

Challenges in Recognizing Late-Onset Neuromuscular Diseases (LONDs)

Demonstrate Need for Education & Tools to Support Early Detection and Referral

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Background

Reaching a rare disease diagnosis can take more than six (6) years and 17 clinical encounters on average.¹ For LONDs, the journey is similar: time to diagnosis is nearly one (1) year for amyotrophic lateral sclerosis (ALS)² and myasthenia gravis (MG)³ and up to five (5) years in 13% of MG cases.³

Earlier recognition and referral to neuromuscular specialists (NMS) is key to shortening the time to diagnosis and improving care for LONDs; however, we must first understand physician challenges in achieving this. This information will then guide creation of relevant and practical education, resources, and tools.

The Late-Onset Neuromuscular Disease Consortium (LONDC), a project of the American Neuromuscular Foundation (ANF), conducted a survey to measure physician:

- Ability to recognize a LOND
- Referral behaviors for suspected LONDs
- Perceived barriers to timely diagnosis
- Interest in a practical decision-support tool

Using these results and other research on early signs and symptoms across LONDs, the LONDC will create a screening tool to help physicians spot potential LONDs sooner and send patients to appropriate NMS for more testing.

The LONDC defines late-onset neuromuscular disease as any neuromuscular disease that typically manifests in adulthood. A list of these conditions can be found on the LONDC website.

londc.neuromuscularfoundation.org



Methods

An online survey of 120 US-based physicians likely to see patients with LONDs:

40
Advanced practice
providers (APPs)

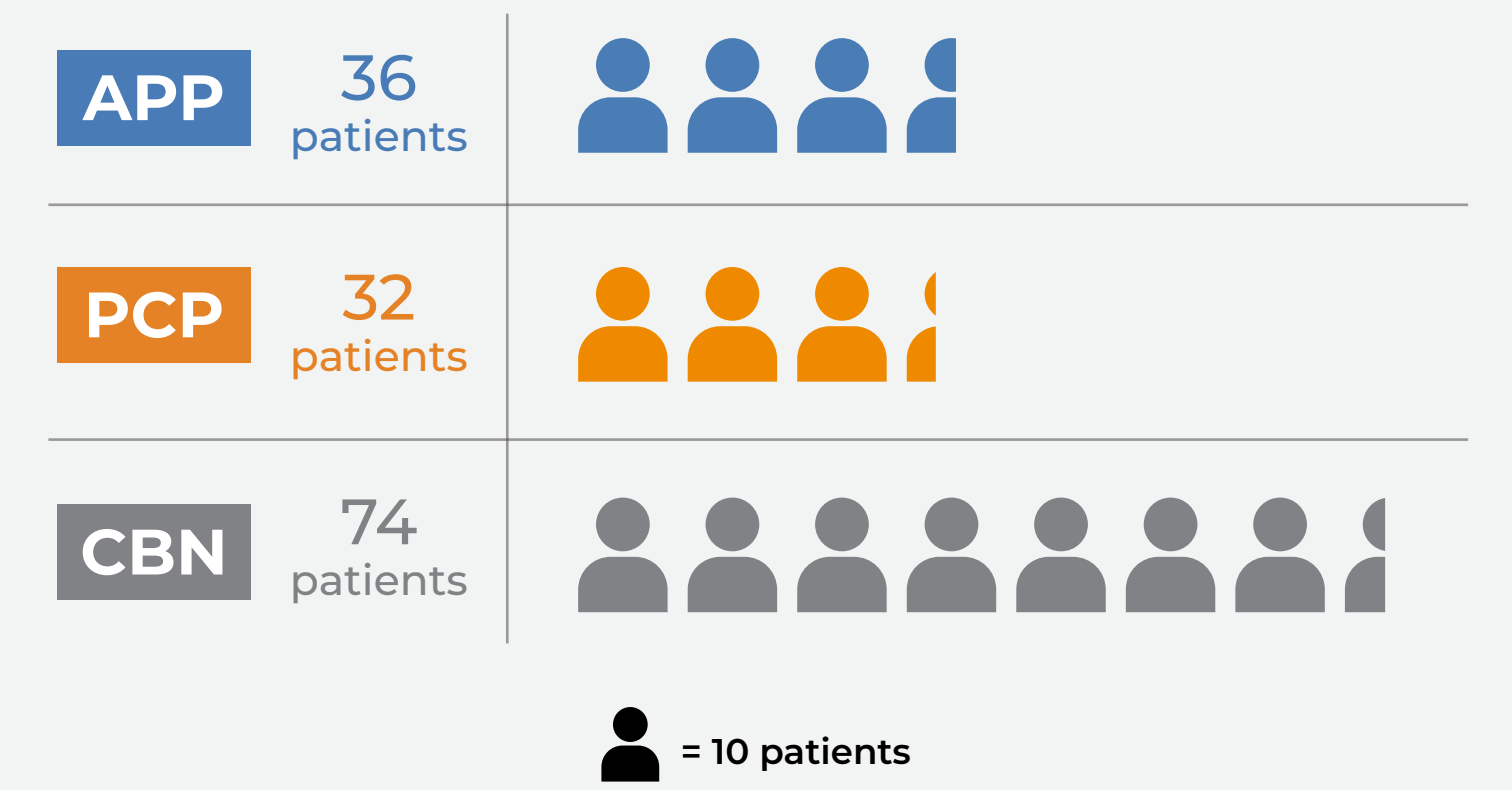
40
Primary care
physicians (PCPs)

