

## Introduction

Rare diseases are defined by the National Institutes of Health as diseases that affect fewer than 200,000 people. In total, rare diseases affect as many as 30 million people in the United States alone and are often responsible for protracted diagnostic journeys associated with significant morbidity.<sup>1</sup>

To better understand the obstacles impeding diagnosis, we surveyed 101 clinicians and 150 patients and caregivers in the rare disease community. In this study, we identified some of the barriers and their impact on patients and clinicians traversing the rare disease diagnostic process.

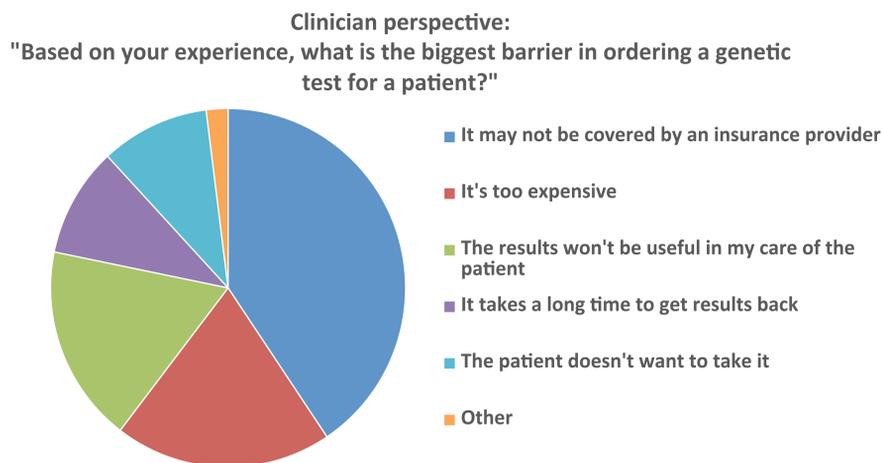
## Methods

The survey was conducted between September 2015 and October 2015. Survey participants were ascertained from a pool of candidate individuals who had previously opted to participate in research surveys. One hundred fifty patients or caregivers of patients who self-identified as being diagnosed with a rare disease and 101 clinicians who self-identified as caring for patients with rare disease responded to an email invitation for an online survey. This study was approved for exemption by the Western Institutional Review Board.

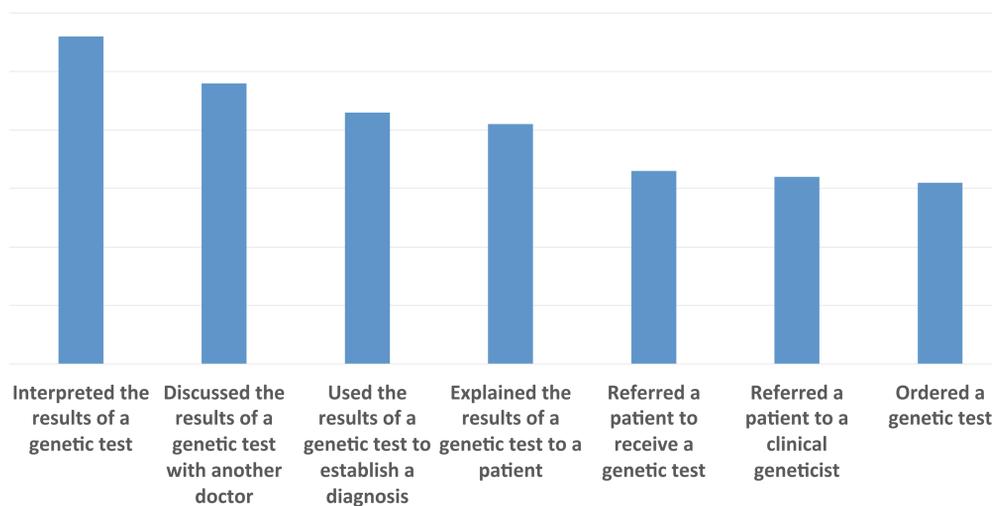
## Clinician findings

Physicians observed that patients with a rare disease received, on average, four to five diagnoses and 14 diagnostic tests or procedures during their diagnostic journey. They reported the lack of information about genetic testing (54%) as an obstacle, with 83% agreeing that such testing is useful and 69% observing that it shortens the time to a diagnosis.

