

2022 REPORT

THE LONG AND WINDING ROAD TO CARE:

Today's Journey to
Diagnosis and Treatment
for Patients Living with
Complex Conditions,
including Rare Diseases



MediFind



Introduction

The COVID-19 pandemic has driven many changes in the healthcare system, both positive and negative.

On the upside, telehealth adoption and access has been dramatically accelerated, making primary care in particular more accessible and flexible. On the downside, patients receiving specialized care have faced many new challenges, including [delayed procedures](#), travel restrictions preventing them from accessing care, and the [cancellation or delay of promising clinical trials](#) in which they may have participated. These issues are especially problematic for families facing serious, life-threatening, or rare diseases.

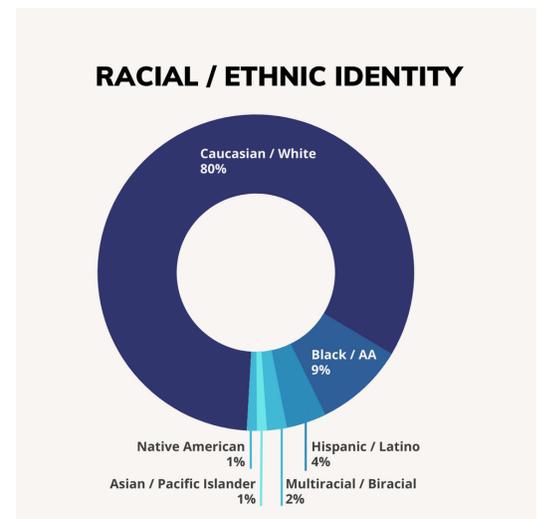
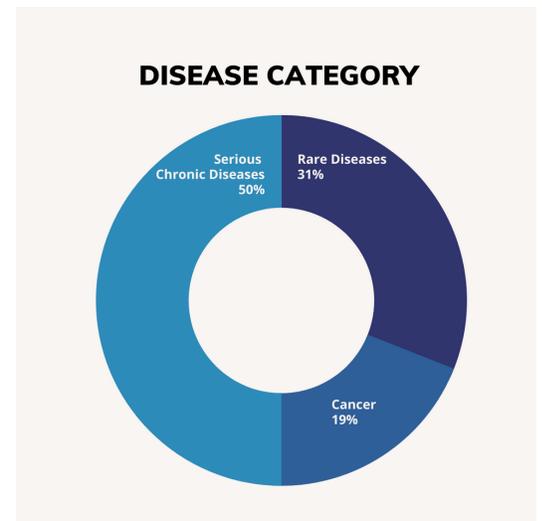
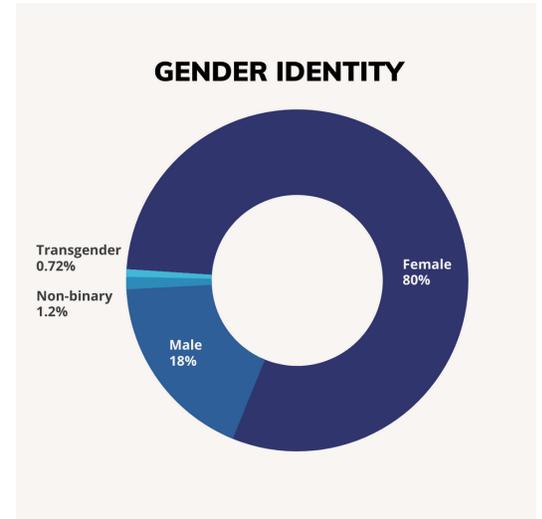
While these challenges are finally beginning to be resolved, other more persistent challenges remain for these patients and their families. To gain a better sense of these issues, MediFind and Rare Patient Voice – two companies dedicated to helping patients living with serious, life-threatening, or rare diseases pursue the best healthcare – joined together to conduct a survey of over 1000 such patients in late 2021.

About this study

This report is based on a survey of 1,229 patients living with a wide range of health conditions, all of whom are members of the [Rare Patient Voice](#) community in the United States. The respondents overwhelmingly identify as female (80%), in line with the general [finding](#) that women tend to be more proactive and engaged in healthcare decisions. The remaining respondents who chose to provide information about their gender identify as male (18%), non-binary (1.16%), and transgender (0.72%).

