

Rare Expertise

• WHITE PAPER

# Adult Learning Theory in **Rare Disease** Education

Applying Andragogy Principles to Close the  
**Rare Disease**  
Knowledge Gap Among Healthcare Professionals

A Rare Expertise White Paper

For rare disease marketing and medical affairs professionals

[rareexpertise.com](http://rareexpertise.com)

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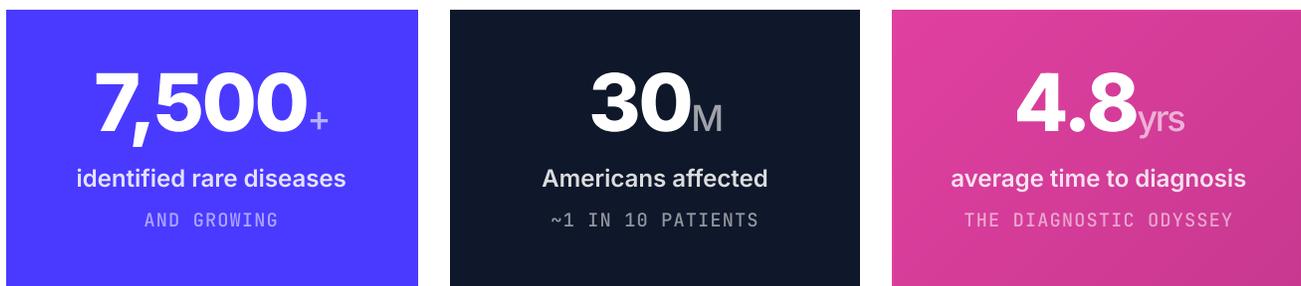
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## Executive Summary

Adult learning theory provides a powerful framework for designing medical education programs that address the rare disease knowledge gap. By understanding how adult learners — particularly healthcare professionals — acquire, retain, and apply new knowledge, educators can create more effective and engaging programs that improve rare disease awareness and diagnostic outcomes.

The principles of andragogy, pioneered by Malcolm Knowles, emphasize that adult learners are self-directed, experience-driven, and motivated by relevance and practical application. When applied to rare disease education, these principles point toward learning experiences that are clinician-centered, problem-based, and embedded in the daily workflows of practicing physicians.



This white paper examines why traditional approaches to rare disease education have fallen short, introduces the core principles of adult learning theory, and demonstrates how applying andragogy to medical education can help close the rare disease knowledge gap — ultimately shortening the diagnostic odyssey for millions of patients.

# 1 in 10

patients seen by the average US physician may have a **rare disease** — but most physicians have not been trained to consider that possibility.

GLOBAL GENES · RARE DISEASE FACTS

## The Need for Better **Rare Disease** Education

There are at least 7,500 identified rare diseases, affecting approximately 30 million people in the United States. That means roughly 1 in 10 patients seen by the average physician may have a rare disease. Yet the average physician has not been trained to consider this possibility.

In medical school and residency, physicians are rightly taught to first consider common diagnoses. The well-known clinical adage — "when you hear hoofbeats, think of horses, not zebras" — reflects a logical approach to everyday practice. But this training, reinforced by years of clinical experience, creates a systematic blind spot: physicians consistently default to common conditions even when the clinical picture does not fully fit.

### ● KEY FINDING

A December 2021 survey by Definitive Healthcare found that healthcare providers believe the lack of rare disease education for physicians (40%) and lack of awareness of symptoms related to rare diseases (39.3%) are the greatest challenges they face around rare diseases today.

## The Knowledge Gap Is Wide — and Acknowledged

The rare disease knowledge gap is not just a problem identified by researchers. Healthcare professionals themselves recognize it. A 2019 survey by NORD found that 50% of patients and caregivers attributed diagnostic delays to a lack of HCP disease awareness. And the physicians agree: research indicates that most physicians — more than 80% of specialists and more than 60% of primary care physicians — want to learn more about rare diseases.

