



Just how rare are rare diseases on Ada, a medical symptom checker?

Vanessa Lemarié, Simon Ronicke, Paul Wicks, Fiona Pick
Ada Health GmbH, Berlin, Germany

Background

When encountering a health problem, people increasingly turn to the internet. One digital tool is Ada, a conversational text-based app that uses AI with a database of doctor-curated medical knowledge to alert people to the possibility of diseases (including those that are rare) based on a constellation of health factors and user-reported symptoms.

We set out to identify what proportion of diseases suggested to our users during symptom assessments were classified as 'rare' under the common European definition of $<1 / 2,000$.¹

Method

Retrospective analysis of online health app database.

Results

To date, Ada has over 8 million users, who have completed over 15 million assessments. In 4% of assessments, rare diseases were suggested as the most likely and were among the top five suggestions in 17%.

The most commonly suggested rare diseases were infectious conditions (e.g. shigella), rheumatological conditions (e.g. systemic lupus erythematosus), and endocrine conditions (e.g. Cushing syndrome).² A subset of the estimated 7,000–10,000 rare diseases has so far been coded in our medical knowledge database, including those diseases coded in response to user feedback.

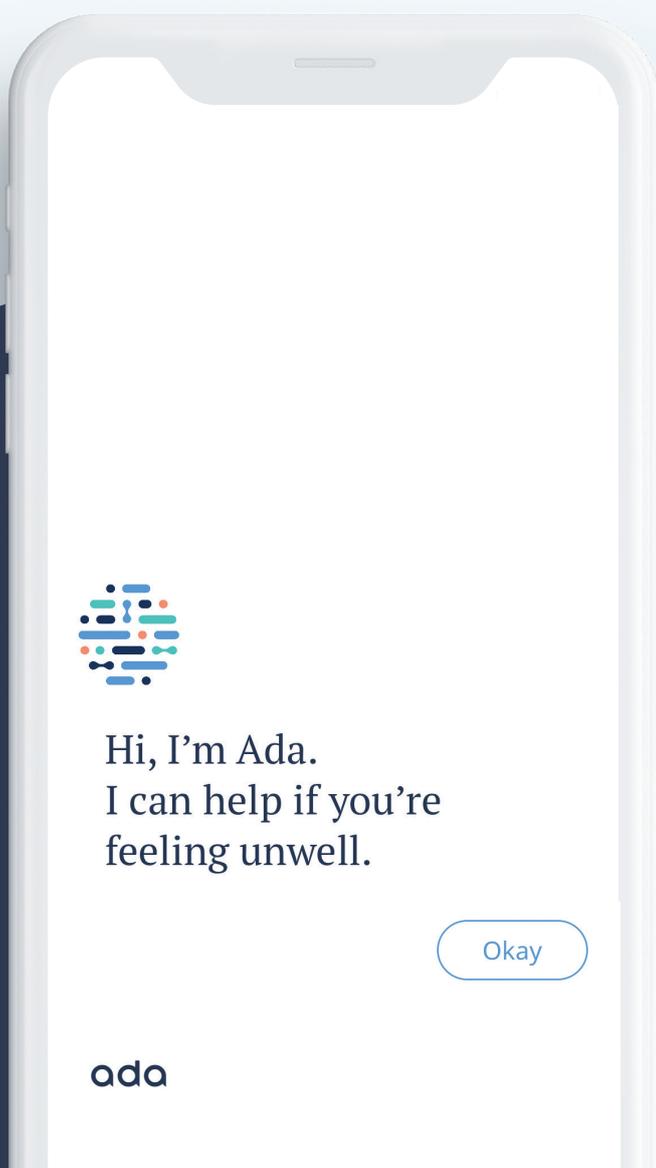
Discussion

People with a rare symptom presentation or complex diagnostic journey might be more likely to search for information through digital tools as they encounter long waiting times and multiple referrals across medical professionals.³

Epidemiological studies suggest around 2–4%¹ of the general population is living with a rare disease, which is about in line with how frequently rare diseases were suggested for each assessment. Signposting to appropriate care pathways in line with national guidance on standards of care may positively influence the diagnostic odyssey.³ Ada shows the potential to shorten the diagnostic odyssey of people living with rare diseases.

References:

1. Wakap et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* 28, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>
2. Haendel et al. How many rare diseases are there? *Nature Reviews Drug Discovery* 19, 77–78 (2020)
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