

BACKGROUND



- No individual clinician can be familiar with the > 7000 known rare diseases; however, recognizing when a patient may have a rare disease and appropriate referral is critical in shortening the pathway to diagnosis
- Unfortunately, there continues to be a significant delay in diagnosis across rare diseases, which may lead to inappropriate or harmful testing or medication, and disease progression

METHODS

N = 927
physicians
specializing
in 16 different
therapeutic
areas

- A 14-question online survey to assess clinician knowledge, experience, and educational needs in rare disease was emailed to Medscape member physicians.
- Questions related to confidence and experience were rated on a Likert-type scale.
- The survey launched on March 31, 2021 and closed on August 2, 2021.

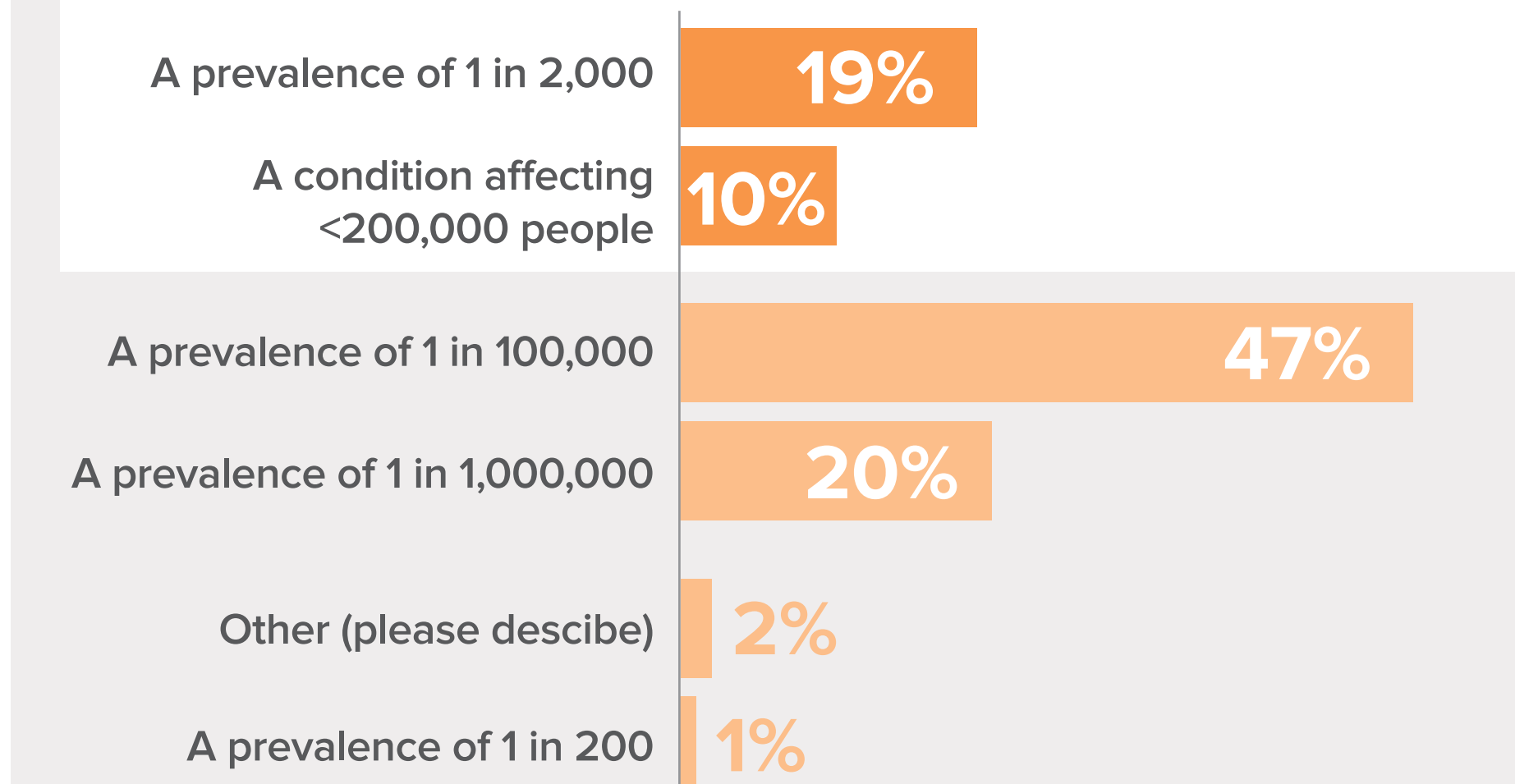
RESULTS

PREVALENCE OF RARE DISEASE

Approximately 71% of physicians considered rare diseases on the order of 10- to 100-fold rarer compared with standard definitions in the EU (1 in 2,000) or US (< 200,000).

