Rare Expertise

WHITE PAPER

Raising the Baseline:

From Rare Awareness to Clinical Suspicion

Why Specialists Need Specialty-Specific Rare Disease Education — and How It Changes Outcomes

A Rare Expertise White Paper

For rare disease marketing and medical affairs professionals

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SECTION 01

Executive Summary

Approximately 300 million people worldwide live with one of more than 7,000 identified rare diseases. Despite this enormous collective burden, the average time to diagnosis for a rare disease patient remains nearly five years. The primary driver of this delay is not a lack of available treatments or diagnostic technologies — it is a lack of clinical suspicion among the physicians these patients see first.

The challenge is not that physicians are indifferent. It is that the sheer number and heterogeneity of rare diseases makes it impossible for any individual clinician to know them all. What is possible — and what published research increasingly supports — is to raise the baseline level of rare disease awareness within each medical specialty, so that specialists are more attuned to the possibility that a patient presenting with unusual or treatment-resistant symptoms may have an underlying rare condition.

300M
people worldwide
LIVING WITH RARE DISEASES

7,000+
identified diseases
AND GROWING

4.7 yrs
average diagnosis time
RARE BAROMETER 2024

This white paper examines the evidence behind the rare disease knowledge gap among specialists, explores what motivates physicians to seek rare disease information and how they prefer to learn, and makes the case that specialty-specific education — delivered through channels specialists already trust — is the most effective strategy for shortening the diagnostic odyssey and improving outcomes for rare disease patients.

90%

of the total diagnostic delay occurs *within* the healthcare system — not before patients first seek medical attention.

RARE BAROMETER SURVEY, 2024 · 6,500+ PATIENTS · 41 COUNTRIES

SECTION 02

The Diagnostic Odyssey: A Problem of Clinical Suspicion

The term "diagnostic odyssey" has become shorthand for one of the most painful realities in rare disease: the prolonged, often years-long journey patients endure from first symptoms to correct diagnosis. This is not merely an inconvenience. It carries profound medical, psychological, and financial consequences.

A 2024 Rare Barometer survey of more than 6,500 patients across 41 European countries found that the average total diagnosis time is 4.7 years, with 56% of respondents waiting more than six months after their first medical contact. Critically, the study found that 90% of the total diagnostic delay occurs within the healthcare system itself — not in the period before patients first seek medical attention.

> KEY FINDING
>
> This finding reframes the problem. Patients are seeking help early. The delay happens after they enter the medical system, as they move between providers, receive misdiagnoses, and undergo inappropriate treatments — often because no clinician along the way considers the possibility of a rare disease.

The Knowledge Gap Is Real — and Measurable

A landmark 2023 multinational survey of 978 clinicians across 16 specialties, published in a collaboration between Medscape Education and leading rare disease researchers, quantified the scope of the problem. The findings were striking.

2/3

of clinicians believed rare diseases are 50-500x rarer than they actually are

59%

said they never or rarely encountered rare disease patients

Despite a point prevalence of 3.5-5.9% — with estimates reaching 10% in the general population — the majority of respondents said they never or rarely encountered patients with rare diseases, suggesting not that the patients are absent from their practices, but that they are going unrecognized.

The barriers physicians reported are instructive: insufficient knowledge of signs and symptoms (reported by 38-44% of respondents), insufficient time to investigate properly, lack of diagnostic guidelines, limited access to diagnostic tests, and uncertainty about where to refer patients. These are not barriers of motivation — they are barriers of infrastructure and education.

The Specialist's Paradox

A Belgian study of 295 physicians published in the Orphanet Journal of Rare Diseases revealed a critical distinction in how different types of physicians relate to rare disease information. General practitioners reported needing rare disease information only when they had a rare disease patient in front of them — a reactive, case-by-case approach. Specialists, by contrast, indicated that they needed more rare disease information in general.

> *40-44% of specialists stated that they only needed rare disease information that related to their own specialty. Specialists do not want to learn about all 7,000 rare diseases. They want to know about the rare diseases most likely to present in their clinics.*
>
> Orphanet Journal of Rare Diseases, 2019

This creates what might be called the "specialist's paradox." Specialists are the physicians best positioned to recognize rare diseases within their domain, yet their training and continuing education rarely equip them to do so. The neurologist who sees a puzzling case of progressive weakness may not think of Pompe disease. The cardiologist managing unexplained left ventricular hypertrophy may not consider Fabry disease. The rheumatologist treating recurrent fevers may not suspect autoinflammatory syndromes. Not because these conditions are outside their scope — but because no one ever taught them to look.

