Research Square

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Development of a Telemedicine Platform with Artificial Intelligence-Assisted Care Diary for Rare Disease Management

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Abstract

Background

Globally, approximately 400 million individuals live with rare diseases, undergoing prolonged diagnostic journeys and requiring ongoing multidisciplinary care. Specialist care is often centralized in major cities, resulting in significant access barriers. Telemedicine demonstrates potential in bridging geographical gaps and facilitating continuous care delivery. Therefore, we developed and tested such a platform at a tertiary care center in Korea.

Methods

We adapted the commercial telemedicine system Olla Care through three development phases: (1) requirements analysis and planning, (2) system design and implementation, and (3) platform validation and evaluation. The platform facilitates remote consultations to reduce travel burden and improve care accessibility for patients with rare diseases and their families. Pilot testing enabled the collection of direct user feedback.

Results

The final platform included five main features: a care diary where families record patients' symptoms and upload photos or videos, automated summaries of diary content using a large language model (GPT-4, OpenAI) designed for future implementation, a module for physician instructions with self-reported adherence tracking, links to the national rare disease information database, and video consultations accommodating up to six members. During pilot testing with eight caregivers and three physicians, technical issues were identified and resolved. The development team and participants resolved technical issues collaboratively. Participants reported improved accessibility and support for family involvement in care but recommended refining login procedures and certain diary functions.

Conclusions

We developed a telemedicine platform that integrates care documentation, AI-assisted review, and multi-party consultations for rare disease care. Beyond providing a practical tool for patient and family engagement, this platform may be a promising approach to improving care coordination and accessibility in similar populations.

Background

Rare diseases collectively affect an estimated 400 million individuals worldwide. In Korea, the Ministry of Health and Welfare classifies diseases affecting fewer than 20,000 individuals as rare diseases. As of

2023, 1,338 diseases are designated as rare, with the count increasing annually [1]. Patients with rare diseases often undergo a prolonged diagnostic journey, consulting with at least seven experts over more than five years before receiving an accurate diagnosis [2]. Even post-diagnosis, patients face several challenges, including limited effective treatments, high-cost medications, and inadequate guidance and support for managing disease progression. Despite the need for coordinated long-term management and multidisciplinary care, patients with rare diseases face structural barriers to healthcare access owing to the concentration of specialized medical personnel in major metropolitan hospitals [3]. This challenge is reflected in the "2nd National Plan for Rare Diseases Management (2022–2026)" by the Ministry of Health and Welfare, which outlines ten strategic priorities for improving the quality of life of patients with rare diseases and their families [4].

In Korea, the Korea Disease Control and Prevention Agency operates a national rare disease management system comprising 17 specialized institutions across 13 provinces and metropolitan cities, including one central support center, to improve healthcare accessibility for patients with rare diseases. However, these resources remain inadequate to support this population [5]. Most rare diseases require long-term monitoring of symptoms, clinical changes, and developmental status under specialist supervision. Nevertheless, follow-up care and prescriptions often necessitate repeated institutional visits, resulting in substantial inconvenience and increased socioeconomic burden [3].

The Coronavirus Disease 2019 (COVID-19) pandemic accelerated the demand for and adoption of telemedicine, driving rapid expansion of its utilization [6]. From a value-based digital healthcare perspective, telemedicine is associated with improved accessibility, lower socioeconomic costs, and enhanced decision-making [7]. Globally, telehealth platforms have been developed to support these shifts by enabling shared treatment records and virtual consultations with specialists, thereby improving access to expert care and facilitating family involvement [8, 9].

In Korea, telemedicine was institutionalized as a pilot program in June 2023, following the COVID-19 pandemic [10]. However, its application remains insufficient for follow-up care and selected patients with chronic conditions [11]. In parallel, digital health interventions have primarily targeted older adults with dementia or severe mental illnesses in long-term care hospitals or psychiatric settings [12]. Considering the importance of family involvement, telemedicine services designed for patients with rare diseases and their families remain underdeveloped.

Although the International Rare Diseases Research Consortium Telehealth Task Force emphasizes that telehealth models incorporating wearable-based monitoring, patient- and caregiver-reported diaries, and mobile health applications can generate real-world data and enhance patient engagement [13], most existing studies examine isolated interventions such as remote monitoring systems, virtual reality applications, or telephone-based care models [9, 13], rather than integrated platforms. Furthermore, platform competition, medication delivery, and legal liability concerns hinder the development of sustainable telemedicine service models [14].

Therefore, this study aims to develop an integrated digital platform that promotes engagement and sustained collaboration with healthcare professionals, and evaluate its technical feasibility and preliminary user experience.

