

Emotions of Physicians Elicited by their Patients' Rare Disease Diagnoses: A Cross-sectional Study in Europe and the United States

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Background

- Approximately 300 million people worldwide are living with rare diseases¹, which require the involvement of multiple medical specialties.
- This presents a unique challenge for physicians, especially regarding their need to gain knowledge and experience in caring for such patients.² This challenge may include an emotional aspect.
- Understanding the emotional impact of rare diseases on physicians could inform how best to provide them with support, potentially improve their relationships with patients and, consequently, clinical outcomes.

Objective

To investigate the emotional impact on primary care physicians (PCPs) and specialists when a patient is newly diagnosed with a rare disease.

Methods



Physicians (including PCPs and specialists) were recruited via physician panels to participate in a web-based cross-sectional survey.



The survey was conducted in January 2023 across France, Germany, Italy, Spain, the United Kingdom (UK) and the United States (U.S.).

Results

In total, 2,292 physicians in Europe (1,610) and the U.S. (682) participated. Table 1 displays the numbers and percentages of physicians by region.

Overall, physicians estimated 6.6% of their current patients to have a rare disease (Table 2). Proportions were similar between Europe (6.6%) and the U.S. (6.8%). Estimates were highest among neurologists (11.6%) and hematologists/oncologists (11.5%) and lowest among obstetricians/gynecologists (3.6%).

Table 1. Participating physicians: Counts and proportions overall and by region

Medical focus	Overall		Europe		U.S.	
	(N=2,292)		(N=1,610)		(N=682)	
	n	%	n	%	n	%
Primary Care	687	30.0	478	29.7	209	30.6
Specialties						
Hematology/Oncology/HemOnc	204	8.9	176	10.9	28	4.1
Cardiology	164	7.2	106	6.6	58	8.5
Pediatrics	154	6.7	88	5.5	66	9.7
Neurology	138	6.0	99	6.2	39	5.7
Eye care – Ophthalmology	126	5.5	77	4.8	49	7.2
Dermatology	125	5.5	82	5.1	43	6.3
Internal medicine*	112	4.9	112	7.0	0	0.0
Obstetrics & Gynecology	110	4.8	63	3.9	47	7.0
Endocrinology & Diabetology	104	4.5	77	4.8	27	4.0
Gastroenterology/Hepatology	102	4.5	67	4.2	35	5.1
Rheumatology	101	4.4	71	4.4	30	4.4
Urology	72	3.1	53	3.3	19	2.8
Nephrology	56	2.4	41	2.6	15	2.2
Infectious diseases (including HIV)	37	1.6	20	1.2	17	2.5