EU5 + US Physicians (n = 927)

Question: How do you define rare disease?

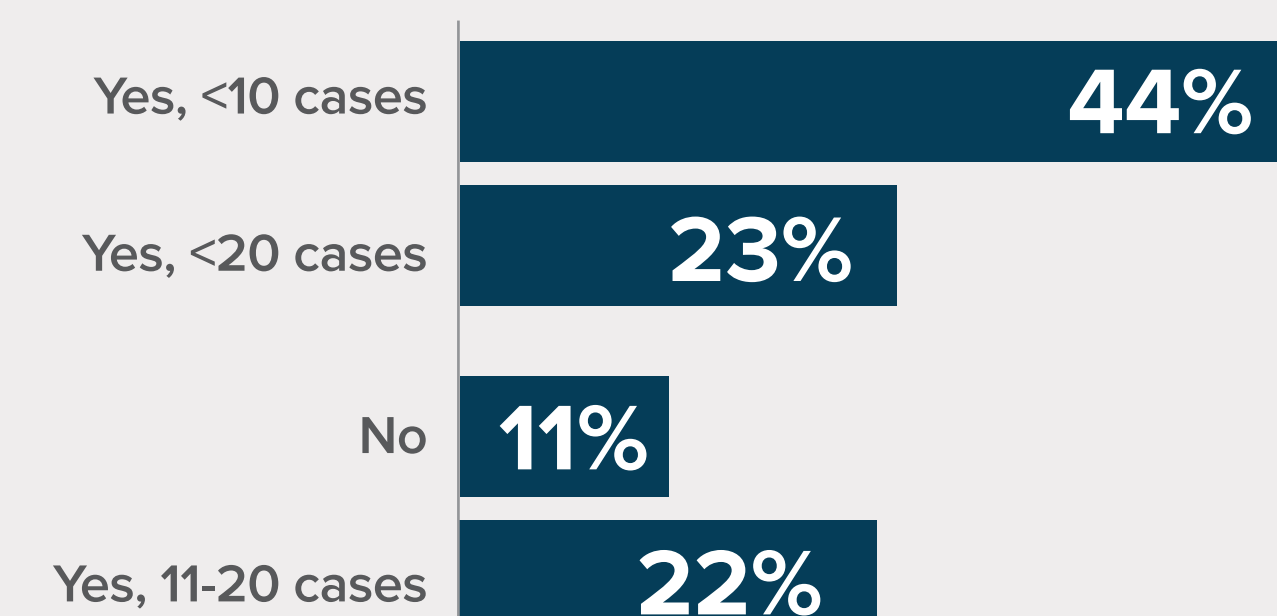


RARE DISEASE DIAGNOSIS

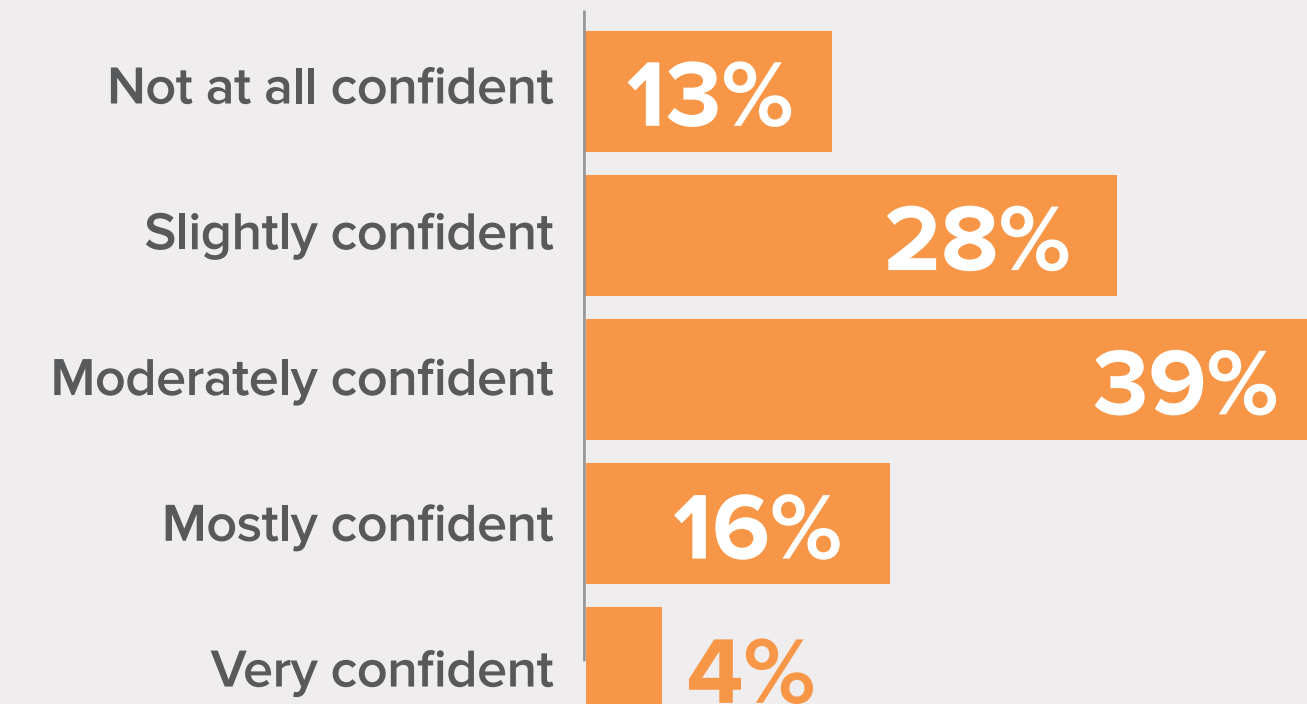
Although 89% have been involved in a rare disease diagnosis, only 20% were mostly or very confident in making such a diagnosis.