Methods

We developed the telemedicine platform by adapting the external Olla Care Telemedicine platform, following the Waterfall model Software Development Life Cycle (SDLC), a widely adopted software development framework [15]. In collaboration with KB Healthcare, we utilized the Olla Care platform as a foundation, progressing through three major phases: requirements analysis and planning, system design and implementation, and testing and validation.

Phase 1: Requirements Analysis and Planning

1.1. Research Objective Definition and Participant Selection

We defined the research objectives and established eligibility criteria for patients with rare diseases for telemedicine. The attending physician primarily determined eligibility based on patients' health status, generally limited to cases requiring routine medication prescriptions and regular check-ups without in-person examination. Through selecting partner institutions and companies and structured meetings, we established a collaborative framework for platform development.

1.2. System Requirements Analysis

Through consultations with rare disease specialists at Seoul National University Hospital (SNUH), we established the clinical requirements and identified key monitoring symptoms and care plans to inform the platform's clinical functionality. Based on stakeholder input, we developed step-by-step user interaction scenarios. System architecture requirements including technical specifications for mobile-based telemedicine delivery, were defined. We identified the core functional requirements encompassing patient management and healthcare provider workflow integration. Data management and security requirements were established in accordance with healthcare regulations and best practices.

Phase 2: System Design and Implementation

2.1. System Architecture Design

To ensure scalability and accessibility, we designed a cloud-based client-server architecture. We adopted a relational database for data storage and management. The technology stack comprised Next.js for front-end development, React for user interface components, Spring Boot for back-end services, and Java Persistence API (JPA) for data management. We implemented the system using RESTful API design principles, with HTTPS communication protocol for encryption and JSON Web Token authentication for security measures.

2.2. User Interface and Experience Design

To optimize user interface and experience, we developed scenario-based prototypes. The platform incorporated an artificial intelligence (AI)-based summarization system designed to generate concise overviews of care diary entries for physician review before consultations. This feature was intended to facilitate rapid assessment of patient status, while ensuring that all clinical decisions would be made by licensed physicians based on comprehensive patient assessment, including direct patient-family communication. Visualization dashboards supported intuitive patient monitoring and management.

2.3. Platform Implementation

Using existing API service modules from the Olla Care platform, we developed specialized features for rare disease management. Patient-facing features included appointment scheduling, access to physician instructions, and care diary functionality. The care diary allows patients and caregivers to attach symptom-related photos and videos, enabling visual symptom assessment even in a telemedicine setting. Provider-facing features included patient registration, telemedicine consultation, care diary review and documentation, and comprehensive patient management tools. To facilitate platform adoption, we developed a step-by-step manual for patients and caregivers along with supplementary educational materials covering administrative procedures. Throughout implementation, we emphasized enhancing user convenience and improving healthcare provider workflow efficiency.

Phase 3: Platform Validation and Evaluation

3.1. Multi-Tiered Technical Validation Protocol

To ensure technical stability and functional adequacy of the developed platform, we established a systematic validation framework. Unit testing verified the functional accuracy of individual modules. Integration testing evaluated the interoperability between system components and the integrity of data flow. System testing comprehensively assessed overall platform performance and compliance with initial requirements under scenarios simulating real-world use.

3.2. Clinical Environment-Based Pilot Testing

In collaboration with the KB Healthcare development team, we designed and conducted pilot testing in a clinical environment in two sessions at the Rare Disease Center at SNUH. Test participants included patients at the Rare Disease Center and their caregivers, as well as faculty physicians providing clinical care. Given that prior usability research has shown that testing with five to eight participants is sufficient to detect substantial problems [16]. Considering the recruitment constraints, we enrolled eight patient-caregiver pairs who participated in two sessions simulating the telemedicine consultation process.

Each test session was designed to promptly identify and resolve technical issues, and evaluate system stability, user accessibility, clinical workflow compatibility, and technical interoperability. We systematically recorded technical issues and system performance data for each session.

3.3. User Experience Evaluation

Qualitative feedback was collected through semi-structured discussions with caregivers and healthcare providers after each session. Two researchers independently reviewed the feedback and collaboratively organized it into descriptive categories reflecting perceived advantages, areas for improvement, and overall user experience. We also evaluated system performance across various device environments (PC, tablet, smartphone) and network conditions. Compatibility with clinical workflows was verified through real patient-provider interaction scenarios.

Results

Phase 1: Requirements Analysis and Planning

1.1. Research Objectives and Target Disease Selection

A comprehensive literature review confirmed the need for a telemedicine platform tailored to patients with rare diseases and their caregivers. Based on expert recommendations, we initially selected three conditions as eligible for patient inclusion: epilepsy, Duchenne Muscular Dystrophy, and Polycystic Kidney Disease. However, to extend telemedicine services to a broader patient population, we subsequently expanded the scope to include all rare diseases.

