

# Motivations of Technology Use in Undiagnosed Rare Disease Patients

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I am a third year PhD student at Swansea University working with Amicus Therapeutics to research how technology can empower rare disease patients to play a more active role in their diagnosis. My research uses machine learning approaches for peer matching through an iterative design process with patients.

We conducted a systematic review on decision support systems (DSSs) for rare disease diagnosis. We did this by firstly collating sources which illustrated a typical rare disease patient's journey to diagnosis. We then reviewed the literature using this journey as a lens to evaluate the suitability of DSSs in a rare disease context. This revealed that a DSS for rare disease diagnosis needs to accommodate for the number and variety of clinicians involved in a rare diagnosis as well as continued use since the first consultation rarely leads to a diagnosis. Since patients are the constant throughout this diagnostic process, let us consider how technologies can support patients pre-diagnosis.

Health technologies empower patients to play a more active role in their health and encourage better healthcare decisions. This is particularly important for rare disease patients who, out of necessity, become experts in their condition. However, patient engagement with technology is hard to encourage if it is not perceived as beneficial. To understand what is beneficial to rare disease patients, we ran two workshops with five rare disease patients to investigate the key challenges that they face and explore how technology can help alleviate these burdens. A thematic analysis of the first workshop revealed that some of the key challenges faced by rare disease patients were: a perceived lack of care from healthcare professionals, with one participant stating “*no one's listening to me*” and difficulty explaining experiences to peers as they “*still do not understand*” ten years after their diagnosis.

The second workshop explored how technology can alleviate these problems. A symptom logger which could be shared with the users' clinician was suggested in order to alleviate issues with healthcare professionals. Collaborative filtering could then be used to match patients to people like them based on their logged symptoms. Participants were keen on these concepts with one participant expressing that this “*would be beneficial*”, despite previously stating they were against using pre-diagnostic technology. Therefore, by exploring user-needs and then discussing what would be beneficial to patients, we can motivate patients to engage with technology who were previously uninterested.

To further this work, we will evaluate the peer matching collaborative filtering algorithm we created in order to assess how it affects patients' experiences of diagnosis and their abilities to find relevant health information. It is also important to conduct future work on how to maintain engagement from clinicians' perspective as well as patients for technologies involving patients and clinicians to be effective in practice.