40
Community-based
neurologists (CBNs)

Participants had to be actively practicing and treating, or have treated, a meaningful number of LOND patients. CBNs averaged the most LOND patients at 74, followed by APPs with 36 and PCPs with 32 (Figure 1).

We presented blinded, hypothetical MG and chronic inflammatory demyelinating polyneuropathy (CIDP) patient case studies in an alternating manner to prevent order bias and asked about their differential diagnosis, and their intent to treat or refer.

Figure 1 – Average Number of LOND Patients Seen, by Provider Type



Results

Figure 2 – Self-Reported Recognition of LONDs, by Provider Type

LOND recognition is inconsistent: APPs and PCPs had less awareness, knowledge, and comfort recognizing presenting signs and symptoms of LONDs compared with CBNs.

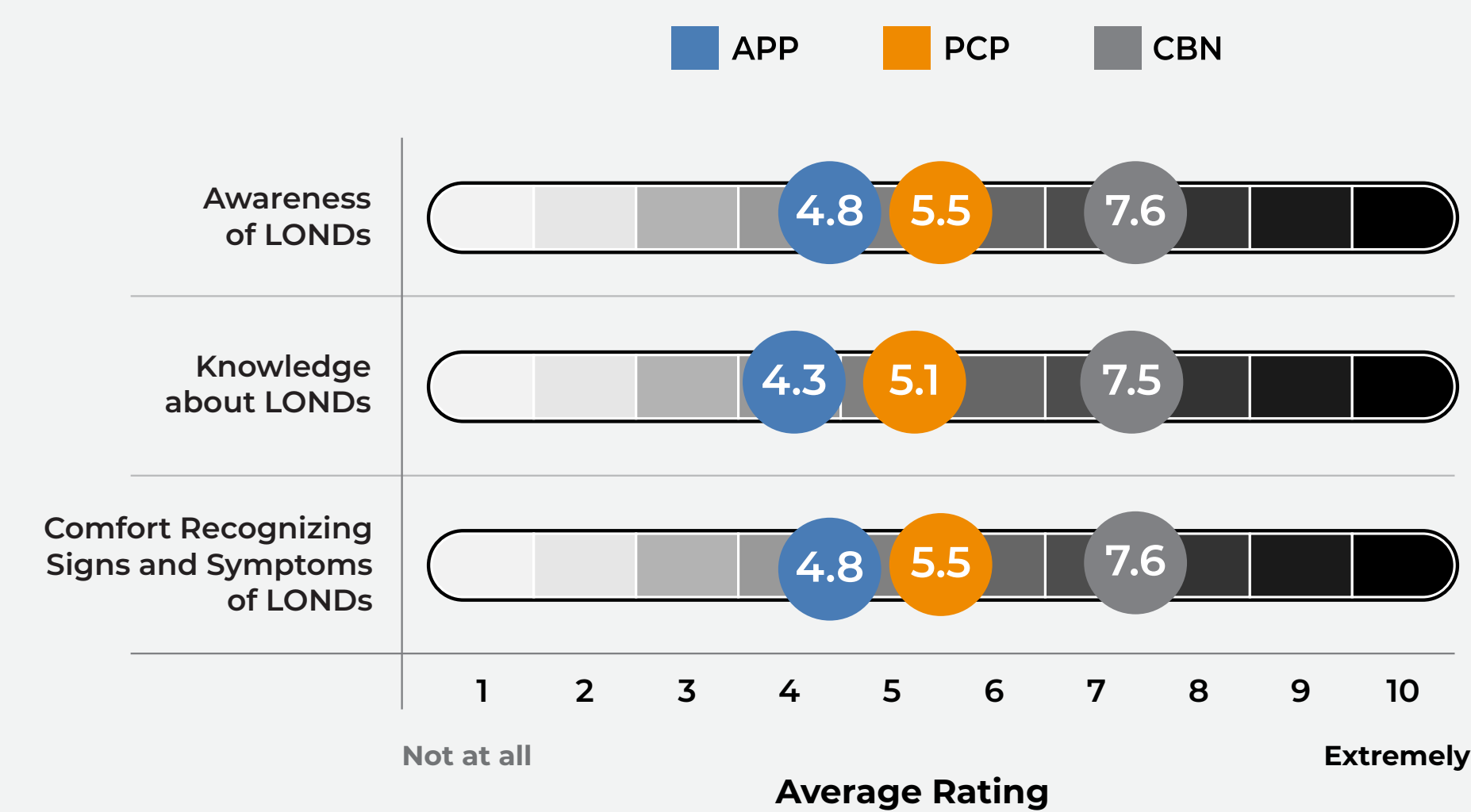


Figure 3 – Diagnostic Accuracy of Blinded MG and CIDP Cases, by Provider Type

CBNs' diagnostic accuracy varied by condition: 88% correctly identified the blinded MG case, but only 40% recognized the blinded CIDP case. Accuracy among other providers was lower: 45% of PCPs and 35% of APPs identified the MG case, while almost none recognized CIDP (0% of PCPs, 3% of APPs).