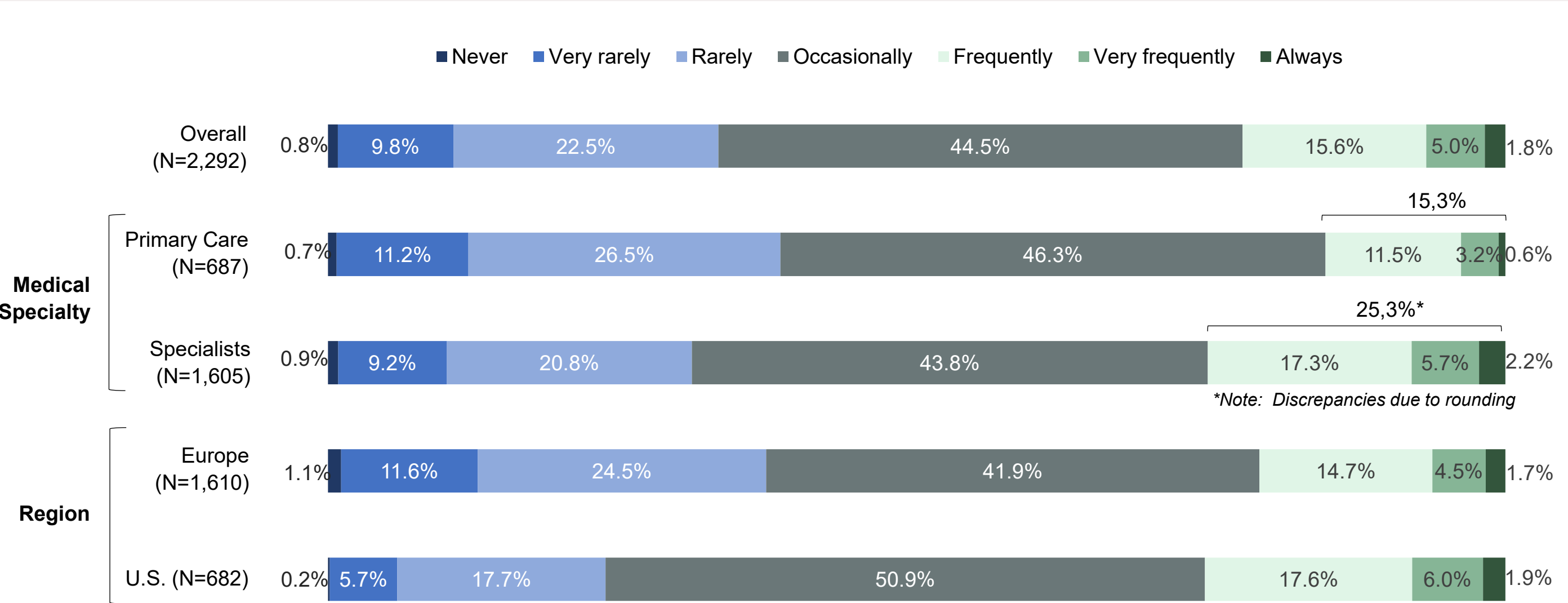
Table 2. Mean proportion of current patients that physicians estimated having a rare disease: Mean percentages (and standard deviations) overall, by region, and by specialty.

	Mean percentage of patients % (SD)
Overall (N=2,292)	6.6 (10.6)
Region	
Europe (N=1,610)	6.6 (10.4)
U.S. (N=682)	6.8 (10.8)
Medical focus	
Primary care (N=687)	4.8 (8.1)
Specialties	
Neurology (N=138)	11.6 (14.5)
Hematology/Oncology/HemOnc (N=204)	11.5 (15.2)
Rheumatology (N=101)	8.9 (9.1)
Endocrinology & Diabetology (N=104)	8.0 (12.7)
Internal medicine* (N=112)	7.8 (11.0)
Gastroenterology/Hepatology (N=102)	7.6 (11.9)
Nephrology (N=56)	7.4 (9.0)
Infectious diseases (including HIV) (N=37)	7.0 (12.9)
Pediatrics (N=154)	6.2 (13.0)
Dermatology (N=125)	5.7 (7.1)
Cardiology (N=164)	5.4 (7.5)
Eye care – Ophthalmology (N=126)	5.2 (9.4)
Urology (N=72)	4.4 (6.4)
Obstetrics & Gynecology (N=110)	3.6 (5.9)

*In the U.S., specialists in internal medicine were categorized as primary care physicians.

The frequency with which physicians suspect a rare disease when they were unable to diagnose symptoms is displayed in Figure 1. Specialists more frequently (frequently/very frequently/always) suspected a rare disease when they were unable to diagnose symptoms (25.3% vs. 15.3% PCPs).

Figure 1. Frequency with which physicians suspected a rare disease when they were unable to diagnose symptoms: Percentage of physicians by medical specialty and by region.



