Rare Expertise

WHITE PAPER

Best Practices in Rare Disease Commercialization

Principles, Strategies, and Expert Insights for Marketing and Medical Affairs Professionals

A Rare Expertise White Paper

Based on interviews with rare disease commercialization leaders

rareexpertise.com

Contents

SECTION 01

Executive Summary

Rare disease commercialization stands apart from traditional pharmaceutical launches in ways that are both profound and practical. With approximately 7,000 identified rare diseases affecting an estimated 300 million people worldwide, these conditions represent an enormous collective unmet medical need — yet each individual disease community is small, often tightly knit, and demands a level of engagement and authenticity that standard commercial playbooks simply cannot deliver.

This white paper distills insights from experienced rare disease commercialization leaders — executives, medical affairs professionals, market access specialists, and patient advocacy experts — into a practical framework of ten best practices. These practices span the full arc of rare disease commercialization, from early patient journey mapping through post-launch persistence and real-world data collection.

7,000+
rare diseases
IDENTIFIED WORLDWIDE

300M
patients affected
GLOBAL RARE DISEASE POPULATION

10
best practices
EXPERT-DRIVEN FRAMEWORK

The central finding is clear: success in rare disease requires deeper relationships, earlier engagement, greater patience, and a more genuine commitment to patients than virtually any other area of the pharmaceutical industry. Companies that approach rare disease with standard commercial thinking — treating advocacy groups as marketing channels, underinvesting in patient support, or rushing timelines — consistently underperform. Those that earn the trust of rare disease communities through authentic partnership and sustained commitment are the ones that achieve both meaningful patient outcomes and commercial success.

20-30%

of ultra-rare disease patients may be connected to advocacy organizations — making these groups uniquely powerful partners in patient identification, education, and clinical development.

INSIGHTS FROM RARE DISEASE COMMERCIALIZATION LEADERS

SECTION 02

Overarching Principles for Rare Disease Launches

Before examining specific best practices, it is essential to understand the foundational principles that distinguish rare disease commercialization from more conventional pharmaceutical launches. While many of these principles echo familiar industry tenets — patient-centricity, stakeholder engagement, sound clinical strategy — their application in rare diseases is materially different in scope, depth, and urgency.

Principle 1: Patient-Centricity as the Core

Patient-centricity is the foundation of all rare disease research and commercialization. Unlike in large therapeutic areas where market dynamics can sometimes overshadow individual patient needs, rare disease work is inherently personal. The patients are identifiable, their stories are known, and the urgency of their conditions demands a responsiveness and agility that goes well beyond what typical pharmaceutical development requires.

> *In rare disease, the agility, the responsiveness, the urgency, the unencumbered way to solve the 'just do it' ness is huge. It's because people are dying. People are sick. You don't go into rare diseases for headaches. The only way a smaller population becomes an identified need is because it's so significant, severe, life-changing in an unfavorable way.*
>
> Rare Disease Industry Expert

Principle 2: Commitment to Each Rare Disease Community

Building meaningful relationships with rare disease advocacy groups is critical to both clinical development and commercial success. Companies must be prepared to engage with multiple advocacy organizations — and in diseases where no formal groups exist, they may need to help catalyze the formation of new ones. This commitment must be genuine, sustained, and visible.

> *I don't think it's about a lot of money. You have to show up. We go to walks; we go to meetings. The patients are there, the parents are there. Lots of times, the doctors are there.*
>
> Rare Disease Industry Expert

Principle 3: Deep Key Opinion Leader Relationships

In rare disease, relationships with key opinion leaders and centers of excellence run deeper than in conventional drug launches. KOLs are not simply endorsers or speakers — they are partners in clinical development, trial design, and patient identification. Building these relationships requires clinical credibility, mutual respect, and a long-term perspective.

> *There needs to be some KOL advocacy as well. Partnering, collaborating, or developing KOLs so that our voice can be amplified from a legitimized, credible source out in the community is really important.*
>
> Rare Disease Industry Expert

Principle 4: Managing Expectations Internally and Externally

Clinical and commercial success in rare disease typically takes longer than anticipated. Internally, leadership teams and investors must understand that timelines are inherently less predictable. Externally, patients and families who have learned to manage their conditions may be cautious about new therapies and reluctant to accept unfamiliar risks. Honest, transparent communication — about what a product can and cannot do — is essential at every level.

