

Education Needs in Diagnosing Rare Diseases in Psychiatry: a Clinician Survey

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BACKGROUND

Neuropsychiatric symptoms occur commonly in rare diseases. As many as 74% of rare genetic disorders are associated with central nervous system (CNS) symptoms.¹ Patients presenting with such symptoms to psychiatrists may be misdiagnosed with primary psychiatric disorders, considered to have conversion disorder, or a reaction to having an unexplained condition.² Psychiatrists may play a key role in identifying these patients early; however, there continues to be a significant delay in diagnosis across rare diseases.



METHODS

n = 927
physicians
specializing
in 16
different
therapeutic
areas

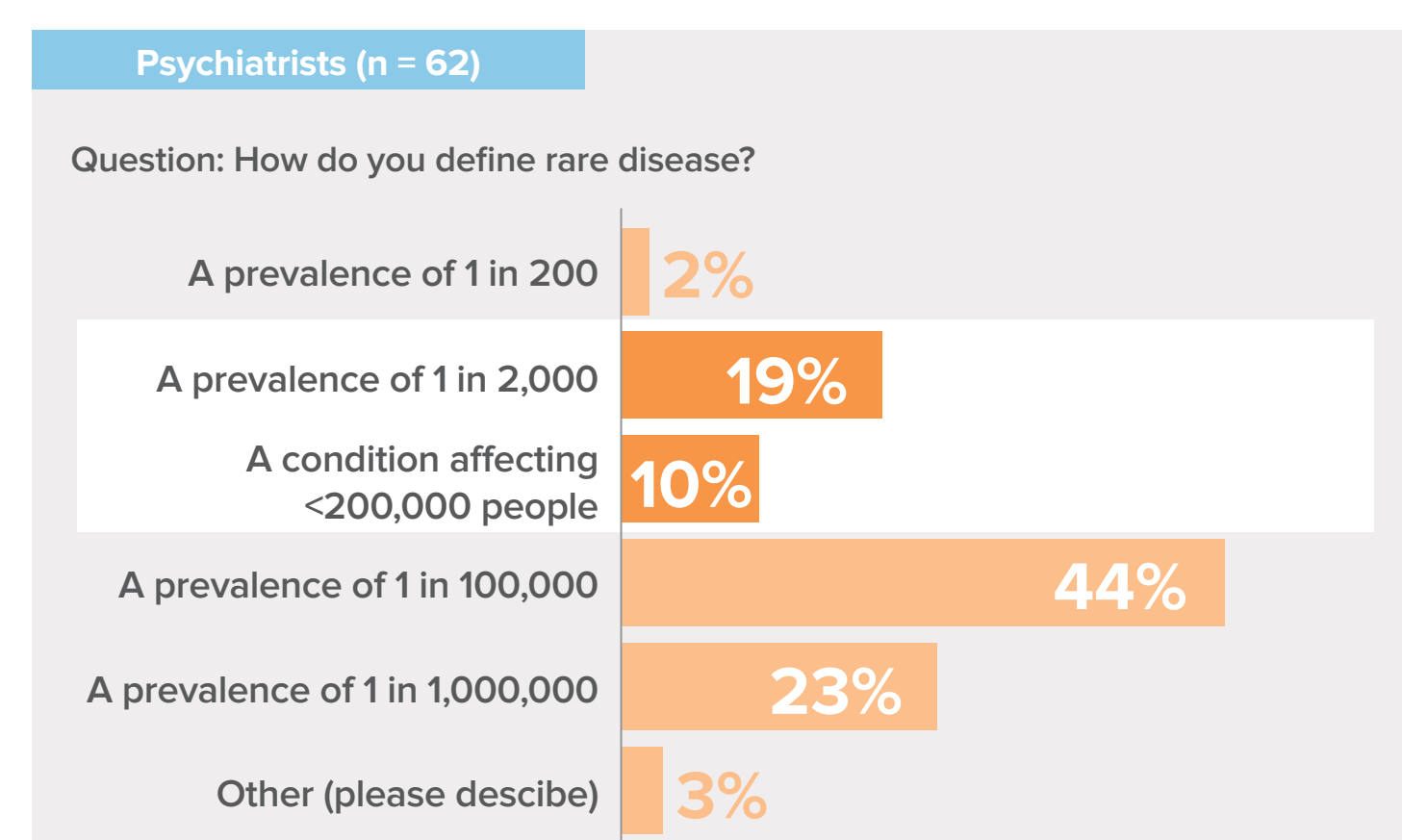
A 14-question online survey to assess rare disease knowledge, experience, and educational needs was emailed to EU5 and US Medscape member physicians. It launched on March 31, 2021 and closed on August 2, 2021. Respondent confidentiality was maintained, and responses were de-identified and aggregated prior to analyses.

