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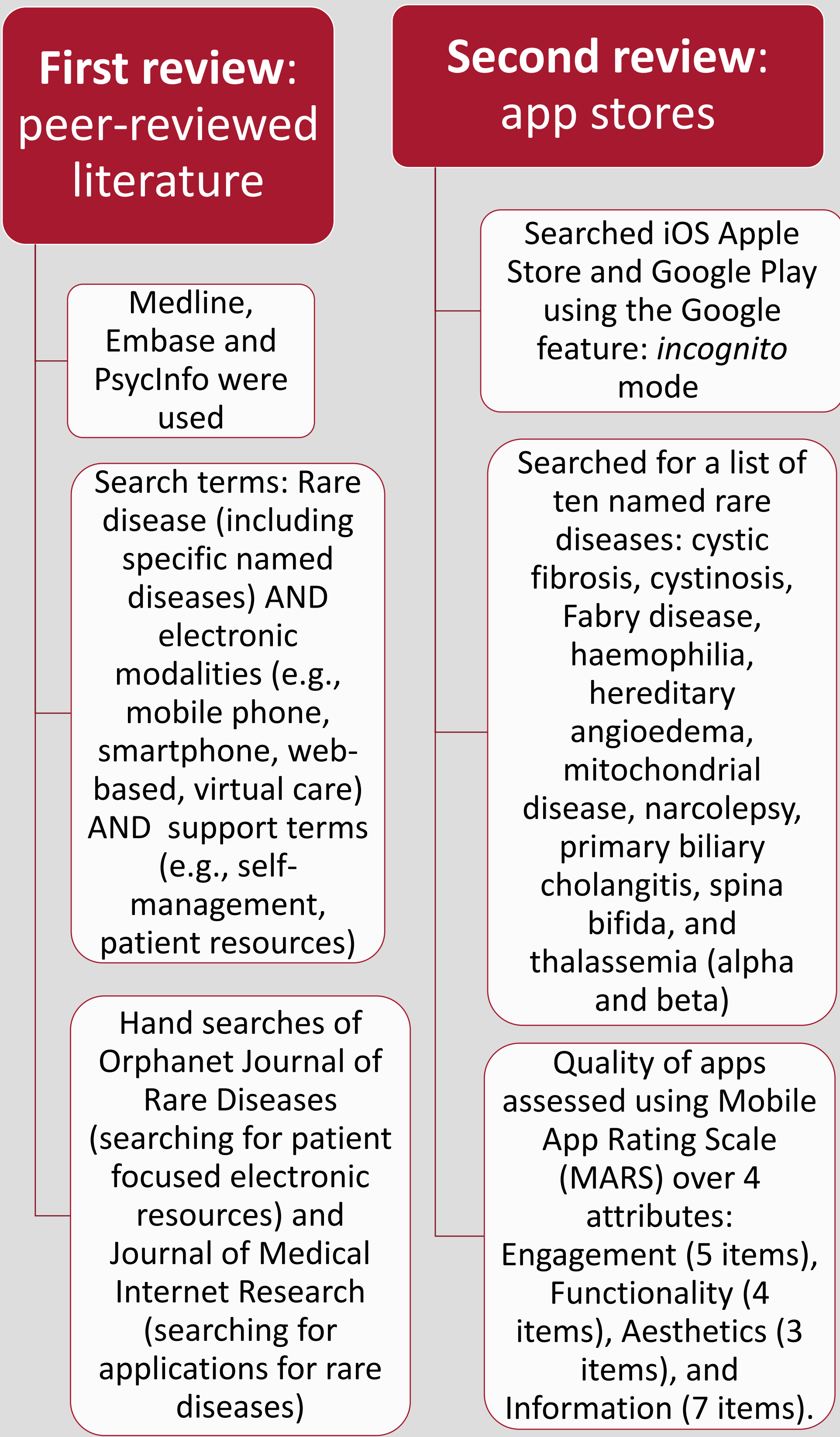
INTRODUCTION

Rare diseases are defined as having an incidence of less than 1:2,000 live births. While there are thought to be around 7,000 individual rare diseases, many health professionals may only see one case of a particular rare disease in their whole career. This leads to a number of unique issues for people with rare diseases. This study looked at these needs, and electronic resources that sought to meet this need.

METHODS

The **first** was a **scoping review** of Medline, Embase and PsycInfo, supplemented by hand searches of selected journals. We searched for articles using terms for rare disease, electronic modalities (eg, mobile phone, telehealth) and patient support terms. From this we extracted key patient needs that resources were addressing by examining the reasons that authors gave for developing the technologies.

The **second review** was of the two biggest mobile phone app stores, iOS Apple Store and Google Play. Since “rare disease” in this context returned very few apps, we searched for a list of ten named rare diseases: including cystic fibrosis, haemophilia, primary biliary cholangitis, Fabry disease, and spinal muscular atrophy. We assessed the quality of each app in terms of function, ease of use, and quality of content using the Mobile App Rating Scale (MARS).



RESULTS

The searches found 72 articles and 29 apps (screened for relevance from 383 papers and 2272 apps respectively). In the first review, the articles described 56 different electronic resources in five broad categories and identified four needs. Most common rare diseases addressed were **cystic fibrosis** and **haemophilia**. Types of electronic resources were:



- Apps for mobile phones or tablets
- Social networking platforms
- Telehealth and virtual care platforms
- Websites with interactive content
- Websites with just information

The needs of people with rare diseases were



1. Support for self-management
2. Access to high quality information
3. Access to appropriate specialist services
4. Social and emotional support

The second review found 29 apps that addressed 14 rare diseases or disease groups. The most common conditions that had an app were cystic fibrosis (n=6), haemophilia (n=5), and thalassemia (n=5). Apps had been developed to meet the four identified needs plus an additional one around participation in research. Most apps provided factual and visual information, providing tools for self-monitoring and resources to help improve interactions during health consultations. The quality of the apps varied greatly. Quality scores were mostly high for accuracy of information but low for engagement (lack of interactive features, interesting design).

CONCLUSIONS

We conclude that compared to people with common conditions (e.g., breast cancer or heart disease), people with rare diseases have additional needs including the lengthy odyssey to find a diagnosis, then identify appropriate specialists, the lack of evidence around effective treatments, guidelines or access to knowledgeable general health service providers. Electronic resources can meet many of these needs.

Developers of resources are recommended to consider ways to make appropriate apps more easily identifiable to consumers, to always include high-quality information, improve engagement, provide qualitative evaluations of the app, and include consumers and clinicians in the design. Otherwise, high quality care to patients who are most in need of it will not be provided in a sustainable, patient-centred way.

To read more ...



Mobile Apps for People With Rare Diseases: Review and Quality Assessment Using Mobile App Rating Scale

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