1.2. Care Plan Model and Research Scenario Development

Through expert consultations, we established a care plan model incorporating care diary documentation, physician instruction entry, and patient self-assessment of adherence to instructions. The research process was structured into six stages, with scenarios guiding systematic progression from informed consent to the final survey.

1.3. System Requirements Definition

We analyzed the functionality and technical environment of the Olla Care platform to distinguish reusable components from elements requiring new development. We defined essential data elements for a data-driven personalized care system, and mapped key user flows to inform the user-centered interface design.

Phase 2: System Design and Implementation

2.1. Platform Technical Architecture

We developed an integrated platform comprising a Next.js-based user interface, Web Real-Time Communication-enabled video consultation, Spring Boot-based RESTful Application Programming Interface (API), MySQL and Redis database integration via Java Persistence (JPA) API, and real-time

communication through the KakaoTalk Channel (Figure 1). The AZURE cloud infrastructure ensured scalability and reliability. We organized the overall system architecture into three service layers: patient services (mobile application), provider services (web-based consultation), and administrator services (integrated management portal).

The system infrastructure was deployed on Microsoft AZURE cloud services, with web application servers hosted on AZURE Virtual Machine (Linux) instances using Docker containers. User files and images were stored in AZURE Storage, while the operational database used the AZURE Database for MySQL to ensure high availability and scalability. This cloud infrastructure facilitated efficient load distribution and rapid response to system failures.

Figure 1. Platform software architecture

Abbreviations: API, application programming interface; RDBMS, relational database management system; DB, database; WebRTC, Web Real-Time Communication.

2.2. User-Centered Design and Institutional Coordination

Platform development followed user-centered design principles integrated within the SDLC methodology. The design process included iterative consultations with rare disease specialists, nursing staff, and potential end users to identify functional requirements specific to rare disease care. Scenario-based prototypes were developed to visualize functional workflows before implementation. The technical architecture employed a RESTful API framework, and user experience flows tailored to roles, distinguishing patient and caregiver interfaces from healthcare provider administrative systems. An AI-based summarization feature was incorporated to establish a data framework for the integrated collection and analysis of care diary entries, enabling efficient clinical review of patient-documented information.

The provider, especially managing the system interface, integrated appointment management, telemedicine consultation, and patient management functions into a streamlined administrative dashboard.

2.3. Rare Disease-Specific Features

Building on the existing Olla Care infrastructure (login/membership management, basic video call functionality), five core features were developed to address the specific needs of patients with rare diseases and their caregivers. (1) Dedicated appointment scheduling: A specialized KakaoTalk channel ("Rare Disease Self-Management") was created for patient appointments and caregiver registration, offering administrative functions for scheduling, modification, and confirmation. (2) Care diary: A module that enables caregivers to document symptom changes, medication adherence, and daily living status, with photo and video attachment capabilities. Care diary data were synchronized in real time between the server database and the provider web interface (Figure 2).

Figure 2. Care diary interface.

(a) Entry point for accessing the care diary from the application menu.
(b) Initial screen for entering patient identification information.
(c) Calendar-based interface for selecting and reviewing diary records.
(d) Form for entering author information and patient status.
(e) Care diary entry screen including free-text documentation, image upload, and self- assessment.

(3) Physician instruction delivery: A feature for documenting personalized instructions, enabling patients to review instructions and track adherence (Figure 3).

Figure 3. Physician instruction delivery interface.

(a) Physician input of patient-specific instructions.
(b) Automated delivery of instructions to the patient via KakaoTalk.
(c) Patient interface for reviewing assigned tasks.
(d) Documentation of physician instructions within the care diary.

(4**)** Rare disease information integration: Linkage to the Korea Disease Control and Prevention Agency rare disease information database (Helpline) to provide reliable disease-specific information (Figure 4).

Figure 4. Integration with the Korea Disease Control and Prevention Agency Rare Disease Helpline.

(a) App-based interface linked to the helpline service.
(b) Webpage of the national rare disease information portal operated by KDCA.
(c) Educational video resources provided for patients and families.

(5) Multi-party video consultation: A solution supporting up to six simultaneous participants to accommodate multiple caregivers. Three virtual consultation rooms per physician were used to minimize patient wait times. Systematic workflows were established across hospital departments to operationalize telemedicine services. In addition to these platform features, cross-departmental coordination was essential for operationalizing telemedicine services, including telemedicine slot creation within the Electronic Medical Record (EMR) (Clinical Operations), patient appointment guidance (Nursing), external network installation (IT), billing and electronic prescription systems (Administrative Services), educational material development, and KakaoTalk notification channel creation by the research team, as well as electronic prescription networking with 15 partner pharmacies (Pharmacy) and partnerships with external service providers (Table 1).

