THE EVOLVING WORLD OF ARTIFICIAL INTELLIGENCE: RARE DISEASE IMPACT

A Q&A White Paper with **Oodaye Shukla,** *Chief Data & Analytics Officer,* HVH Precision Analytics, now EVERSANA™

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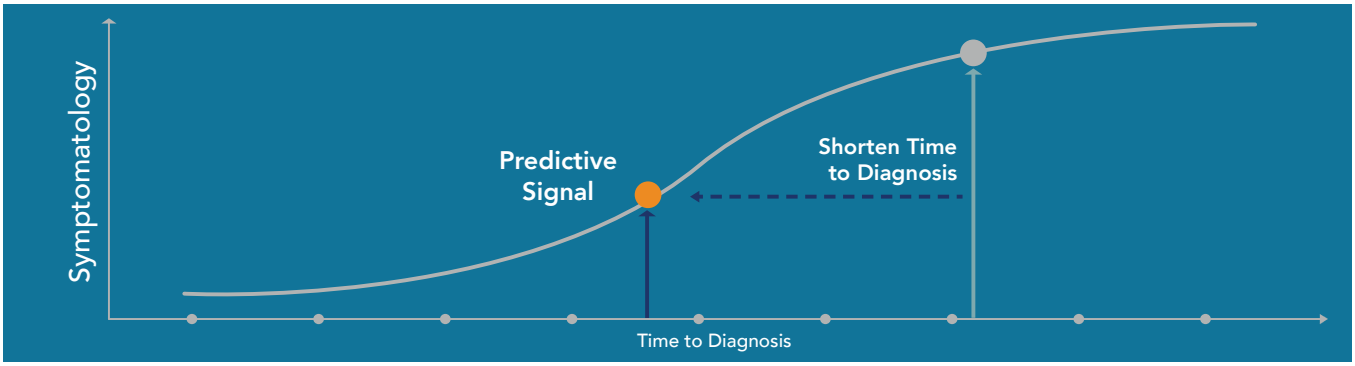
Q: From a data science perspective when we talk about analytics being used in the healthcare setting what does that mean? What is happening behind the scenes?

A: Massive volumes of healthcare data, petabytes and exabytes, are being matched with predictive and machine learning algorithms. These healthcare claims data repositories are mathematically compatible in terms of ingestion and scalability with the vast collection of machine learning and predictive algorithms available. This includes a suite of algorithms ranging from logistics regression to numerous variants of deep learning algorithms. It is the combination of a continuously growing amount of healthcare data with predictive technology that allows unexpected patterns and relationships to be uncovered. This fusion of medicine and technology has created the perfect storm for disruption in the healthcare space.

Q: How can predictive modeling impact the time and accuracy of rare disease diagnosis?

A: Searching for a rare disease diagnosis is often a frustrating process that can take many years. Applying predictive modeling techniques to healthcare data has accelerated the accuracy and speed of rare disease diagnosis. Imagine two scenarios, one where a physician (human) is diagnosing a patient and the other where a predictive algorithm (machine) trained to detect rare diseases is assisting a physician in diagnosing a patient. In first case, the lone physician relies on her medical knowledge and years of experience treating patients. But keep in mind that a physician may only see one rare disease patient (at most) in her entire career given the prevalence of rare diseases. Contrast this scenario with the one where the physician is leveraging the 'learnings' of a predictive algorithm (or model) that has been 'trained' on rare diseases. This predictive model during its training phase has learned the noisy patterns of care from the longitudinal health records of 100's of millions of rare disease and healthy patients. It can use this information to determine if the patient looking for a diagnosis matches the noisy patterns of care of a rare disease patient. During the diagnosis of a patient, this predictive model provides insights on the patient being diagnosed. The model would be able to alert the physician to the similarities of the patient to any one of 1,000's of rare diseases. Unlike the predictive model, the average physician cannot glean from her finite exposure to patients (numbering in the 1,000's) the subtle signs and symptoms indicative of a rare disease that develop over time. How can predictive modeling impact the time Q and accuracy of rare disease diagnosis? A 4 Using predictive modeling by leveraging healthcare data assets available today can significantly accelerate the accurate diagnosis of rare disease patients. With the right predictive model and data resources, it is possible to find patients hidden in healthcare databases and diagnose certain rare diseases in weeks compared to years. Imagine what this could do for patients and their families who are waiting for answers. The figure below is an illustration of the impact of earlier diagnosis with the assistance of predictive models.



