Are Pediatric Neurologists Who Participate in Rare Disease Education More Likely to Order Genetic Tests for Rare Diseases?

S. Christy Rohani-Montez, PhD; Karen Reid, MPharm; Meng Yuan; Jake Cohen:
Medscape Education Global, London, United Kingdom

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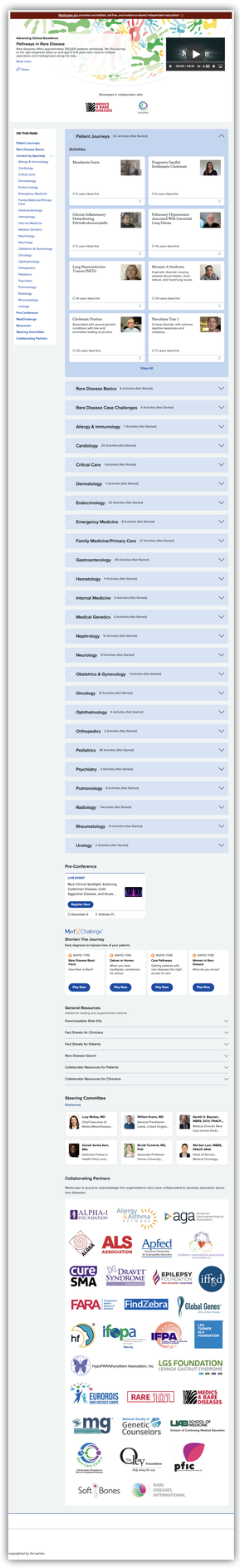
BACKGROUND

Approximately 70% of rare diseases present exclusively in pediatric practice[1]; however, there remains a significant diagnostic delay of an average 4-5 years. We previously reported that of 978 HCPs surveyed, 59% said they never or rarely (1× or 2× per year) see patients with rare diseases, and specifically for paediatricians, this number rises as high as 70%,[2] suggesting that pediatricians are often missing or misdiagnosing patients with rare diseases.

We sought to evaluate the impact of online accredited medical education in reducing diagnostic delays in rare diseases in pediatric practice by evaluating whether pediatric neurologists who participated in rare disease education vs those who didn't were more likely to order genetic tests for rare diseases.

METHODS

We evaluated total claims using 30 CPT codes for specific rare diseases, multigene panels, whole exome and whole genome sequencing, cytogenetic testing, biochemical genetic testing, pharmacogenomics & carrier screening, genetic counseling between 2021 and 2024 amongst pediatric neurologists who participated in one or more of 24 online CME activities (n = 301) compared with a cohort of same specialists who submitted claims in the same time frame (n = 341) but had not participated in the education.



Pediatric Neurologist Learners (n = 301)

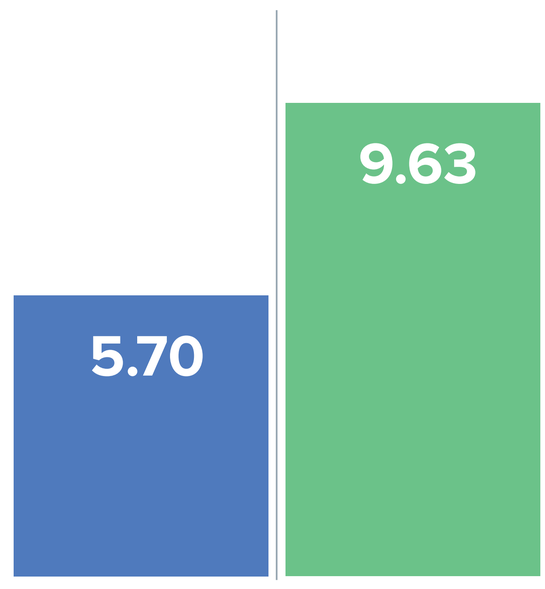
Pediatric Neurologists Non-learners (n = 341)

RESULTS

Pediatric neurologists who participated in rare disease education ordered genetic tests for an average of 9.63 patients, as compared with 5.70 patients amongst those who did not participate in education. This represents a 69% increase in ordering genetic tests amongst those educated ($P < .001$).

AVERAGE GENETIC TESTS ORDERED

↑69%
RELATIVE INCREASE
$P < .001$



CONCLUSION

This study demonstrates that online rare disease education for pediatric neurologists is associated with increases in ordering genetic tests, suggesting that education has the power to directly reduce delays in the diagnosis of rare diseases.

MEDSCAPE'S RARE DISEASE LEARNING CENTER
https://www.medscape.org/advances/rare-disease-2025a100013m

For more information contact:
S. Christy Rohani-Montez, PhD
Rare Disease Education Lead
Senior Director of Clinical Strategy
Medscape Education Global
srohani@medscape.net

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