About 1/3 of respondents (31%) have an officially classified rare disease, using the EU definition of a disease affecting fewer than 1 in 2000 individuals and validated against the OrphaNet database. About 1/5 (19%) have some form of cancer (some of which themselves are rare diseases). The vast majority of the remainder have serious chronic diseases. We refer collectively to the group of patients living with a serious chronic disease, cancer, or rare disease as having a “complex” condition, which characterizes the nature of the respondents to this survey.

In terms of insurance coverage, nearly half of respondents (49%) have private insurance, while most of the remainder have government-provided insurance (47%). In terms of race and ethnic identity, the vast majority of respondents identify as White or Caucasian (81%), followed by Black or African-American (9%), Hispanic or Latino (4%), Multiracial or Biracial (2%), Asian or Pacific Islander (1%), and Native American or Alaska Native (1%).



Highlights

1 The path to successful diagnosis and care can be particularly burdensome for some patient groups

More than half of all patients (57%) were misdiagnosed before receiving the correct diagnosis, with this percentage rising to nearly two-thirds (65%) for patients with a rare disease. Rates of misdiagnosis are even higher for respondents identifying as female (51%), transgender (60%), or non-binary (56%) compared to male (37%), and those identifying as multiracial or biracial (67%) compared to around 40–50% across all other racial identities.

2 Genetic testing is changing the game of diagnosis, but not for all

The rate of genetic testing for diagnostic purposes has doubled in the last 5 years alone, primarily among those diagnosed with cancer within the last 2 years. But less than 20% of those living with rare diseases report receiving genetic testing.

3 Clinical expertise drives provider choice, but patients (and even many doctors) have no reliable way of assessing it

While general health consumers prioritize insurance coverage as the most important factor when choosing a doctor, both [physicians](#) and those living with complex disease (that is, a serious, life-threatening, or rare disease) overwhelmingly prioritize clinical expertise in their conditions. For these patients, insurance coverage, appointment availability, location, provider ratings and reviews, and even reputation of the health system are not near the top of the list. Unfortunately, patients (and [doctors](#) in many cases) have no clear way of judging clinical expertise.

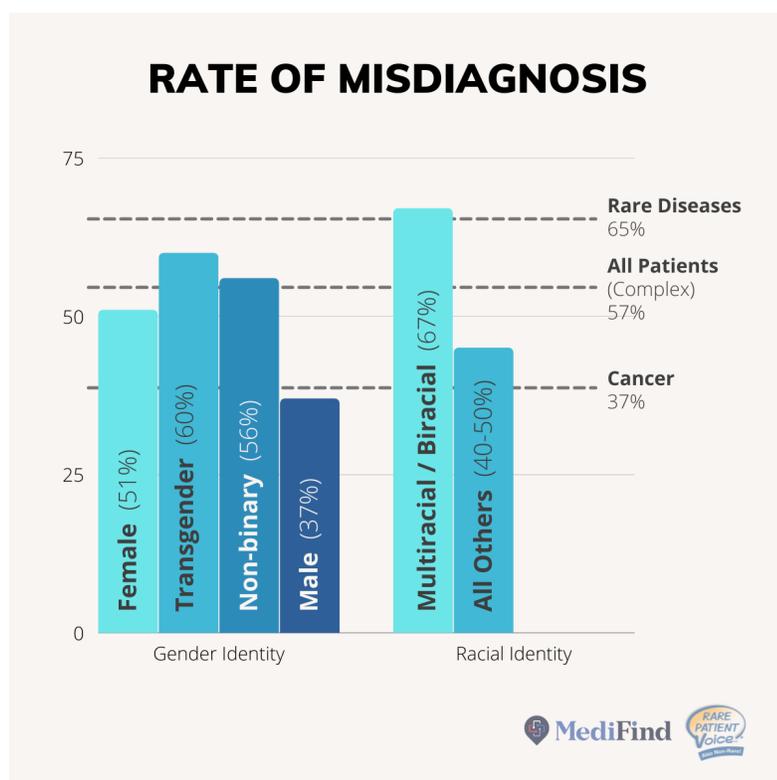
Discussion

The diagnostic journey remains burdensome, but unequally so

Almost one third (31%) of respondents were properly diagnosed within three months, while about one-fifth (21%) took five or more years to get correctly diagnosed. As expected, patients with rare diseases took significantly longer to be correctly diagnosed: only 23% were diagnosed within 3 months, while it took five or more years for 28% of patients. This is in stark contrast to patients with cancer, where two-thirds were diagnosed within 3 months, and only 1% took five or more years to be properly diagnosed.