Lack of information about the cost of testing (49%) and about indications for referral to a clinical geneticist (31%) were also obstacles. Sixty-seven percent (67%) of physicians indicated that the absence of a confirmed diagnosis for rare disease patients hinders their ability to care for these patients, and approximately 45% indicated that more resources are needed for effective diagnosis of rare diseases.



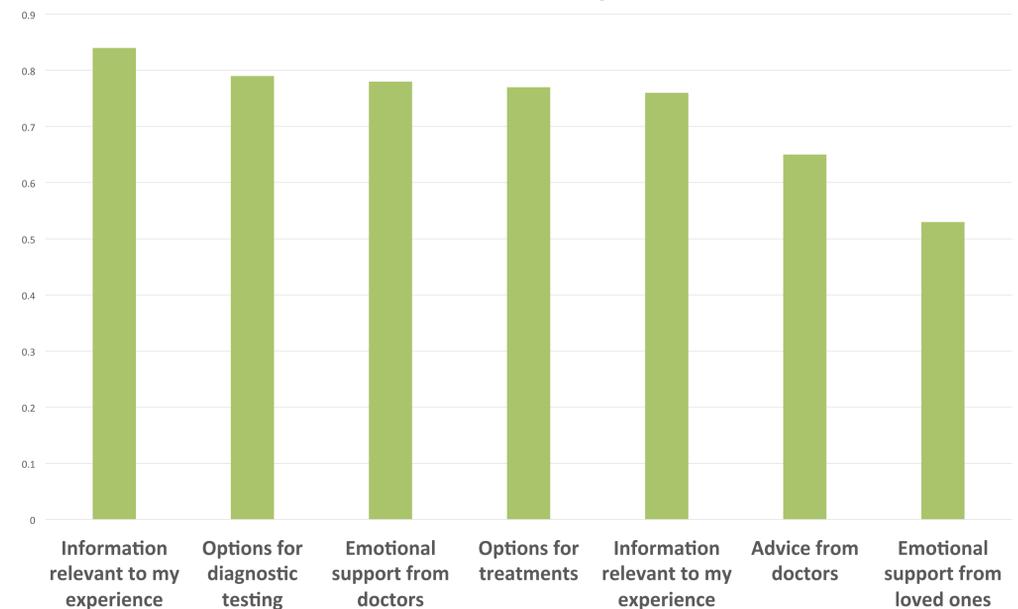
**Clinician perspective: "I have never..."**



## Patient findings

Patients reported receiving, on average, two to three preliminary diagnoses and six to seven diagnostic tests and/or procedures during their diagnostic journeys. These journeys lasted an average of 3.9 years. Approximately 23% of patients received several genetic tests and 29% of patients felt that they had paid for ineffective treatments or interventions that had been based on an incorrect diagnosis. Obstacles for patients on the diagnostic journey included lack of information (76%) and inadequate insurance coverage (21%), resulting in 60% of patients incurring unexpected financial burden and 29% going into debt.

**"During the time it took for me to receive a correct diagnosis, I don't feel that I received enough..."**



Selected queries	Average of patient Responses
Number of incorrect diagnoses before correct one obtained	3
Time to arrive on a diagnosis	4 years
Number of diagnostic tests	7
Longest period of time to receive a diagnostic test result	15 days

## Conclusion

This study observed that both patients and clinicians face significant obstacles navigating the diagnosis of rare diseases, including lack of information, time required, and cost. These observations suggest a need for more educational resources for patients and clinicians regarding the role of diagnostic genetic testing, for earlier implementation of appropriate genetic testing, and for mechanisms to make genetic testing more accessible financially.

As the field of molecular diagnostics continues to expand, the need for education, accessibility, and clinical support becomes ever more critical for patient care. Although this study's cohorts are relatively small, we propose that they highlight important challenges of the rare disease community as a whole. These observations suggest that further research is needed to understand the obstacles faced and to identify solutions that expedite the successful completion of a patient's personal diagnostic journey.

## Reference