Table 1. Departmental roles and responsibilities for telemedicine platform implementation

Department	Responsibilities
Clinical Operations	Creation of telemedicine slots within the hospital EMR system
Nursing	Patient appointment scheduling and guidance for telemedicine slots in EMR
Information Technology	Installation of external hospital internet network (hospital admin system operated separately as EMR integration with Olla Care was not feasible)
Administrative Services	Establishment of administrative networking infrastructure: billing system, electronic prescription system, and documentation processing
Research Team	Development of educational materials for research staff and participants; creation of KakaoTalk notification channel for research participants
Pharmacy	Establishment of in-hospital pharmacy networking; partnership with 15 external pharmacies enabled for electronic prescriptions
External Partners	Networking with electronic prescription developer (Lemon Healthcare) and notification service provider for automated payment and messaging services

Note. **EMR: Electro Medical Record.**

2.4. Educational Materials Development

To facilitate platform adoption, the research team developed educational materials for staff and participants: 1) a workflow guide covering six implementation phases (Supplementary File 1), 2) a step-by-step video consultation manual for patients and caregivers (Supplementary File 2), and 3) supplementary guides on channel usage, payment procedures, prescription pickup, and device settings (Supplementary File 3).

Phase 3: Platform Validation and Evaluation

3.1. Testing Process

A systematic multi-tiered testing protocol was implemented. Unit testing was conducted concurrently with development to ensure functionality and maintainability. Integration testing verified interface functionality and defect management. System testing comprehensively evaluated functionality, interface performance, and reliability. Pilot testing was conducted during the system testing phase to identify improvements in stability and usability.

3.2. Pilot Test Verification Items and Optimization

We systematically evaluated the verification items across three scenarios: (1) user scenarios (personal identification and clinical information input), (2) medical service scenarios via hospital admin (personal information accuracy, care plan usability, disease code accuracy, adherence metrics, and all patient-facing functions), and (3) integrated management system scenario (application-web synchronization,

care plan operations, video consultation functionality, and error analysis). To resolve video disconnection during multi-party consultations, we adjusted resolution settings and improved browser compatibility by switching from Arc to Chrome on specific devices.

3.3. Real-Time Issue Response and Optimization

We promptly addressed issues identified during the consultations (Table 2). For infrastructure, we allocated a dedicated IP within Seoul National University Hospital to facilitate video consultation services. For privacy protection, we implemented two-factor authentication and patient number-based verification for administrator login. For telemedicine operations, we provided pre-consultation education; caregivers assisted with smartphone setup, and we assigned dedicated support staff for rapid response.

Table 2. Issues Identified During Pilot Testing and Optimization Measures

Category	Issue	Resolution
Infrastructure	Network issues for real-time video consultation	Dedicated IP allocation within the hospital
Privacy Protection	Need for enhanced security measures	Two-factor authentication for admin login; patient number-based verification
Telemedicine	Patient reluctance toward telemedicine	Pre-consultation education sessions
	Difficulty with platform registration and usage	Professional guidance by research staff
Technical	Smartphone configuration issues	Caregiver-assisted device setup and verification
	Consultation failure owing to unstable internet	Dedicated support staff for rapid response
	Camera/microphone permission settings	Pre-consultation device configuration support

Note. IP: Internet protocol

3.4. Pilot Test

3.4.1. Participant Characteristics

Patients had a median age of 3 years (IQR: 1.5–5.0, male 4, female 4), with diagnoses with diagnoses including epilepsy (n=5; including Lennox-Gastaut syndrome, n=1), muscular dystrophy (n=1), autism spectrum disorder (n=1), and MECP2-related disorder (n=1).

Caregivers were aged 31–40 (n=2), 41–50 (n=3). 51–60 (n=2), and 61–70 (n=1). Three physicians from the Rare Disease Center participated as healthcare providers.

3.4.2. Implementation Process and Identified Issues

Prior to implement this study, written informed consent was obtained from all adult caregivers. For pediatric participants under 16 years of age, written permission was obtained from their parents or legal guardians. Assent was not obtained from the children because of their age and developmental level.

The research team, together with the KB Healthcare development team, conducted two pilot test sessions on September 24 and 26, involving eight patient-caregiver dyad in a real-world telemedicine environment.

Session 1 (September 24): Four patient-caregiver dyad from the Rare Disease Center participated. Key issues included consultation interruption due to unstable internet connection (resolved by switching to telephone consultation), audio feedback (resolved same day by the development team), video mirroring (resolved), and duplicate link errors when caregivers booked on behalf of patients (resolved through system modification). Prescriptions for controlled substances were impossible via telemedicine due to regulatory restrictions, requiring rescheduling to in-person visits. Payment notification failures for patients with special reimbursement status were addressed by establishing alternative payment processes.

Session 2 (September 26): A browser access issue occurred on a Samsung S22 device, which required the consultation to be conducted via a standard video call. The development team contacted the patient directly and resolved the issue within four days.

