QuintilesIMS™

Real-World Evidence Solutions
A breakthrough in rare diseases

Using real-world data and advanced analytics to find undiagnosed patients with rare diseases

Patients with rare diseases already face huge challenges in accessing treatments that will help them. Often, they aren't diagnosed with their condition until the later stages where even exceptional treatments are less effective. What if we could change that? What if we could find those patients at an earlier stage, from anonymous patient-level data that is already being captured in clinical practice?

Rare diseases are not that rare, yet accurate diagnoses can be

350m people worldwide have a rare disease[1]

7,000 rare diseases have been identified[1]

50% of rare disease sufferers are children[1]

40% of patients are misdiagnosed initially[2]

7.3 physicians (avg.) are seen before diagnosis[3]

4.8 years (avg.) before an accurate diagnosis[3]

Finding the needle in the haystack

Increased availability of RWD and advances in pattern recognition techniques mean that rare disease detection algorithms now represent a potentially highly effective way of finding undiagnosed patients.

- **Detailed RWD** on symptomology, diagnoses, treatment history, lab tests and more, which is routinely collected from patients anonymously, presents a rich foundation that can be harnessed exactly for this purpose
- **Predictive analytics** can exploit often complex, subtle patterns in the data of diagnosed patients with rare conditions to identify new undiagnosed sufferers of a disease

Pattern recognition techniques exploiting machine learning methods have been successfully applied on routine healthcare data to find 'the needle in the haystack'. These advanced approaches have revolutionized facial recognition systems and online search engines and when applied with clinical insight are now helping to solve complex problems in healthcare.

Under-diagnosis is a major barrier to the successful treatment of rare diseases. New tools applying real-world data and innovation in advanced analytics are creating potential for dramatic improvements in bringing hope for hard-to-find patients.

Ground-breaking solutions for detecting rare diseases

IMS Health is at the forefront of innovative research combining RWD and predictive analytics to help detect undiagnosed cases of rare diseases.

- **Largest international fit-for-purpose RWD** portfolio of 500 million+ patients, with expertise in data management and sourcing
- **Dedicated rare disease detection strategy team** supported by highly qualified biostatisticians, data scientists, epidemiologists and clinical experts providing deep disease understanding
- **Pioneering methodologies** that incorporate predictive analytics in healthcare, applying modern machine learning methods to solve complex problems
- **Driving clinically relevant insights,** partnering with providers to improve clinical practice for rare diseases

Demonstrating dramatic improvements

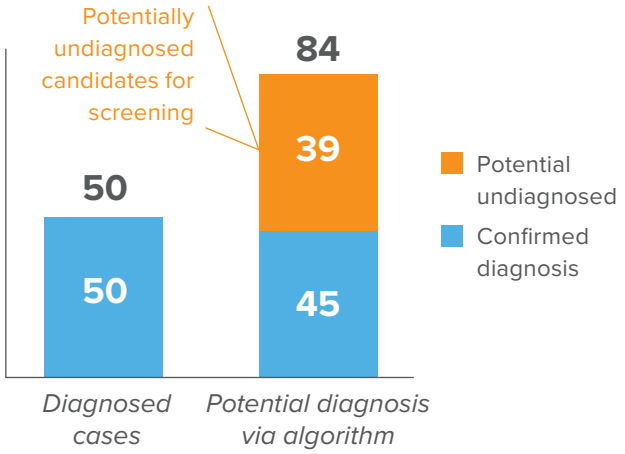
Pioneering work from IMS Health is already demonstrating the promise of diagnostic algorithms in rare diseases, including at-risk patients in oncology. Our analyses of patient care pathways using RWD are also shedding light on barriers in health systems causing delays in diagnosis. This research illustrates the power of these techniques within the context of broader efforts to reach individuals with potentially curative treatments.

Finding high-risk cancer patients earlier

Challenge: To identify patients with a rare cancer that is substantially under-diagnosed and where up to 40% of identified patients are diagnosed late, often by decades.

