

Identifying Drivers for the Timely Referral of Patients for a Rare Disease Diagnosis: A Cross-Sectional Survey of Physicians in the United States

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Background

- The diagnostic journey for a patient with a rare disease is often long, with an average of 5 years between the onset of symptoms and a definitive diagnosis.¹
- A diagnostic delay may result in worsening symptoms in the absence of treatment and has detrimental psychological effects on patients and caregivers.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 assess the factors which physicians would find most useful to facilitate the referral of patients with suspected rare diseases.

Objectives

To describe physicians' perspectives on factors that would expedite the referral of suspected cases of rare diseases to rare disease specialists.

Methods

In this cross-sectional study, primary care physicians (PCPs) and specialists were recruited via physician panels in the United States (US). They completed an online questionnaire in May 2024.

Results

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

Medical focus	Physicians (N=479)	
	n	%
Primary care physicians*	158	33%
Specialists (net, overall)	321	67%
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%

*Includes Family Medicine, General Practice, and Internal Medicine

Barely two-thirds of physicians are familiar with the US definition of a rare disease (<200,000 people), and only a minority are familiar with details on worldwide prevalence and genetics in Table 2. **Correct answers are shown in green.**

A total of 479 US physicians participated; 158 PCPs and 321 specialists, as per the specialty designation in Table 1.

Table 2 Proportion of physicians who indicated true/false/don't know for each statement on rare diseases (n=479)

	True	False	Do not know / Not sure
A rare disease is defined as a disease which affects fewer than 200,000 people	61%	12%	27%
The proportion of rare diseases which are exclusively pediatric is 70%.	29%	31%	39%
The worldwide prevalence of rare diseases is estimated to be 4% to 6%.	39%	31%	29%
The worldwide prevalence of rare diseases is estimated to be 1% to 2%.	53%	23%	24%
The proportion of rare diseases which are genetic is approximately 70%.	41%	26%	33%
The proportion of rare diseases which are genetic is approximately 20%.	29%	37%	35%