**80%+**

of specialists want to learn more about rare diseases

**50%**

of patients attribute diagnostic delays to lack of HCP awareness

## **Beyond Awareness: The Complexity Challenge**

But there are more than 7,500 identified rare diseases, and perhaps as many as 10,000. Genetic and phenotypic variability add to the incomplete knowledge of rare diseases, which complicates the process of diagnosis. No healthcare professional could be expected to have the knowledge level needed to consider and accurately diagnose every rare disease that has been identified.

Physicians may also not be aware of what tests to order — and when. The rapid technological advancements in genomics, sequencing, and genetic testing make it virtually impossible for the average clinician to stay up-to-date on the availability and potential value of genetic tests for hundreds of diseases. Even when clinicians consider the option of genetic testing, most will refer patients to a medical geneticist — a specialty that currently faces extremely high demand, with typical wait times of more than 3 months for appointments and another 2-3 months to receive test results.

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*The result is that, on average, the journey that rare disease patients and caregivers go through to get an accurate diagnosis takes 4-8 years. This "diagnostic odyssey" is a hallmark of the rare disease patient and caregiver experience, and clear evidence that there is a gap in rare disease education that must be addressed.*



When you hear hoofbeats, think of horses, not zebras — but what happens when the patient *is* the zebra, and no one has been taught to look?

# Malcolm Knowles' Five Principles of Andragogy

Those teaching adults should understand and practice the five principles of andragogy espoused by Malcolm Knowles, a pioneer in the study of adult learning. Knowles posited that adults learn best under specific circumstances — and these principles have profound implications for how rare disease education should be designed and delivered.

## ● THE FIVE PRINCIPLES

Knowles identified five conditions under which adults learn most effectively: when learning is self-directed, experiential, relevant to current roles, problem-centered, and driven by intrinsic motivation. Each principle maps directly to the challenges and opportunities of rare disease medical education.

## 1. Self-Directed Learning

One of the most important differences between teaching children and teaching adults is the self-concept of adult learners. Adult learners are usually mature and self-confident enough to know how they learn best, what their areas of strength and weakness are, and how to go about learning. They do not require much help acquiring resources or developing goals because, in most cases, they have done this before.

Adult educators need to grant their students plenty of space and be there to support rather than guide. Another benefit of self-directed learning is that students can design their studies around their preferred learning style — visual, auditory, or kinesthetic — allowing them to absorb and retain information in ways that work best for them individually.

## 2. Experiential Learning

Adult educators need to utilize each learner's background knowledge as a resource. No matter how old adult learners are or what type of professional life they have led, every learner will have acquired an extensive cache of experiences that can be drawn on to enrich instruction.

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*Rather than behaving as if the classroom should be a level playing field and ignoring irregular stores of background knowledge, use them to enrich instruction. The moments of authenticity and spontaneity that come from sharing experiences will prove to be some of the most powerful.*

### **3. Relevance to Current Roles**

Adult students are most likely to want to learn about subjects that will have immediate payoffs in their life, especially as it pertains to their professional roles. Adults have little use for material that is not relevant to the roles they already occupy — and this is another reason for allowing students to play a part in designing their own curriculum.

The job of adult educators is to get to know students well enough to be able to teach to their roles. The goal of adult education is to fit the needs of students, who are more often than not opting to be there because they identified an area of need for themselves.

### **4. Problem-Centered Instruction**

Adult learners do not desire to learn about material that does not fit into their lives, and they do not usually want their learning to be abstract. Adults are practiced, knowledgeable, and flexible learners with a lot of problems to solve. Unlike young students, they do not usually need long to think about unfamiliar subjects before trying a skill out for themselves because they exercise their problem-solving skills every day.

Adult educators need to tailor instruction to specific problems that students face rather than approaching teaching one subject at a time. Andragogy is about spending more time doing than learning, and the quality of instruction is much more important than topic coverage.

## **5. Motivation to Learn**

"When the student is ready, the teacher appears" is a proverb that applies well to all areas of education. No matter how hard a teacher tries, learning only begins once a student is ready. Many adult educators find that their students are eager to grow their knowledge — adults who have chosen to continue their education are probably already motivated to learn, or would not have made the choice to continue.

