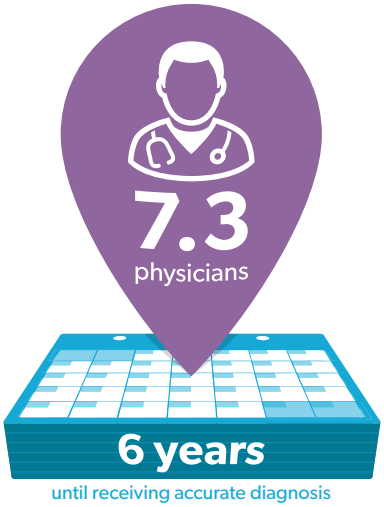
Finding patients and building rare communities



The problem

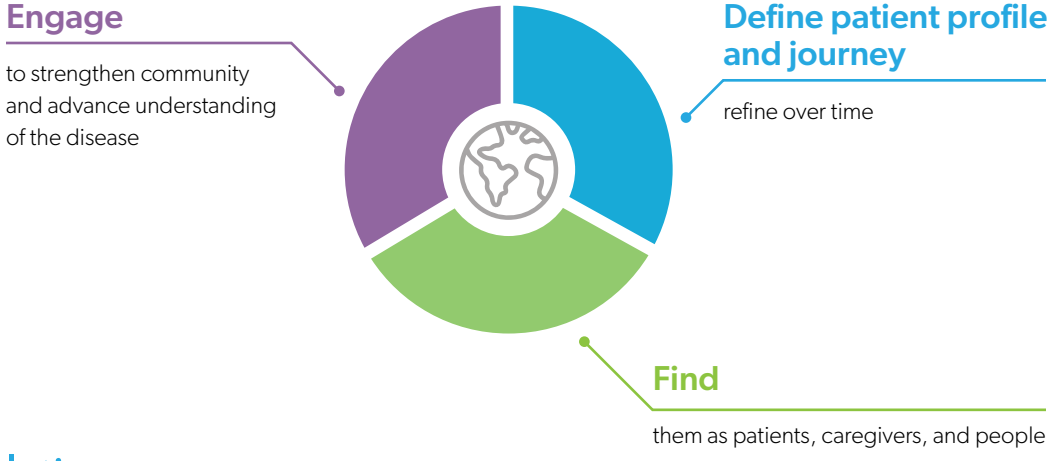
In rare diseases, patients experience an average delay of 6 years from onset of symptoms to receiving an accurate diagnosis. Along the way, they see an average of 7.3 physicians, often receiving incorrect diagnoses.[1] As a result of this protracted diagnostic odyssey, patients are often treated for conditions they don't have while their actual condition progresses unchecked. Along with the health burden associated with the condition and any treatments, patients and their families also experience associated psychosocial and financial burdens.

For pharmaceutical and biotech companies working in rare diseases, finding patients to serve as advisors, define the patient journey, participate in trials, or receive approved therapies is a major challenge. With limited numbers of diagnosed patients and diagnosed patients with more advanced disease, industry often struggles to enroll trials for and demonstrate effectiveness of investigational products. Inability to efficiently conduct trials negatively impacts all people living with the disease.

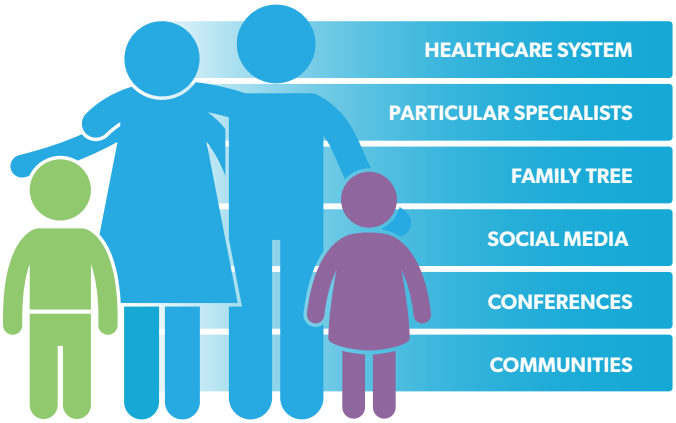
Finding and diagnosing patients early in the journey benefits the entire rare disease community. While artificial intelligence and big data are growing as ways to potentially find patients sooner, sometimes—especially in rare—a more personal approach is required.

Rare challenges call for rare experts

Because we are immersed in rare diseases, both professionally and personally, our team understands the intricacies of rare. Patient finding and community building are cornerstones of rare disease problem solving as they directly benefit individuals living with a rare disease and the rare community at large, while enabling improvements in patients' experience and care. Our proprietary iterative process illustrated below includes 3 steps that repeat over time as we learn and find more patients. To complement patient-finding efforts, rareLife also deploys educational initiatives so that healthcare providers (HCPs) are better equipped to suspect and diagnose people with a rare disease—here we focus on finding people directly.