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SECTION 03

What Motivates Physicians to Seek Rare Disease Information?

Understanding why physicians seek rare disease information — and what prevents them from doing so — is essential to designing effective education strategies. The research reveals several distinct motivational patterns.

The Puzzling Patient

The single most powerful trigger for rare disease information-seeking is the encounter with a patient who does not fit standard diagnostic categories. When a patient's symptoms are unexplained, treatment-resistant, or multisystem in nature, clinicians experience what researchers describe as "pattern failure" — the recognition that their existing diagnostic frameworks are insufficient.

Research on physician diagnostic strategies shows that clinicians commonly begin with "inductive foraging" (open-ended questioning), progress to "triggered routines" (system-specific inquiry), and then apply probabilistic reasoning to arrive at a diagnosis. When these standard strategies fail, physicians either continue cycling through common diagnoses — a phenomenon researchers call "particularising" — or they escalate their search to consider less common possibilities. The key question is whether rare diseases are accessible in the physician's mental library when that escalation occurs.

Professional Identity and the Desire for Mastery

Beyond the immediate clinical trigger, physicians are motivated by a deeper professional drive: the desire to be excellent clinicians within their specialty. Specialists take pride in their ability to diagnose and manage the full range of conditions within their domain.

IMPORTANT DISTINCTION

This professional motivation is distinct from the CME compliance motivation that drives much continuing education. Physicians seeking rare disease knowledge are typically not checking a box — they are trying to solve a clinical puzzle or fill a gap they have recognized in their own expertise. Education designed for this audience must be clinically compelling, not merely accredited.

The Emotional Dimension

Research has also identified an underappreciated emotional dimension to the rare disease diagnostic experience. Physicians who encounter patients with undiagnosed rare diseases report frustration, self-doubt, and stress — emotions that mirror the distress of the patients themselves.

This emotional burden can be both a barrier and a motivator. For some physicians, the frustration of repeated diagnostic failure leads to disengagement — the patient is referred elsewhere, or their symptoms are attributed to psychosomatic causes. For others, it creates a powerful drive to learn more. Effective education should acknowledge this emotional reality rather than treating rare disease diagnosis as a purely intellectual exercise.

Peer Influence and Collegial Learning

Physicians do not learn in isolation. A substantial body of evidence shows that peer interaction — through case conferences, grand rounds, informal consultations with colleagues, and specialty society discussions — is one of the most influential channels through which clinicians update their diagnostic thinking.

This peer-to-peer dynamic is particularly powerful in rare disease, where a single memorable case discussion can permanently alter a specialist's index of suspicion. The challenge is that such interactions are sporadic and unscalable — unless they are deliberately facilitated through educational platforms that create similar dynamics in a more structured and accessible format.

SECTION 04

How Physicians Search for Rare Disease Information

When physicians do seek rare disease information, their search behaviors follow predictable patterns — patterns that have significant implications for how educational content should be designed and delivered.

Reading vs. Interaction: Two Complementary Pathways

Physician learning broadly divides into two modes: **reading** (self-directed consumption of written content) and **interaction** (learning through dialogue with peers, experts, or patients). Both are essential, but they serve different purposes in the rare disease context.

READING

Builds baseline awareness. When a specialist reads about a rare disease in their field — even without immediate clinical need — it creates a cognitive "bookmark" that activates when a matching patient presents.

INTERACTION

Converts awareness into action. When a specialist hears a colleague describe a rare disease case, the learning is experiential and memorable in a way reading alone rarely achieves.

The multinational clinician survey found that the most preferred educational formats were **case-based content**, **short text-based formats of 15 minutes or less**, and **online learning platforms taught by recognized experts**. These preferences suggest that physicians want rare disease education to fit into the natural rhythms of their professional reading habits.

The most effective rare disease education strategies do not force a choice between these two modes. They combine regular, specialty-relevant content delivery (raising baseline awareness through reading) with the clinical specificity and narrative power of case-based learning (creating the memorable associations that change diagnostic behavior).