Ominously, more than half of patients (57%) were misdiagnosed before receiving the correct diagnosis. And this percentage jumps to nearly two-thirds (65%) for patients with a rare disease. Notably, while still unacceptably high, cancer patients were dramatically less likely to be misdiagnosed, with only 37% of patients with cancer having been misdiagnosed first.

Respondents who identify as female report being misdiagnosed at a higher rate than male (51% vs. 37% respectively). While sample size was extremely limited, respondents who identify as transgender or non-binary report even higher rates of misdiagnosis (60% and 56% respectively). When looking at racial differences, respondents who identify as multiracial or biracial report the highest rate of misdiagnosis (67%), compared to a rate of around 40–50% across all other racial identities.



Even when a patient is not misdiagnosed, they still see multiple doctors on their path to finding the right diagnosis. On average, patients report seeing 4 different doctors before getting correctly diagnosed. This number jumps to almost 6 different doctors when the patient has a rare disease.

This is generally in line with findings from the [2021 National Economic Burden of Rare Disease Study](#), which reports that rare disease patients see an average of 4.2 primary care physicians and 4.8 specialists prior to receiving the correct diagnosis. Interestingly, our data shows that a patient only needs to see 2-3 (average = 2.5) different doctors to get correctly diagnosed with cancer.

PATIENTS TELL US:

WHAT COULD HAVE LED TO A FASTER CORRECT DIAGNOSIS?

1. DOCTORS WHO ACTUALLY LISTEN

"Doctor dismissed me time after time when I sought treatment or answers. Proper diagnostic questioning and proper diagnostic testing would have made all the difference"

"Doctors who are willing to actively seek out an answer"

"If my doctor had listened and not decided it was psychosomatic"

"If the family doctor did not think he was God. He would not give me any referrals when I kept asking"

"Doctors taking patients seriously and listening"

"Doctors who actually listened"

"Doctors who LISTEN, not assume"

"Not assuming that because I'm a woman my problems are expected, exaggerated or just anxiety"

"Less reliance on strict criteria for diagnosis and more listening to the patient's experience and symptoms"

"Doctors who are dedicated to the well being of their patients, rather than punching a ticket. Too few MDs enter their profession with a drive to help people"

2. BETTER ACCESS TO EXPERTS

"Visiting the right specialist, more education among doctors"

"Getting to the right doctor"

"Willingness to think outside the box and consider rare conditions rather than brush them off because they were taught not to "think horses, not zebras"

"I changed doctors to get better care"

"Having doctors who were familiar with other diseases, rather than just the most common ones. Also having doctors who spend time with you during appointments and really listen to you so that they can get to the bottom of things"

"Easier access to specialists"

"More awareness of my rare disease in the medical field"

3. A MORE COORDINATED APPROACH

"If all the doctors I saw at different locations could share information and history so they could see the whole picture instead of pieces"

"Better testing and a holistic approach to analyzing my condition in correlation with other health issues"

"Doctors and Specialists all talking together and working together"

"Doctors working together better"

"A holistic approach instead of one body part at time"

"Coordination of services - if a patient is being sent to multiple, diverse specialists - time to get escalated/referred to a place like Mayo Clinic"

4. BETTER, FASTER TESTING

"Earlier testing"

"More testing covered by insurance"

"More relevant testing, being more open-minded"

"Accurate tests. Accurate readings. Better equipment"

"Standardized genetic testing"

"Just wish you could get tests faster and results back faster"

Q: "What do you think could have led to a faster correct diagnosis, if anything?"



Genetic testing is shortening time-to-diagnosis, but progress is uneven

The good news is that advances in diagnostic capabilities and technologies have led to a shorter time-to-diagnosis over the past decade. For example, over half (51%) of the patients diagnosed within the last year were correctly diagnosed within 3 months. This is up considerably from 5 years ago, where only one quarter (26%) of patients report being correctly diagnosed within 3 months.

One possible reason for shortened time-to-diagnosis is the increasing use of genetic testing. Over the past two years, almost 4 in 10 patients (38%) received genetic testing. This is up from the rate of 3 in 10 who had genetic testing when diagnosed 3–4 years ago. The share of patients receiving genetic testing has essentially doubled over the past 5 years as only 2 in 10 patients had genetic tests when diagnosed five or more years ago.

