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Recognising patient expertise in the search for a rare diagnosis

DESIGN INSIGHT

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SUMMARY
Enola is a tool to help with the diagnosis of rare diseases. It was initially designed for use by clinicians, but testing with the rare disease community provided two key design insights which changed the deployment strategy: 1) people in the rare disease community often learn the medical terminology relevant to their conditions and 2) out of necessity, many people with rare disease become the local, if not international, expert on their disorder. This led to the decision to deploy the tool as a web-based application which can be accessed by anyone.

Key Words
Rare disease; diagnostic odyssey; patient empowerment; diagnostic assistance; human cognitive limits.

INTRODUCTION

Physicians learn approximately 200 diseases in their medical training, yet greater than 10,000 rare diseases are known to exist.[1] This massive knowledge gap leaves patients who have rare disease searching for an average of 4–5 years before they reach a correct diagnosis.[2] Although any given rare disease is by definition rare, collectively rare diseases are conservatively estimated to affect 1 in 20 people.[3]

SUMMARY

Diagnoses are missed when clinicians do not recognise the significance of a collection of signs and symptoms. This is especially problematic for rare diagnoses, where clinicians may have little to no knowledge of or experience with the patient's particular disease or syndrome.

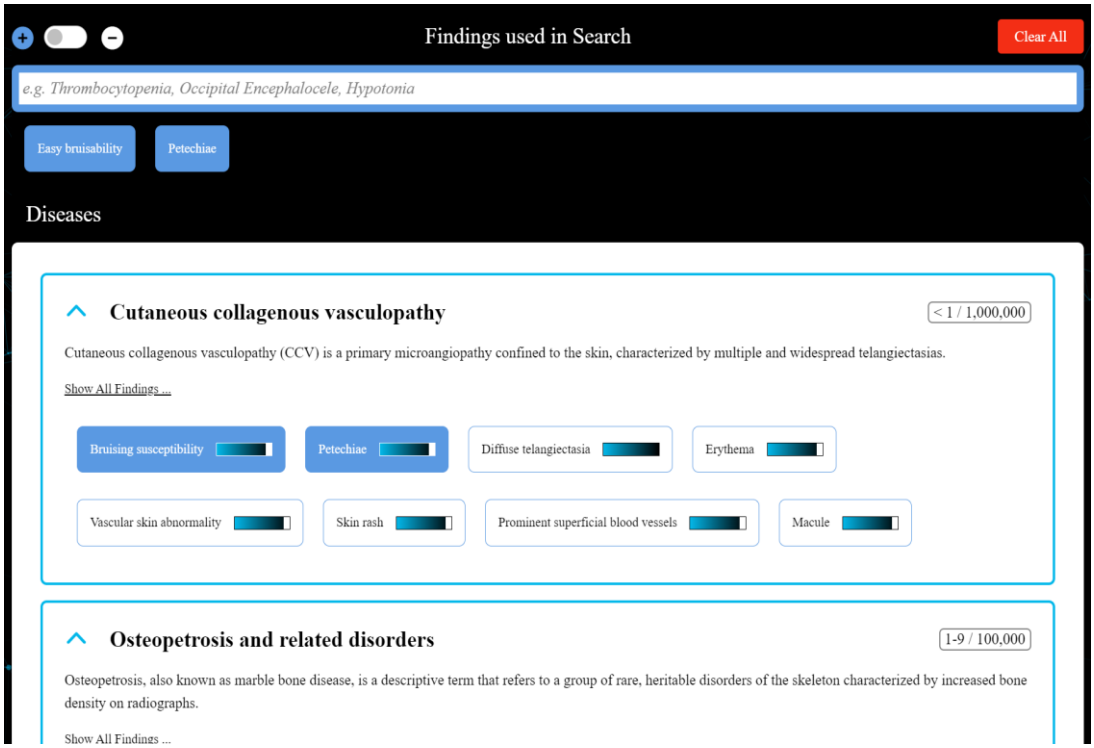
To address this problem, an application named Enola was developed, which accepts clinical findings as input and returns a list of all rare diseases that have any association to the input findings, sorted by the probability of disease. Users review the suggested diagnoses and modify the search by confirming or denying their associated signs and symptoms. See an example search with an explanation of the interface (Figure 1).

Enola draws on well-established data sources that include Orphanet[4] and the Human Phenotype Ontology.[5]

LESSONS LEARNED

Enola was initially designed for use by clinicians, but testing with the rare disease community provided a critical design insight that completely changed the deployment strategy. When lay persons with rare diseases who already knew their diagnoses had access to Enola, they were often quite successful in finding their diagnosis again, simply by typing in their signs and symptoms.

Figure 1. Enola rare disease search tool



There is a search box at the top, with a toggle button that allows the user to specify whether the clinical finding they are adding to the list is present (positive sign) or absent (negative sign). All of the clinical findings included in the current search appear just below the search box. The search results appear on the remainder of the screen, under the heading "Diseases". Each disease appears in a blue-bordered box, with the disease name in bold at the top left, the incidence and/or prevalence value(s) at the top right, with a paragraph describing the disease underneath followed by associated clinical findings. Each clinical finding has a blue bar beside it indicating how frequently that finding is associated with the disease.

The key insights gained from these encounters were as follows:

1. People in the rare disease community often learn the medical terminology relevant to their conditions; and
2. Out of necessity, many people with rare diseases become the local, if not international, expert on their disorder. While their expertise may not come with the imprimatur of a medical school diploma, it often does empower them to make diagnoses that are later confirmed by clinicians, and to make well-informed decisions regarding their medical care.

Sometimes the physician's challenge is to educate the patient, and sometimes it is to recognise the patient's expertise, and to listen.

Thanks to these insights, Enola, the web-based rare disease search tool was deployed for beta testing.

DESIGN INSIGHT

This AI-powered application, which suggests possible rare diseases based upon a person's symptoms, integrates AI with a very *human-centered design approach* into the diagnosis process. What an interesting association between AI and ***human-powered*** design. Many people with rare

diseases, through their own dogged research, become the local or international experts on their disorder. In using a human-centered approach to finding and solving problems, designers seek out these types of *expert-users* to help uncover unmet needs that might never be noticed without their collaboration.

The authors continue to explore and deepen the robustness of their system by reviewing existing "solved" cases of rare diseases. Their user interface with its simple + and – to identify and filter symptoms makes this software appear easy to approach for clinicians and friendly to a user that might have a lesser knowledge of technical/medical language.

Although doctors are still considered the most reliable source of health information, studies reported on in 2006[1] and 2018[2] suggest people who face serious health needs turn to the internet—roughly 69 per cent of adults in the U.S. chose the internet and 15 per cent chose healthcare providers as their first source of health information, respectively. These findings suggest a strong need for reliable information for these *expert-users* and, indeed, the broader population beyond to help find solutions for rare diseases. Why not expand this AI-powered application to include the 200 diseases that are taught in medical schools to become a resilient system for clinical users as well as *expert-users*?

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PEER REVIEW

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CONFLICTS OF INTEREST

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None