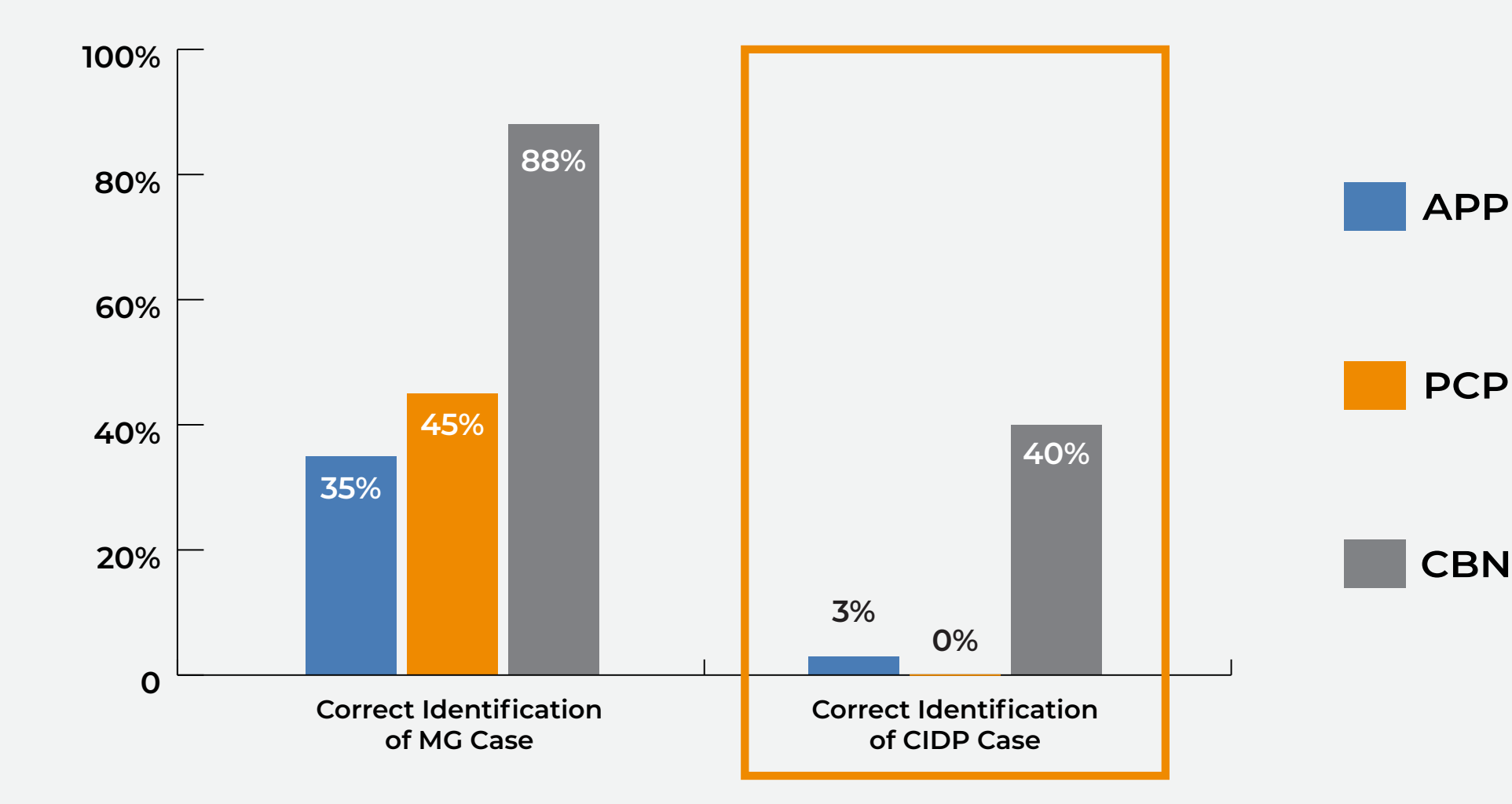


Figure 4 – Referral Intentions for MG and CIDP Patients, by Provider Type

NMS referrals are rare: Referral intentions were low for both MG and CIDP cases, with providers highly likely to retain patients, suggesting the referral rate is low in general. Just over one quarter (27.5%) of APPs referred MG patients, and this figure was even lower for CIDP patients at 15%.