> *You have to manage clinical expectations. Parents are looking for a cure, and we have a good drug, but it's not a cure. Then you have to manage the leaders of the company, who need to convince the street that we have a revenue-generating drug.*
>
> Rare Disease Industry Expert

Principle 5: Starting Early and Investing in Understanding

Each rare disease and its patient population is unique, which means that building the necessary relationships, designing appropriate clinical trials, recruiting patients, identifying key experts, and gaining a deep understanding of the patient experience all take longer than expected. Companies that invest early — particularly in understanding the lived patient experience — position themselves for significantly stronger outcomes.

KEY INSIGHT

Investment in a deep understanding of a rare disease, especially the patient experience, must be a priority from early on in development. Companies that wait until Phase III or later to engage with patient communities find themselves at a significant disadvantage.

In rare disease, trust is the most valuable asset a company can build. Companies that earn that trust — through transparent community engagement, authentic KOL relationships, and patient-centered support — achieve both meaningful patient outcomes and commercial success.

SECTION 03

Ten Best Practices in Rare Disease Commercialization

The following ten best practices represent a comprehensive framework for rare disease commercialization. Each is grounded in the overarching principles described above and is supported by direct insights from experienced industry professionals. Together, they provide a roadmap for marketing and medical affairs teams preparing for or currently managing rare disease programs.

1. Patient Journey Mapping

The patient journey is the foundational element of rare disease market analysis. Every insight gathered for market and customer understanding should relate back to the lived experience of patients and their caregivers. In rare disease, this experience is often profoundly stressful and emotional — patients frequently endure years of misdiagnosis, multiple specialist visits, and significant psychological burden before arriving at a correct diagnosis and appropriate treatment.

Companies that want to succeed in rare diseases must go beyond standard qualitative research. They need to engage patients directly — through advocacy organizations, qualitative interviews, patient questionnaires, and ongoing dialogue — to develop an authentic understanding of how the disease affects daily life. Patients are experts in their own conditions, and companies that fail to meet them where they are will lose ground to competitors who do.

> *The experience of rare disease patients and caregivers is often very stressful. And emotional. Companies that want to work in rare diseases need to understand how the struggle of coping with rare disease impacts everyday life.*
>
> Rare Disease Industry Expert

> *Patients understand; patients know their disease. And if we as an industry don't meet them where they are, they've got choices, and if they have a choice, they'll go with somebody that wants to engage them.*
>
> Rare Disease Industry Expert

2. Commitment to the Rare Disease Community

Authentic community engagement is a hallmark of successful rare disease companies. This means building relationships with advocacy groups — sometimes one key organization, sometimes several — on the basis of mutual respect rather than transactional marketing. In ultra-rare diseases, a significant percentage of the total patient population (often 20-30%) may be connected to advocacy organizations, making these groups uniquely powerful partners in patient identification, education, and clinical development.

20-30%

of ultra-rare patients

CONNECTED TO ADVOCACY GROUPS

Trust

most valuable asset

IN RARE DISEASE COMMERCIALIZATION

Earning the trust of advocacy communities takes time, particularly given the skepticism many advocacy leaders feel toward pharmaceutical companies. Companies must be transparent about why they are engaging, what they hope to gain, and how they will use patient information.

> *You have to be very honest with them and upfront about why you're engaging them, what you're hoping to get out of the engagement and the experience, and then how you're going to use their information.*
>
> Rare Disease Industry Expert

REALITY CHECK

You'd be surprised at how long it takes certain people in the advocacy world to sort of warm up to that because you're pharma, you're bad, you're just trying to make money. Earning trust requires patience, transparency, and genuine commitment over time.

3. Patient Identification and Activation

Identifying and activating patients in rare diseases presents unique challenges. There are no "typical patients" — the variability of the patient journey means each individual's path to diagnosis and treatment is different. Advocacy organizations are often the most efficient channel for reaching patients, and building relationships with advocacy leaders should be an early priority.

Data challenges compound the difficulty. Most commercial data warehouses are not designed to capture rare disease patient data effectively, and claims datasets are coded primarily for billing purposes rather than outcomes research. Patient activation also requires nuance: while an initial wave of active seekers may be eager for treatment at approval, reaching the broader population — particularly older patients who have adapted to their condition — demands more creative engagement strategies.