The Specialty Filter

One of the most consistent findings across the rare disease education literature is that physicians filter information by specialty relevance. A neurologist is unlikely to engage with content about rare endocrine disorders; a cardiologist will not attend a webinar on rare dermatologic conditions. This is not a limitation — it is entirely rational behavior in a world where every physician faces information overload.

> STRATEGIC IMPLICATION
>
> Rare disease education organized by specialty — delivered through specialty-specific channels, written in the clinical language of that specialty, and focused on the rare diseases most likely to mimic common conditions within that specialty — will consistently outperform generic rare disease education in terms of engagement, retention, and clinical impact.

CLINICAL RED FLAGS

Seven Pre-Diagnostic Indicators of Rare Disease

1. **Multisystem involvement** affecting three or more organ systems
2. **Genetic inheritance patterns** observed in family history
3. **Symptom continuity** from childhood through adulthood
4. **Multiple specialist referrals** without resolution
5. **Extended unexplained symptoms** with no clear diagnosis
6. **Prior misdiagnosis** or attribution to common conditions
7. **Treatment resistance** to standard therapeutic approaches

Source: Busbridge et al., Orphanet Journal of Rare Diseases, 2024

SECTION 05

The Case for Specialty-Specific Rare Disease Education

The evidence presented above converges on a single strategic insight: the most effective way to improve rare disease diagnosis is not to teach all physicians about all rare diseases. It is to ensure that within each medical specialty, practitioners have sufficient awareness of the rare conditions most relevant to their practice to maintain clinical suspicion when standard diagnoses fail.

The "Red Flags" Approach

Rare disease experts have long advocated for a "red flags" approach to education — teaching physicians not to diagnose rare diseases per se, but to recognize the clinical patterns that should trigger suspicion. Belgian rare disease experts emphasized that physicians' continuing education should focus on red flags that increase rare disease attentiveness in daily clinical practice.

This approach aligns with how diagnostic reasoning actually works. Physicians do not diagnose by running through a mental checklist of 7,000 conditions. They diagnose by recognizing patterns and, when patterns break, by escalating their investigation. The goal of specialty-specific education is to ensure that rare diseases are included in the set of possibilities a specialist considers when they encounter treatment-resistant or unexplained presentations.

Meeting Specialists Where They Already Are

The challenge of rare disease education is not just content — it is distribution. Physicians are overwhelmed with educational offerings, and rare disease content must compete with updates on common conditions, new treatment guidelines, and mandatory CME requirements.

> *When rare disease content appears alongside other clinical updates within a specialist's field, it is perceived as relevant rather than tangential. A neurologist encountering an article about a rare neuromuscular condition in a neurology-focused newsletter processes it differently than the same article in a generic rare disease publication.*

This is particularly important for what might be called "ambient awareness building." Not every piece of rare disease content will be immediately actionable. But repeated, specialty-relevant exposure creates a cumulative effect on clinical awareness. The specialist who has read articles, scanned lists, or taken a quiz about rare diseases over time is meaningfully more likely to consider a rare diagnosis when a puzzling case appears.

From Individual Articles to an Ecosystem of Awareness

The logic of specialty-specific rare disease education suggests a systematic rather than ad hoc approach. Rather than sporadic articles placed in general medical journals, what is needed is a sustained, multi-specialty educational ecosystem — one that consistently delivers rare disease content tailored to each specialty's clinical context, diagnostic vocabulary, and practice patterns.

THE CORE INSIGHT

The goal is not to transform every specialist into a rare disease expert, but to shift the entire distribution of rare disease awareness slightly upward across all specialties. Even a modest increase in the probability that any given specialist will consider a rare disease when faced with an atypical presentation can, at population scale, dramatically reduce the number of patients trapped in the diagnostic odyssey.

The goal is not to transform every specialist into a rare disease expert. It is to ***raise the baseline*** — shifting the entire distribution of awareness upward across all specialties.

SECTION 06

Implications for Rare Disease Marketing and Medical Affairs

For professionals working in rare disease marketing and medical affairs, the research and arguments presented in this paper carry several practical implications.

Invest in non-branded disease education

The most impactful intervention for many rare disease companies is not promotion of a specific therapy — it is ensuring that the specialists most likely to encounter their patient population can recognize the condition in the first place. Non-branded, specialty-specific disease awareness education serves this purpose more effectively than traditional HCP marketing.