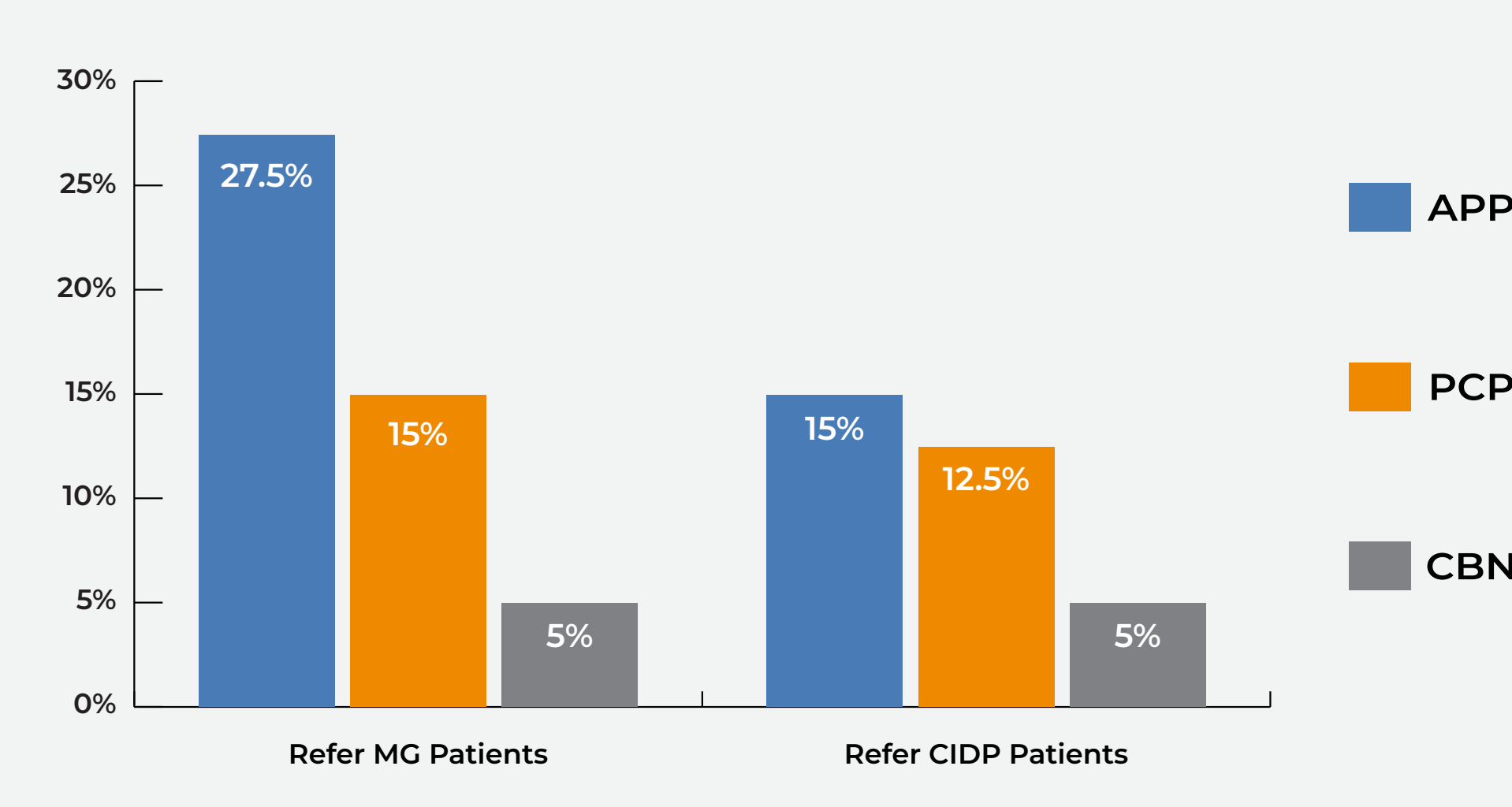


Figure 5 – Ease of Referral to Neuromuscular Specialists

Despite reported ease, referrals to NMS are low: Clinicians of all types rated referrals to a NMS as easy, with an average rating of 7.4 on a 1-10 scale with 10 being very easy. But referrals were rare (Figure 4), and when they did occur, all but one referral was to a neurologist, indicating low likelihood of referral to an NMS.