The educator's role is to encourage this motivation and help students maintain positivity toward learning. Listen carefully for teaching moments and take advantage of them — when a student says or does something that cues a new topic, be flexible and discuss it, even briefly, to show that their interests are important.

# Knowles' Five Principles Applied to Medical Education

- 1 Self-directed learning** — clinicians choose what and how they learn based on their own identified gaps
- 2 Experiential foundation** — prior clinical experience serves as the basis for integrating new knowledge
- 3 Role relevance** — education must connect directly to the clinician's specialty and daily practice
- 4 Problem-centered design** — learning organized around clinical puzzles, not abstract disease taxonomy
- 5 Intrinsic motivation** — the desire to solve diagnostic mysteries and help patients drives engagement

# Applying Andragogy to **Rare Disease** Education

The five principles of adult learning theory map remarkably well to the unique challenges of rare disease education. When applied thoughtfully, andragogy transforms rare disease education from a one-size-fits-all lecture into a clinician-centered, problem-driven experience that produces lasting changes in diagnostic behavior.

## Understanding the Learner

Medical educators can use adult learning theory to understand the needs and characteristics of adult learners. Healthcare professionals are typically self-directed and motivated, with specific learning goals and extensive prior clinical knowledge. Effective rare disease education begins by recognizing these characteristics and designing programs that respect the learner's autonomy and expertise.

### ● LEARNER-CENTERED DESIGN

A key principle of adult learning theory is that it is critical to understand the learning needs and goals of each individual and tailor education accordingly. For rare disease, this means specialty-specific content delivered through channels clinicians already trust.

### ● ACTIVE ENGAGEMENT

Medical educators can incorporate active engagement strategies — discussion, problem-solving, and hands-on case analysis — into their teaching practices. These strategies promote active learning and enhance retention of rare disease knowledge.

## Developing Relevant and Practical Content

Another key tenet of adult learning theory is the importance of providing relevant and practical content that is applicable to the learner's real-world experiences. For rare disease education, this means presenting information in the context of clinical scenarios that physicians actually encounter — not abstract disease descriptions disconnected from daily practice.

This approach helps to promote the transfer of learning from the educational setting to real-world clinical decision-making. When a neurologist learns about a rare neuromuscular condition through a case that mirrors a patient they might see next week, the knowledge becomes immediately actionable.

- FEEDBACK AND REFLECTION

Medical educators can use adult learning theory to incorporate feedback and reflection into their teaching methods. This approach helps learners recognize their progress, identify areas for improvement, and build confidence in their ability to consider rare disease diagnoses. Interactive case-based assessments and peer discussion are particularly effective mechanisms.

## The Power of Case-Based Learning

Case-based learning sits at the intersection of multiple andragogy principles. It is problem-centered, experiential, relevant to clinical roles, and engages the learner's intrinsic motivation to solve diagnostic puzzles. When a physician works through a complex case and arrives at a rare disease diagnosis, the learning is memorable in a way that reading a disease monograph rarely achieves.

This is particularly powerful in rare disease, where a single memorable case discussion can permanently alter a specialist's index of suspicion. The challenge has historically been that such interactions are sporadic and unscalable — unless they are deliberately facilitated through educational platforms designed with andragogy principles in mind.

### SECTION 05

## The Emotional Dimension of Rare Disease Diagnosis

A dimension of the rare disease diagnostic challenge that is often overlooked is the frustration and emotional impact on healthcare professionals. Trained to identify and solve problems and empathetic to the suffering of patients and families, HCPs are often as frustrated by the mysteries of rare diseases as patients and families themselves.

Diagnosing rare pediatric diseases can be especially difficult, complex, and fraught with sensitive emotional issues. The age of many patients, the uncertainty surrounding the diagnosis, the fear of disease progression, and the potential lack of treatments all contribute to an extremely stressful environment for healthcare professionals.