3.4.3. Feasibility Outcomes

Despite the wide range of technical and administrative challenges, all scheduled consultations were completed (completion rate: 100%, 8/8 sessions). Mean consultation duration was 4.0 minutes (IQR: 2.0–7.5). Technical issues requiring real-time intervention occurred in 3 of 8 sessions (37.5%), all of which were resolved within the session or samedays. Platform fidelity was achieved in 75% of the sessions; however, all deviations were managed through predefined adaptive strategies, such as alternative communication modalities or manual administrative workflows. However, the AI-based care diary summarization function was not tested because of the single-day pilot. Care diaries are intended for continuous documentation between scheduled consultations over days or weeks, which could not be assessed within the brief pilot period.

3.4.4. User Feedback

Qualitative feedback was collected from the patients, caregivers, and healthcare providers after the two pilot tests. Overall, caregivers and healthcare providers reported positive experiences with the platform through multi-party video consultations, particularly in terms of improved accessibility, reduced travel

burden, and enhanced family participation. Usability-related issues such as registration processes and data entry procedures were identified as areas requiring further refinement (Table 3).

Table 3. User Feedback from Pilot Testing

Stakeholder	Advantages	Areas for Improvement
Caregiver	• Convenient care access without hospital visits • Flexible participation during daily activities • Multiple caregiver participation • Reduced waiting time and economic burden • Advance communication via care diary	• Complex process for correcting registration errors • Date restrictions in care diary prevented backdated entries • Absence of auto-fill function for patient registration number and name, requiring manual entry for each session
Healthcare Provider	• Improved consultation efficiency • AI-based care diary summaries • Quick identification of significant events • Enhanced patient trust through advance review • Multiple virtual rooms reduced transition time	• Manual patient number entry required

NOTE. AI: Artificial intelligence

Discussion

We developed and pilot tested a telemedicine platform for patients with rare diseases at a Korean tertiary care center. The platform integrates a care diary with photo and video attachments, AI-based summarization, and video consultations, supporting up to six participants per patient. Pilot testing with eight patient-caregiver pairs revealed perceived benefits including reduced travel burden, simultaneous multi-caregiver participation, and efficient clinical information review, while highlighting areas for refinement in login procedures and diary functionality. From a clinical decision-making perspective, the care diary with multimedia attachments enables physicians to review longitudinal symptom patterns and visual documentation prior to consultations, potentially supporting more informed diagnostic and therapeutic decisions. The AI-based summarization feature, once fully implemented, is intended to reduce cognitive load by highlighting clinically significant changes, thereby facilitating efficient

information processing during time-limited consultations. The care diary component of this platform aligns with previous evidence of the clinical utility of electronic symptom tracking, as Fisher et al. reported clear advantages of electronic seizure diaries compared with paper-based records [17]. The photo and video attachment feature extends this concept; Ricci et al. concluded that home video recordings provide diagnostic value [18], and Tatum et al. reported 89.1% accuracy in expert interpretation of smartphone-recorded seizure videos [19].

The multi-party consultation feature addresses needs identified by Haynes and Marcin, who noted that telemedicine enhances shared decision-making by enabling multiple caregivers to participate [7]. Scherr et al. reported similar benefits in their telehealth model for patients with Batten disease [8]. While our platform does not directly address the diagnostic odyssey documented by Cho et al. [2], the structured documentation and telemedicine capabilities address geographic access barriers contributing to care fragmentation.

This study contributes to health information technology for rare disease management in three ways. First, it describes a systematic development process, providing a methodological template for similar efforts. Second, it documents practical implementation challenges, including dedicated IP allocation requirements and browser compatibility issues, offering actionable guidance for institutions. Third, qualitative feedback identifies specific benefits and areas for improvement, directing future platform refinement.

Korea's 2nd National Plan for Rare Diseases Management (2022–2026) identified limited accessibility to specialist care as a key challenge [1]. Our findings indicate that telemedicine platforms can address this gap by enabling remote consultations while maintaining care continuity. The multi-party consultation feature facilitates family-centered care through simultaneous caregiver participation. However, implementation requires extensive institutional coordination across multiple departments and external partners. The observation that telemedicine cannot accommodate controlled substance prescriptions under current regulations highlights regulatory considerations requiring attention.

Strengths and Limitations

The strengths of this study include a user-centered design approach that incorporated input from healthcare professionals during initial platform development, followed by iterative refinement based on real-world feedback from participants during pilot testing.

The limitations include the small sample size (eight caregivers), absence of formal usability assessment using standardized instruments, and lack of clinical outcome evaluation. The platform could not be integrated with hospital EMR, thereby requiring separate workflows. Although the overall user experiences were positive, several usability issues, including registration procedures, data entry constraints, and administrative workflow integration, were not fully resolved during the pilot phase and require further refinement in future platform iterations. Additionally, the AI-based care diary summarization feature, a key component of the platform design, was not evaluated during pilot testing

due to the brief testing period. Future studies should specifically assess the accuracy, clinical utility, and physician acceptance of this feature. Finally, the single-center design and reliance on institution-specific technological infrastructure may limit generalizability.