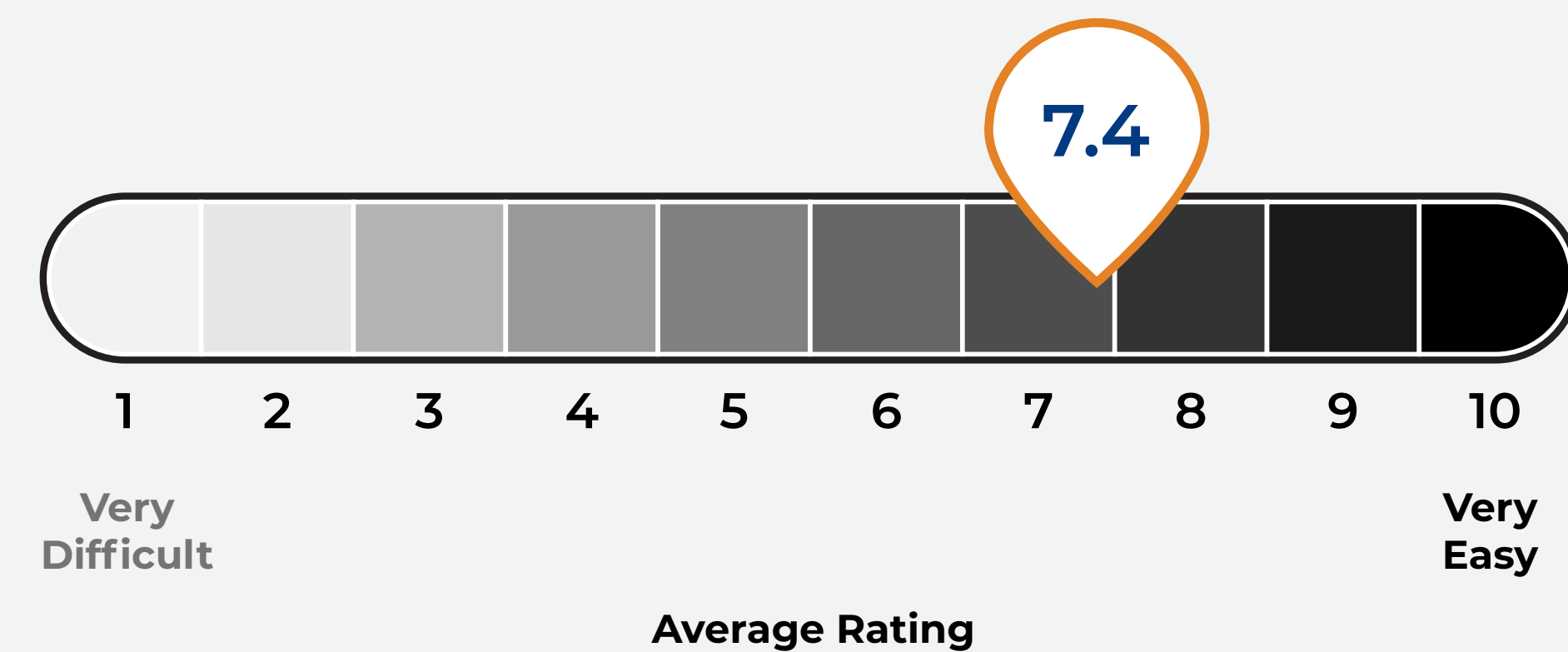


Figure 6 – Clinician-Reported Barriers to NMS Referrals

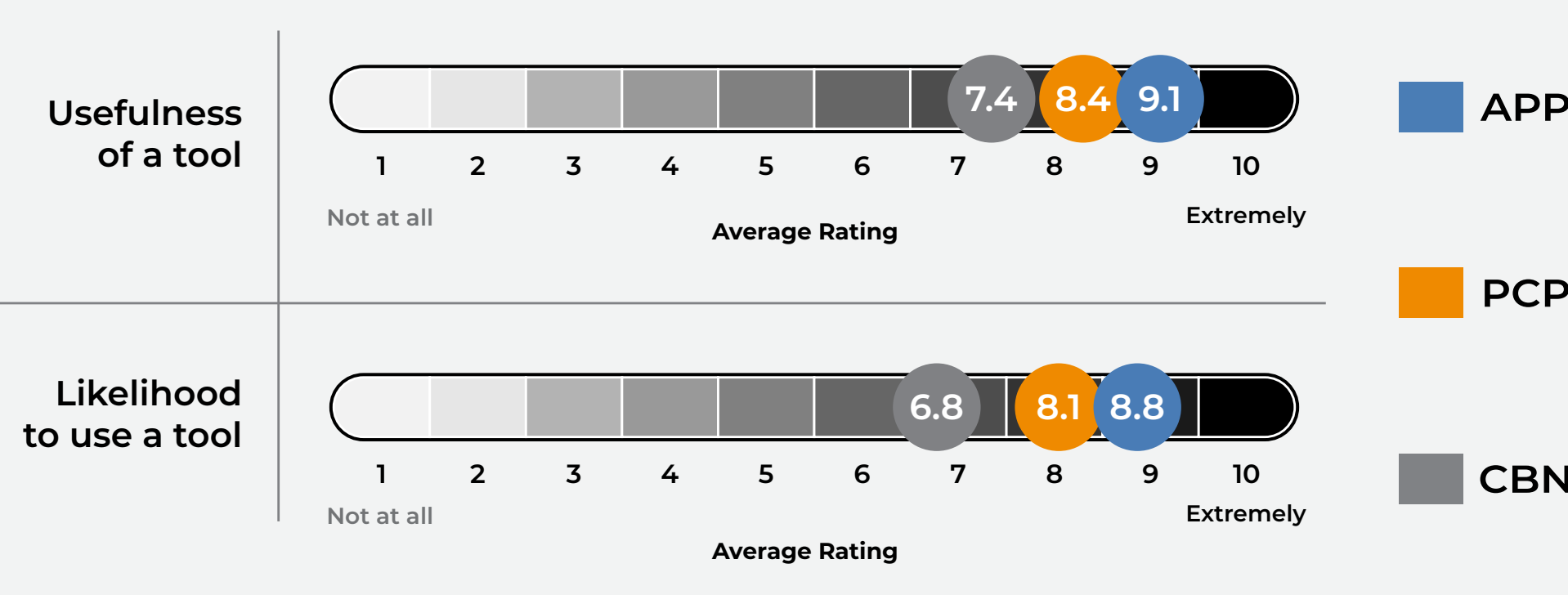
Barriers hindered timely referrals: Physicians reported several barriers including: difficulty getting appointments, lack of exposure to LONDs, and not recognizing symptoms, among others.

