Continuing Medical Education is Significantly Associated With Increased Rare Neurological, Endocrine and Liver Disease Diagnoses in Real World Practice

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

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BACKGROUND

The odyssey to confirm a rare disease diagnosis often lasts many years. This can be attributed to poor familiarity with rare diseases amongst healthcare providers (HCPs) as well as non-specific symptoms associated with most rare diseases.

We previously reported that of 978 HCPs surveyed, 59% said they never or rarely (1× or 2× per year) see patients with rare diseases,[1] despite a point prevalence of 3.5% to 5.9% globally[2].

We sought to evaluate the impact of online accredited medical education in reducing diagnostic delays in rare diseases by evaluating diagnostic code use amongst educated and non-educated physicians as a surrogate measure.

METHODS

14,996 neurologists, **8,389 paediatricians**, and **28,421 primary care physicians (PCPs)** who participated in at least one of 24 online CME activities on rare neurological conditions

3,604 endocrinologists, **3,002 PCPs**, and **447 rheumatologists** who participated in at least one of 9 online CME activities on rare endocrine conditions

5,585 gastroenterologists/hepatologists (GASTROs/HEPs), **5,677 paediatricians**, and **619 emergency medicine physicians (EMPs)** who participated in at least one of 9 online CME activities on rare liver conditions

We compared total rates of diagnostic code use (ICD-10 codes) amongst physicians educated versus not educated on between 2020 and 2025 across

- **11 rare neurological conditions:** amyotrophic lateral sclerosis, chronic inflammatory demyelinating polyneuropathy, muscular dystrophies, rare epilepsies, Friedreich's ataxia, frontotemporal dementia, idiopathic hypersomnia, Lambert-Eaton myasthenic syndrome, multifocal motor neuropathy, Rett syndrome, and Wilson's disease
- **6 rare endocrine conditions:** X-linked hypophosphatemia, tumour-induced osteomalacia, acromegaly, Cushing syndrome, adrenocortical insufficiency, and hypophosphatasia
- **8 rare liver conditions:** acute intermittent or hepatic porphyria, alpha-1 antitrypsin deficiency, hepatorenal syndrome, Alagille syndrome, progressive familial intrahepatic cholestasis, biliary atresia, and primary biliary cholangitis

RESULTS

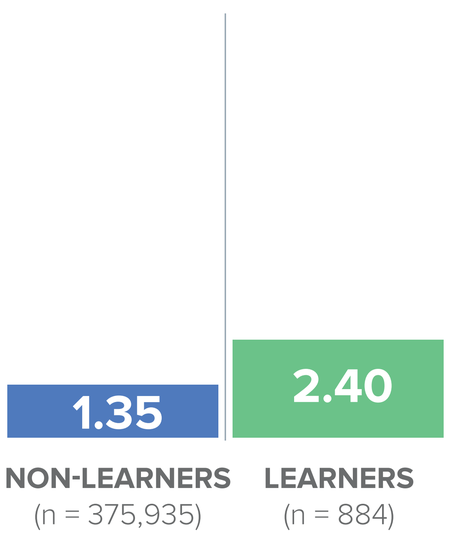
RARE NEUROLOGICAL CONDITIONS

A significant increase in diagnostic claims for rare neurological disorders amongst those who participated in at least one of 24 online accredited activities between 2020 and 2025

There were **376,819 total HCPs** with ICD claims for these conditions between 2020 through July 2025.

Physicians participating in **rare neurology activities** who had NPI numbers included 9,267 neurologists, 3,703 pediatricians, and 13,046 PCPs. Within this cohort, for those who also used an ICD10 code associated with the rare neurological conditions listed above, there was a significant difference in average ICD claims per physician depending on whether they participated in rare disease education (learners) or did not (non-learners).