Future research should conduct larger-scale evaluations using standardized usability measures and clinical outcome assessments. The AI-based summarization feature warrants specific evaluation for accuracy and clinical utility. Cost-effectiveness analysis would inform policy decisions regarding telemedicine reimbursement for rare disease care.

Conclusions

This study describes the development and pilot testing of an integrated telemedicine platform incorporating care diaries with multimedia capabilities, AI-based summarization, and multi-party video consultation. Although formal efficacy evaluation was beyond the study's scope, participant feedback indicates potential benefits in reducing travel burden, enabling family-centered care, and facilitating clinical information review. Future research should evaluate clinical outcomes to inform broader implementation.

Abbreviations

AI: Artificial Intelligence

API: Application Programming Interface

COVID-19: Coronavirus Disease 2019

EMR: Electronic Medical Record

JPA: Java Persistence API

REST: Representational State Transfer

SDLC: Software Development Life Cycle

SNUH: Seoul National University Hospital

Declarations

Ethics approval and consent to participate

This study was conducted in accordance with the principles of the Declaration of Helsinki. Ethical approval was approved by the Institutional Review Board (IRB) of Seoul National University Hospital (IRB No. 2404-047-1525). Written informed consent was obtained from all adult caregivers. For pediatric

participants under 19 years of age, written permission was obtained from their parents or legal guardians. Assent was not obtained from the children because of their age and developmental level.

Consent for publication

Not applicable.

Availability of data and materials

The datasets generated and/or analyzed during this study are not publicly available because of confidentiality agreements and proprietary considerations but are available from the corresponding author upon reasonable request.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

YSJ contributed to validation, formal analysis, data curation, visualization, writing – original draft, review, and editing of the manuscript. JSB conducted the investigation and contributed to data curation. THP and GMC contributed to software development and technical implementation. JHC, SBL, and SYK provided clinical resources and contributed to validation. YJK conceptualized and designed the study, developed the methodology, supervised the research, managed project administration, acquired funding and editing of the manuscript. All the authors have read and approved the final version of the manuscript.

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Figures

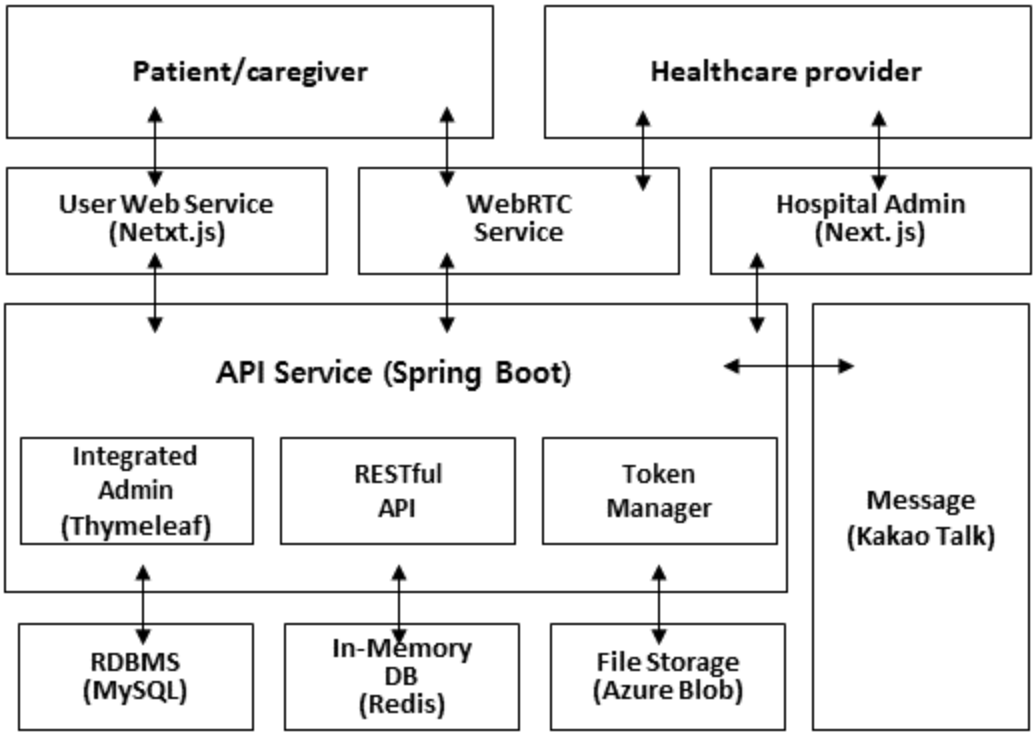

Figure 1

Platform software architecture

Abbreviations: API, application programming interface; RDBMS, relational database management system; DB, database; WebRTC, Web Real-Time Communication.