Think in terms of specialties, not just rare diseases

The natural unit of rare disease education is not the disease — it is the specialty. Every rare disease lives within one or more clinical specialties, and the physicians who will diagnose it are specialists first. Educational strategy should begin by mapping each rare disease to the specialties most likely to encounter it, and then delivering education through specialty-appropriate channels.

Prioritize clinical pattern recognition

Specialists do not need to become experts in rare diseases to improve diagnostic outcomes. They need to know the red flags: when a common presentation may actually be a rare one, what additional questions to ask, and where to refer the patient. Education designed around clinical scenarios and differential diagnosis is more effective than encyclopedic disease overviews.

Leverage the power of sustained exposure

One-time educational interventions have limited impact on diagnostic behavior. Sustained, regular delivery of rare disease content — through specialty newsletters, website content, case discussions, and similar channels — builds the ambient awareness that changes clinical practice over time.

> **Recognize that peer influence amplifies education**
>
> When a specialist learns something clinically valuable about a rare disease, they are likely to share it with colleagues. This multiplier effect means that reaching even a subset of specialists within a field can shift awareness across a much larger group. Educational content designed to be shareable and discussable will have disproportionate impact.

SECTION 07

Conclusion

The rare disease diagnostic odyssey is, at its core, a problem of clinical suspicion. Patients enter the healthcare system and are seen by competent, well-intentioned specialists who simply do not consider the possibility that a rare disease may explain what they are observing.

The research is clear that physicians want to learn about rare diseases relevant to their specialties. They prefer short, case-based, clinically actionable content delivered through trusted channels. They are motivated by the puzzling patient, by professional pride, and by the emotional weight of diagnostic uncertainty. And they filter everything through the lens of specialty relevance.

Raising the baseline of rare disease awareness across medical specialties is not a matter of asking every physician to memorize thousands of conditions. It is a matter of consistent, specialty-specific education that keeps rare diseases visible in the differential diagnosis when standard explanations fail. This is achievable. It is evidence-based. And for the patients who are waiting — sometimes for years — for someone to think of the right question to ask, it is urgent.

References

1. Faye F, Crocione C, de Pena RA, et al. Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. *European Journal of Human Genetics.* 2024.
2. Vandeborne L, van Overbeeke E, Dooms M, et al. Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in Belgium. *Orphanet Journal of Rare Diseases.* 2019;14:99.
3. Marwaha S, Engel PA, Engel JA, et al. Educational needs in diagnosing rare diseases: A multinational, multispecialty clinician survey. *Genetics in Medicine Open.* 2023.

4. Walkowiak D, Domaradzki J. Are rare diseases overlooked by medical education? Awareness of rare diseases among physicians in Poland. *Orphanet Journal of Rare Diseases*. 2021;16:400.
5. Benito-Lozano J, Arias-Merino G, Gomez-Martinez M, et al. Diagnostic Process in Rare Diseases: Determinants Associated With Diagnostic Delay. *International Journal of Environmental Research and Public Health*. 2022;19(11):6456.
6. Busbridge C, Adams E, Howard A, et al. A quantitative and qualitative analysis of patient group narratives suggests common biopsychosocial red flags of undiagnosed rare disease. *Orphanet Journal of Rare Diseases*. 2024;19:166.
7. Brando JL, Sohail AM, Nweke E, et al. Rare disease education in medical schools: patient-centered and innovative strategies. *Orphanet Journal of Rare Diseases*. 2025;20:60.
8. Amiculum. Enhancing the patient-HCP relationship in rare diseases. 2023.

Rare Expertise

Rare Expertise is a strategic consultancy focused on helping companies developing and marketing products for patients with rare diseases. Our mission is to shorten the diagnostic and treatment journey in people with rare diseases through better education.

Rare Expertise and the Rare Medical Network work at the intersection of rare disease knowledge, clinical practice, and trusted professional networks. Our focus is on supporting healthcare professionals with credible information and access to expertise — when it matters most.

FOUNDED

Rare Expertise was founded in 2015 by Jack Davis and Jeff Sweeney, who are parents of children with rare diseases, and who both have extensive professional experience in marketing communications and medical education in rare disease markets.