A list of emotions was developed and displayed to physicians. Different emotions can be experienced by physicians when they first learn about their patients' rare disease diagnosis (Table 3):

- Overall, more than half of the physicians (50.3%) felt challenged when patients were diagnosed with a rare disease, and almost a third felt excited (28.9%).
- Specialists were significantly more likely to feel excited (31.2%) than PCPs (23.7%) ($p<0.001$) and less likely to feel helpless (7.8% vs 10.6%) ($p=0.03$).
 - Physicians in the U.S. were more likely than their European counterparts to feel challenged (71.0% vs 41.6%, $p<0.001$), confident (16.3% vs 12.2%, $p=0.008$), nervous (13.5% vs 7.0%, $p<0.001$), sad (10.9% vs 7.0%) and intimidated (10.7% vs 4.5%, $p<0.001$).
 - Physicians in Europe more often felt helpless (10.6% vs 4.0%, $p<0.001$), indifferent (5.1% vs 2.5%), and bitter (2.9% vs 0.2%).

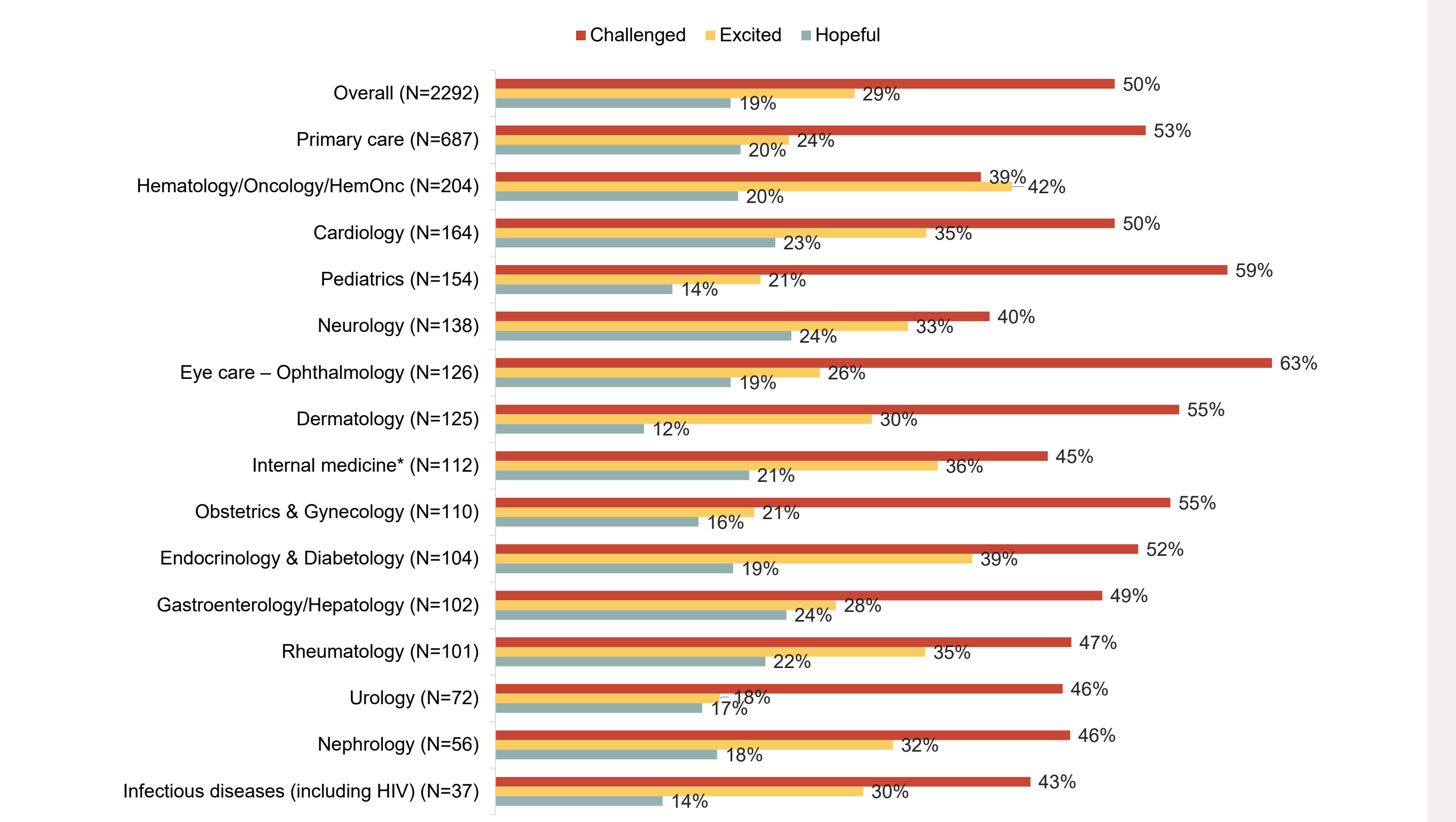
Table 3. Emotions experienced by physicians when they first learned about their patients' rare disease diagnosis: Counts and proportions of physicians and statistical significance, overall and by medical specialty.