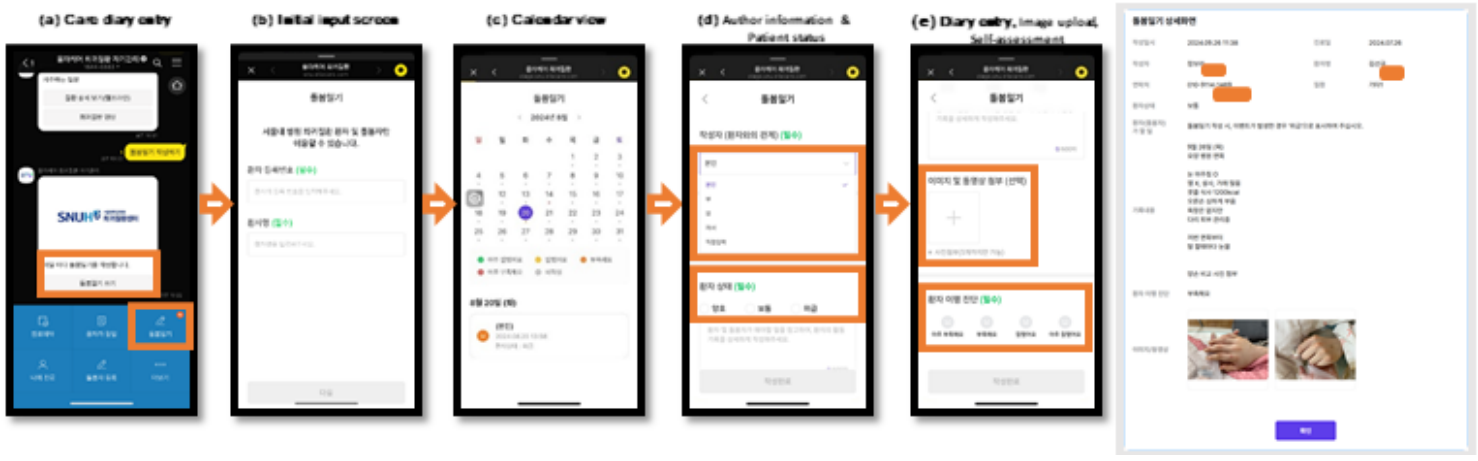

Figure 2

Care diary interface.
(a) Entry point for accessing the care diary from the application menu.
(b) Initial screen for entering patient identification information.
(c) Calendar-based interface for selecting and reviewing diary records.

(d) Form for entering author information and patient status.
(e) Care diary entry screen including free-text documentation, image upload, and self- assessment.