62 were psychiatrists from the EU or US

RESULTS

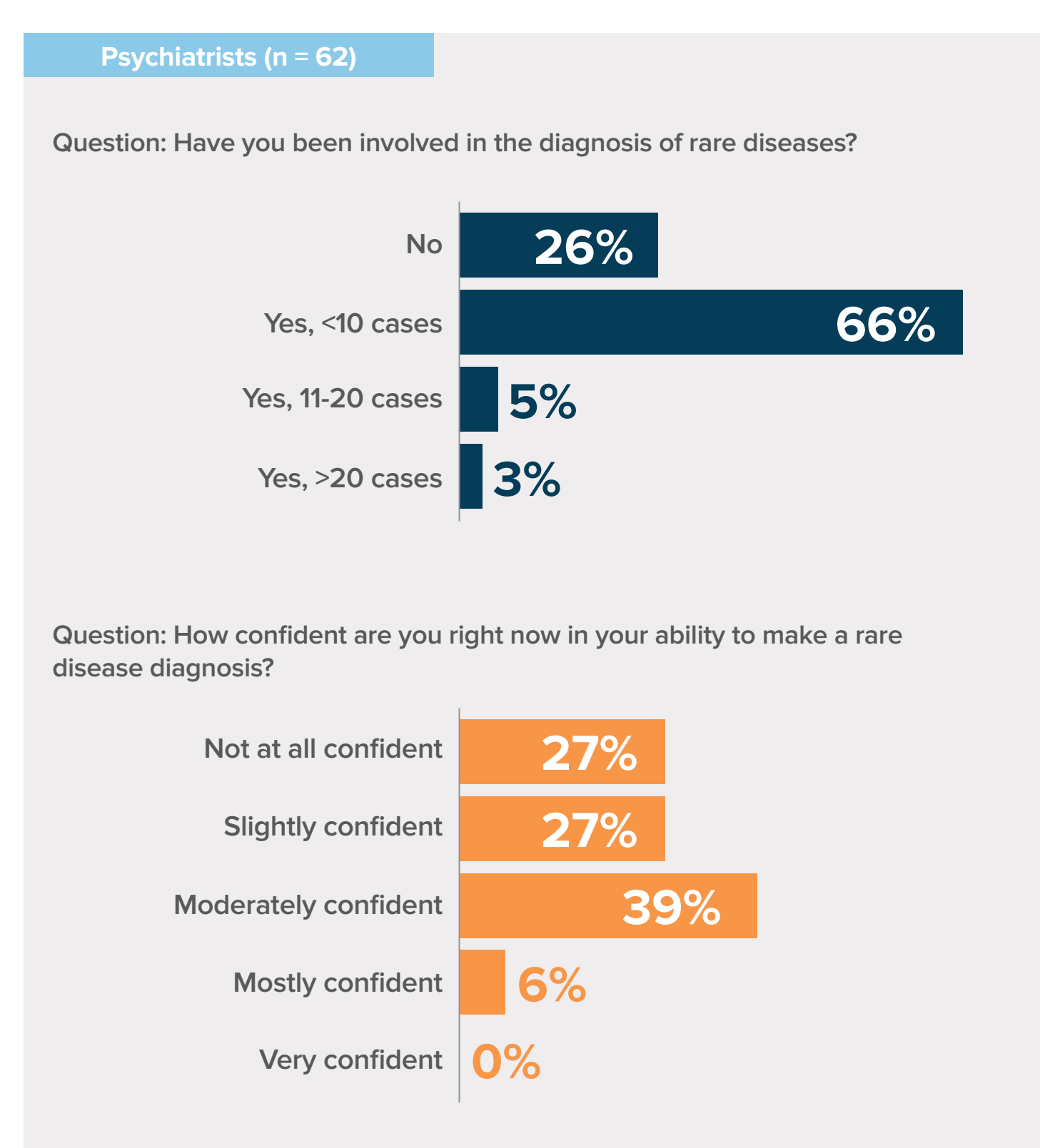
PREVALENCE OF RARE DISEASE

67% of psychiatrists believed rare diseases to be much rarer as compared with standard definitions in the EU (5 per 10,000) or US (<200,000).