Emotion	Overall		Primary care		Specialists		p-value	Europe		U.S.		p-value
	(N=2,292)		(N= 687)		(N=1605)			(N=1,610)		(N=682)		
	n	%	n	%	n	%		n	%	n	%	
Challenged	1153	50.3	361	52.5	792	49.3	0.2	669	41.6	484	71.0	<0.001
Excited	663	28.9	163	23.7	500	31.2	<0.001	477	29.6	186	27.3	0.3
Hopeful	441	19.2	136	19.8	305	19.0	0.7	302	18.8	139	20.4	0.4
Confident	307	13.4	78	11.4	229	14.3	0.061	196	12.2	111	16.3	0.008
Nervous	205	8.9	65	9.5	140	8.7	0.6	113	7.0	92	13.5	<0.001
Helpless	198	8.6	73	10.6	125	7.8	0.027	171	10.6	27	4.0	<0.001
Sad	187	8.2	67	9.8	120	7.5	0.068	113	7.0	74	10.9	0.002
Intimidated	146	6.4	49	7.1	97	6.0	0.3	73	4.5	73	10.7	<0.001
Frustrated	145	6.3	50	7.3	95	5.9	0.2	110	6.8	35	5.1	0.13
Overwhelmed	143	6.2	44	6.4	99	6.2	0.8	99	6.2	44	6.5	0.8
Distressed	126	5.5	42	6.1	84	5.2	0.4	90	5.6	36	5.3	0.8
Indifferent	99	4.3	36	5.2	63	3.9	0.2	82	5.1	17	2.5	0.005
Frightened	70	3.1	20	2.9	50	3.1	0.8	56	3.5	14	2.1	0.07
Heartbroken	59	2.6	19	2.8	40	2.5	0.7	30	1.9	29	4.3	<0.001
Hopeless	52	2.3	15	2.2	37	2.3	0.9	42	2.6	10	1.5	0.093
Bitter	47	2.1	14	2.0	33	2.1	>0.9	46	2.9	1	0.2	<0.001
Appalled	35	1.5	7	1.0	28	1.7	0.2	30	1.9	5	0.7	0.044
Repelled	14	0.6	5	0.7	9	0.6	0.8	10	0.6	4	0.6	>0.9
Furious	8	0.4	4	0.6	4	0.3	0.3	6	0.4	2	0.3	>0.9

Feeling challenged was reported by 62.7% of ophthalmologists and 59.1% of pediatricians. Feeling excited was common in hematology/oncology (41.7%) and endocrinology/diabetology (38.5%). Feeling hopeful was reported by neurologists (23.9%), gastroenterologists/hepatologists (23.5%), and cardiologists (22.6%).

The top three most common emotions physicians experience when learning about their patients' rare disease diagnosis are displayed by medical specialty in Figure 2.

Figure 2. Top three most common emotions experienced by physicians upon learning about their patients' rare disease diagnosis, by specialty.



*In the U.S., specialists in internal medicine were categorized as primary care physicians.

Region-specific differences were noted among some specialties when learning of their patients' rare disease diagnosis

Figure 3. Percentages of pediatricians who felt challenged or nervous when learning of their patients' rare disease diagnosis, by region.

Pediatricians in the U.S. were more likely to feel challenged (U.S.: 80%, Europe: 43%) ($p<0.00001$), nervous (U.S.: 21%, Europe: 9%) ($p<0.05$).

