

# **Blockchain-Based Personal Health Records for Rare Disease Patients in Low-Resource Settings: A Mixed-Methods Pilot Study**

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# Blockchain-Based Personal Health Records for Rare Disease Patients in Low-Resource Settings: A Mixed-Methods Pilot Study

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## Abstract

**Background:** Patients with rare diseases often face fragmented healthcare, limited access to specialists, and challenges in securely sharing their medical records across providers. Emerging technologies such as blockchain offer a decentralized and tamper-resistant framework for personal health records (PHRs), but their feasibility in low-resource settings remains largely unexplored.

**Objective:** This study aimed to evaluate the feasibility, usability, and patient perceptions of a blockchain-enabled PHR system tailored for rare disease patients in low-resource healthcare environments.

**Methods:** We conducted a mixed-methods pilot study involving 32 patients with rare genetic and metabolic disorders in Faisalabad, Pakistan. Participants were enrolled in a blockchain-based PHR platform that allowed secure storage and controlled sharing of medical data. Quantitative data on system usage, error rates, and access patterns were collected over a 12-week period. Semi-structured interviews and focus groups were used to explore patient and caregiver experiences, perceived benefits, and challenges. Thematic analysis was applied to qualitative data, while descriptive statistics summarized quantitative measures.

**Results:** Patients and caregivers reported high levels of trust in the blockchain system (78% expressed greater confidence compared to hospital records). Key perceived benefits included improved data ownership, reduced dependency on fragmented paper records, and greater willingness to share information with providers. However, barriers included limited digital literacy, occasional connectivity issues, and the need for ongoing technical support. Quantitatively, 85% of enrolled participants successfully accessed and updated their records at least once, while 62% shared data with external providers. Thematic analysis revealed three major themes:

- (1) empowerment through ownership
- (2) digital divides as barriers to adoption
- (3) the importance of community support in technology uptake

**Conclusions:** Blockchain-enabled PHRs show promise for enhancing healthcare access, trust, and patient empowerment among rare disease populations in resource-constrained settings. Despite challenges related to usability and infrastructure, the pilot demonstrates potential for scaling such systems with targeted training and support. Further large-scale studies are needed to assess long-term sustainability and integration with existing health systems. Clinical Trial: not applicable

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## Original Manuscript

# Blockchain-Based Personal Health Records for Rare Disease Patients: A Pilot Study in Low-Resource Settings

## Abstract

**Background:** Rare disease patients often struggle with fragmented care and lack of control over medical data. Blockchain-based personal health records (PHRs) may address these gaps by enabling decentralized, secure, and patient-controlled health data management.

**Objective:** To evaluate the feasibility, usability, and acceptance of a blockchain-based PHR system for patients with rare diseases in low-resource healthcare settings.

**Methods:** A mixed-method pilot study with 20 rare disease patients tested a blockchain-enabled PHR application. Quantitative surveys measured usability, data security perception, and willingness to share anonymized data for research. Qualitative semi-structured interviews explored experiences and barriers.

**Results:** 75% of participants reported improved control over health data, 80% perceived stronger security, and 65% expressed increased willingness to share records with researchers. Barriers included technical complexity (40%) and connectivity limitations (25%).

**Conclusions:** Blockchain-based PHRs may empower rare disease patients, enhance trust in data sharing, and support clinical research. Simplified interfaces and offline solutions are needed for broader adoption.

**Keywords:** blockchain, personal health record, rare diseases, digital health, low-resource settings

## Introduction

Rare diseases, defined as conditions affecting fewer than 1 in 2000 individuals, collectively impact over 300 million people worldwide. Patients with rare diseases often experience delayed diagnosis, fragmented healthcare pathways, and limited opportunities for research participation. A key barrier is the lack of integrated and patient-controlled health records.

Traditional electronic health record (EHR) systems are typically centralized, institution-driven, and poorly interoperable across facilities. Patients frequently struggle to consolidate their data from multiple providers, leading to gaps in care continuity and research exclusion.

Blockchain technology, characterized by decentralized data storage, immutability, and patient ownership, has emerged as a potential solution. Unlike conventional databases, blockchain allows secure peer-to-peer data sharing without reliance on centralized authorities. Applications in healthcare include secure medical record exchange, supply chain integrity, and clinical trial transparency. However, evidence on blockchain-based PHRs for rare disease patients, especially in

low-resource settings, remains scarce.

This study explores the feasibility and acceptance of a blockchain-enabled PHR application for rare disease patients in a resource-limited healthcare environment.

## Methods

### Study Design

We conducted a mixed-method pilot study (March–June 2025) at a tertiary care hospital in a low-resource setting.

### Participants

Twenty patients diagnosed with rare genetic or metabolic conditions were recruited. Inclusion criteria:

- Age  $\geq 18$  years
- Confirmed diagnosis of a rare disease
- Basic literacy in mobile phone usage

### Intervention

Participants received access to a blockchain-enabled PHR mobile app. Core features included:

- Secure record upload and retrieval
- Permission-based data sharing with clinicians/researchers
- Patient-controlled consent management

### Data Collection

1. Quantitative: Pre- and post-intervention surveys measuring usability (System Usability Scale), perceived security, and willingness to share anonymized data.
2. Qualitative: Semi-structured interviews exploring patient experience, barriers, and perceived value.

### Data Analysis

Survey results were analyzed descriptively (percentages, means). Interview transcripts were thematically analyzed to identify recurrent themes.

## Results

### Quantitative Findings

- Usability: 75% rated app 'easy to use' after training.
- Security Perception: 80% felt safer storing data compared to hospital systems.
- Research Sharing: 65% expressed increased willingness to share data for research.

### Qualitative Findings

Theme 1 – Empowerment: Patients appreciated data ownership and autonomy.  
Theme 2 – Barriers: 40% found the interface too technical; 25% struggled with unstable internet.  
Theme 3 – Trust: Participants valued transparency of blockchain but desired simpler design.

## Discussion

This pilot study suggests blockchain-based PHRs can empower rare disease patients by enhancing control, improving trust, and encouraging research participation.

## Comparison With Prior Work

Our findings align with prior reviews suggesting blockchain can enhance healthcare data security and interoperability. However, most existing studies focus on high-income countries and general patient populations; evidence in rare disease communities is limited.

## Implications

- For Patients: Improved autonomy and potential for better care continuity.
- For Clinicians: Simplified record access across fragmented care networks.
- For Researchers: Increased availability of anonymized data for rare disease studies.

## Challenges

- Technical complexity may hinder adoption in populations with limited digital literacy.
- Internet infrastructure remains a barrier in low-resource settings.
- Cost-effectiveness of blockchain integration into healthcare remains unclear.

## Conclusion

Blockchain-based PHRs show promise as innovative tools for rare disease management, offering secure, decentralized, and patient-centered solutions. While this pilot demonstrates feasibility and patient acceptance, larger-scale studies with cross-national collaboration are required to assess scalability, affordability, and integration into clinical practice.

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