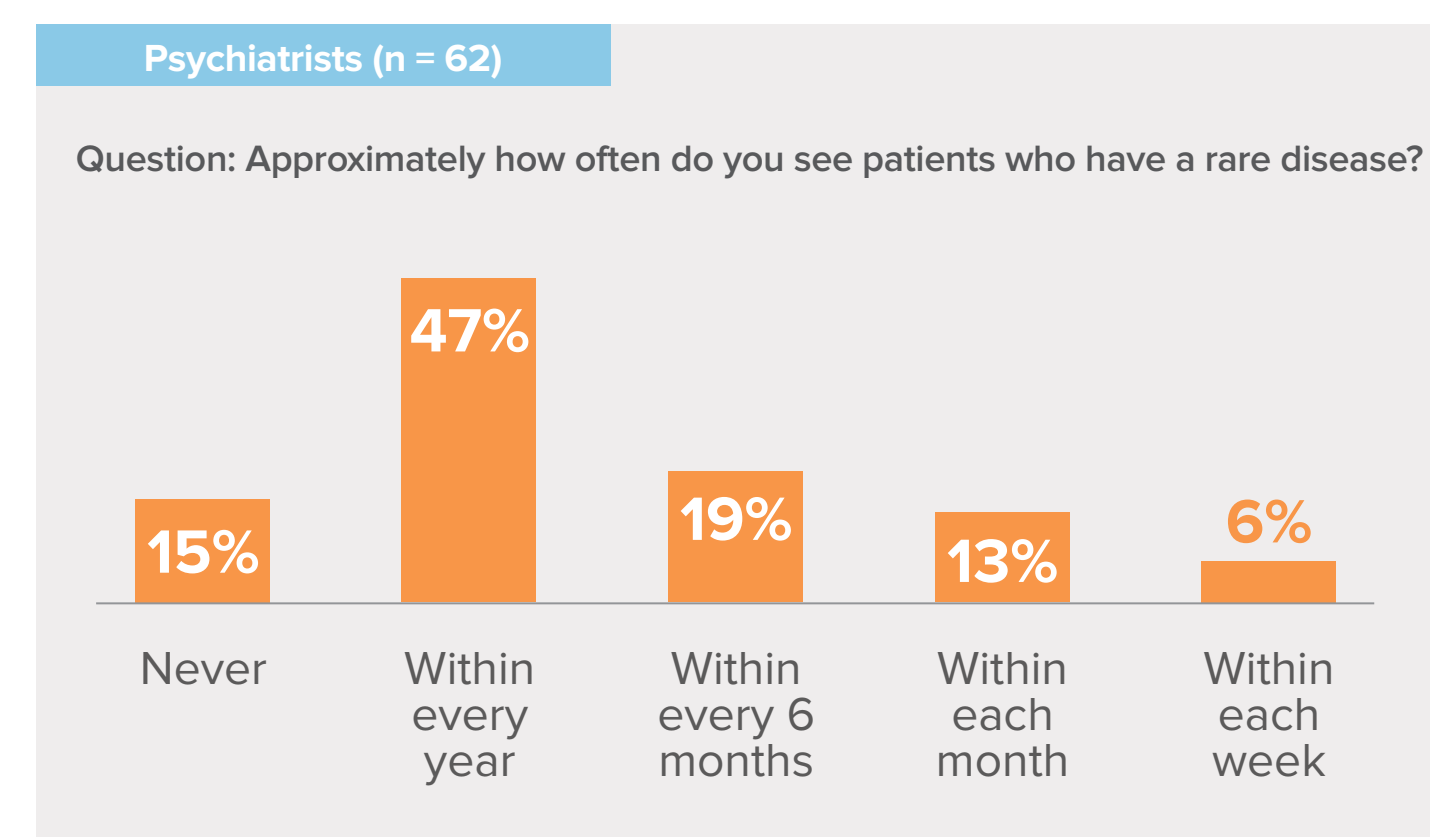
RARE DISEASE DIAGNOSIS

Although 74% said they have been involved in a rare disease diagnosis, only 6% were 'mostly' confident and none 'very' confident in making such a diagnosis.



