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Assessing gaps in the timely referral of patients for a rare disease diagnosis: **A cross-sectional survey of physicians in Europe**

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Objective

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

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

Methods

In this cross-sectional study, primary care physicians and specialists were recruited via physician panels in France, Germany, Italy, Spain, and the United

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- 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 main factors which physicians would find most useful to facilitate the referral of patients with suspected rare diseases.

Results

A total of 1,082 physicians in five European countries participated, including 320 primary care physicians (29.6%) and 762 specialists (70.4%) (Table 1). Table 1 : Participating physicians: Numbers and percentages of physicians by medical specialty

Medical focus	Physic (N=1,0	Physicians (N=1,082)			
	n	%			
Primary care physicians	320	29.6			
Specialists (overall)	762	70.4			
Cardiology	82	7.6			
Hematology/oncology	63	5.8			
Pediatrics	61	5.6			
Dermatology	61	5.6			
Pulmonology	61	5.6			
Obstetrics/gynecology	60	5.5			
Endocrinology/diabetology	54	5.0			
Neurology	48	4.4			
Gastroenterology	46	4.3			
Rheumatology	46	4.3			
Internal medicine	43	4.0			
Ophthalmology	42	3.9			
Urology	42	3.9			
Nephrology	33	3.0			
Infectious diseases	20	1.8			

Figure 1: Percentages of physicians who agreed that access to readily available practice guidelines would be most useful for facilitating the timely

Kingdom (UK). They completed an online questionnaire in May 2024.

- Overall, physicians ranked readily available clinical practice guidelines, better access to diagnostic tests, and better communication between centers as factors they would find most useful to expedite referrals.
- Differences were observed between types of physicians, with specialists in ophthalmology and endocrinology/diabetology most likely to report the need for clinical practice guidelines (Figure 1). Specialists in nephrology sought better access to diagnostic tests (Figure 2), while pediatricians felt that better communication between physician settings and rare disease centers would be beneficial in facilitating referrals (Figure 3).

Table 2: Factors which physicians would find most useful to facilitate the timely referral of patients suspected of having a rare disease, overall and by type of physician

		Total		Primary care		Specialists	
Factors	(N=1082)		(N=320)		(N=762)		
	Ν	%	Ν	%	N	%	
Readily available clinical practice guidelines	135	12.5	44	13.8	91	11.9	
Better access to diagnostic tests	130	12.0	42	13.1	88	11.5	
Better communication between my center and (for PCPs) hospitals / specialized rare disease centers (for specialists) Rare Disease Centers of Excellence	126	11.6	33	10.3	93	12.2	
Training in medical procedures (e.g., screening protocols, routine monitoring tests)	121	11.2	38	11.9	83	10.9	
Access to a rare disease online learning platform	102	9.4	33	10.3	69	9.1	
Telephone consultations with rare disease specialists	86	7.9	37	11.6	49	6.4	
More time to investigate clinical aspects	75	6.9	18	5.6	57	7.5	
Rare disease educational campaigns in my center	68	6.3	18	5.6	50	6.6	
Longer patient visits	64	5.9	19	5.9	45	5.9	
Computerized medical expert systems	62	5.7	13	4.1	49	6.4	
Online research database (e.g. MEDLINE) search capabilities	45	4.2	11	3.4	34	4.5	
Consultation with a trusted colleague	38	3.5	10	3.1	28	3.7	
Ability of patients to articulate symptoms clearly	30	2.8	10	3.1	20	2.6	
Availability of a health educator in my center	11	1.0	1	0.3	10	1.3	

referral of patients with suspected rare diseases, overall and by physician specialty



Figure 2: Percentages of physicians who agreed that better access to diagnostic tests would be most useful for facilitating the timely referral of patients with suspected rare diseases, overall and by physician specality



Figure 3: Percentages of physicians who agreed that better communication between physician settings and rare disease centers would be most useful for facilitating the timely referral of patients with suspected rare diseases, overall and by physician specality



The most pronounced differences between countries in relation to factors which could facilitate referrals included **training in medical procedures** (range: 5% Germany and the UK, 15% Italy), the availability of clinical practice guidelines (range: 10% Germany, 15% UK), access to computerized medical expert systems (range: 2% UK, 15% in Germany), more time to investigate clinical aspects (range: 3% France, 14% Germany), and longer patient visits (range: 2% France and Italy, 10% Spain) (Figure 4).

Spain

United Kingdom

Figure 4: Country-level differences in the factors which physicians estimated would be most useful to facilitate the timely referral of patients with suspected rare diseases

Ital\

France

Germanv



Better communication between physician settings and rare disease centers

Conclusion

Overall, physicians considered that readily available clinical practice guidelines, better access to diagnostic tests, and improved communication between health care centers would best improve the timely referral of patients with suspected rare diseases.

Substantial differences were observed between medical specialties and countries, which highlights the need for tailored support strategies where gaps have been identified.

Understanding the resource needs of physicians may shorten the diagnostic journey of patients with rare diseases and improve the lives of patients and their caregivers.

References

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