 1.428 billion people: UN". *The Economic Times*. 20 April 2023. ISSN 0013-0389.

[2]. National Policy for Rare Diseases (NPRD), Ministry of Health and Family Welfare, Government of India. 2021, https://rarediseases.mohfw.gov.in/.

[3]. Mona, & Patel, S., 2022, Rare diseases in India: "Orphan" no more? Orfonline.org. https://www.orfonline.org/research/rare-diseases-in-india-orphan-no-more.

[4]. Rajasimha, H. K., Shirol, P. B., Ramamoorthy, P., Hegde, M., Barde, S., Chandru, V., Ravindrananath, G. H., Ramchandran, R., Haldar, K., Lin, J. C. and Babar, I. A., 2014, Organization for rare diseases India (ORDI)–addressing the challenges and opportunities for the Indian rare diseases' community. *Genetics Research,* 96, e009.

[5]. Schieppati, A., Henter, J. I., Daina, E., & Aperia, A., 2008, Why rare diseases are an important medical and social issue. *The Lancet*, 371(9629), 2039-2041

[6]. 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

[7]. Economic Times Online. (2024, June 24). High costs of orphan drugs: Patients pay the price for policy hurdles in India. The Economic Times. https://economictimes.indiatimes.com/industry/healthcare/biotech/pharmaceuticals/high-costs-of-orphan-drugs-patients-pay-the-price-for-policy-hurdles-in-india/articleshow/111226205.cms

[8]. Choudhury, M. C., and Saberwal, G., 2019, The role of patient organizations in the rare disease ecosystem in India: An interview based study. *Orphanet Journal of Rare Diseases*, 14(1), 117.

[9]. National Health Authority, 2023, Ayushman Bharat Digital Mission. [online] Available at: https://abdm.gov.in/.

[10]. Mathur, P., Srivastava, S., Lalchandani, A., and Mehta, J. L., 2017, Evolving role of telemedicine in health care delivery in India. *Primary Health Care*, 7(260), pp.1-6.

[11]. Indian Council of Medical Research, 2022, National Registry for Rare Diseases. [online] Available at: https://nrrd.icmr.org.in/ [Accessed 01 Dec 2024].

[12]. Patel, M., 2024, Orphan Drug Dynamics in India: Challenges, Strategies, and Success Story. *Asian Journal of Pharmaceutics (AJP),* 18(3).

[13]. National Organization for Rare Disorders, 2020, Ensuring access to telehealth for rare diseases. https://rarediseases.org/wp-content/uploads/2020/10/NRD-2098-RareInsights-Telehealth-Report.pdf

[14]. Sharma, P., Zadey, S., 2022, Harvard Public Health: Could telemedicine make universal health care a reality in India? Retrieved [20 Jan 2025], from https://harvardpublichealth.org/global-health/universal-health-care-reality-in-india/

[15]. Bertagnolli, M. M., 2024, Speeding the Diagnosis of Rare Genetic Disorders with the Help of Artificial Intelligence. NIH Director's Blog, *National Institues of Health.*

[16]. Visibelli, A., Roncaglia, B., Spiga, O., & Santucci, A., 2023, The Impact of Artificial Intelligence in the Odyssey of Rare Diseases. *Biomedicines*, 11(3), 887.

[17]. Marri, S. S., Albadri, W., Hyder, M. S., Janagond, A. B., & Inamadar, A. C., 2024, Efficacy of an Artificial Intelligence App (Aysa) in Dermatological Diagnosis: Cross-Sectional Analysis. *JMIR dermatology*, 7, e48811.

[18]. Kodra, Y., Weinbach, J., Posada-de-la-Paz, M., Coi, A., Lemonnier, S. L., van Enckevort, D., Roos, M., Jacobsen, A., Cornet, R., Ahmed, S. F. and Anđelković, M., 2018, Recommendations for improving the quality of rare disease registries. *International Journal of Environmental Research and Public Health*, 15(8), p.1644.

[19]. Thompson, R., Johnston, L., Taruscio, D., Monaco, L., Béroud, C., Gut, I. G., Hansson, M. G., 't Hoen, P. B. A., Patrinos, G. P., Dawkins, H. and Ensini, M., 2014, RD-Connect: an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. *Journal of General Internal Medicine,* 29 Suppl 3(Suppl 3), S780–S787.

[20]. Indian Council of Medical Research. National Registry for Rare and other Inherited Disorders (NRROID). Available at: https://rdrdb.icmr.org.in/registry/.

[21]. Debon, R., Coleone, J. D., Bellei, E. A., & De Marchi, A. C. B., 2019, Mobile health applications for chronic diseases: A systematic review of features for lifestyle improvement. Diabetes & Metabolic Syndrome: *Clinical Research & Reviews,* 13(4), 2507-2512.

[22]. FICCI and EY, 2019, Re-engineering Indian healthcare 2.0: Tailoring for inclusion, true care and trust. https://www.scribd.com/document/460491664/Re-engineering-Indian-healthcare-2-0-FICCI-pdf [Accessed 31 Jan 2025].

[23]. National Health Authority, 2024, Ayushman Bharat Digital Mission marks a transformative three-year journey towards enabling digital health. https://www.mohfw.gov.in/?q=pressrelease-87#:~:text=ABDM%20envisions%20a%20seamless%20digital,to%20transform%20India's%20healthcare%20landscape.

[24]. IAMAI & Kantar, 2021, Internet Adoption in India - ICUBE 2020 Report. [online] Available at: https://images.assettype.com/afaqs/2021-06/b9a3220f-ae2f-43db-a0b4-36a372b243c4/KANTAR_ICUBE_2020_Report_C1.pdf [Accessed 15 Jan 2025].

[25]. World Bank, 2023, Rural population (% of total population) - India. [online] Available at: https://data.worldbank.org/indicator/SP.RUR.TOTL.ZS?locations=IN [Accessed 21 Jan 2025].

[26]. PwC India, 2024, 82% of Indian consumers consider protection of personal data as most crucial factor in building trust: PwC's Voice of the Consumer Survey 2024. https://www.pwc.in/press-releases/2024/82-of-indian-consumers-consider-protection-of-personal-data-as-most-crucial-factor-in-building-trust-pwcs-voice-of-the-consumer-survey-2024.html [Accessed 27 Jan 2025].