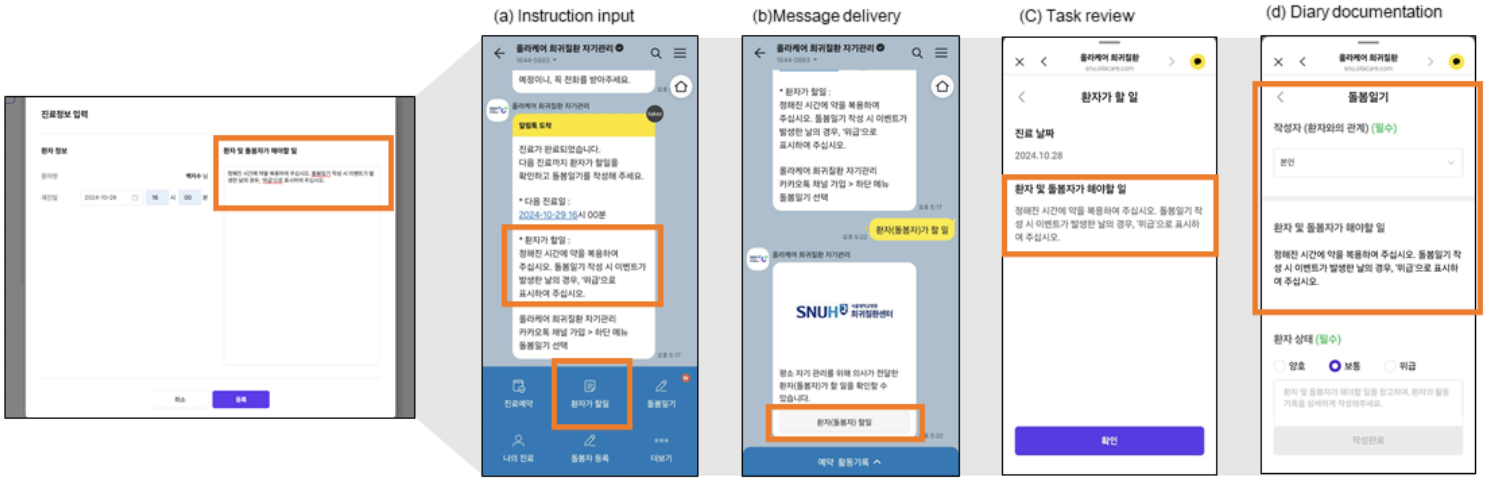

Figure 3

Physician instruction delivery interface.
(a) Physician input of patient-specific instructions.
(b) Automated delivery of instructions to the patient via KakaoTalk.
(c) Patient interface for reviewing assigned tasks.
(d) Documentation of physician instructions within the care diary.

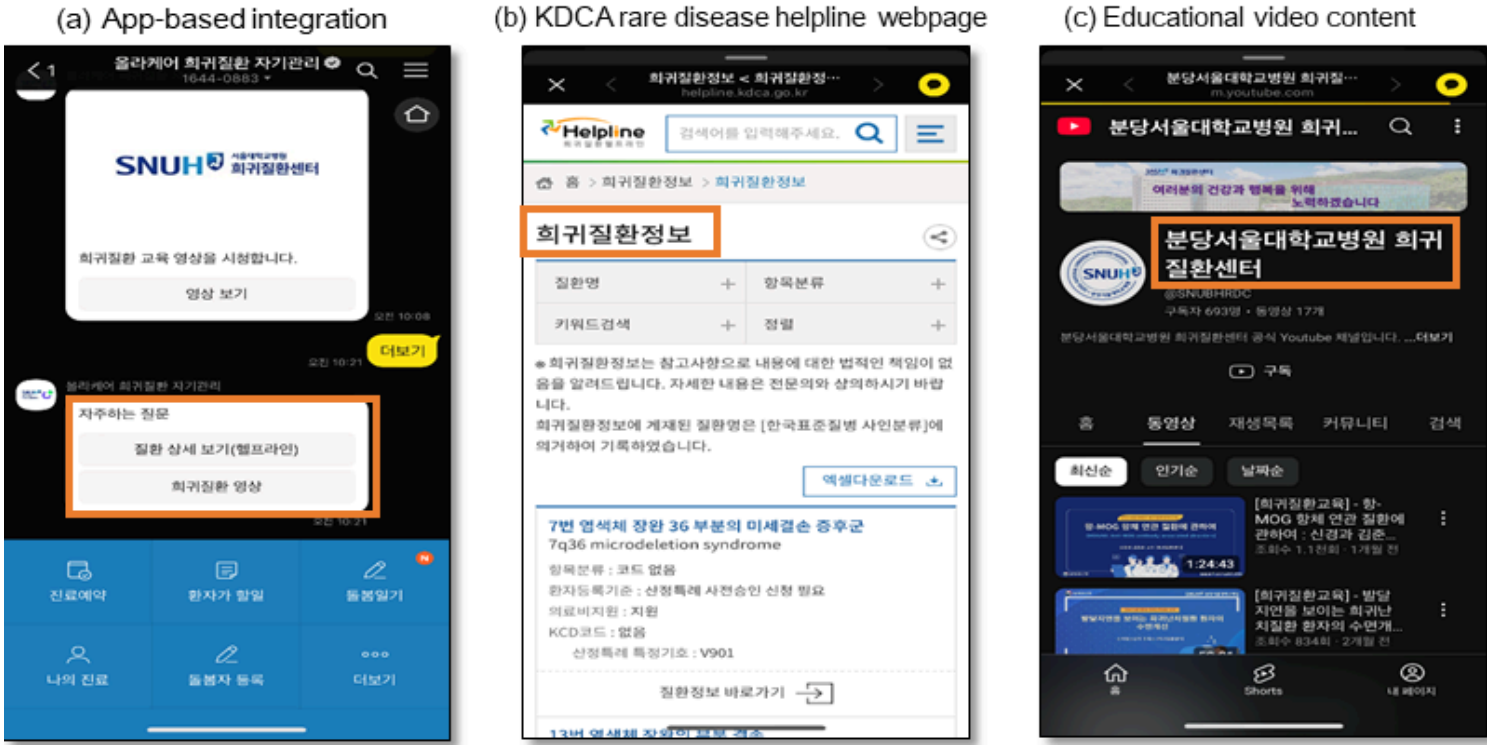

Figure 4

Integration with the Korea Disease Control and Prevention Agency Rare Disease Helpline.

(a) App-based interface linked to the helpline service.
(b) Webpage of the national rare disease information portal operated by KDCA.
(c) Educational video resources provided for patients and families.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- SupplementaryFile1Revised.pdf
- SupplementaryFile2Revised.pdf
- SupplementaryFile3Revised.pdf