Genetic testing has become particularly prevalent for patients with cancer as three quarters (75%) of patients diagnosed with cancer over the last two years have been genetically tested. Given the rapid development of precision medicine including targeted treatments, the corresponding rise in genetic testing is perhaps not surprising. What is more surprising is that genetic testing is not a mainstay of rare disease diagnosis, with only 20% of patients diagnosed with a rare disease in the past two years reporting that they received genetic testing. This suggests opportunity for improvement, particularly among the [39%](#) of rare diseases that have an identifiable genetic cause.

How clinical expertise drives provider choice

Provider referrals still rule in terms of finding a skilled specialist, regardless of disease category

Patients find their doctor through multiple paths, but nearly half (45%) of patients found the doctor who correctly diagnosed them through a referral from another doctor, while 20% of patients did so through their own research. Both figures remain stable over time, as patients implicitly trust the long-standing referral process. These findings are consistent with an [annual study conducted by Kyruus](#), which surveys health consumers to assess the generational differences in attitudes and behaviors related to finding a physician. Though not specifically reported by Kyruus, we are operating under the assumption that the health status of their respondents is largely in line with that of the greater US population. As such, we take their data as a useful baseline comparator to our study population, which over-indexes on serious, life-threatening, and rare diseases. In their most recent analysis (2020), Kyruus reports that 39% of respondents found their specialist through a recommendation from a provider, while 22% did so through their own research.

When we look specifically at patients living with a rare condition, physician referral is still the number one way that patients find their doctor. However, there is also a large jump in the percentage who find their doctor through their own research (going from 18% for non-rare patients to 24% for rare patients), indicating some potential dissatisfaction in the existing system.

WHAT DRIVES PHYSICIAN REFERRALS?

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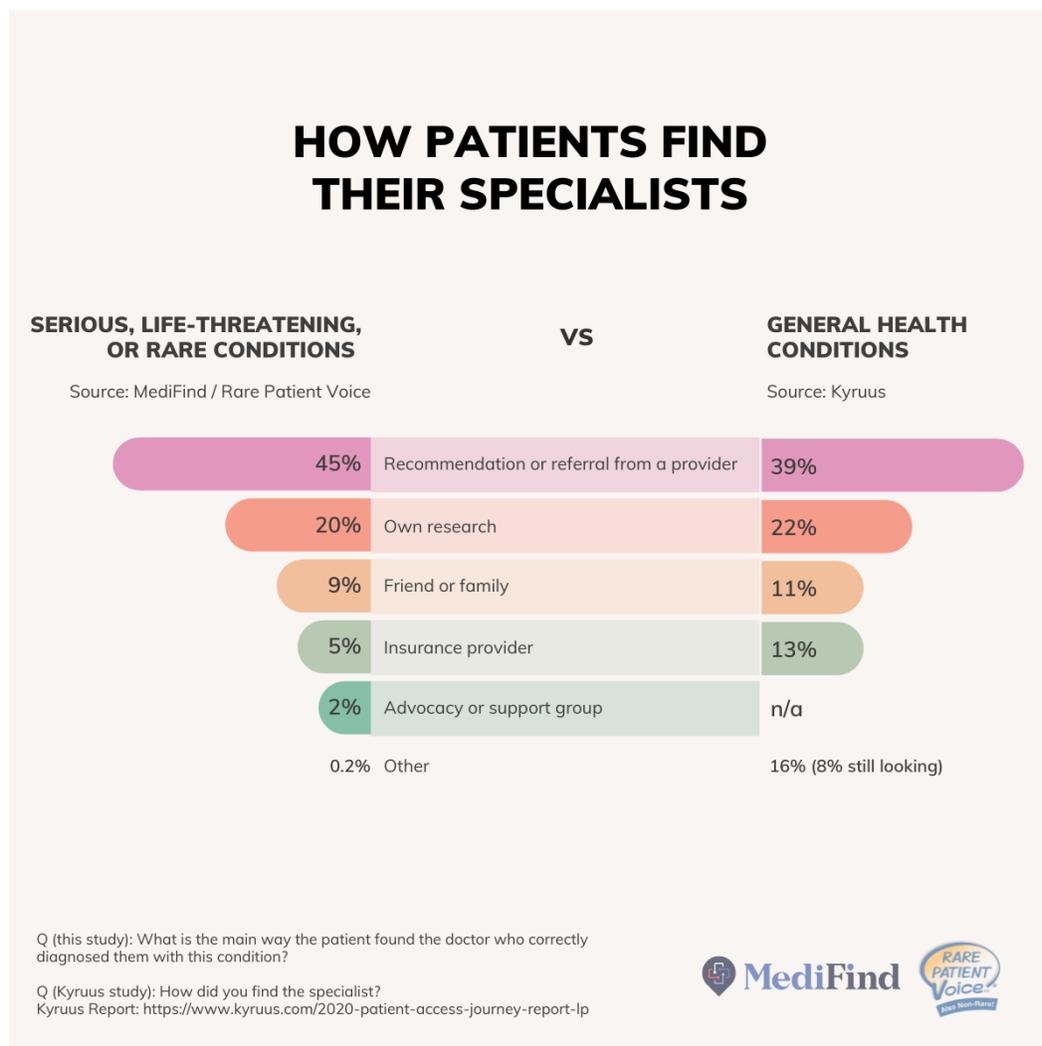
Although medical skill was of greatest importance, how physicians make the determination of the specialist’s skill is not completely clear.

