Rare Expertise

● WHITE PAPER

Content Marketing and Education for Rare Diseases

A Strategic Approach to Educating Patients, Caregivers, and Healthcare Professionals in the Rare Disease Market

A Rare Expertise White Paper

For rare disease marketing and medical affairs professionals

rareexpertise.com

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SECTION 01

Summary

Content marketing is a strategic approach to creating and distributing educational content that is designed to attract and engage a specific target audience. The goal of content marketing is to establish a relationship with the audience and build trust through the provision of useful and relevant information.

In the rare disease segment of the biopharma industry, content marketing can be used to educate and inform patients, healthcare providers, and other stakeholders about various topics including symptoms, diagnosis, testing, disease management, and clinical trials. Content marketing uses various communication channels, including videos, e-books, infographics, blog posts, newsletters, and social media updates. Because there is a significant gap in rare disease education, content marketing programs are an excellent way to improve the knowledge of rare disease patients, caregivers, and healthcare professionals.

7,500+
identified rare diseases
AND GROWING

30M
Americans affected
~1 IN 10 PATIENTS

5%
of rare diseases have
FDA-APPROVED THERAPIES

Overall, content marketing can be a valuable tool for the pharmaceutical industry to educate and inform patients, healthcare providers, and other stakeholders about various topics related to the industry. By providing valuable content that is relevant and useful, pharmaceutical companies can build trust and establish themselves as thought leaders in their respective areas of expertise.

1 in 10

patients seen by the average physician in the United States may have a rare disease.

GLOBAL GENES · RARE DISEASE FACTS

SECTION 02

The Need for Better Rare Disease Education

There are at least 7,500 identified rare diseases, affecting about 30 million people in the United States. That means that approximately 1 in 10 patients seen by the average physician in the United States may have a rare disease. But the average physician hasn't been trained to consider the possibility that many of their patients may have an unknown rare disease.

In medical school and residency, physicians are rightly taught that they should first consider a common diagnosis, not a rare one. Consequently, although most physicians will have to diagnose or manage patients with a rare disease at some point in their professional lives, many assume that they are unlikely to ever see a rare disease patient in their practice.

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

In addition, a 2019 survey by NORD found that 50% of patients and caregivers attributed diagnostic delays to a lack of HCP disease awareness. A majority of physicians indicated that they need to know more about rare disease. Research indicates that most physicians — more than 80% of specialists and more than 60% of PCPs — want to learn more about rare diseases.

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> *For decades, medical students and residents have been taught the phrase "when you hear hoofbeats, think of horses, not zebras" — rightly indicating that the symptoms that appear most commonly in clinical practice are usually the result of common conditions, not rare syndromes.*

That training is reinforced by years of clinical practice, which provides healthcare professionals with consistent reinforcement that most sets of symptoms they see — and most test results — are indicative of common conditions. The result is that physicians are often the last to consider a rare diagnosis, even when the evidence is mounting.

SECTION 03

The Knowledge Gap: What Physicians Don't Know

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.

40%

cite lack of rare disease education for physicians as greatest challenge

39%

cite lack of awareness of symptoms as the greatest challenge

The Testing Challenge

Physicians may also not be aware of what tests to order for patients — and when. The rapid technological advancements in genomics, sequencing, and genetic testing in this field 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.

> ● WAIT TIMES
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> 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 for appointments of more than 3 months, and another 2-3 months to receive test results.

The Symptom Overlap Problem

Because the number of people affected with an individual disease is relatively small and the number of rare diseases is so large, the average healthcare professional is unlikely to consider that an individual patient may have a rare disease. The challenge of diagnosing rare disease patients is further complicated by the fact that so many of the symptoms that rare disease patients exhibit overlap with symptoms that are similar to many of the common conditions that physicians see every day.

> ● STRATEGIC IMPLICATION
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> This is where content marketing becomes essential: well-designed educational content can help bridge the knowledge gap by presenting rare disease information in formats that are accessible, specialty-relevant, and clinically actionable for healthcare professionals.

> Often neither the doctor nor the patient knows it. The average healthcare professional is unlikely to consider that an individual patient may have a rare disease — ***because so many rare disease symptoms overlap with common conditions physicians see every day.***

SECTION 04

The Diagnostic Odyssey and Its Human Cost

The result of low rare disease awareness is that, on average, the journey that rare disease patients and caregivers go through to get an accurate diagnosis takes 4-8 years. The average time it takes for rare disease patients to receive an accurate diagnosis is 4.8 years. This "diagnostic odyssey" is a hallmark of the rare disease patient and caregiver experience.

4.8yrs

average time to diagnosis

RARE DISEASE PATIENTS

50%

of patients cite lack of

HCP DISEASE AWARENESS

80%+

of specialists want to

LEARN MORE ABOUT RARE DISEASES

This diagnostic odyssey is clear evidence that there is a gap in rare disease education that must be addressed if the diagnostic journey is to be improved. Low rare disease awareness and knowledge among HCPs is one of the most important reasons why many rare disease patients face late and numerous misdiagnoses.