AVERAGE CLAIMS PER PHYSICIAN



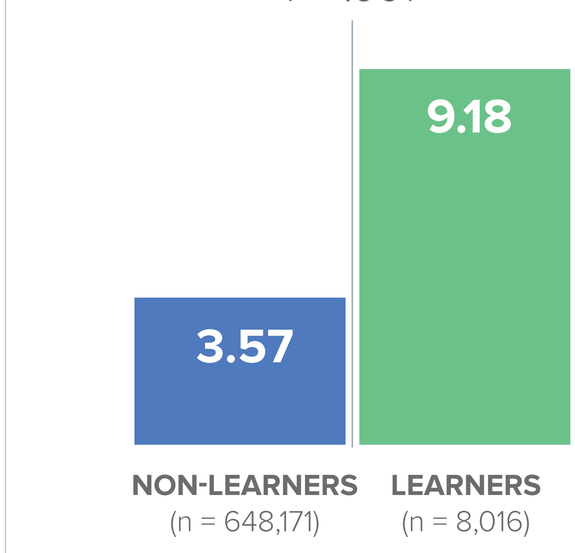
RARE ENDOCRINE CONDITIONS

A significant increase in diagnostic claims for rare endocrine disorders amongst those who participated in at least one of 9 online accredited activities between 2020 and 2025

There were **656,187 total HCPs** with ICD claims for these conditions between 2020 through July 2025.

Physicians participating in **rare endocrine activities** who had NPI numbers included 1,618 endocrinologists, 1,334 PCPs, and 197 rheumatologists. Within this cohort, for those who also used an ICD10 code associated with the rare endocrine conditions listed above, there was a significant difference in average ICD claims per physician depending on whether they participated in rare disease education (learners) or did not (non-learners).

AVERAGE CLAIMS PER PHYSICIAN



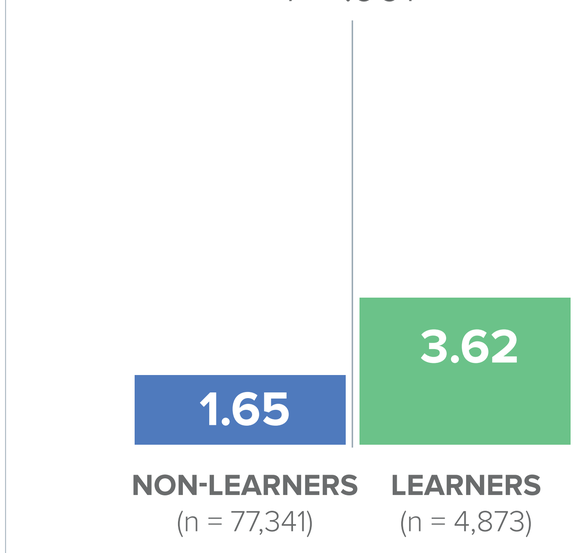
RARE LIVER CONDITIONS

A significant increase in diagnostic claims for rare liver disorders amongst those who participated in at least one of 9 online accredited activities between 2020 and 2025

There were **82,214 total HCPs** with ICD claims for these conditions between 2020 through July 2025.

Physicians participating in **rare liver activities** who had NPI numbers included 2,622 gastroenterologists/hepatologists, 632 paediatricians, and 242 emergency medicine specialists. Within this cohort, for those who also used an ICD10 code associated with the rare liver conditions listed above, there was a significant difference in average ICD claims per physician depending on whether they participated in rare disease education (learners) or did not (non-learners).

AVERAGE CLAIMS PER PHYSICIAN



CONCLUSION

This study demonstrates that online rare disease education is associated with increased rates of rare neurological, endocrine, and liver disease diagnoses, suggesting that education has the power to directly reduce diagnostic delays.

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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REFERENCES

1. Rohani-Montez, SC et al. Educational needs in diagnosing rare diseases: A multinational, multispecialty clinician survey. *Genet Med Open*. 2023;1:100808.
2. Nguengang Wakap, S et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* 2020; 28:165-173.