ORACLE Life Sciences

HSD91

Rare disease awareness among physicians in five European countries: Results of a cross-sectional survey

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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]
- An awareness of rare diseases and the breadth of available educational resources could enhance the ability of physicians to diagnose and treat rare diseases.
- The aim of this survey was to assess the rare disease awareness of physicians and to describe their primary sources of rare disease information.

Objective

To assess physicians' knowledge of rare diseases and to describe their primary information resources.

Methods

In this cross-sectional study, primary care physicians and specialists were recruited via physician panels in France, Germany, Italy, Spain, and the United Kingdom (UK). They completed an online questionnaire in May 2024.

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	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

- Overall, physicians reported a mean of 20.3 years of clinical, post-qualifying experience (with a standard deviation [SD] of 9.8). Primary care physicians had a mean of 22.6 years (SD=10.6) compared to specialists with 19.4 years (SD=9.3). Differences were observed between countries: France (22, SD=8), Germany (21, SD=8), Italy (24, SD=11), Spain (18, SD=9) and the UK (16, SD=9).
- Physicians estimated that 4.2% (SD=5.5) of their current patients had a rare disease. Specialists reported a higher percentage of 4.8% (SD=5.9) compared to primary care physicians with 2.9% (SD=4.1).
- Physicians were asked questions concerning rare disease **epidemiology, etiology** and the predominantly affected **patient population**.
- Overall, fewer than half of participating physicians were aware of the worldwide prevalence estimate of rare diseases, the percentage which are genetic in origin, or the proportion which affects the pediatric population exclusively (Table 2).
- In total, 48.9% considered an erroneous definition of rare diseases to be correct (46.6% of primary care physicians and 49.9% of specialists).
- There were no substantial differences in rare disease awareness between primary care physicians and specialists.

Table 2: Numbers and percentages of physicians who provided correct responses to questions about rare diseases, overall and by type of physician

	Total		Primary care		Specialists	
	N=1,082		N=320		N=762	
	n	%	n	%	n	%
Proportion of rare diseases which are genetic is approximately 20% (FALSE)	487	45.0	139	43.4	348	45.7
Proportion of rare diseases which are genetic is approximately 70% (TRUE)	442	40.9	134	41.9	308	40.4
Worldwide prevalence of rare diseases is estimated to be 4% to 6% (TRUE)	418	38.6	130	40.6	288	37.8
Proportion of rare diseases which are exclusively pediatric is 70% (TRUE)	282	26.1	92	28.8	190	24.9
Worldwide prevalence of rare diseases is estimated to be 1% to 2% (FALSE)	269	24.9	82	25.6	187	24.5

- Country-level results are presented in Figure 1. Fewer than half of the physicians in each country correctly identified the worldwide **prevalence** of rare diseases (ranging from 27% of physicians in France to 44% in the UK) or the proportion which are exclusively **pediatric** (ranging from 16% of physicians in Germany to 31% in Italy). Percentages of physicians who were aware of the predominantly **genetic nature** of rare diseases ranged from 28% in Italy to 56% in Spain.

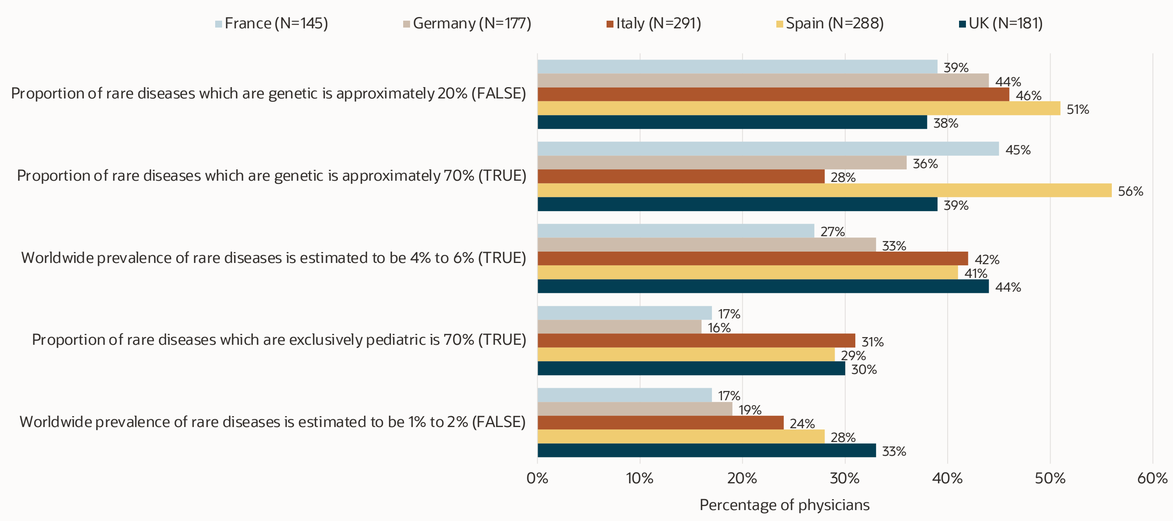

Figure 1 Percentages of physicians within each country who correctly answered questions about rare disease characteristics