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Source: Kinchen KS, Cooper LA, Levine D, Wang NY, Powe NR. Referral of patients to specialists: factors affecting choice of specialist by primary care physicians. *Ann Fam Med.* 2004;2(3):245-252. doi:10.1370/afm.68



The fact that referral is still the dominant way that patients find doctors is likely unsurprising. In fact, both [doctors](#) and patients (as discussed in the next section) report medical/clinical expertise as the most important factor in choosing a physician (in the case of doctors, they're choosing the specialist to whom to refer their patients). What is more surprising is that neither doctors nor patients [have a reliable way to gauge expertise](#) even though it's clearly (and appropriately) the most important factor in this decision.



A [study](#) on referral patterns of primary care physicians to specialists states: “Although medical skill was of greatest importance, how physicians make the determination of the specialist’s skill is not completely clear.” Further, [conflicting incentives](#) now shaping referral patterns at large threaten to undermine patient trust in the current system. The opportunity exists to provide objective data that can help both patients and their doctors make more informed decisions about referrals based on clinical expertise.

Patients with cancer and rare diseases value clinical expertise even more strongly

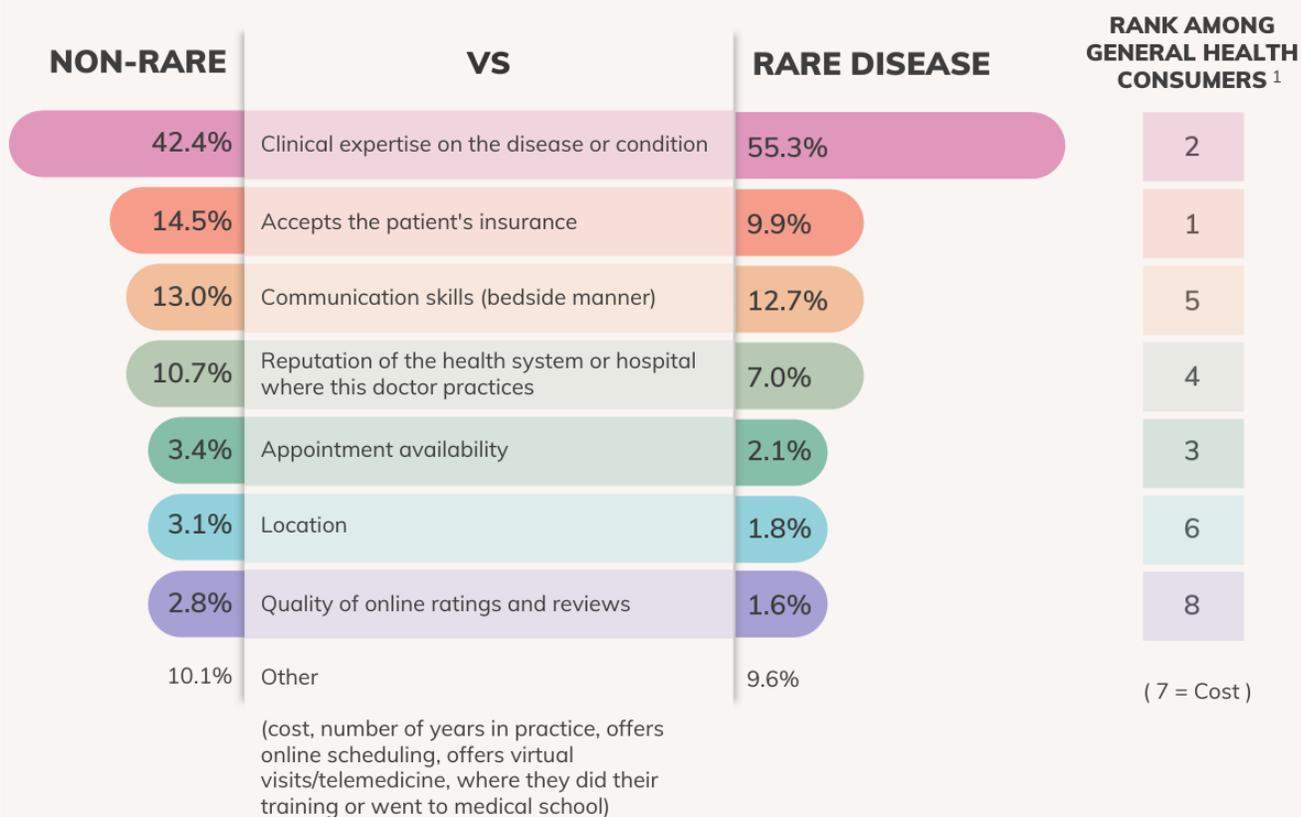
In terms of choosing their treating physician, clinical expertise is far and away the primary factor driving this important decision among our respondents, most of whom are living with complex health conditions. Nearly one-half (47%) of patients identify expertise in their specific disease/condition as the single most important factor in choosing a doctor. For patients with a rare disease, expertise is even more critical, as 55% indicate it to be the most important attribute when choosing a doctor. Insurance coverage and good communication skills (bedside manner) are the next most important reasons for choosing a treating physician among our respondents.

