Exploring physicians' perspectives on the diagnosis journey of rare diseases in the United States (US): findings from a cross-sectional survey

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Background

- Diagnosing rare diseases is a complex and prolonged process, with patients often enduring diagnostic odysseys that can last for decades.[1]
- An analysis by NORD revealed that, in 2019, 36% of patients had been diagnosed with a rare disease within the first year of symptom onset, while 28% reported that it took seven or more years to receive a diagnosis. Additionally, 38% of individuals experienced a misdiagnosis during their diagnostic journey and 3% remained undiagnosed at the time they completed the survey. [1]
- The delay in diagnosing rare diseases leads to worse health outcomes and places a significant financial strain on families, as they incur unnecessary costs for medical appointments, treatments, equipment, and travel. [2]
- Results from a previous survey described the referral behavior of primary care physicians and specialists when they encountered patients with undiagnosed symptoms and the reasons for referral delays.[3]
- The current survey aimed to gather insights from healthcare professionals in the United States (US) regarding the significant challenges they encounter in diagnosing rare diseases, including prolonged diagnostic delays, limited awareness, and the complexity of reaching an accurate diagnosis.

Objective

To understand the perceptions of primary care physicians and specialists regarding the diagnosis pathway for rare diseases in the US.

Methods

- In this cross-sectional study, primary care physicians and specialists from the US were recruited via physician panels to complete an online survey in May 2024.
- Physicians rated their perceptions of the diagnosis pathway for rare diseases using a seven-point Likert scale. The results were categorized as follows : Disagree (answers of 1 - 3); Neutral (4); and Agree (5 - 7).

Results

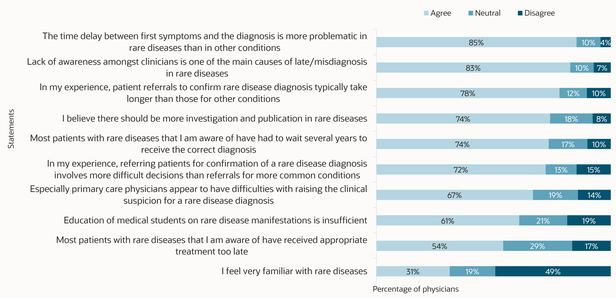
- A total of 479 physicians in the United States (US) participated, including 158 primary care physicians (33%) and 321 specialists (67%) (Table 1).

Table 1 Participating physicians: Numbers and percentages of physicians by medical specialty

Medical focus	Physicians (N=479)	
	n	%
Primary care physicians (FM/GP/IM)	**158**	**33.0**
Specialists (overall)	**321**	**67.0**
Cardiology	33	6.9
Dermatology	32	6.7
Pulmonology	31	6.5
Obstetrics/gynecology	29	6.1
Ophthalmology	28	5.8
Pediatrics	28	5.8
Neurology	23	4.8
Urology	21	4.4
Gastroenterology	20	4.2
Endocrinology/diabetology	19	4.0
Rheumatology	18	3.8
Hematology/oncology	17	3.5
Infectious diseases	12	2.5
Nephrology	10	2.1

- Physicians generally showed a high level of agreement with most statements about the challenges in diagnosing rare diseases. However, an exception was noted regarding familiarity with rare diseases, where only 31% of physicians expressed agreement, while nearly half disagreed (Figure 1).

Figure 1 Physicians' level of agreement with statements related to rare diseases



Overall, the same opinion trends were observed among primary care physicians and specialists (Figure 2). The top 3 affirmations with the highest level of agreement were:

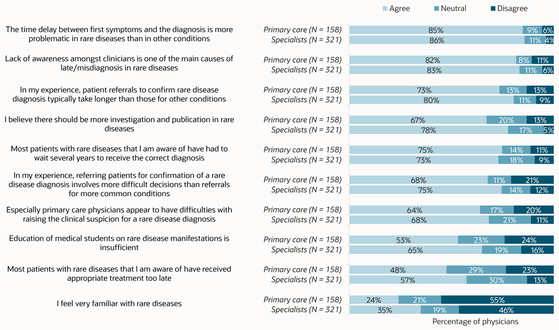
- Both primary care physicians (85%) and specialists (86%) largely agreed that the time delay between first symptoms and diagnosis is more problematic in rare diseases than in other conditions. This reflected a widespread concern about the challenges in timely diagnosis across both groups.
- A majority of both groups supported that a lack of awareness among clinicians is one of the main causes of late or missed diagnoses in rare diseases, emphasizing the need for targeted educational initiatives and improved diagnostic resources.
- A significant proportion of primary care physicians (73%) and specialists (80%) agreed that patient referrals for rare disease diagnoses tend to take longer than those for more common conditions, suggesting potential inefficiencies in the referral process.

