Rare Expertise

WHITE PAPER

The Impact of Personal Experience on the Business of Rare Diseases

How Personal Connections to Disease Are Shaping Research, Commercialisation, and Patient Outcomes

A Rare Expertise White Paper

For rare disease marketing and medical affairs professionals

rareexpertise.com

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Executive Summary

In most areas of medicine, the professionals who research, develop, and commercialize treatments are separated from the patients they serve by training, institution, and experience. Rare disease is different. Across the biotechnology, pharmaceutical, and patient advocacy sectors, it is strikingly common to encounter researchers, executives, founders, and commercial leaders who have a direct personal connection to the condition they are working on — as patients themselves, as parents or caregivers of affected children, or as family members who have lost someone to a disease for which no treatment existed.

This is not coincidence. The rare disease field has, over decades, been shaped by the particular intensity that personal experience brings: a willingness to take risks that conventional industry logic would reject, a depth of understanding that no clinical training alone can provide, and a motivation that sustains effort through the prolonged timelines and frequent failures that characterize rare disease drug development.

50%

of rare disease foundations

FOUNDED BY A PATIENT OR FAMILY MEMBER

10+

years typical timeline

FROM RESEARCH TO APPROVAL

7,000+

rare diseases identified

MOST WITH NO APPROVED TREATMENT

This white paper examines the nature and prevalence of personal connections in the rare disease industry, the ways in which they influence behavior and outcomes, the genuine advantages they confer, and the challenges — some rarely discussed — that they also create. Understanding this dimension of the rare disease ecosystem is essential for anyone seeking to work effectively within it.

~50%

of all rare disease advocacy foundations were established by a patient or family member following a diagnosis with *no clear treatment pathway* and no existing support network.

MANY OF THE MOST INFLUENTIAL ORGANIZATIONS IN THE FIELD WERE FOUNDED AT A KITCHEN TABLE BY A PARENT.

SECTION 02

How Common Is the Personal Connection?

Precise figures are difficult to establish — personal experience is rarely disclosed in professional biographies or on company websites, and many individuals are selective about when and how they share it. But anecdotal and qualitative evidence from across the rare disease sector strongly suggests that personal connections are far more prevalent here than in any other area of the pharmaceutical and biotech industry.

Patient Advocacy Organizations

Patient advocacy organizations provide the clearest data point. It is estimated that roughly half of all rare disease advocacy foundations were established by a patient or a family member following a diagnosis with no clear treatment pathway and no existing support network. Many of the most influential organizations in the field — including those that have directly funded the research leading to approved treatments — were founded at a kitchen table by a parent who had just been told their child had a condition affecting fewer than a thousand people worldwide.

Biotech Founders

Among biotech founders, the pattern is also striking. A disproportionate number of rare disease biotechnology companies have been founded by or in close collaboration with individuals who have a personal stake in the outcome. Several of the most significant rare disease approvals of the past two decades trace their origins to a founding moment that was as much personal as it was scientific.

> ● WITHIN LARGER COMPANIES
>
> Within larger pharmaceutical and biotech companies, personal connections are less visible but remain present. Medical affairs professionals, clinical development leads, commercial directors, and patient engagement teams in rare disease divisions frequently include individuals who were drawn to the field by direct experience. Many will say, privately, that they cannot imagine working on anything else.

SECTION 03

What Personal Experience Brings to the Field

The advantages conferred by personal connection to rare disease are multiple, deeply rooted, and have been demonstrated repeatedly across the history of the field.

Exceptional Motivation and Persistence

Rare disease drug development is among the most demanding endeavors in medicine. Development timelines are long, patient populations are small, regulatory pathways are uncertain, and failure rates are high. The sustained effort required — often over a decade or more from initial research to approval — demands a quality of motivation that is difficult to manufacture through professional incentives alone.

> *For a parent who has watched a child's condition progress while conventional medicine offers nothing, the urgency is not abstract. For a patient-founder who is racing against their own disease timeline, persistence is not a professional virtue but a survival instinct.*

Personal experience provides a different quality of motivation. This type of motivation has repeatedly proven capable of sustaining effort through setbacks that would cause a purely commercially motivated enterprise to redirect its attention elsewhere.