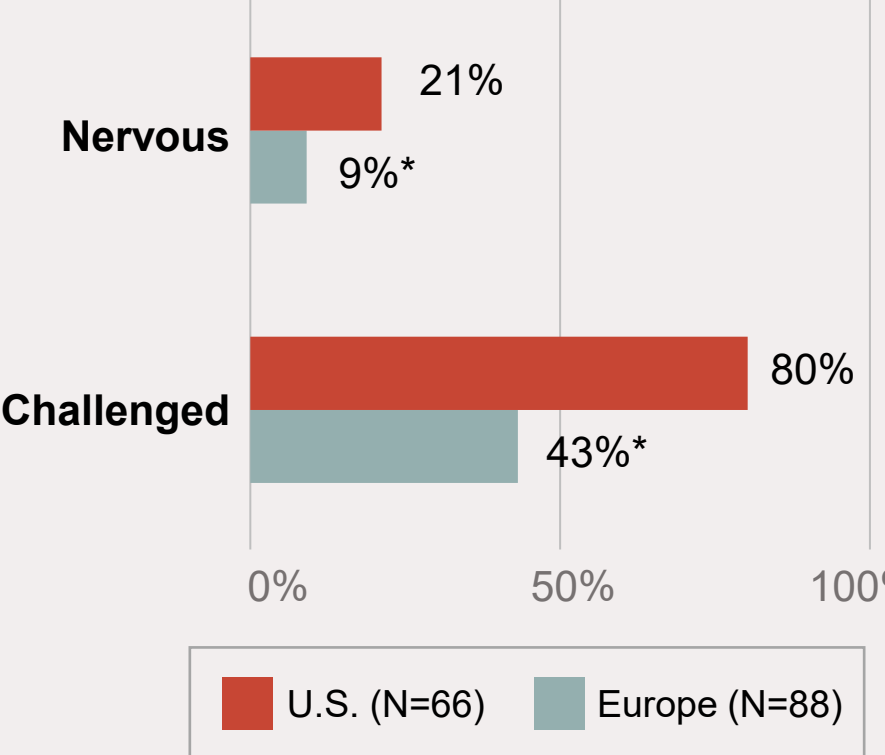


Figure 4. Percentages of obstetricians/gynecologists who felt intimidated when learning of their patients' rare disease diagnosis, by region.

Obstetricians/gynecologists in the U.S. were more likely to feel intimidated than their counterparts in Europe (19% and 6%, respectively) ($p<0.05$).

