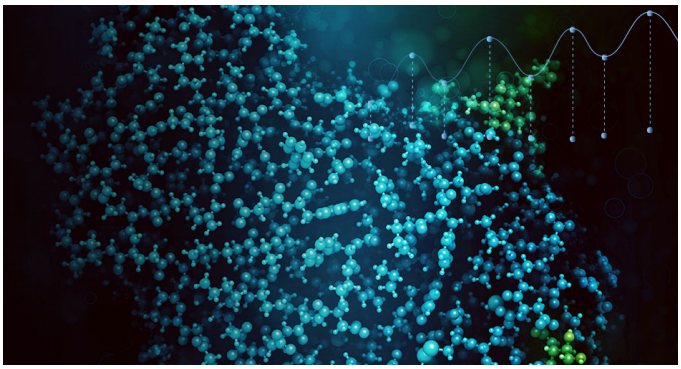
Case Study: Patient Identification in Rare Diseases

Challenges

A global biotechnology company has developed a powerful therapy for rare metabolic disease patients, but many potential beneficiaries don't know they have the disease until it's progressed significantly. Our partner asked us to use our data assets and AI capabilities to identify more patients.



Solution

We built our proprietary OM1 Patient Finder to identify key patterns in medical histories that signal undiagnosed disease. Using OM1 Patient Finder for our client, we achieved:

- Very strong overall predictive performance (AUC of 0.82)
- Identification of patient subgroups at much higher risk than average, with more than 20x positive patients in the riskiest 1% of the population
- Clarification of diffuse symptoms – like GI distress – that clinicians recognize but often aren't specific enough to us in diagnosis
- New hypotheses for better understanding and predicting disease progression and complications

Obstacle 1	Lack of quality data for understanding and identifying rare disease patients	The OM1 Patient Finder is integrated with the OM1™ Real-World Data Cloud of over 250 million patients, which allows us to achieve strong predictive capability with high quality research-grade data.
Obstacle 2	Limited explainability of AI technologies	At OM1, we aim to achieve strong predictive performance while also maintaining clinical explainability. We can describe what's going on in the technology, so you're not left in the dark.
Obstacle 3	Lack of real-world application in clinical practice	At OM1, we conceptualize and calibrate Patient Finder from the start to be useful in clinical practice to find actual patients.

Results

The OM1 Patient Finder delivered tangible results, accurately identified patients, and improved clinical decision making:

- The combination of strong data assets and a purpose-built AI platform delivered transparent and implementable results
- We independently identified all the major clinical characteristics of this disease – and some that patients frequently bring up as underappreciated, like mental health and addiction issues
- Because Patient Finder is built to accept a range of data inputs, it can be implemented in a variety of environments

Key Milestones

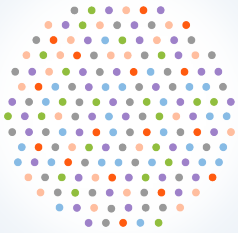
- Rapid initial calibration (3-4 weeks)
- Go / no-go decision for both teams
- Analytic outcomes and scenario simulation
- Deep clinical insight into explanability
- Clinical implementation into workflows to find patients

Cutting-edge AI technology integrated with high quality RWD can deliver real value and rare insights.

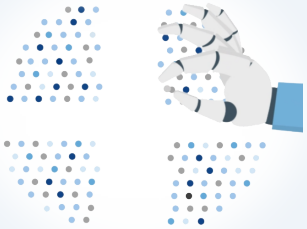
The OM1 Patient Finder has demonstrated the ability to improve detection and diagnosis of rare disease patients.

Diseases with long paths to diagnosis and diffuse, heterogeneously symptomology are especially good candidates.

Platform Calibration



Learns from millions of data points to understand **common themes**



Organizes data into '**signals**' that help distinguish the target patients

Uses these signals to **find patients** most likely to have undiagnosed disease

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