Deep Patient Insight

The gap between clinical understanding and lived experience of rare disease is significant. Clinicians may understand a disease mechanistically and prognostically, but the texture of daily life with a rare condition — the impact on family dynamics, the emotional weight of diagnosis and

uncertainty, the specific frustrations with existing care pathways, the things that patients and caregivers most need from a treatment — is knowledge that clinical training does not reliably provide.

> PATIENT-CENTERED IMPACT
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> **Professionals with personal experience of rare disease carry this knowledge as a matter of course. It informs how they design clinical trials, how they communicate with patient communities, how they frame value propositions for payers, and how they prioritize endpoints that matter to real people living with the condition. The result, in the best cases, is research and commercial programs that are genuinely patient-centered rather than merely claiming to be.**

Authentic Patient Advocacy Relationships

Trust between industry and patient advocacy communities is a critical and often underestimated asset in rare disease. Patient advocacy organizations control access to patient registries, biospecimen collections, natural history data, and — critically — the patient communities whose participation makes clinical trials possible. In rare diseases, where the entire eligible trial population may number in the hundreds globally, these relationships can be the difference between a program that can enroll and one that cannot.

Industry professionals who share personal experience with patient communities often find that these relationships form more quickly and run more deeply. There is a recognition of common ground — a shared language and understanding — that can take years to build through professional engagement alone. This is a genuine competitive advantage in a field where program success can depend on the quality of patient community relationships.

Credibility With Investors and Stakeholders

The rare disease investment community has become increasingly sophisticated in assessing management teams. Experienced rare disease investors know that the field's particular demands — patient recruitment challenges, regulatory complexity, commercial constraints of small populations — require a specific combination of scientific, clinical, and operational knowledge. A founder or executive whose personal connection to a disease is matched by deep expertise in that disease's biology, natural history, and treatment landscape presents a compelling combination of motivation and capability.

Patient and family advocates who have built relationships across the rare disease community over years or decades also bring a form of stakeholder credibility that cannot easily be hired in. Their presence in leadership positions signals to patients, clinicians, payers, and partners that the organization understands what is at stake — not only commercially, but humanly.

For a parent who has watched a child's condition progress while conventional medicine offers nothing, the urgency is not abstract. For a patient-founder racing against their own disease timeline, persistence is not a professional virtue — ***it is a survival instinct.***

SECTION 04

Challenges and Tensions

The advantages of personal experience in rare disease are real and well-documented. Less often discussed are the challenges that personal connection can introduce — challenges that are worth examining frankly, because unacknowledged they can limit the effectiveness of individuals and organizations.

The Risk of Blurred Boundaries

Personal connection is a strength when it fuels motivation and insight. It can become a liability when it makes objective decision-making more difficult. A founder who is also the parent of an affected child may find it harder to make a dispassionate assessment of clinical data that is ambiguous, to discontinue a program that is unlikely to succeed, or to weigh the interests of all patients equitably when personal relationships create differential loyalties.

> GOVERNANCE MATTERS
>
> These are not abstract risks. In a field where the stakes are high and the patient populations are small and often known personally, the pressures on objective judgment are real. The best organizations in rare disease acknowledge this openly and build governance structures and team compositions that complement personal passion with the capacity for rigorous and independent assessment.

Emotional Sustainability

Working in rare disease is emotionally demanding for everyone in the field. For those with personal connections, the weight can be considerably heavier. The death of a patient in a clinical trial is a professional and scientific event; for someone who knew that patient personally, or who sees their own child in every trial participant, it is something more. Over the course of a career, this accumulation of personal investment and personal loss takes a toll that organizations rarely acknowledge or adequately support.

> *Burnout is a known challenge in rare disease. Among those with personal connections to the diseases they work on, the risk is amplified by the difficulty of separating professional setbacks from personal ones. A failed clinical trial is also a failed hope; a program discontinuation is also, in some sense, an abandonment.*

The rare disease sector would benefit from a more open conversation about emotional sustainability and the support structures that individuals and organizations can put in place.