Q: What are the key data sets being analyzed and signals being detected to identify undiagnosed patients? How can machine learning and predictive modelling impact this process?

A: Leveraging technology allows therapies to proactively find patients rather than patients needing to seek out therapies. Machine learning, and more specifically knowledge transfer techniques, enable the integration of seemingly disparate data sets to provide greater insight into patients' journeys and their disease progression. These data sets include healthcare administrative claims data, EMR/EHR data, genomic data, imagery, consumer, environmental and geospatial data. Building predictive models that deliver high fidelity predictions and actionable signals is of significant value. The end goal in leveraging predictive modeling is to derive a set of actionable signals that directly lead to an earlier accurate diagnosis and improved patient health. Given the current availability of large databases, machine learning and predictive modeling techniques generate insights from the data without any user bias or intervention. For example, it is now possible to determine the specific set or collection of data elements that are present months or years before a patient is diagnosed with a rare disease. These data elements are collected in an administrative claims database and are thus actionable in the sense that the treating physician can be identified who can then intervene to positively change the trajectory of a patient's health journey.

A: The amount of healthcare data collected is growing exponentially and shows no sign of slowing down. A typical analyses of healthcare data can determine the incidence and prevalence of a disease and indicate the number of patients with that specific diagnosis. However, incidence and prevalence based on real-world data now drives a number of business decisions such as, how many patients are available to support the investment required to develop the drug, execute a successful clinical trial, and bring the drug to market and whether an orphan designation can be sought for a therapy. Access to orphan drugs requires both awareness and education. The data sources and machine learning and predictive modeling tools enable the discovery of undiagnosed rare disease patients. Discovering undiagnosed rare disease patients drives efforts to increase awareness to all stakeholders. The most effective technique to raise awareness is to precisely target educational efforts that will have the greatest impact on getting undiagnosed rare disease patients on the right therapy.

Q: How are new data sources and analytic tools going to impact the way rare disease patients access orphan drugs?

Q: Technology continues to become more sophisticated and there seems to be no end in sight. Where is healthcare technology headed?

A: Technology is and will continue to be an enabler. The deployment of technology solutions will continue to accelerate. In addition, the promise of reduced healthcare costs and improved patient care will serve as the driver of technology solution adoption. Technology will allow healthcare to be conducted at the edge, meaning at home, in the community and outside of healthcare facilities. Routine health and wellness checkups will be significantly technology driven and will be a resource of patient generated data. One ubiquitous example is smartphone health applications that can capture various data points including physical activity, heart rate, ECG, blood glucose levels and measure gait. This is data that can be collected and processed continuously with the ability to provide real time feedback to patients. Overlaying this data generation and collection process, the application of machine learning and predictive modeling will enable proactive interventions reducing costly treatments. Other examples of patient generated data are digital therapeutics and potentially, smart speakers and electronic personal assistants. Healthcare technology is headed towards more patient autonomy and independence with less reliance on healthcare providers for routine care. The democratization of healthcare data is taking place that will enable, for example, the integration of personally generated data (smart home and smart personal devices) with data collected by healthcare professionals and an individual consumer's purchasing patterns. In addition, predictive modeling will enable more effective and prospective interventions that will lead to better disease management and reduced cost.

About Oodaye Shukla

Oodaye Shukla is the Chief Data and Analytics Officer at HVH Precision Analytics. He has broad experience in the Intelligence Community, Healthcare, Telecom and DoD industries spanning over 20 years.

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