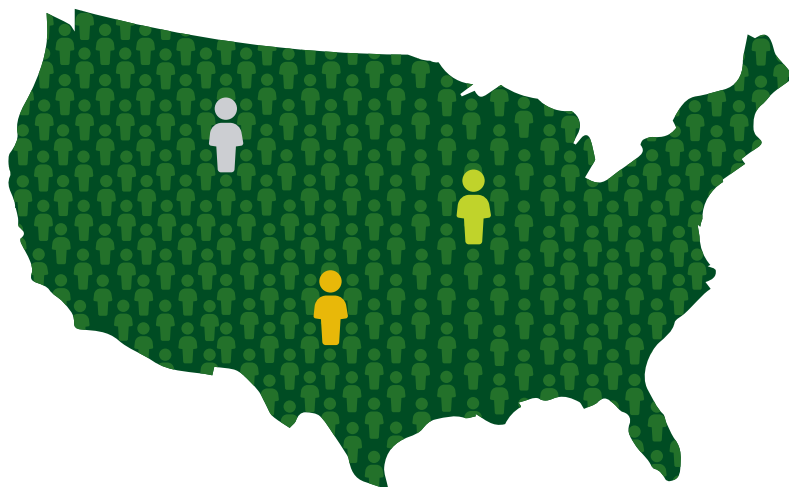


BIOPHARMA SERVICES | COMMERCIALIZATION

Data-driven physician engagement for ultra-rare diseases

Rare diseases, defined as conditions affecting fewer than 200,000 individuals in the United States, present unique challenges in diagnosis and treatment. Within this category, ultra-rare diseases are even more elusive, with a **prevalence often less than 1 in 50,000 people (or approximately 6,700 people in the United States)**. Despite their rarity, these diseases collectively impact a significant portion of the population—up to 10% globally.¹



The introduction of new therapies for such conditions is a monumental task, particularly when identifying eligible patients is akin to finding a needle in a haystack. This case study explores how Quest Diagnostics, led by its Healthcare Analytics Solutions (HAS) team, leveraged advanced data analytics to overcome these challenges and support the successful commercialization of a groundbreaking therapy for an ultra-rare disease **which constitutes 0.44% of all hematological malignancies detected each year.**²

Strategic collaboration

To address the challenges inherent in identifying eligible patients for a new therapy targeting an ultra-rare and aggressive form of cancer, Quest Diagnostics partnered with a leading pharmaceutical company.



The collaboration, led by Quest's Healthcare Analytics Solutions (HAS) team, was designed to leverage Quest's vast data resources and analytical expertise to support the commercialization efforts for this breakthrough treatment.

The team focused on two key objectives: identifying physicians who might have patients with the disease and engaging them with actionable information about the new therapy. **Given the absence of a single definitive diagnostic test for this rare condition, the collaboration required innovative approaches to pinpoint potential patients based on clinical markers and deidentified patient data.**



By integrating advanced analytics with clinical insights, Quest developed a tailored algorithm to locate physicians treating patients who might exhibit the subtle and often overlooked signs of the disease. **This not only supported initial outreach and education efforts but also proved valuable for broader omnichannel orchestration and salesforce operations, enhancing the pharmaceutical company's ability to coordinate its commercial strategy.**

Data-driven methodology

The success of this collaboration hinged on the ability to identify patients with an ultra-rare and aggressive form of cancer in the absence of a definitive diagnostic test. Quest Diagnostics leveraged its extensive data resources, drawing on 500 million lab tests performed annually by more than 50% of US healthcare providers, to address this challenge.

The HAS team, consisting of experienced data scientists, developed a sophisticated algorithm to analyze clinical markers and deidentified patient data. This algorithm was designed to identify subtle indications of the disease, which are often missed due to the rarity of the condition.

The HAS team collaborated closely with Quest's clinical and lab scientists, tapping into the expertise of its network of 700+ MDs and PhDs. This collaboration ensured that the algorithm was both precise and effective in identifying potential patients. The results were then reviewed, and patients who fit the criteria were flagged for further evaluation by their physicians.

This approach demonstrated the powerful role that data can play in overcoming the challenges of diagnosing ultra-rare diseases, highlighting how targeted, data-driven strategies can identify patients who might benefit from new therapies, even in the most complex scenarios.

Outcomes and impact

The data-driven approach implemented by Quest Diagnostics led to significant and meaningful outcomes in a challenging therapeutic area.

Over the course of 18 months, Quest successfully identified 158 patients whom physicians suspected might have the ultra-rare disease.

These patients were flagged for further examination, which was critical given the aggressive nature of the condition.

Notably, 20 patients were identified as eligible candidates for the new therapy—a remarkable achievement given the rarity of the disease.* This success significantly enhanced the pharmaceutical company's ability to strategically target its commercialization efforts, ensuring that the therapy reached those who could benefit the most. **The pharmaceutical company continues to run this program several years after its initial launch, further demonstrating its effectiveness in identifying hard-to-reach patients in need of this life-changing therapy.**

Broader implications and future directions

The success of this collaboration highlights the growing importance of data-driven approaches in the commercialization of therapies for ultra-rare diseases. By effectively identifying patients who might benefit from a novel treatment, Quest Diagnostics demonstrated how advanced analytics can help overcome the inherent challenges of these complex conditions.

This case sets a precedent for future projects, showcasing the potential of leveraging large-scale deidentified data resources to support the development and market introduction of therapies for other rare and challenging diseases. As orphan drug development continues to expand, the ability to harness data in this way will be increasingly critical, not only for ultra-rare diseases but across a broad spectrum of medical conditions.

Looking forward, the methodologies developed and refined through this collaboration can be adapted and applied to new challenges, further enhancing the role of data in transforming patient outcomes. The collaboration also underscores the importance of interdisciplinary efforts, where data scientists, clinical experts, and healthcare providers work together to deliver targeted, effective solutions in even the most complex scenarios.

* Initially, the prevalence of this disease was believed to be approximately 600 cases annually in the US. However, after a more extensive evaluation following the program's implementation, the pharmaceutical company determined that the actual prevalence was closer to 120-150 patients per year.



Through our breadth and depth of deidentified data, we can help support your therapeutic development and commercialization efforts.



To learn more visit Biopharma.QuestDiagnostics.com/CommercialAlerts

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