EU5 + US Physicians (n = 927)

Question: Have you been involved in the diagnosis of rare diseases?



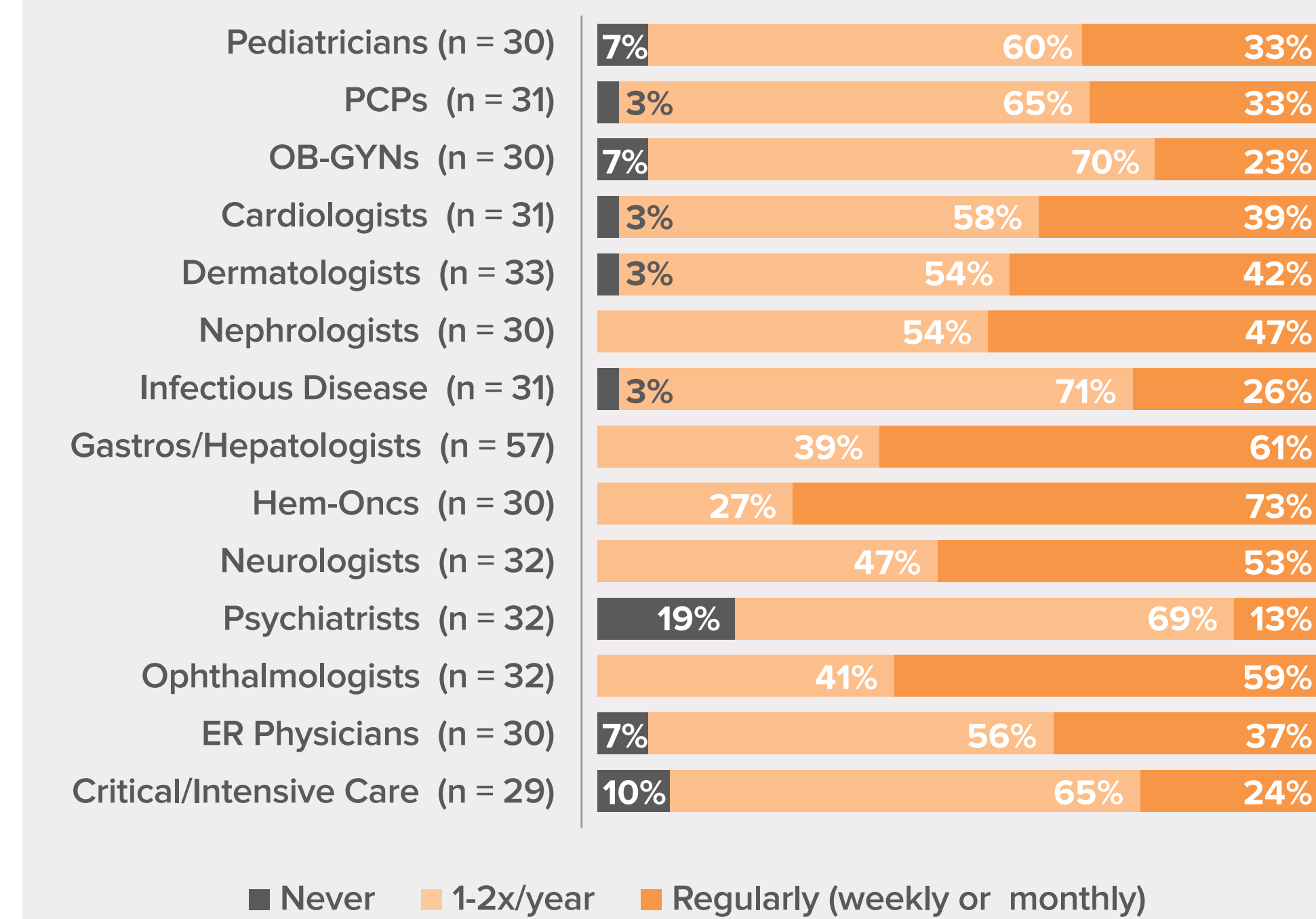
Question: How confident are you right now in your ability to make a rare disease diagnosis?