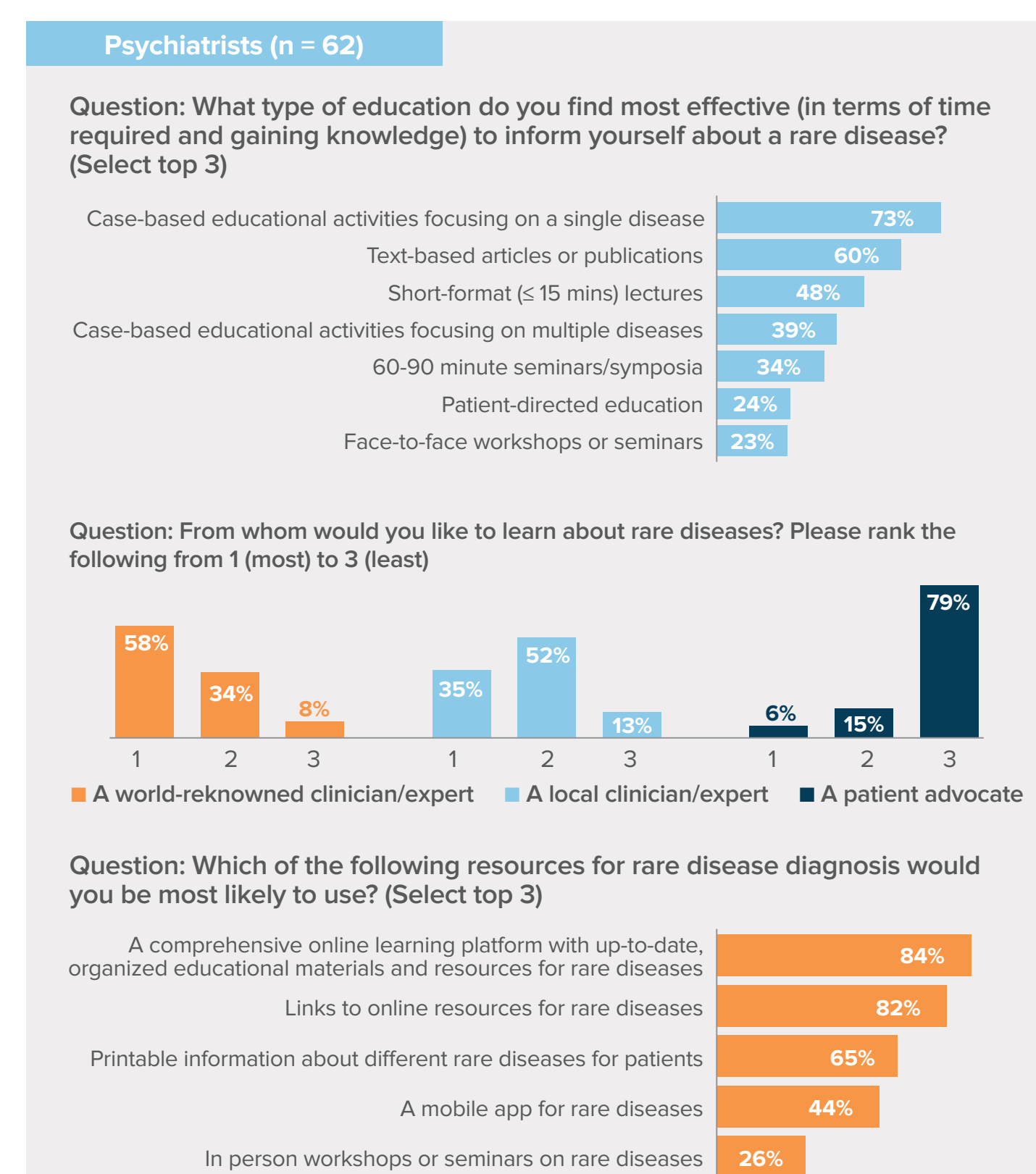
EXPERIENCE IN RARE DISEASE

15% of psychiatrists said they never see rare disease patients in their practice, by far more than any other specialty surveyed; and 66% said they see them only rarely (every 6 months to 1 year).



WHAT KIND OF RARE DISEASE EDUCATION DO PSYCHIATRISTS WANT?

When asked about how they wanted to learn about rare diseases, psychiatrists noted a preference for having access to a comprehensive online learning platform with educational materials (particularly case-based, text-based, and short-format education), given by world renowned as well as local clinician experts. There was also interest in having online resources for different rare diseases, and printable information on different rare diseases for their practice.