Again, the Kyruus study offers a useful comparator from a more general population, reporting that the most important criterion to consumers when selecting a provider is insurance coverage, followed by clinical expertise on their condition, appointment availability, reputation of health system or hospital, communication skills, location, cost, and so on. (Note: While overall rankings can be compared between our study and Kyruus', care should be taken when comparing percentages due to differences in question structure: our study required respondents to make a forced choice of "the most important factor" while the Kyruus study allowed for rating of each factor individually.)



Priorities for patients living with rare diseases look to be decidedly different from the general population. As mentioned, 55% report that clinical expertise in their disease/condition is the most important factor for choosing a doctor. Insurance is even less important to rare disease patients than non-rare patients, with 10% reporting insurance as the most important factor compared to 15% for non-rare patients. Fully 93% of respondents in the Kyruus study rated insurance coverage as “extremely” or “very important,” making it the overall most important factor driving physician choice.

MOST IMPORTANT CRITERIA TO PATIENTS WHEN CHOOSING A TREATING PHYSICIAN



Q: What is the most important characteristic in terms of choosing a doctor for diagnosing or treating the health condition with which the patient was diagnosed?

1. Kyruus Report: <https://www.kyruus.com/2020-patient-access-journey-report-lp>



Given the high stakes, rare disease patients may be keen to seek out expertise regardless of where it resides. They consider reputation of the health system less important than non-rare patients (7% versus 11%; 83% of Kyruus respondents rated health system reputation as “extremely” or “very important”). Location is a distant consideration, particularly for rare disease patients; it’s the primary factor in physician choice for only 1.8% of rare disease patients compared with 3.1% of non-rare patients. Given that a [study from NORD](#) reports that 17% of rare disease patients have permanently relocated to access medical care related to their disease, and the Burden of Rare Disease Study reports that rare disease patient take an average of 2.4 out-of-state trips related to their diagnosis, location seems to be an obstacle that rare disease patients are willing to conquer when necessary. In contrast, 76% of Kyruus respondents rated location as “extremely” or “very important.”

Interestingly, in neither our study nor Kyruus’ did quality of online patient ratings and reviews rank highly in terms of driving provider choice (8th most important factor in both studies). While many patients consult online review platforms, and Google prioritizes such platforms in their search results, physician ratings and reviews have actually been shown [not to correlate](#) with quality of care. Clearly the ability to assess providers based on more important criteria, such as expertise and insurance coverage, would be welcome.

PATIENTS TELL US:

WHAT DIFFERENCE CAN FINDING THE RIGHT DOCTOR MAKE?

