Abstract

Introduction: Rare diseases pose unique challenges in diagnosis due to their complex and diverse nature. Collaborative efforts involving primary care doctors, experts, patients, and external resources are crucial for accurate diagnoses and effective treatment strategies.

Objective: This study aims to develop a framework incorporating digital health technologies into primary care settings to improve the diagnosis of rare diseases.

Methodology: A qualitative phenomenological methodology was employed to address the challenges of diagnosing rare diseases in primary care. The study consisted of three phases: exploring difficulties faced by primary care professionals, identifying potential digital health solutions, and the development of a theoretical framework. Semi-structured face-to-face interviews were conducted with participants from primary care units in the Colombo District of Sri Lanka. Thematic analysis was applied to the data collected, providing insights into the unique perspectives and experiences of the participants.

Results: The study findings identified four primary areas of challenges and corresponding solutions: unusual presentations and diagnostic uncertainty, obstacles in the referral process, knowledge gaps, and the need for professional development.

Conclusion: This research offers a comprehensive framework to enhance the diagnosis and management of rare diseases through collaborative practices, knowledge sharing, and technology integration. The proposed framework emphasizes the establishment of a Community of Practice, bringing together primary care doctors, experts, and patients (along with their families) to facilitate effective communication and collaboration. It advocates implementing an electronic referral system, telemedicine platform, and forums to enhance stakeholder communication. The framework also emphasizes the importance of interoperability standards for efficient data exchange. Local implementation of investigation and specialist registries and using international rare disease databases and rare disease diagnosis support systems support rapid and accurate diagnoses. The generated knowledge during the diagnostic process is shared within the community and rare disease databases, advancing collective knowledge. By implementing this framework, healthcare practitioners can effectively collaborate, utilize available resources, and improve diagnoses of rare diseases.