- Overall, physicians most often relied upon **scientific literature** (69.7%), the **internet** (56.1%) and **conferences/symposia** (47.0%) to gain knowledge about rare diseases (Table 3).
- Differences were observed between types of physicians, with specialists much more likely than primary care physicians to rely on **scientific literature** (73.0% and 61.9%, respectively), **conferences/symposia** (52.9% and 32.8%, respectively) and **Centers of Expertise/European Reference Networks** (20.6% and 14.7%, respectively). Meanwhile, primary care physicians made greater use of **continuing education courses** for their rare disease knowledge (49.1% versus 41.6%).
- Specialists differed in relation to the resources they consulted: scientific literature (ranging from 57% of urologists to 81% of ophthalmologists), the internet (ranging from 40% of urologists to 72% of pediatricians), and conferences/symposia (ranging from 36% of urologists to 70% of pulmonologists) (Figure 2).

Table 3: Primary sources of information physicians use to learn about rare diseases, overall and by primary care physicians and specialists

	Total		Primary care		Specialists	
	N=1,082		N=320		N=762	
	n	%	n	%	n	%
Scientific literature	754	69.7	198	61.9	556	73.0
Internet (e.g., Orphanet, NORD)	607	56.1	191	59.7	416	54.6
Conferences/symposia	508	47.0	105	32.8	403	52.9
Continuing education courses	474	43.8	157	49.1	317	41.6
Colleagues/peers	339	31.3	106	33.1	233	30.6
Webinars	305	28.2	86	26.9	219	28.7
In-house hospital/practice training	240	22.2	66	20.6	174	22.8
Centers of Expertise/European Reference Networks	204	18.9	47	14.7	157	20.6
Patient organization training programs	117	10.8	38	11.9	79	10.4