Managing Conflicts of Interest

Personal connection creates potential conflicts of interest that must be managed carefully. A patient-executive whose company is developing a treatment for their own condition faces questions — from investors, regulators, and patient communities — about how they manage decisions where their personal interest and their fiduciary duty may diverge. A patient advocate on a company's scientific advisory board who is also receiving a treatment from that company occupies a complex position.

These conflicts are manageable, but they require transparency and clear governance. Organizations that handle them well tend to do so by acknowledging them openly rather than minimizing them, and by establishing frameworks that protect the integrity of decision-making without asking individuals to deny the personal experience that makes them effective.

Representation and Equity

The personal connection model has historically tended to amplify the voices of those with the social capital, education, and resources to found organizations, build networks, and engage with industry. This has served some rare disease communities very well. It has left others — those affecting populations with less access to these resources, or diseases that attract less visibility — comparatively underserved.

EQUITY IMPERATIVE

As the rare disease field matures, there is growing recognition that the personal connection model needs to be consciously broadened to ensure that the communities with the greatest need are not the ones least represented in research prioritization and commercial development decisions.

CHALLENGES

Four Tensions of Personal Connection in Rare Disease

1. **Blurred Boundaries** — when passion compromises objective decision-making on ambiguous data
2. **Emotional Sustainability** — the accumulated toll of personal investment and personal loss over a career
3. **Conflicts of Interest** — managing divergence between personal stakes and fiduciary duties
4. **Representation Gaps** — ensuring underserved communities are not left behind by a resource-dependent model

SECTION 05

Implications for Industry and Investors

For professionals working in and around the rare disease sector, understanding the role of personal experience has practical implications across hiring, investment, and partnership strategy.

Hiring and Team Building

For rare disease organizations, understanding the role of personal experience in team motivation and capability has practical implications for hiring. Personal connection should neither be treated as a prerequisite nor as irrelevant — it is one dimension of what makes a professional effective in this field, alongside scientific rigor, commercial capability, and operational experience.

Organizations that understand this well tend to build teams that consciously combine individuals with deep personal investment with those who bring external rigor and independence. They create cultures in which personal motivation is recognized and valued, emotional wellbeing is actively supported, and governance structures ensure that passion does not compromise objectivity.

Investor Due Diligence

For investors in rare disease biotechnology, understanding the role of personal experience in a management team's motivation and capability is a legitimate and important part of due diligence. A founder's personal connection to a disease is not a replacement for scientific or commercial capability, but it is evidence of a quality of motivation and depth of knowledge that is relevant to assessing the team's likelihood of sustained performance under the pressures of rare disease development.

Equally, investors should consider how personal connections are being managed, not just whether they exist. Organizations that are transparent about these dynamics and have thoughtful governance in place are likely to navigate the associated challenges more effectively than those that treat personal connection as purely a marketing asset.

Patient Advocacy Partnerships

For organizations without deeply personal connections to the communities they serve, understanding this dynamic is important context for building effective patient advocacy partnerships. Patient communities can sense the difference between engagement that is driven by genuine understanding and engagement that is performative. Closing that gap — through genuinely patient-centered research design, equitable data sharing, meaningful participation in governance, and consistency of engagement through setbacks as well as successes — is the work of building the kind of trust that personal connection often provides more easily.

SECTION 06

Conclusion

The rare disease field has been built, to a remarkable degree, by people with skin in the game — scientists, executives, advocates, and investors whose engagement with the work is personal as well as professional. This is one of the defining features of the rare disease ecosystem, and it has been a genuine force for progress: driving research into diseases that the mainstream industry had neglected, sustaining effort through the prolonged timelines of rare disease development, and building the patient community relationships on which successful programs depend.

Understanding this dimension of the field is not merely interesting context. It has practical implications for how organizations hire and develop talent, how they build governance structures, how investors assess teams, and how patient communities and industry engage with one another. The personal connection in rare disease is at once one of the field's greatest assets and one of its most underexamined dynamics.

Acknowledging it clearly — its power, its value, its risks, and its limits — is part of the maturation of a field that is increasingly capable of delivering on the transformative promise it has always carried.

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.