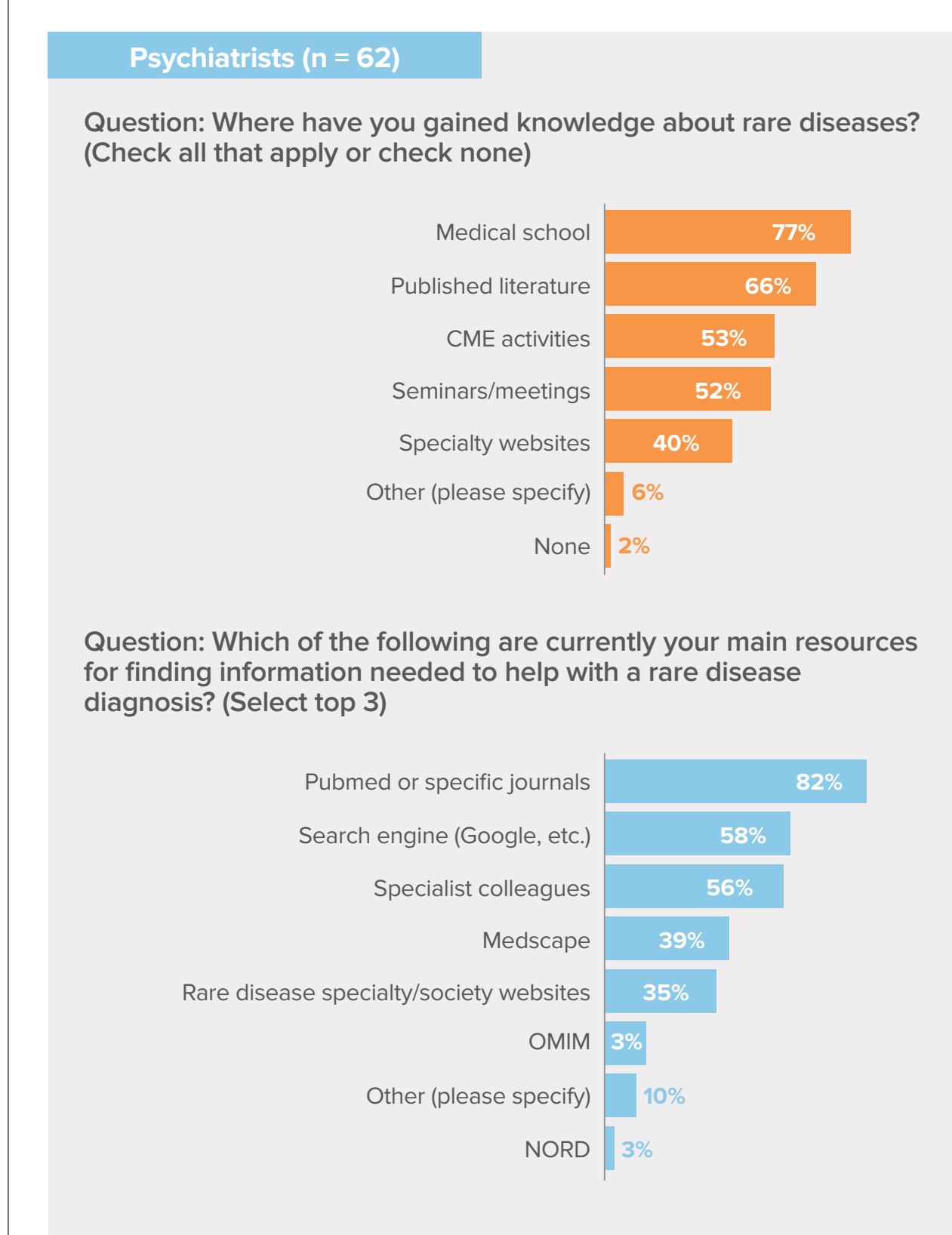
BARRIERS IN RARE DISEASE

Barriers that psychiatrists often or always encountered included sufficient time to investigate properly (47%), availability of diagnostic guidelines (47%), access to diagnostic tests (41%), knowing where to refer the patient (40%), and sufficient knowledge of signs and symptoms that should trigger suspicion of a rare disease (34%).



RARE DISEASE KNOWLEDGE AND INFORMATION

Psychiatrists said they have gained knowledge about rare diseases from medical school (77%), published literature (66%), CME activities (53%), seminars or meetings (52%), or specialty websites (40%). Published literature remains the current top resource for finding information needed to help with a rare disease diagnosis, much more so than specialist colleagues or rare disease websites.



CONCLUSIONS

This educational research study identifies gaps in knowledge regarding the prevalence of rare diseases, low confidence in diagnosing rare diseases, and a low rare disease patient load for psychiatrists, especially considering the proportion of rare diseases that present with neuropsychiatric symptoms. This study supports the need to develop education for psychiatrists on recognizing rare diseases earlier to reduce the very common diagnostic odyssey often associated with unnecessary diagnostic testing or treatment, frustration, and missed opportunities for the right treatments.

ACKNOWLEDGEMENTS

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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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