Barrier*	APP	PCP	CBN
Getting an appointment with an NMS	●	●	●
Lack of exposure to LONDs	●	●	●
Not recognizing symptoms		●	●
Lack of diagnostic testing			●

*Filled circle denotes that at least 25% of respondents reported these respective barriers

Figure 7 – Provider Interest in a Decision-Support Tool for LONDs

Strong interest in tools: Across all provider types, there was substantial interest in a decision-support tool to aid in recognizing LONDs and guiding referral and potential diagnostic next steps. APPs rated both usefulness and likelihood of use the highest and PCPs followed with slightly lower ratings. CBNs had the lowest ratings of the three specialties, but interest was still fairly strong.



Key Insights and Outcomes

- **Recognition of LONDs is low:** CBNs show higher awareness and accuracy than APP and PCPs, but among CBNs, diagnostic accuracy differs by condition.
- **Referrals to neuromuscular specialists (NMS) are rare:** Most providers prefer to retain patients for further testing rather than refer. When referrals happen, they usually go to general neurologists, not NMS.
- **Multiple barriers slow timely referral:** Difficulty obtaining NMS appointments, limited provider exposure to LONDs, gaps in knowledge and training, and challenges in symptom recognition are key obstacles to NMS referrals.
- **Strong support exists for decision-support tools:** All provider types expressed significant interest in tools that assist with LOND recognition and diagnostic guidance, with APPs showing the highest enthusiasm.

Conclusions

In this study of primary care providers (APPs and PCPs) and general neurologists, awareness, knowledge, and recognition of LONDs are insufficient, contributing to a low rate of referrals from front-line providers to NMS. Gaps in recognition and understanding of LONDs are key barriers to referrals that potentially contribute to diagnostic delays.

There is significant room for improvement in LOND referrals to NMS by developing resources that address neuromuscular conditions collectively. A practical, cross-condition decision-support tool could meaningfully improve early recognition and appropriate referral to NMS, ultimately reducing time to diagnosis and improving patient care.



About LONDC

The American Neuromuscular Foundation (ANF)'s Late-Onset Neuromuscular Disease Consortium (LONDC) is intended to bring the community together and foster collaboration to improve care, education, and quality of life.

To learn more, visit londc.neuromuscularfoundation.org or scan here:



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Discussion

- Delayed diagnosis of LONDs is a challenge that could be addressed with earlier and better referrals to NMS.
- An effective solution to the lack of LOND referrals to NMS may be less about extensive condition-specific education and more about providing a practical, cross-condition screening tool.
- Raising suspicion of LONDs in general may help providers recognize when a patient falls into this broader category, thereby facilitating timely and effective care through earlier referral and a quicker, more accurate diagnostic journey.
- The survey data show that providers are interested in a decision aid and would potentially use it in their practice.

Limitations

- Referral rates reported by physicians surveyed were low; regardless of specialty, providers more often retain possible LOND cases for additional testing to reach a diagnosis. This could be because of their prior experience with LOND patients.
- This survey did not assess the time interval between initial patient presentation and eventual referral, so we cannot determine how quickly identified cases would reach a NMS.
- In some situations, retaining patients for additional evaluation before referral may be clinically appropriate and necessary to ensure informed decision-making. A tool would not be meant to replace clinical judgement.

References

1. Yang, G., Cintina, I., & Pariser, A. (2022). The national economic burden of rare disease in the United States in 2019. Orphanet J, 17, 163. 2. Paganoni, S., Macklin, E. A., Lee, A., Murphy, A., Chang, J., Zipf, A., Cudkowicz, M., & Atassi, N. (2014). Diagnostic timelines and delays in diagnosing amyotrophic lateral sclerosis (ALS). Amyotrophic lateral sclerosis & frontotemporal degeneration, 15(5-6), 453-456. <https://doi.org/10.3109/21678421.2014.903974> 3. Bubuic, A-M., Vitale, L. (2023). Time to diagnosis of myasthenia gravis: A tertiary care center analysis. Journal of the Neurological Sciences, Volume 455, Supplement, 122017, ISSN 0022-510X. <https://doi.org/10.1016/j.jns.2023.122017>.

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