Rare Expertise

WHITE PAPER

Why HCPs Seek Rare Disease Information

Triggers, Uncertainty, and Curiosity

A Rare Expertise White Paper · #3 in a series

For rare disease marketing and medical affairs professionals

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SECTION 01

Executive Summary

Healthcare professionals do not seek rare disease information out of academic curiosity. Information-seeking is most often triggered by moments of uncertainty, clinical failure, or perceived risk — to the patient, to outcomes, and to the clinician's own professional responsibility. Yet HCPs increasingly recognize that rare disease is not a niche concern: an estimated 10% of the population is likely to have a disease that qualifies as rare.

Rare diseases represent a stress test for traditional medical knowledge pathways. Low prevalence, heterogeneous presentations, limited evidence, and fragmented expertise create situations where standard diagnostic and treatment frameworks fall short. In these moments, HCPs are forced into just-in-time learning under pressure.

> RARE DISEASE REALITY CHECK
>
> **Rare diseases do not fail because clinicians lack intelligence or diligence. They fail because medical systems are optimized for what is common — not for what is rare, evolving, or atypical.**

This white paper examines the clinical triggers, emotional drivers, and professional risks that motivate the search for rare disease information — and explains why rare disease information-seeking differs fundamentally from routine medical education. Understanding these motivations is foundational to improving how rare disease information is created, delivered, and trusted.

SECTION 02

Rare Disease as a Knowledge Stress Test

Modern medicine is built on pattern recognition, guidelines, and probability. Most clinical encounters rely on conditions that are common, well-characterized, and supported by robust evidence. Rare diseases disrupt this model.

By definition, rare diseases sit outside the boundaries of routine clinical exposure. Many HCPs encounter only a handful — if any — cases during training or practice. Symptoms are often nonspecific, multi-system, or progressive. Diagnostic criteria may be evolving, and treatment options limited or unfamiliar.

As a result, rare diseases expose a fundamental tension in clinical practice: clinicians are expected to deliver confident, evidence-based care in situations where evidence is sparse and experience is limited. In this context, information-seeking is not a background activity. It becomes an urgent, situational response to uncertainty.

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It's not that you immediately think 'rare disease.'
It's that nothing else makes sense anymore.

— HCP interview

SECTION 03

The Moment of Motivation: What Triggers the Search

Rare disease information-seeking almost always begins with a trigger. These triggers are rarely abstract; they arise directly from patient care.

KEY INSIGHT

Rare disease information-seeking is not proactive learning. It is a reaction to clinical friction — when standard pathways stop working.

Diagnostic Uncertainty

The most common catalyst is a case that does not fit expected patterns. Symptoms persist without explanation. Test results conflict or remain inconclusive. The clinical picture evolves in ways that challenge initial assumptions. HCPs often describe this moment simply as: "Something isn't adding up." This realization prompts a shift from routine problem-solving to broader hypothesis generation. Rare disease enters consideration not because it is likely, but because common explanations have failed.

> *You reach a point where you've ruled out the usual things, but the patient is still getting worse. That's when you start thinking, "Okay — what am I missing?"*
>
> — HCP interview

Clinical Failure

Information-seeking is also triggered when established treatments do not produce expected results. Patients fail to respond, deteriorate unexpectedly, or relapse despite adherence to standard protocols. In these cases, rare disease information-seeking is motivated by the need to reassess foundational assumptions.

> *When a treatment that should work doesn't, you start questioning the diagnosis — not the drug.*
>
> — HCP interview

Patient-Initiated Triggers

Increasingly, patients and caregivers play an active role in initiating rare disease searches. Many arrive with extensive information gathered from online communities, advocacy groups, or social media. When a patient raises a rare disease by name — accurately or not — it creates immediate pressure for the HCP to evaluate the possibility. This dynamic shifts information-seeking from optional to obligatory.