> *As the patient gets older, they become less likely to initiate a new therapy unless something really happens. You're going to get a big group when you're approved, so those are the active seekers — then you're going to have to go after the rest.*
>
> Rare Disease Industry Expert

4. KOL Involvement and Centers of Excellence

Key opinion leaders should be engaged as early as possible — ideally from the moment a company begins thinking about protocol design or IND submission. KOLs bring invaluable understanding of the disease, its impact on patients, and the unmet needs that a new therapy must address. They are also critical partners in clinical trial design and patient recruitment.

The relationship between a rare disease company and its KOLs goes beyond conventional thought leader management. It requires gaining genuine respect from clinicians deeply invested in their patient communities. Medical Science Liaison teams play a particularly important bridging role, requiring a blend of scientific credibility and relationship-building skill. Centers of Excellence, while essential to clinical trials, vary significantly in trial execution experience and may need additional support.

> *I can't stress enough the importance of that relationship. Building relationships with the advocacy groups. Building relationships with the clinicians and KOLs. And actually, I think the most important thing is not just building a relationship. It's gaining the respect of those different groups.*
>
> Rare Disease Industry Expert

MSL STRATEGY

That whole thought leader development thing is a skill that is really the head of your MSL team that will pass it over to the sales force. But being able to have a medical affairs department that's half sales in their brain, half medical affairs is important.

5. Clinical Trial Design and Long-Term Expansion

Clinical trial design in rare diseases requires careful balancing of scientific rigor with practical constraints. Because patient populations are small, a high proportion of rare disease trials are multicenter and multinational. Identifying the most appropriate primary endpoint and target population for a pivotal trial is crucial for regulatory approval.

Advocacy organizations often play a decisive role in patient enrollment, sometimes producing more enrolled patients than clinical centers themselves. A critical and sometimes overlooked element of trial design is the collection of patient-reported outcomes. Many treatment benefits are not fully captured through laboratory tests, imaging, or biomedical measurements — patient-reported outcomes provide the evidence needed to demonstrate meaningful improvements in quality of life.

> *The centers are key to our clinical trials, but advocacy probably produces the most enrolled patients.*
>
> Rare Disease Industry Expert

CRITICAL DESIGN ELEMENT

It is critical to collect patient-reported outcomes as part of clinical trials in rare diseases. Many treatment outcomes are not evident through laboratory tests, imaging, or other biomedical measurements. These outcomes provide the real-world evidence needed for both regulatory approval and payer negotiations.

"

We considered outsourcing our patient call center, but we wanted people who 'lived it.' Keeping the call center inside was one of the smartest decisions the company made. Our people would spend an hour or more on the phone crying with parents and listening to their stories. ***An outside company wouldn't have done that.***

RARE DISEASE INDUSTRY EXPERT

SECTION 03 (CONTINUED)

Ten Best Practices *(continued)*

6. Organizational Structure and Team Composition

The structure and composition of rare disease teams differ meaningfully from traditional pharmaceutical organizations. Successful companies typically employ specialized KOL liaison roles, dedicated advocacy relations functions, and cross-functional teams that integrate HCP marketing, consumer marketing, analytics, and market access under unified leadership.

Collaboration among customer-facing staff is paramount. Field teams must operate as integrated units, recognizing that the small, interconnected nature of rare disease communities means all company representatives contribute to a single, cumulative impression. Reimbursement account managers have also evolved, becoming more patient-focused and education-oriented, with increasing involvement in pre-approval planning.

> *There's got to be that collaboration between customer-facing staff. I don't want them out there by themselves. They have to realize it's a team effort.*
>
> Rare Disease Industry Expert

GUIDING PRINCIPLE

Advocacy groups should be treated as partners, not as marketing channels, and sales representatives should work to build long-term relationships based on trust and mutual respect.

7. Patient Education and Support Programs

Patient support programs are central to rare disease commercial success. Because therapies often do not produce immediate improvement, sustained support is necessary to keep patients on therapy long enough to realize benefit. Programs should encompass disease education, treatment support, emotional support, and practical assistance with access and reimbursement.

Experienced patients and caregivers — sometimes called "veterans" — represent a powerful resource, offering real-world insights and peer support to newly diagnosed individuals. An especially impactful decision is whether to manage patient call centers internally. Companies that keep call centers in-house, staffed with people who deeply understand the disease and its emotional toll, consistently report stronger patient relationships and better persistence outcomes.