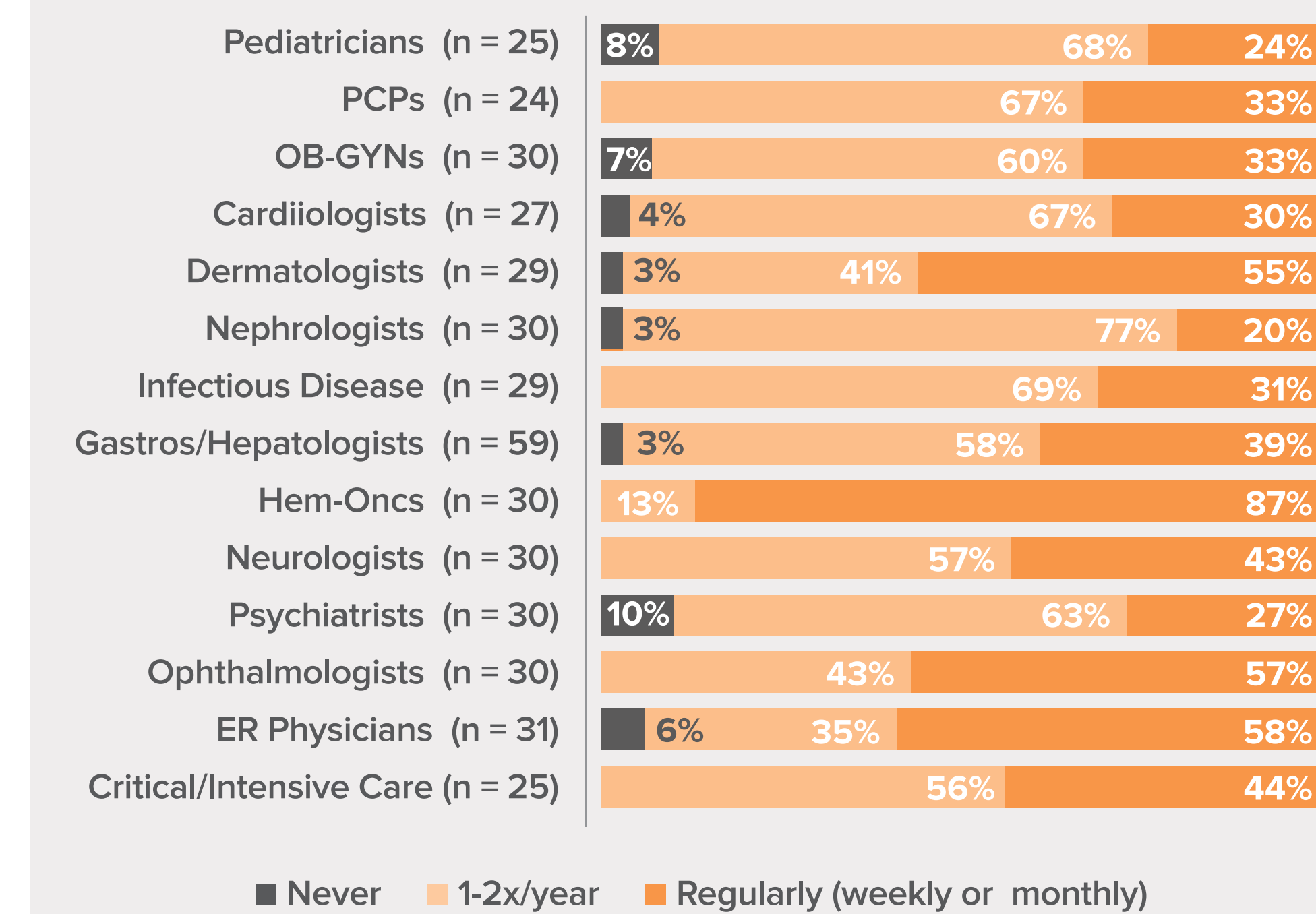
EXPERIENCE IN RARE DISEASE

Overall, 57% said they never or rarely (1-2x/year) see rare disease patients. Despite most rare diseases originating childhood, as many as 70% of pediatricians across regions (n = 55) said they never or rarely see rare disease patients.

EU5: How often are physicians seeing patients with rare diseases?



US: How often are physicians seeing patients with rare diseases?

