Finding Patients With Rare Diseases

TWO TRUTHS AND A LIE

The hope for many companies developing drugs for rare diseases is this: **the significant unmet need, given a dearth of treatment options, will drive demand and thus product uptake.** Unfortunately, the reality is more challenging.

While we have seen a significant increase in the number of drugs for rare diseases, accurate diagnosis rates of rare diseases continue to trail behind those of more prevalent diseases.

The average time to diagnosis for a patient with a rare disease is 5 to 7 years. The delay is driven by:

- Lack of awareness among health care professionals (HCPs) of symptoms and markers
- Patient education deficits
- Heterogeneity of both patients and symptoms

In response to this delay, companies have invested in disease awareness and diagnosis programs, including artificial intelligence (AI)-driven patient-finding initiatives. While heralded as the solution to address the diagnosis lag, results from these programs have been mixed and are the source of several challenges, including:

- How to encourage HCPs to diagnose these patients when they do not appear to have the disease markers highlighted in disease awareness materials
- How to assess patients when results have low-precision numbers
- How to reconcile differences between patient-finding models

At 81qd, we take a unique approach by leveraging AI to identify specific physician practices where undiagnosed patients with the highest probability of having a rare condition are currently being managed for other conditions.

Over the years, we have partnered with many companies to help them overcome the challenges with their patient-finding initiatives. To guide our clients forward, we offer a simple framework based on the popular icebreaker game:

[1]National Organization for Rare Disorders (NORD) -National Organization for Rare Disorders (NORD) Launches Free CME Program to Educate Medical Professionals about Rare Diseases - https://www.prnewswire.com/news-releases/national-organization-for-rare-disorders-nord-launches-free-cme-program-to-educate-medical-professionals-about-rare-diseases-300453583.html

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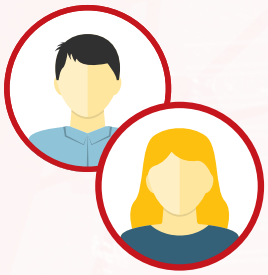
TWO TRUTHS AND A LIE

Truth: Undiagnosed patients with rare diseases will not look like the patients you know

Be prepared to be surprised. Undiagnosed patients identified through AI-driven approaches will frequently not look like diagnosed patients. If they did, and if they were that simple to find, why would we need AI? Because most marketers are guilty of confirmation bias, they often look for attributes that confirm their preexisting knowledge about the market or a disease. When they see something contrary to their experience, they naturally refute it. AI should be designed to identify patients that may not look like "typical patients." However, the inherent heterogeneity of the patient journey experienced by those with rare diseases means there are no "typical patients."

In developing models, data scientists must work actively to avoid confirmation bias, rather than collecting and analyzing data with the goal of supporting existing beliefs.

The Lesson: Be prepared to be surprised; be wary if you are not.



Truth: Identifying patients is only a third of the battle

If the final output from a patient-finding initiative is a list of deidentified undiagnosed patients with locations, you are setting yourself up for failure.

Patient-finding solutions that enable patients to be mapped to actual clinician practices where they are currently being managed for other conditions considerably increase actionability of the results. The ability to find undiagnosed patients along their disease journey and to understand the HCPs who are currently treating them has the potential to dramatically change 2 critical factors:

- How we identify HCPs who encounter yet-to-be-diagnosed patients
- How we engage effectively with these HCPs

A more effective approach to identifying patients includes a closed-loop solution that engages with patients and with HCPs to drive diagnosis, which comprises the other two-thirds of the battle.

The Lesson: Ensure you have an integrated strategy for engaging HCPs and supporting diagnosis, not just finding patients.

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TWO TRUTHS AND A LIE

Lie: Data science alone can solve every problem.

Rare disease patient finding is hampered by the very nature of rare diseases: they are rare and complex.

First, a lesson in data science to better understand the impediments to finding patients with rare diseases: the concept of "precision" is critical. Precision is the number of True Positives (patients known to have the disease) divided by the sum of True Positives + False Positives (patients inaccurately predicted to have the disease).

Given disease epidemiology, precision levels of data models within the rare disease space can be lower in comparison with more prevalent diseases. The clinical journeys for patients with rare diseases are the core reason for this. These journeys are often heterogeneous in terms of progression rates and symptoms. More times than not, data scientists who build the models do not have the medical expertise needed to understand the nuances of the disease and create the ensemble of models required to drive up precision. This is why we at 81qd ensure our data scientists partner with medical experts to develop models and data inputs.

Precision aside, one must be able to build an actionable communications plan based on the patient-finding models. Doing so can drive earlier diagnosis through more effective targeting and messaging. When biopharmaceutical company executives hear about the promise of AI for diagnosis, they hope to walk into an HCP office and jump right into a conversation about undiagnosed patients. Good luck with that. This approach has been met with mixed success at best. Since patients are deidentified, the discussion is necessarily general. A meaningful discussion has to be insightful without being judgmental, which is difficult to achieve without specific patient information.

The Lesson: Data science alone is not the answer to the problem. One needs to integrate medical and marketing expertise along with data science to create an end-to-end approach that facilitates earlier diagnosis.

The potential patient impact is clear. As the almost 600 biopharmaceutical companies work to develop life-saving treatments, we can harness the opportunities that AI-driven analytics provide to shorten the diagnosis journey and ensure that more of the 30 million people in the United States living with rare diseases have access to treatment.