

# RARE DISEASE IN THE U.S. 2025

By Susannah Fox

## Introduction

People living with rare diseases and their families are often at the forefront of technology, science, and health policy advancements:

- Rare disease advocates were instrumental in passing the 1983 Orphan Drug Act, the world's first law incentivizing drug development for diseases affecting fewer than 200,000 people in the U.S.
- People living with rare diseases were early internet users, forming geographically-dispersed communities that could never have been created offline.
- They pioneered genetic testing and patient-led data registries.

Rare disease families' lives are characterized by extreme stress, often matched by their resourcefulness. Convenience samples and community-based surveys have yielded important insights but no probability-based, nationally representative survey has measured this population. Until now.

A new survey finds that U.S. adults living in rare disease households are more likely than other people to have, in the past year, seen a clinician via telehealth, gone online to find other people who might have health concerns similar to their own, and used an AI tool such as ChatGPT or Gemini to learn about a health condition or treatment options. Once again, people living in rare disease households are at the forefront of the patient-led revolution in medical care.

## Findings

The first probability-based national survey to measure the rare disease population has found that 8% of U.S. adults say they or someone they live with has ever been told by a doctor or other health professional that they have a rare disease (defined as one affecting fewer than 200,000 people in the U.S.).

This finding reinforces a long-held assertion that one in 10 Americans is affected by rare disease.<sup>1</sup> Advocates and experts in the field estimate the number is even higher since more rare diseases are identified and diagnosed each year.<sup>2</sup> Some people may also live with a diagnosis

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<sup>1</sup> Global Genes: Rare Disease Facts: <https://globalgenes.org/rare-disease-facts/>

<sup>2</sup> RARE-X: The Power of Being Counted: <https://rare-x.org/case-studies/the-power-of-being-counted/>

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that they do not know is a “rare disease” because their clinician did not tell them their condition is rare. This survey captures a snapshot of those who identify with the term “rare disease” as a measure of awareness. With this data we can further understand how these households interact with health care resources.

An additional 7% of U.S. adults who do not live in rare disease households say they or someone they live with is currently experiencing an illness that doctors or other health care professionals have not been able to diagnose. Together, they represent 15% of U.S. households, nearly 20 million, affected by rare disease or an undiagnosed illness.

People living in rare disease and undiagnosed illness households are from all walks of life and regions of the country. Our survey found no significant differences among age groups, people with various levels of education or income, or political party identification among respondents who report living in a household dealing with rare disease or undiagnosed illness. Rare disease and undiagnosed illness do not discriminate in the types of households they affect.

The survey found that people affected by rare disease or undiagnosed illness are more likely than others to use telehealth options, AI tools to learn about a health condition or treatment options, and to go online to find other people with similar health concerns.

Looking only at survey respondents who reported living in a rare disease household:

- 63% have, in the past year, seen a doctor or other health care provider, such as a specialist, online or via a telehealth appointment, compared with 45% of those not living in a rare disease household.
- 50% have, in the past year, gone online to find other people who might have health concerns similar to their own, compared with 37% of those not living in a rare disease household.
- 38% have, in the past year, used an AI tool such as ChatGPT or Gemini to learn about a health condition or treatment options, compared with 21% of those not living in a rare disease household.
- 29% have, in the past year, seen a mental health professional or therapist online or via a telehealth appointment, compared with 18% of those not living in a rare disease household.
- 28% have, in the past year, seen a mental health professional or therapist in their office or health care clinic, compared with 19% of those not living in a rare disease household.

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Since it is particularly challenging for rare disease household members to connect with peers in their offline communities, it is heartening to see that they are likely to be successful when they look online. Of those who tried, eight in ten were successful in finding people who share their same health concerns.

When it comes to seeing a doctor or other health care provider, such as a specialist, in their office or health care clinic in the past year, eight in ten respondents said yes, whether they live in a rare disease household or not. The only difference was frequency: 19% of those living in rare disease households reported seeing a clinician in person seven or more times in the past year, compared with 9% of non-rare-disease household members.

Rare diseases test both the affected families as well as the communities and health systems that support them. This study shows that people living in rare disease households continue to lead the way in using technology to pursue connection, information, and well-being.

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## Methodology

This study was conducted by SSRS on its Opinion Panel Omnibus platform for Susannah Fox and ARCHANGELS, with support from the Association on Aging in New York. The SSRS Opinion Panel Omnibus is a national, twice-per-month, probability-based survey. Data collection was conducted on two waves of the omnibus, from January 17 to January 21, 2025 and from February 7 to February 10, 2025, among a total sample of  $N = 2,012$  respondents. The survey was conducted via web ( $n = 1,952$ ) and telephone ( $n = 60$ ) and administered in English ( $n = 2,012$ ). More information about the SSRS Opinion Panel can be found at [www.ssrs.com](http://www.ssrs.com).

All SSRS Opinion Panel Omnibus data are weighted to represent the target population of U.S. adults ages 18 or older. The margin of error for total respondents is +/- 2.6 percentage points at the 95% confidence level.