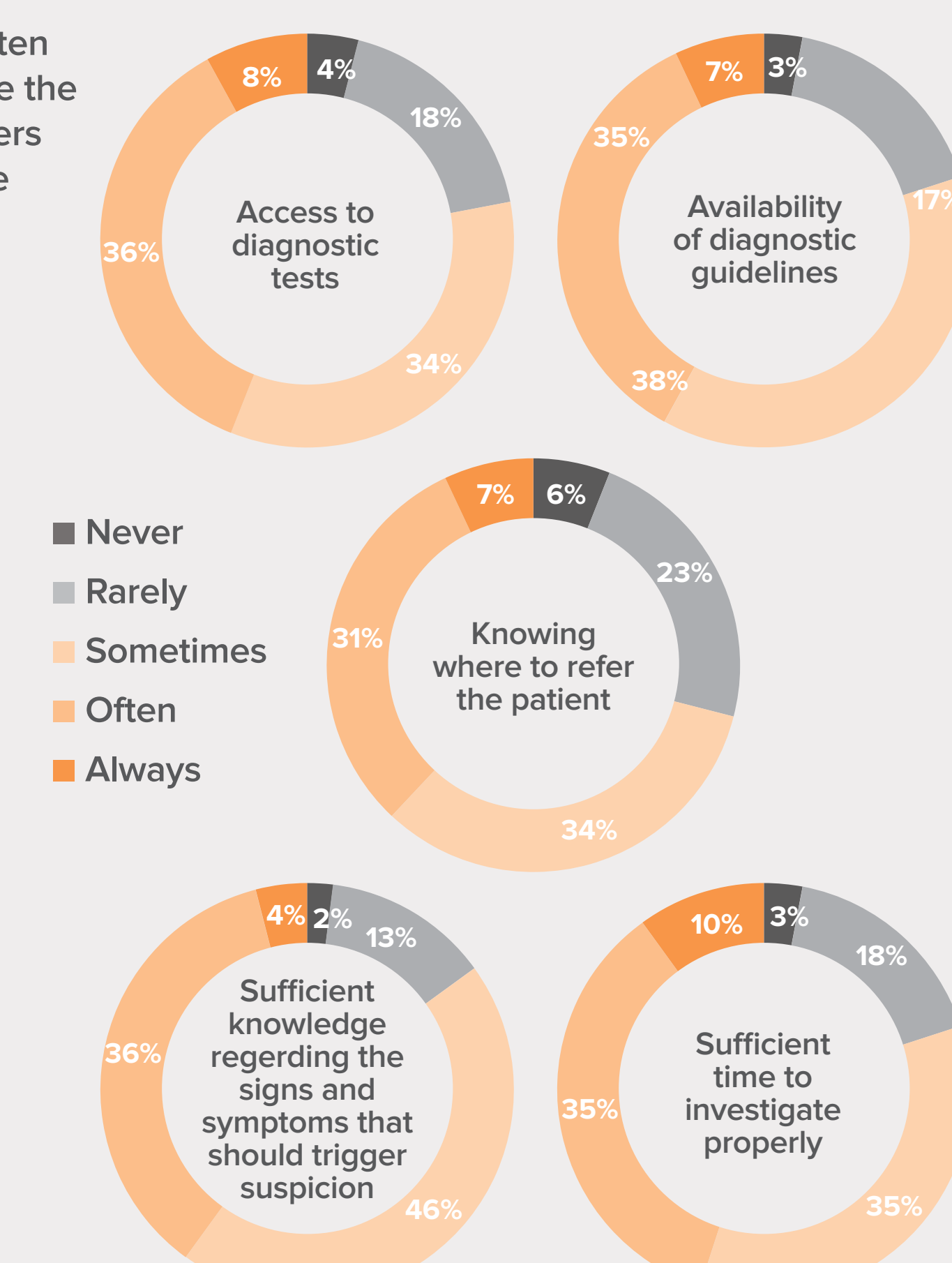


BARRIERS IN RARE DISEASE

Barriers between US and EU physicians were similar, with 35% to 45% indicating sufficient knowledge regarding signs and symptoms that should trigger suspicion, sufficient time to investigate properly, availability of diagnostic guidelines, access to diagnostic tests, and knowing where to refer the patient were all barriers in their practice.

EU5 + US Physicians (n = 927)

Question: How often do you experience the following as barriers to diagnosing rare diseases in your practice?

