Precision (Personalized) Medicine:

Data, Applications, Benefits and Challenges

This is the second article in a series focused on understanding Precision Medicine. In this offering, we discuss the different types of data that can be used to build a Precision Medicine program and impact member/patient outcomes.

Much of the success in reducing morbidity and mortality over the past half century has been due to earlier detection of disease, in tandem with public health measures such as smoking cessation and weight management; as well as wider application of proven therapies for primary and secondary prevention.

Fast forward to today, the significant advances in data collection and data platforms (including the advent of AI and sophisticated algorithms) have raised interest in leveraging expanded data and information to create a more comprehensive (all-encompassing) view of individual risk markers and other health risk characteristics outside the realm of more traditional clinical (biometric) measures. These data can be derived from various sources including electronic health records (EHRs), genetic markers (genomics), wearable devices, patient surveys, lifestyle information, environmental factors, and even social media. The aim is to create a comprehensive picture of an individual's health profile to deliver a treatment plan that is more likely to yield the best outcomes, as well as preventative strategies.

Expanded Data Types Collected					
Clinical Data Medical history, diagnoses, medications, lab results, imaging reports – all typically sourced from EHRs	Genomic Data DNA sequencing information providing insights into genetic variations that may influence disease risk	Biometric Data Measurements like blood pressure, heart rate, weight – collected via wearable devices or clinical assessments	Lifestyle Data Diet, exercise habits, sleep patterns, stress levels – often gathered via patient self-reports	Environmental Data Exposure to pollutants, air quality, geographic location	Social Determinants of Health (SDOH) Data Socioeconomic factors like income, education, access to healthcare

There have emerged two pathways for moving forward with leveraging expanded data to both improve outcomes stemming from more targeted application of known tried and true therapies, along with opportunities to get ahead of disease and illness with similar targeting of individuals at expected risk.

These two complementary pathways include: relying mainly on biometric and clinical data sometimes combined with genetic testing results versus leveraging greater availability of attributes outside of the realm of biometrics that reflect

not just a person's biological footprint, but also the influence of lifestyle, environment, and social determinants of health.

Moreover, with digital trackers and devices such as the Apple watch, there is no longer the need to simply rely on self-reported actions and behaviors; for example, data recounting the number of steps taken per day, number of hours of sleep, evidence of heart rate variability are now readily available for harvesting.

Rather than getting caught up in trying to tease out differences in the data or source information used to establish a more personalized evaluation of individual health are risk, we can adopt a working definition that encompasses both approaches, as they are more complementary than contradictory. Sometimes it is simply more about what information is available and to a degree, what the specific application or goal is.

Here is a simplified working definition that avoids the debate on what exactly constitutes "precision medicine":

"Precision medicine is a medical model that tailors healthcare to individual patients based on their unique characteristics. These characteristics can include genetic, environmental, and lifestyle factors. The goal of precision medicine is to provide more accurate diagnoses, improve treatment outcomes, and reduce adverse effects by customizing medical care to each individual's unique and specific needs."

Regardless of the approach taken, there are several key concepts that serve as a good foundation when beginning to contemplate moving forward: the role of genomics, translation into working models, integrated data and analysis.

The Role of Genomics

The first set of considerations include taking advantage of advances in genomics and genetic profiling to be used in tandem with more traditional biomarkers.

These advances include **genomic sequencing,** which involves analyzing an individual's genetic code to identify mutations or variations that may influence disease risk, progression, and response to treatment; think popular subscription programs like 23 and Me, AncestryDNA, and CRI Genetics.

There is also the relatively new field of **pharmacogenomics,** or the study of how genes affect an individual's response to medicines. This enables the selection of drug therapies that are more likely to be effective and less likely to cause serious side effects. A well-known example involves the use of statins for management of high cholesterol. Use of biomarkers along with review of individual risk factors (e.g., lifestyle, diet) for potential adverse reactions, can enable tailoring of therapies combining drugs from different classes as a way to address risk while allowing these individuals to benefit from treatment.

Translation of the output of genomic analyses into data to be combined with more traditional biomarkers such as blood pressure, pulse, blood and radiographic testing allows for a broader lens then can be obtained simply by knowing an individual's current biological status as obtained via a clinical examination or cursory review of family history and recent lifestyle changes or environmental exposures.

Transforming genomic sequencing from a diagnostic tool for patients who already have an illness, to one that can find known, treatable diseases that have not produced symptoms yet is an exciting prospect.

This type of applied Precision Medicine was put to use during the Covid-19 pandemic through use of various biomarkers such as IL-6 and c-reactive protein (both derived from simple blood assays) in assessing severity of disease, and as part of the development of therapies such as Paxlovid and determining the efficacy of vaccination. Precision Medicine is also finding its place in management of infectious diseases, chronic diseases such as asthma, cardiovascular disease, diabetes, and obesity.

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Exciting new developments in treatment and management of cancer have emerged from mitigation of conventional adverse effects of chemotherapy, and application of immunotherapy where knowing more about an individual's immune system can be used to help control tumor growth by stimulating an individual's own immune system to fight disease progression.

Translation into Working Models

To make practical use of the combined data for risk assessment, the output of genetic and other data needs to be able to stratify individuals into different risk categories, enabling more precise prevention and management strategies (including subgroups that are more likely to benefit from specific treatments). Today only a small proportion of individuals at high genetic risk for specific conditions know their status, which means they are missing out on recommended screenings.

Integrated Data and Analytics

New avenues for integration of diverse data and datasets as well as advances in analytics and machine learning can be a topic in and of itself, as it pertains to precision medicine. But for our purposes here, the following are the major categories of effort involved.

- **Big Data and AI:** Utilizing large datasets and advanced analytical tools, including artificial intelligence and machine learning, to uncover patterns and make predictions about disease and treatment outcomes.
- **Electronic Health Records (EHRs):** Integrating data from EHRs to provide comprehensive **patient** profiles that inform personalized care plans.
- **Social Determinants of Health (SDOH):** These are the non-medical factors that influence health outcomes, largely shaped by the environment in which people live and work. SDOH encompasses a wide range of conditions and influences that can either promote or hinder health status, well-being, and quality of life.

To date, SDOH and their correlation to health risk and outcomes has translated into the most widely accepted application of "non-clinical" factors that can help predict and categorize individuals with regard to factors outside of more transitional models of health and illness. We will discuss this topic further in an upcoming article that will delve further into SDOH and practical applications.

In conclusion, numerous challenges still exist influencing the future of Precision Medicine such as the cost involved, ethics, Big Data security concerns, and complexity of merging data and building algorithms. However, the promise and wide potential application merits investments to overcome these challenges and concerns.

At the Well Solutions Group and DataWELL Informatics, we have extensive expertise in understanding population health data and developing analytics to stratify populations for which specific programs can be developed. Our approach is comprehensive and specific, focused on improving member/patient outcomes and reducing payer financial impact.

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