Additionally, many physicians acknowledged the importance of further investigation and publication in rare diseases, with 67% of primary care physicians and a higher proportion of specialists (78%) expressing agreement.

While 64% of primary care physicians and 68% of specialists agreed that raising clinical suspicion for a rare disease diagnosis is difficult for primary care physicians, the 20% of primary care physicians who disagreed, compared to only 11% of specialists, suggested a notable variation in perceived challenges.

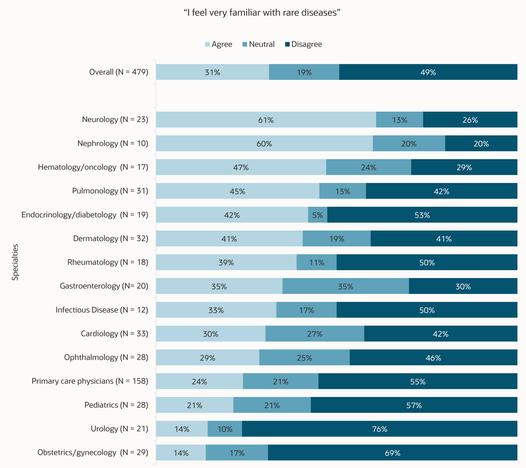
Only a small proportion of primary care physicians (24%) reported feeling very familiar with rare diseases, compared to 35% of specialists. This emphasizes the urgent need for improved training and resources to better equip physicians in recognizing and managing rare diseases.

Figure 2 Physicians' level of agreement with statements related to rare diseases, by specialty



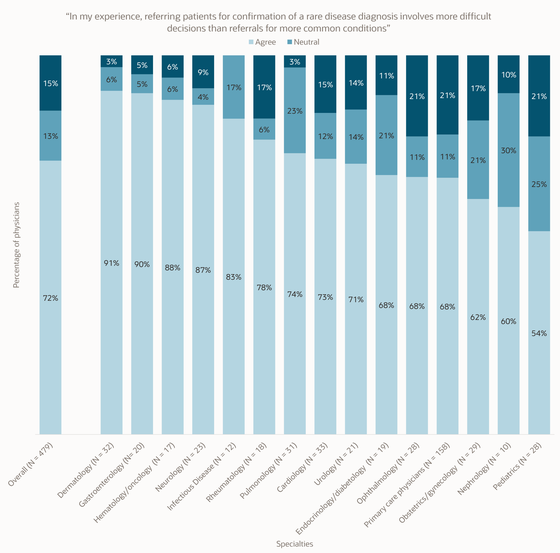
The highest proportion of physicians who agreed with the statement "*I feel very familiar with rare diseases*" was among neurologists, with 61% expressing agreement, followed by nephrologists at 60%. On the other hand, 76% of urologists and 69% of obstetrics/gynecologists, reported disagreeing with the statement. This highlights a large disparity in familiarity with rare diseases across different medical specialties. (Figure 3)

Figure 3 Physicians' level of agreement with the statement related to their familiarity with rare diseases, by medical specialty



Agreement with the statement "In my experience, referring patients for confirmation of a rare disease diagnosis involves more difficult decisions than referrals for more common conditions" was highest among dermatologists (91%), followed by gastroenterologists (90%), hemato-oncologists (88%), and neurologists (87%). In contrast, pediatricians showed the lowest level of agreement at 54%. (Figure 4)

Figure 4 Physicians' level of agreement with the statement related to the difficulty of referral for rare diseases, by medical specialty



Conclusion

Physicians widely acknowledge the challenges in diagnosing rare diseases. However, familiarity with rare diseases remains low, with less than a third expressing agreement and nearly half disagreeing, highlighting a critical gap in knowledge.

The majority agree that limited clinician awareness is a key factor in late or missed diagnosis, emphasizing the need for targeted education and improved diagnostic resources.

Many physicians recognize the importance of further investigation in rare diseases, with primary care physicians and an even higher proportion of specialists supporting more research and publications.

Addressing these issues may better equip healthcare professionals in effectively diagnosing and managing rare diseases.

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Abbreviations

-NORD: National Organization for Rare Disorders

-FM/GP/IM: FM – Family Medicine, GP – General Practitioners, IM – Internal Medicine

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