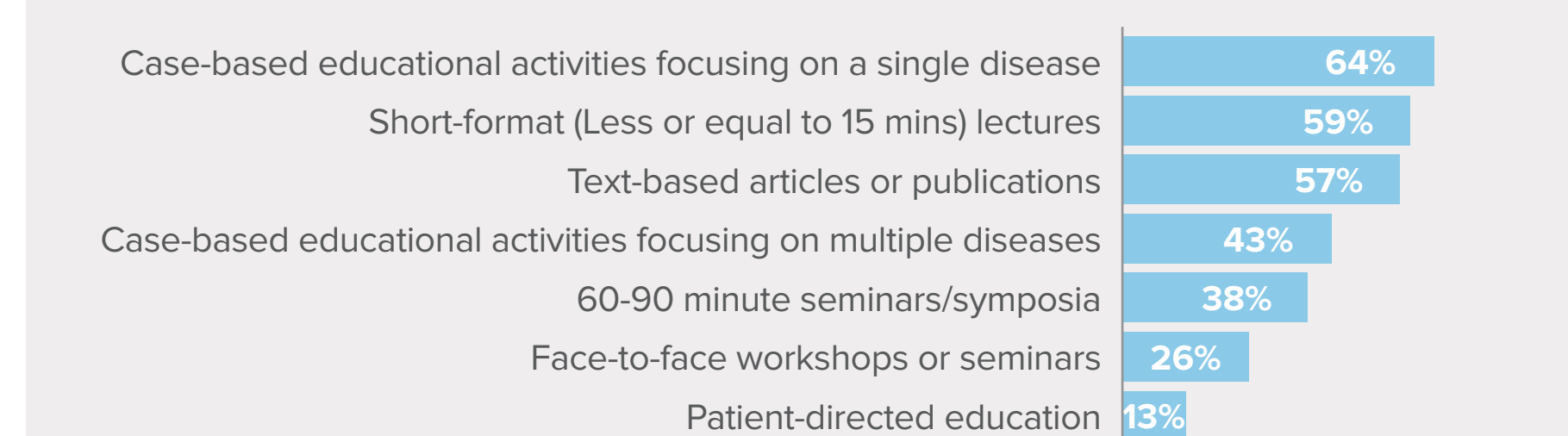


WHAT KIND OF RARE DISEASE EDUCATION PHYSICIANS WANT

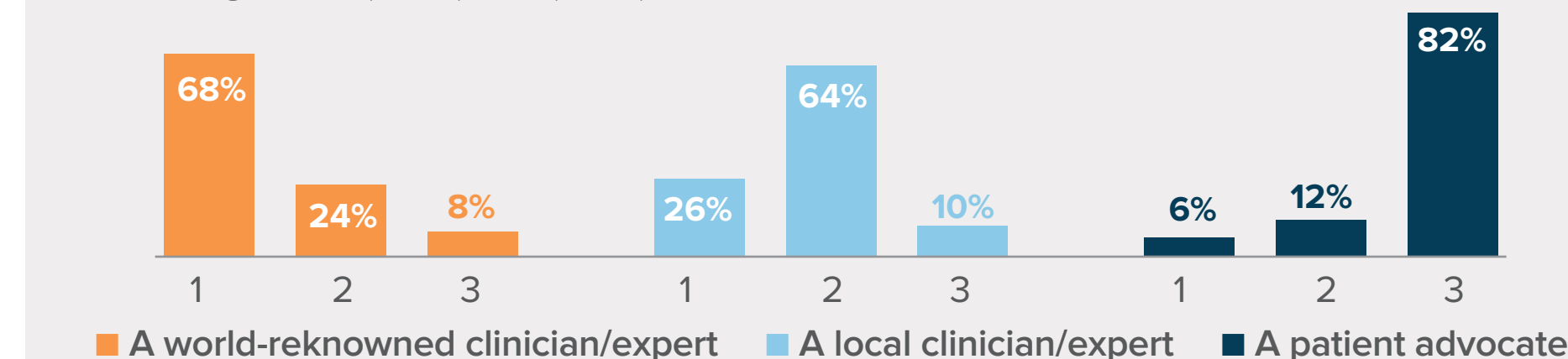
When asked about what they consider most effective in rare disease education, case-based, text-based, and short-format education, given by world renowned clinicians were most popular. And a comprehensive online learning platform with current, organized education and resources was considered most valuable.

EU5 + US Physicians (n = 927)

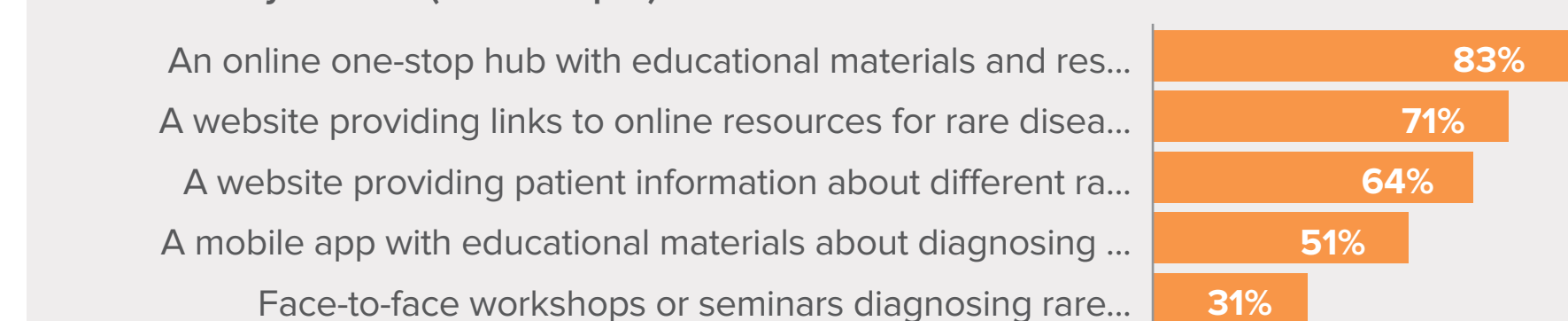
Question: What type of education do you find most effective (in terms of time required and gaining knowledge) to inform yourself about a rare disease? (Select top 3)



Question: From whom would you like to learn about rare diseases? Please rank the following from 1 (most) to 3 (least)



Question: Which of the following resources for rare disease diagnosis would you be most likely to use? (Select top 3)



CONCLUSIONS

This educational research study identifies gaps in diagnosing rare diseases and supports the need to develop education for physicians on recognizing rare diseases earlier, shortening the diagnostic odyssey and enabling earlier referral and treatment.

ACKNOWLEDGEMENTS

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For more information, contact
S. Christy Rohani-Montez, PhD
Clinical Strategist, Medscape Global
srohani@medscape.net

REFERENCE

A Medscape Membership Survey: How Do You Tackle Rare Diseases? Launched on March 31, 2021 and closed on August 2, 2021. Medscape Internal Data.



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