Step 1: Define patient profile and journey—refine over time

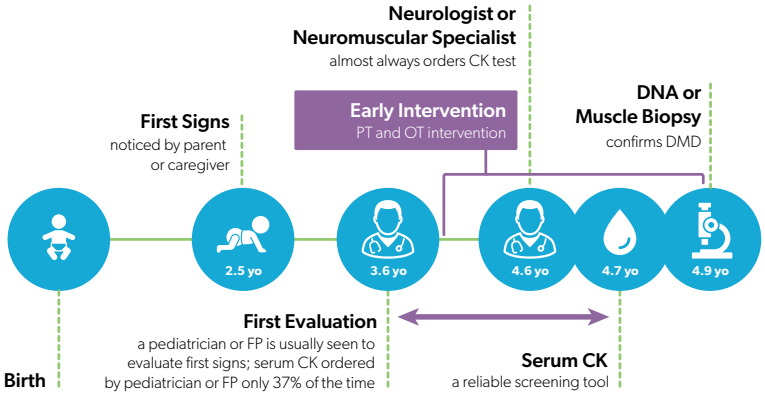


The patient-finding process begins with defining the patient and caregiver profiles and journeys based on available information. We do this to discover where to meet families at specific places along their journey. "Where" to meet them may be within the healthcare system, at particular specialists, within their family tree, on social media, at conferences, and sometimes in certain geographic locations or communities. Patient profile and journey insights may come from any number of places including the scientific literature, social media, advocacy groups, medical charts, HCPs, investigators, or patients and families themselves. The specific tactics we use depend on the disease and situation. Taken together, the clues we find will enable us to build likely journeys, patient profiles, and potential places for us to focus.

Step 2: Find them as patients, caregivers, and people

Here we employ tactics designed to find patients at those places we've identified in Step 1. This might be done through engagement of known treaters and those who have published, "motive-ed " campaigns (that's a campaign created to motivate and educate), support of peer-reviewed publications, pedigree analysis, or Facebook group and advocacy organization engagement, etc.

For example, we discovered in Step 1 of a recent project that early intervention (EI) physical and occupational therapists were involved in assessments and care of patients not yet diagnosed with a rare muscular dystrophy. We designed a motive-ed campaign focused on a specific red-flag symptom that provides the confidence, tools, and resources to help EI therapists recognize patients with this muscular dystrophy and get physicians to order the right diagnostic test. Motivating and educating the EI therapists is expected to shorten the time to diagnosis and enable appropriate care, sooner.



FP - family practitioner
CK - creatine kinase
DMD - Duchenne muscular dystrophy
PT - physical therapy
OT - occupational therapy

Step 3: Engage to strengthen community and advance understanding of the disease

Here we continue to build on what we know and the patients we have found in Step 2. Tactical elements may include HCP, researcher, and patient/advocate/caregiver (PAC) interviews and advisory boards, center of excellence (CoE) certification, building advocacy skills and infrastructure, developing custom digital communities, and executing motive-ed campaigns. This step is critical because through it, we can motivate and educate while we continue to better understand the patient journey and find more patients.

rareLife solutions
collaboration + science + technology

Case Study

Patients with this rare disease typically receive the correct diagnosis fairly quickly; however, there is no disease-specific patient organization or place for them to learn more and receive the support they need. Moreover, our up-and-coming biotech client is planning a Phase 3 trial and will need to be able to recruit appropriate patients efficiently (with no advocacy organization or CoEs to help). In this case, we designed a simple social media campaign for Step 2 and developed a patient engagement strategy with patient/industry/key opinion leader (KOL) collaboration through a patient steering committee, trial design advisory board, surveys, and motive-ed materials for Step 3. By enabling collaboration between patients found in Step 2, industry, and KOLs, we are building a vibrant and diverse community of people who are organized and invested in learning more, sharing ideas, and participating in research when possible. This community will further optimize diagnosis, improve the patient experience, and help get appropriate treatments to the people that need them faster.

Key Takeaways

- Shortening the time to diagnosis is good for all stakeholders in a rare disease community
- Addressing the often-difficult task of finding patients (before and after diagnosis) enables the building of a vibrant, motivated community
- A vibrant community can improve every step of the patient journey and drug development process
- Because rare is personal, it makes sense to employ a humanistic approach to find and engage people living with rare diseases

Coming soon: rarelQ on rare disease collaboration beyond clinical development.
Contact us and we will send it to you as soon as its ready!

To expand your rarelQ, contact Laura Wuerth at rareLife solutions: **LWuerth@rareLifesolutions.com**.

Reference
1. Ronicke S, et al. *Orphanet J Rare Dis*. (2019) 14:69.