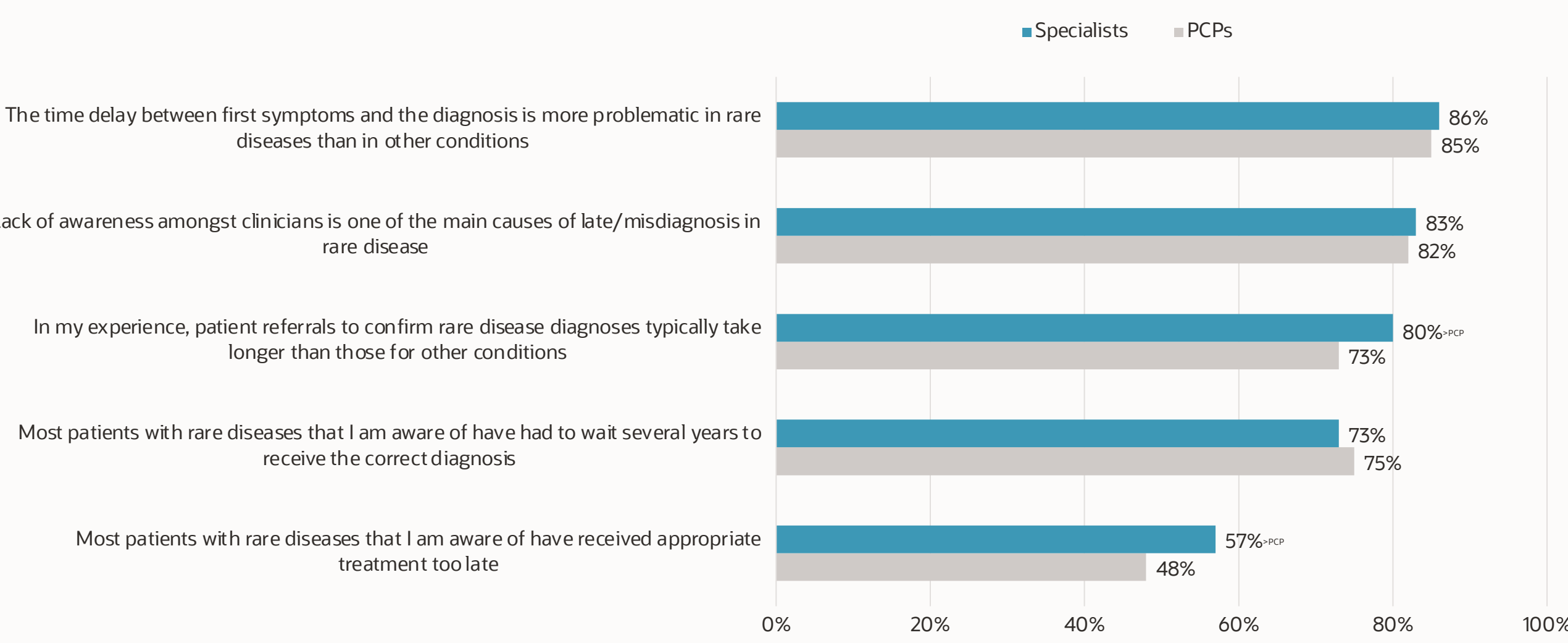
Conclusion

- Significant education on the prevalence and genetics of rare diseases are still necessary for US physicians.
- The majority of physicians recognize the difficulty of diagnosing rare diseases and many agree that patients receive appropriate treatment too late.
- Readily available clinical practice guidelines, better access to diagnostic tests, and better communication between physicians' practices and rare disease centers could help facilitate the timely referral of patients with suspected rare diseases to expedite time to appropriate treatment and ultimately improve patient care.

Physicians largely agreed that the timeliness of rare disease referrals, diagnoses, and treatment take longer than would be optimal (Figure 1).

Physicians ranked readily available clinical practice guidelines, better access to diagnostic tests, and better communication between physicians' practices and rare disease centers as most useful to facilitate the timely referral of patients with suspected rare diseases (Table 3).

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

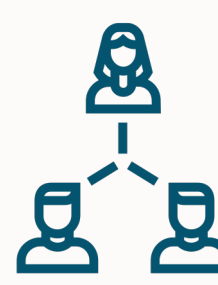


>PCP = significantly higher at the 95% confidence level.

PCPs chose better communication between physicians' practices and rare disease centers (14%), better access to diagnostic tests (10%), readily available clinical practice guidelines (10%).



Training in medical procedures (10%) as the most useful.



Better communication between my center and (for PCPs) hospitals / specialized rare disease centers (for specialists) Rare Disease Centers of Excellence (14%)



Better access to diagnostic tests (10%)



Readily available clinical practice guidelines (10%)

Table 3 Proportion of physicians overall who ranked each factor as most useful

Most useful ranks	N = 479
Readily available clinical practice guidelines	13.6%
Better access to diagnostic tests	12.1%
Better communication between my center and (for PCPs) hospitals / specialized rare disease centers (for specialists) Rare Disease Centers of Excellence	10.0%
More time to investigate clinical aspects	8.4%
Telephone consultation with rare disease specialists	8.1%
Consultation with a trusted colleague	7.3%
Online research database (e.g. MEDLINE) search capabilities	6.9%
Longer patient visit length	6.7%
Training in medical procedures (e.g., screening protocols, routine monitoring tests)	6.1%
Access to an online learning platform about rare diseases	5.6%
Computerized medical expert systems	5.4%
Rare diseases educational campaigns in my center	4.2%
Ability of patients to articulate symptoms clearly	3.8%
Availability of a health educator in my center	1.9%

Specialists chose readily available clinical practice guidelines (16%), better access to diagnostic tests (13%), and more time to investigate clinical aspects (9%) as most useful.



Readily available clinical practice guidelines (16%)



Better access to diagnostic tests (13%)



More time to investigate clinical aspects (9%)

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2. Llubes-Arrià L, Sanromà-Ortíz M, Torné-Ruiz A, Carillo-Álvarez E, García-Expósito J, Roca J. Emotional experience of the diagnostic process of a rare disease and the perception of support systems: A scoping review. J Clin Nurs. 2022 Jan;31(1-2):20-31
3. Omri A, DeCongelio M, Irfan T, Sterzi D, Le Calvé P, Vincent B, Esposito F, Collin-Histed T, Reed S. Towards Diagnosing Rare Diseases: Referral Behavior of Physicians in Europe [abstract]. Value in Health 26(12):S311-S312.