1. DRAMATICALLY BETTER OUTCOMES

"I lived"

"Much better quality of life"

"Getting the appropriate treatments"

"Life or death finding a doctor who knew how to diagnose and then know how to treat"

"Night and day!!! Not so much for the diagnosis - but prescribing the right medicine"

"Relief sooner and the ability to be able to live somewhat normally"

"I finally got the treatment I needed for so long"

"Finding the right doctor has meant being placed on the proper treatment. With the proper treatment, I have been relapse free for 15 years, and been able to improve"

"Finally knowing what's wrong"

"It has literally been life changing. I went from being barely functional and having to be taken care of by family, to being able to be independent again and even go back to work part time. I can stay awake, I can focus, I can learn and remember things, I can drive, all things I couldn't before I found the right doctor. This doctor have he the proper diagnosis and helped me to find the right medication. I don't have the proper words to emphasize just how much of a difference she has made"

"Time is of the essence. Precious time"

"Finding the right doctor can be the difference between living a healthy life and becoming handicapped and confined to a wheelchair"

"Saved my life!"

"Having the right doctor has given me my life back. I am receiving proper treatment, my infection rate is much lower, and I do not fear hospitalization for every infection I now have"

"I feel like my chance of survival is increased in his care"

"The difference has been remission"

"I received an accurate diagnosis and treatment, which has given me back a quality life"

"It made all the difference. I went from suddenly becoming bedridden and unable to work to having an answer for my symptoms with a plan to get me to being more stable and functional"

"Expedited care plan"

"It has made a world of difference. I now drive over 3 hours to a different state once a month just to see a doctor who has been able to help me for the last 10 years or so"

"Finding the right Dr saved my life"

2. MORE TRUST + CONFIDENCE

"The difference is the confidence that the best practices and treatment options are known"

"The right doctor is validating, helps get to the solution and feel improved and hopeful"

"It gave me great confidence in the diagnosis"

"The way that a doctor makes a patient feel is extremely important. The way that the doctor talks to a patient. The way that they explain things to them. The very way that they break the news to them is so important. It can mean the difference between scaring someone for life and giving someone hope that they will still have a life with this disease...Getting the right doctor is so important to your journey"

"A world of difference - I feel comfortable going to my doctor and have confidence in her judgement"

"Ease of mind knowing I have a doctor I can trust in the decision making of helping my disorder"

"Communication, mutual respect, ability to ask any question and get a meaningful answer, mutual interest in research"

"Less worry"

"It makes all the difference. You want someone who understands how you are feeling and that you want to get better soon"

"Their knowledge and reassurance that I am pursuing the correct actions"

"Being believed and taken seriously has helped with confidence overall, and quality of life"

Q: "What difference, if any, has finding the right doctor made to the patient?"



Conclusion

Patients living with serious, life-threatening, or rare diseases face numerous challenges along their diagnostic and treatment journeys. Misdiagnosis is extremely common among patients living with these diseases, but even more so for patients who also identify as female, non-binary, or transgender, as well as those who identify as multiracial or biracial. Genetic testing has dramatically improved the diagnostic and treatment experience for patients living with cancer, but there is still much unrealized potential for similar improvement among those living with rare diseases, where progress has been much slower. In terms of choosing a provider, there is consensus among these complex patients that clinical expertise is of the utmost importance, especially for those living with a rare disease. However, there remains opportunity to help both patients and physicians make more informed assessments of provider expertise to this end.

Given the many differences noted throughout this report, it's clear that the opinions and needs of rare disease patients should be specifically sought out by those working to improve the healthcare system, so it can more equitably serve patients and their families, including those with the highest need.

About the authors

MediFind

MediFind uses advanced analytics to connect patients facing complicated, life-threatening, and rare diseases with leading health expertise. Inspired by a story of deep personal loss and backed by the Merck Global Health Innovation Fund, MediFind launched on Rare Disease Day 2020 with the goal of restoring objectivity to health decision-making. By democratizing access to essential information, the team has already helped over 1 million patients find the best doctors based on real-world medical expertise, not marketing effort.

Visit us: www.medifind.com

Rare Patient Voice

Rare Patient Voice empowers patients and caregivers to share their opinions and experiences share their voices with researchers and companies developing products, devices, and treatments to improve lives. The RPV community includes over 100,000 patients and caregivers across more than 700 diseases, both rare and non-rare, in the US, Canada, UK, France, Germany, Italy, Spain, Europe, Australia, and New Zealand.

Visit us: www.rarepatientvoice.com