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> *The diagnostic odyssey is not merely an inconvenience — it carries profound medical, psychological, and financial consequences for patients and families who spend years searching for answers that their healthcare providers are not equipped to find.*

Content marketing and educational programs represent the most scalable and effective approach to addressing this gap. By systematically delivering rare disease education to healthcare professionals through the channels they already use and trust, pharmaceutical companies can meaningfully reduce diagnostic delays and improve patient outcomes.

CONTENT STRATEGY

Five Content Marketing Channels for Rare Disease Education

1. **Disease education materials** that help patients understand their condition and manage symptoms
2. **Infographics** that enable rapid communication of key information to busy HCPs about hundreds of rare diseases
3. **Product information content** including blog posts and social media updates about medications, administration, and patient expectations
4. **Clinical trial information** helping patients understand the potential benefits and risks of participating in research
5. **Video and multimedia content** including e-books, newsletters, and social media updates that engage diverse audiences

Source: Rare Expertise analysis of biopharma content marketing best practices

SECTION 05

Content Marketing Strategies for Rare Disease

Content marketing in the rare disease segment takes many forms, each serving a distinct purpose in the broader educational ecosystem. The most effective strategies combine multiple content types to reach patients, caregivers, and healthcare professionals through the channels they already use.

Disease Education

Creating educational materials that provide information about specific diseases can help patients better understand their condition and manage their symptoms. Disease education content should be clear, accessible, and medically accurate — serving as a trusted resource for patients navigating the complexity of a rare diagnosis.

Infographics for HCP Education

Infographics provide a channel for rapid communication of key information, which is critical when attempting to educate HCPs about hundreds of rare diseases. Visual formats allow physicians to quickly absorb differential diagnosis cues, symptom patterns, and testing pathways in a way that long-form text cannot achieve.

Product Information

Providing information about a particular medication, including its benefits and potential side effects, can help patients make informed decisions about their healthcare. A pharmaceutical company could create blog posts or social media updates that provide information about how a medication works, how it is administered, and what patients can expect when taking it.

Clinical Trial Information

Clinical trials are the cornerstone of the pharmaceutical industry, and providing information about these trials can help patients better understand the potential benefits and risks of participating. For rare disease patients who often have few or no treatment options, awareness of clinical trials can be life-changing.

● FOR HCPS

Focus on specialty-relevant content: differential diagnosis guides, red flag symptoms, testing algorithms, and case-based learning that fits into clinical workflow.

● FOR PATIENTS

Focus on empowerment: disease education, treatment options, clinical trial awareness, caregiver support resources, and community connection.

SECTION 06

The Emotional Dimension: Impact on Healthcare Professionals

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.

● OFTEN OVERLOOKED

Healthcare professionals experience their own form of the diagnostic odyssey — the frustration of recognizing that something is wrong with a patient but being unable to identify what it is. This emotional burden can lead to disengagement or, when supported by the right educational resources, a powerful drive to learn more.

This is where content marketing plays a critical dual role. Educational content not only helps HCPs build the clinical knowledge needed to recognize rare diseases, but it also provides emotional validation — confirming that the diagnostic challenges they face are real, shared, and addressable. When physicians see that their struggles with rare disease diagnosis are part of a broader, well-documented pattern, it normalizes the challenge and motivates further learning.

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> *Rare diseases represent a significant share of the research and development in the biopharma industry, which provides hope for many rare disease patients. Content marketing can help ensure that the advances being made in laboratories and clinical trials are effectively communicated to the healthcare professionals and patients who need them most.*

Effective content marketing programs acknowledge the emotional dimension of rare disease care. They address not only the clinical knowledge gaps but also the human experience of working at the intersection of uncertainty, urgency, and hope that defines rare disease medicine.

SECTION 07

Conclusion: Closing the Education Gap

The rare disease education gap is one of the most significant — and addressable — challenges in modern healthcare. With more than 7,500 identified rare diseases affecting 30 million Americans, and an average diagnostic journey of nearly five years, the need for better education has never been more urgent.

Content marketing offers a strategic, scalable approach to closing this gap. By creating and distributing educational content that is relevant, clinically useful, and tailored to specific audiences — whether patients, caregivers, or healthcare professionals — pharmaceutical companies can build trust, establish thought leadership, and meaningfully contribute to improved outcomes for rare disease patients.

● THE OPPORTUNITY

Because there is a significant gap in rare disease education, content marketing programs represent one of the highest-value investments a rare disease company can make. Every piece of well-designed educational content has the potential to shorten a patient's diagnostic journey, improve an HCP's clinical suspicion, or connect a caregiver with the resources they need.

The tools are available: videos, e-books, infographics, blog posts, newsletters, and social media. The audiences are receptive: more than 80% of specialists want to learn more about rare diseases. The need is undeniable: millions of patients are waiting for answers. What is required is a strategic, sustained commitment to content marketing that puts education at the center of rare disease commercial strategy.

By providing valuable content that is relevant and useful, pharmaceutical companies can build trust and establish themselves as thought leaders in their respective areas of expertise — and in doing so, make a tangible difference in the lives of rare disease patients and their families.

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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.