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*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 learning.*

For some physicians, the frustration of repeated diagnostic failure leads to disengagement — the patient is referred elsewhere, or symptoms are attributed to psychosomatic causes. For others, it creates a powerful drive to learn more. Effective rare disease education, grounded in adult learning theory, should acknowledge this emotional reality rather than treating rare disease diagnosis as a purely intellectual exercise.



## Motivation Through Mastery

Adult learning theory tells us that motivation to learn is strongest when learners recognize a gap in their own knowledge and believe that closing it will improve their professional performance. For healthcare professionals, the emotional weight of undiagnosed patients is precisely this kind of motivator — a deeply felt recognition that they need tools they do not yet possess.

Education designed for this audience must be clinically compelling, not merely accredited. It must speak to the physician's identity as a problem-solver and diagnostician, offering practical pathways from suspicion to diagnosis rather than abstract disease taxonomies.



Often neither the doctor nor the patient knows it.  
The challenge is not that rare disease patients  
are absent from clinical practice — ***it is that  
they are going unrecognized.***

## Closing the **Rare Disease** Knowledge Gap

The rare disease knowledge gap refers to the lack of information and understanding about rare diseases among physicians, researchers, and the general public. Despite their "rarity," rare diseases collectively affect tens of millions of people, and many are life-threatening or severely debilitating. Closing this gap requires a strategic approach grounded in adult learning principles.

### **Recognition: Consider the Possibility**

The first and most critical missing element is recognition — the awareness that some patients may actually have a rare disease. Adult learning theory suggests that this recognition is best built through repeated, specialty-relevant exposure that creates cognitive "bookmarks" activating when a matching patient presents.

### **Awareness: Know the Clues**

Beyond simple recognition, physicians need awareness and knowledge of the clues that can help uncover a rare disease diagnosis. Problem-centered instruction, a core andragogy principle, positions these clues within clinical scenarios rather than abstract lists — making them immediately memorable and actionable.

### **Education: Accelerate Diagnosis**

Specific education about symptoms and diagnostic pathways that can lead to faster diagnosis completes the framework. By leveraging experiential learning and the clinician's existing knowledge base, rare disease education can build on what physicians already know rather than starting from scratch.

## **From Theory to Practice: Design Principles**

Translating adult learning theory into effective rare disease education programs requires attention to several design principles that emerge directly from andragogy research.

- CONTENT DESIGN

Case-based, specialty-specific content that respects the learner's existing expertise and connects new rare disease knowledge to familiar clinical patterns. Short-format content (15 minutes or less) fits the rhythms of clinical practice.

- DELIVERY STRATEGY

Sustained, regular delivery through specialty-specific channels builds ambient awareness over time. One-time educational interventions have limited impact — consistent exposure changes clinical behavior and diagnostic thinking.

The peer-to-peer dynamic is particularly powerful in rare disease 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 — a principle that aligns with the andragogy emphasis on learning as a social, experiential activity.

SECTION 07

## Conclusion

Adult learning theory provides a valuable and underutilized framework for addressing the rare disease knowledge gap. By designing educational programs that are self-directed, experiential, relevant, problem-centered, and intrinsically motivating, educators can create learning experiences that produce meaningful and lasting changes in how healthcare professionals think about rare diseases.

Doctors and other healthcare professionals have committed their lives to doing everything they can to improve the lives of their patients — and research indicates that they want to do more to help rare disease patients. But training and education about rare diseases has rarely been a priority of the healthcare system. The world has changed: more rare diseases are increasingly being identified and diagnosed, but there is a large gap between the knowledge that exists in academic medicine and everyday clinical practice.

Closing the gap can make a major contribution to improving the lives of millions. Better outcomes are possible with a relatively small investment in rare disease education — if that education is designed according to the principles of how adults actually learn.

● THE PATH FORWARD

The key is not to transform every physician into a rare disease expert. It is to apply andragogy principles — self-direction, experiential learning, role relevance, problem-centered design, and intrinsic motivation — to create specialty-specific education that keeps rare diseases visible in the differential diagnosis when standard explanations fail. This is achievable, evidence-based, and for the patients who are waiting, it is urgent.

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# 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.