Novel approach: A two-step process leveraging de-identified EMRs involved firstly the development of a diagnostic algorithm using classical statistical methods combined with clinical expertise, and secondly the application of advanced machine learning methods to refine and optimize the algorithm.

Impact: A test sample of 70,000 randomly selected patients was risk scored by the initial algorithm, producing a high-risk group containing 8% of confirmed cases. The test sample was then risk scored by the refined algorithm, exploiting machine learning techniques, producing a prevalence of the confirmed diagnosis in the highest risk group of 20.5%. Given that only 0.7% of patients in the test sample had the disease, the study provided evidence that use of the algorithm could dramatically increase the odds of finding high-risk patients earlier.



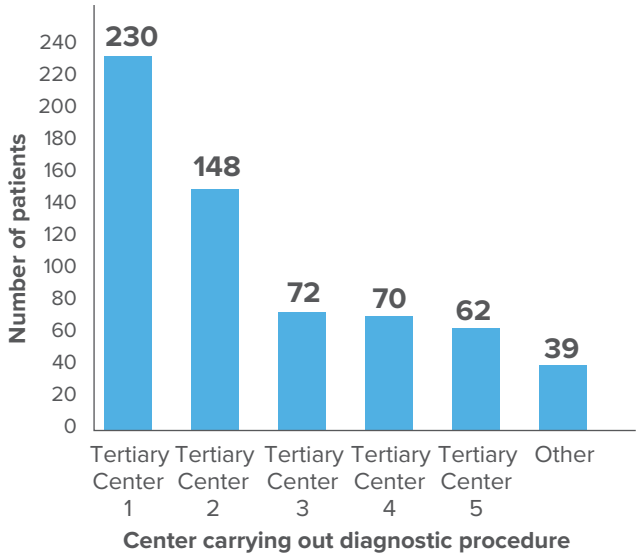
Identifying health system barriers causing under-diagnosis

Challenge: To determine in a complex, multi-center pathway whether a lengthy diagnosis process could be a causal factor in late presentations of patients with a potentially fatal rare cardiac disease that could be reversible or manageable with treatment if detected early.

Novel approach: In a process based on literature and data profiling, a cohort selection algorithm was developed leveraging Hospital Episode Statistics data covering outpatient, inpatient and A&E activity in England. The selected cohort triangulated well with literature incidence and demographic values and enabled investigation of health service usage and diagnosis patterns over more than five years. This revealed a high number of events for three years ahead of a formal diagnosis, with over 90% of patients being known to the hospital system within the three-year time frame, and a wide variety in the types of diagnostic pathways to reach a tertiary center initially. Furthermore, the study identified substantial variability in the incidence rate per 100K population, with regions feeding into the leading diagnosis center having significantly higher levels of incidence, suggesting challenges of under diagnosis in other parts of the country.

Impact: Insights from the analysis were positively received by leading clinical experts in the field as a novel and previously unseen perspective on their patient population. The study also generated hypotheses for further work, served as a basis for building relationships with academic and clinical institutions to build a rich pool of RWD for this therapy area, and will likely provide evidence to engage payers and take business decisions around funding future interventions.

Number of patients diagnosed by center



Source: IMS Health

Learn more

For more information about the application of advanced analytics to RWD to identify undiagnosed patients with rare diseases, and determine whether a rare disease detection algorithm could address your needs in a specific therapy area, please email John Rigg at **John.Rigg@uk.imshealth.com**.

References

1. Global Genes™, https://globalgenes.org/rare-diseases-facts-statistics/

2. Eurordis – Rare Diseases Europe. Survey of the delay in diagnosis for 8 rare diseases in Europe ('EURORDISCARE 2'). www.eurordis.org/IMG/pdf/Fact_Sheet_Eurordiscare2.pdf

3. Engel PA, Bagal S, Broback M, Boice N. Physician and patient perceptions regarding physician training in rare diseases: The need for stronger educational initiatives for physicians. Journal of Rare Disorders, 2013; 1(2): 1-15