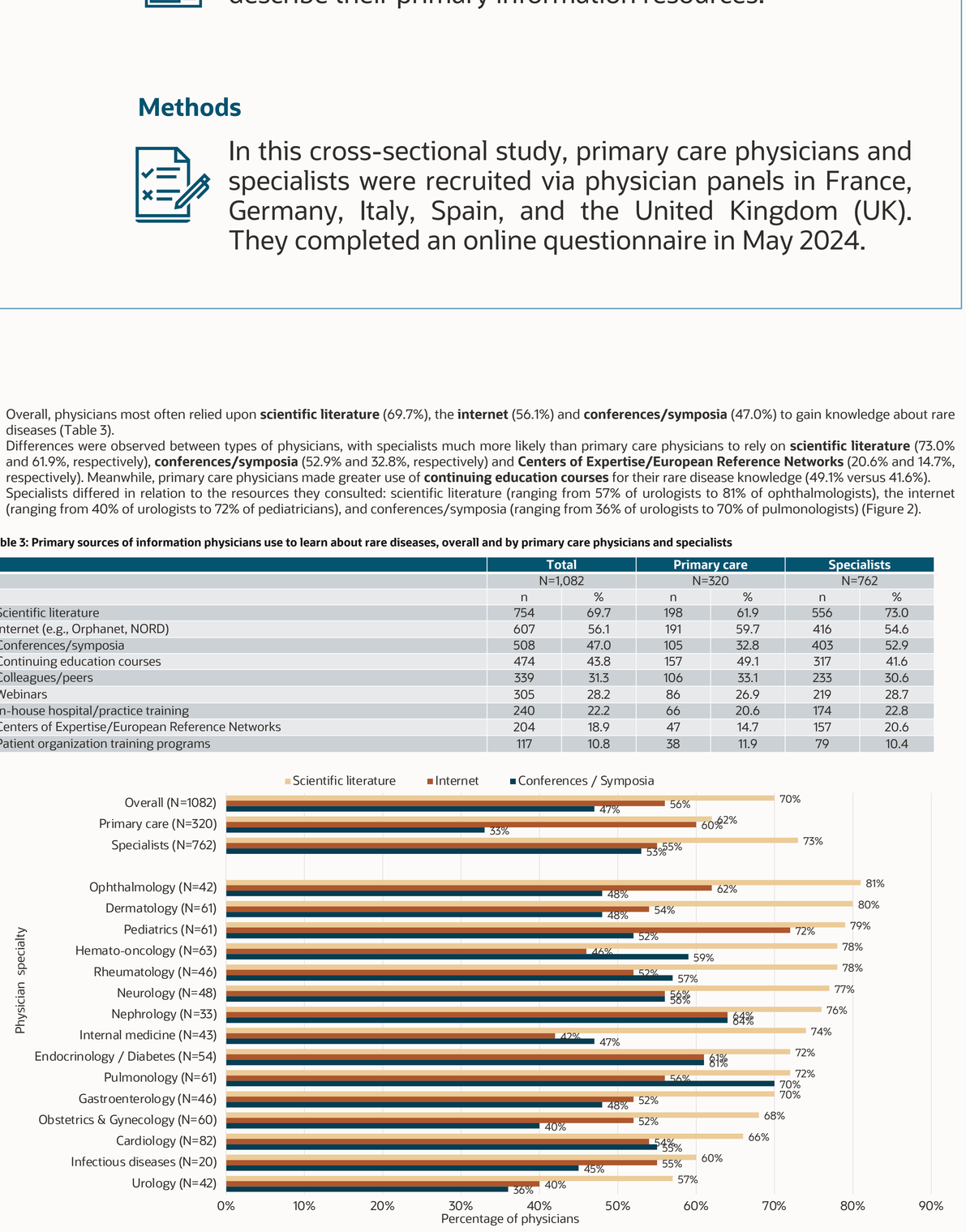

Figure 2: Percentages of physicians who reported scientific literature, the internet or conferences/symposia as their primary rare disease resources

- Use of information resources varied somewhat between countries (Figure 3).
- Physicians in France and Italy were substantially less likely to use **continuing education courses** as a resource (31% and 36%, respectively, compared to ~50% in other countries), while physicians in France and the UK relied heavily on their **colleagues/peers** (40% and 43%, respectively, compared to 23% to 32% in other countries). **Patient organizations** were reported as a resource by a low percentage of physicians (ranging from 4% in Germany to 15% in Spain).

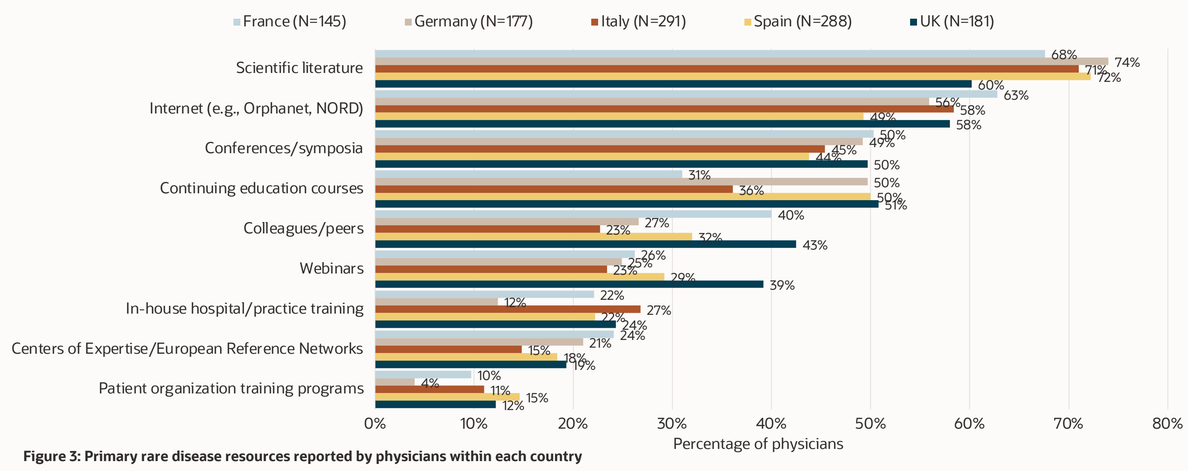

Figure 3: Primary rare disease resources reported by physicians within each country

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Conclusion

- The majority of physicians were unaware of the prevalence of rare diseases, their predominantly genetic origin, and the extent to which the pediatric population is exclusively affected.
- Whilst scientific literature, the internet and conferences/symposia were primary sources of rare disease knowledge for physicians, very few relied upon the expertise of patient organizations, Centers of Expertise or in-house training programs.
- Identifying rare disease knowledge gaps may inform future training of physicians. Improved awareness and increased use of available educational resources could reduce the challenges of diagnosing and treating rare diseases and lead to improved patient outcomes.

References

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