> *We considered outsourcing our patient call center, but we wanted people who 'lived it.' Keeping the call center inside was one of the smartest decisions the company made before and after launch. Our people would spend an hour or more on the phone crying with parents and listening to their stories. An outside company wouldn't have done that.*
>
> Rare Disease Industry Expert

8. Distribution, Access, and Payer Preparation

Market access in rare disease demands early, sustained engagement with payers. While any single rare disease may represent a small population, payers increasingly view rare diseases collectively — and the cumulative budget impact is substantial. Manufacturers must begin payer education at least 18 months before launch, focusing on disease awareness, the patient journey, and clinical evidence.

Clinical trial design has direct implications for market access. Inclusion and exclusion criteria from pivotal trials are frequently mirrored in payer prior authorization requirements. Companies must be strategic about trial design, recognizing that the evidence package they generate will directly shape access conditions post-launch.

> *Any one disease, how much are you paying attention to it? Zero. Rare disease as a whole, how much are you paying attention to it? Ten out of ten. Because as a group it's a massive budget impact.*
>
> Rare Disease Industry Expert

ACCESS STRATEGY

What is kind of flexible and in the manufacturer's control are those inclusion/exclusion criteria for a rare disease because you will definitely see those recreated in the authorizations that the payers have physicians do. Trial design directly shapes post-launch access.

9. Community HCP Education

Educating community healthcare providers is essential to improving diagnosis rates and reducing the diagnostic odyssey rare disease patients endure. The goal is not to transform community physicians into rare disease specialists, but to equip them with enough knowledge to recognize potential cases and refer patients to appropriate specialists or centers of excellence.

This is particularly challenging in diseases requiring genetic testing, as many community physicians have limited familiarity with genetic diagnostics. An often-overlooked dimension is the emotional toll rare diseases take on healthcare professionals themselves — physicians who encounter rare disease patients frequently experience frustration at the limits of their knowledge. Acknowledging this professional experience strengthens the manufacturer-physician relationship.

> *It's a lot to ask community doctors to diagnose these patients. We just want to get them to refer potential patients to the right place.*
>
> Rare Disease Industry Expert

OFTEN OVERLOOKED

One thing that is often overlooked is the frustration and emotional impact of the experience of rare disease patients on healthcare professionals. Acknowledging this reality — and designing education that addresses it — strengthens the relationship between manufacturers and the physician community.

10. Post-Launch: Persistence, Adherence, and Real-World Data

The work of rare disease commercialization does not end at launch — in many ways, it is only beginning. Post-launch activities focused on treatment persistence, adherence support, and real-world data collection are critical to long-term success and to ensuring patients continue to receive therapeutic benefit over time.

Patients need comprehensive, ongoing support including financial assistance, disease education, and adherence programs. Real-world data collection is increasingly important, particularly for therapies receiving accelerated approval based on biomarker endpoints. Payers find it difficult to contextualize biomarker-based evidence, and robust real-world data can bridge the gap between surrogate endpoints and demonstrated clinical benefit.

> *Often accelerated approvals are on a biomarker, less about an actual clinical outcome. And this really drives payers nuts because they can't contextualize that.*
>
> Rare Disease Industry Expert

To ensure adherence and prevent discontinuation, physicians and nurses at treatment centers must be educated on side effect management, and patients must be well-informed about both their disease and treatment. A patient-centered approach to post-launch support ultimately answers many payer questions as well, creating a virtuous cycle of evidence generation and access.

THE VIRTUOUS CYCLE

If you're doing this right, and if your focus is entirely on the patient experience, and going directly to the patient, you are answering a lot of payer questions as well. Patient-centered support creates a virtuous cycle of evidence generation and improved access.

SECTION 04

Conclusion

Rare disease commercialization is not simply a scaled-down version of traditional pharmaceutical marketing. It is a fundamentally different discipline that demands deeper relationships, earlier and more sustained engagement, greater patience, and an unwavering commitment to the patient communities at its center.

The ten best practices outlined in this white paper provide a practical framework for marketing and medical affairs professionals entering or advancing in the rare disease space. From the earliest stages of patient journey mapping through post-launch persistence and real-world

evidence generation, each practice reinforces the same core truth: in rare disease, trust is the most valuable asset a company can build.

Companies that earn that trust — through transparent community engagement, authentic KOL relationships, patient-centered support programs, and thoughtful clinical and commercial strategy — will not only achieve commercial success but will make a meaningful difference in the lives of patients and families who need it most. In rare disease commercialization, doing well and doing good are not competing objectives. They are one and the same.

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.