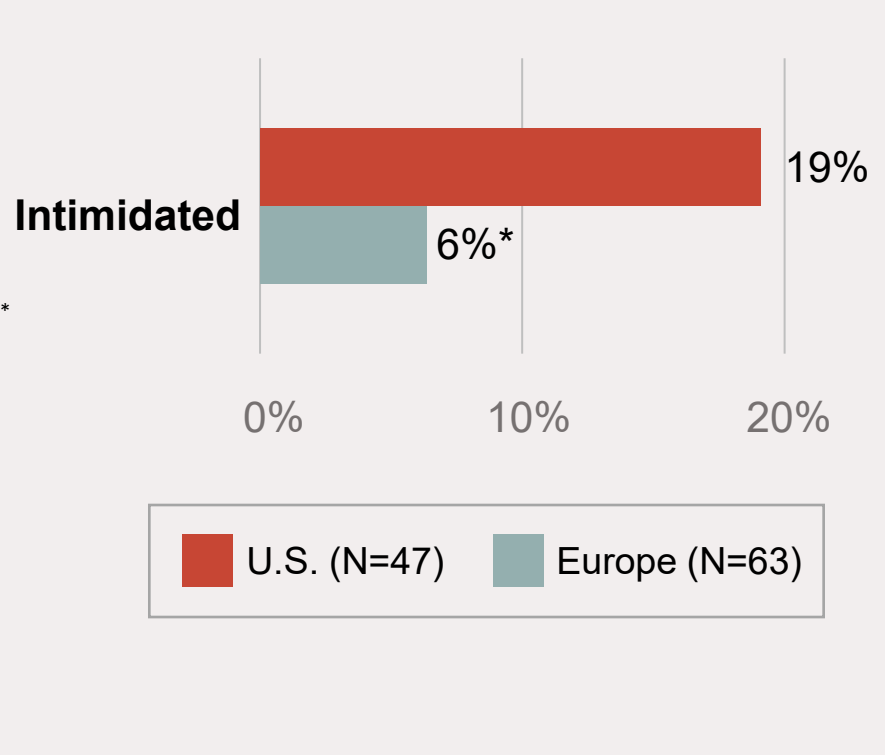
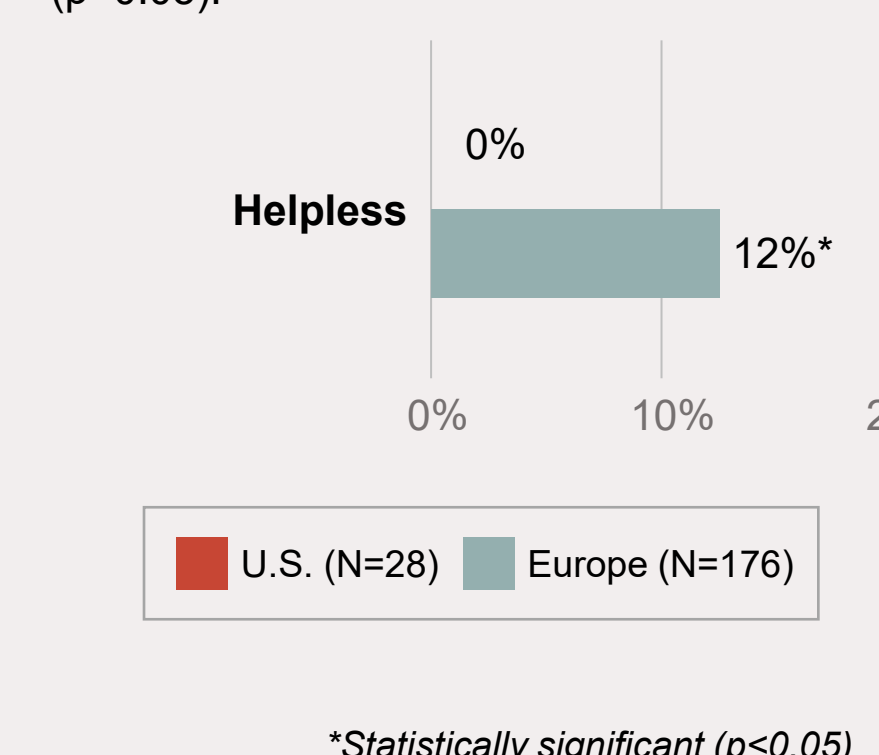


Figure 5. Percentages of hematologists/oncologists who felt helpless when learning of their patients' rare disease diagnosis, by region.

Hematologists/oncologists in the U.S. were significantly less likely to feel helpless toward their patients with a rare disease diagnosis (0.0% and 12%, respectively) ($p<0.05$).



*Statistically significant ($p<0.05$)

Conclusions

- When patients are diagnosed with a rare disease, physicians feel a range of emotions. Feeling challenged is the most common across regions and specialties.
- The emotions experienced vary by region and medical specialty.
- These findings highlight the importance of developing strategies that can help support physicians when faced with a diagnosis of a rare disease in their patients as well as supporting their patient-physician relationships.

References

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Suzanne Reed, Amina Omri, Perrine Le Calvé, and Bastien Vincent, are employees of Oracle. Marc DeCongelio, Tim Irfan, Daniel Sterzi, are employees of Cerner Enviza. Franco Esposito is an employee of All Global. Joana E. Matos is an employee of Rocket Pharmaceuticals. This research was not funded by Rocket Pharmaceuticals.

Abbreviations

PCP = Primary care physician
SD = standard deviation
U.S. = United States