> *Patients come in having read everything. Even if they're wrong, you can't ignore it — you have to check.*
>
> — HCP interview

Referral Pressure and Scope of Care

Rare disease suspicion also forces HCPs to confront questions of responsibility: should this patient be managed locally or referred to a specialist? If so, when? Information-seeking helps HCPs determine whether they are at the limits of their expertise — and whether delayed referral

could have consequences.

SECTION 04

Emotional and Professional Drivers Behind the Search

Clinical triggers initiate rare disease information-seeking, but emotional and professional factors often sustain it. In rare disease, uncertainty is not just clinical — it is personal, professional, and reputational.

Fear of Diagnostic Delay

Many HCPs are acutely aware of the "diagnostic odyssey" experienced by rare disease patients. Delayed diagnosis can mean irreversible progression, lost treatment windows, and significant patient harm. This awareness creates a moral imperative: if this could be rare, I need to know sooner rather than later.

> *You don't want to be the doctor who missed something obvious — especially when the stakes are high.*
>
> — HCP interview

Professional Identity and Reputation

Medicine is deeply tied to professional identity. Not knowing is uncomfortable; admitting uncertainty can feel risky. Rare diseases challenge the expectation that clinicians should have answers — or at least a clear next step. Information-seeking allows HCPs to preserve confidence, both internally and in the eyes of patients and peers.

> *There's definitely a sense of, "I should know this," even when realistically, no one does.*
>
> — HCP interview

Cognitive Dissonance

Rare disease cases often conflict with an HCP's training and experience. There is a psychological gap between what clinicians believe they should know and what the situation demands. Information-seeking becomes a way to reconcile professional expectations with clinical reality.

SECTION 05

The Risk Lens: Why Rare Disease Feels Different

Rare disease information-seeking is shaped by heightened perceptions of risk. Unlike common conditions, rare diseases offer fewer guardrails. Decisions are often made without precedent. In this environment, information-seeking serves a critical function: risk mitigation.

● WHY RARE DISEASE FEELS RISKIER

Fewer guidelines · Limited evidence · Unclear referral thresholds · High consequences of delay

When guardrails disappear, information-seeking becomes a form of protection.

> *You're always thinking about how this will look six months from now if the patient doesn't do well.*
>
> — HCP interview

SECTION 06

Why Traditional Medical Education Falls Short

Most medical education is designed for prevalence, not exception. Rare diseases receive limited attention in training curricula. Continuing medical education is typically structured around common conditions or new therapies, not atypical presentations.

EDUCATION VS. REALITY

HCPs are not failing to remember rare disease information. They were never taught to access it at the moment they need it.

> *You usually learn about rare diseases when you're already dealing with a real patient. It would be good to have a bit more knowledge about them so we are prepared when we see a symptom complex that doesn't make sense.*
>
> — HCP interview

Historically, medical education is longitudinal; rare disease information-seeking is situational. But unless healthcare professionals become more sensitive to the possibility that a patient may have a rare disease — and consider it a real possibility — patients will continue to wait far too long for the right diagnosis.

SECTION 07

Implications for Information Providers

Understanding why HCPs seek rare disease information has direct implications for those who aim to support them. For the average clinician, rare disease information needs to be made easily accessible and engaging, so that they can quickly absorb and retain information that may not be needed immediately — but may be valuable in the future.

When a potential "moment of rare disease need" arrives, the information needs to be structured for rapid orientation and grounded in credible expertise. Most importantly, it must recognize that rare disease information-seeking is not an academic exercise — it is a response to clinical pressure and responsibility. Ease of comprehension, connection to more in-depth information, and trust in where to go for deeper expertise are all critical.

SECTION 08

Key Takeaways

Understanding why — not just what — HCPs search for is foundational to improving rare disease education and support.

1. Raising the average knowledge level about rare diseases among practicing clinicians is the foundation of improving time-to-diagnosis for patients with rare diseases.
2. HCPs are motivated by uncertainty and responsibility, not academic curiosity.
3. Emotional and cognitive factors are as influential as clinical ones in driving information-seeking behavior.
4. Traditional education models do not meet situational rare disease needs.
5. Supporting HCPs requires understanding why they search — not just what they search for.

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.