At Illumina, we’re committed to enabling precision health strategies that use clinical and population data, genomic sequencing, and advanced analytical techniques to help people enjoy longer, healthier lives. Precision health approaches help health systems achieve their quadruple aim by expanding access to personalized treatments, reducing undue burden for all, and improving quality of life for everyone.

Total Healthcare Expenditures

Primary Care 5.4%

To explore the implementation and impact of a real-life example of genomics-powered precision health in action in primary care, we spoke with Peter Hulick, MD, the Janardan D. Khandekar, MD, Chair of Personalized Medicine, and the Medical Director for the Mark R. Neaman Center for Personalized Medicine at NorthShore University Health System.

“Genomics is disrupting healthcare,” says Dr. Hulick. “As costs continue declining rapidly, we can use these novel genomic technologies to deliver better care and engage patients deeply. Our hope is to prevent disease when possible, and otherwise offer precision treatments based on the biology of the disease combined with a

Introducing Broad Genomic Screening to Improve Primary Care

While some personalized medicine pioneers begin their journey with an initial focus on oncology or rare undiagnosed diseases, Dr. Hulick and the team at NorthShore launched a strategic precision health initiative by implementing a genomic screening program as part of routine primary care. A recent review found that general health checks have a positive impact on patient-reported outcomes even when they do not reduce mortality or cardiovascular events. Participants in NorthShore’s screening program submitted DNA for tests that detect genetic markers associated with cancer risk, cardiovascular conditions, or pharmacogenomic outcomes.
We recognized the potential to maximize our interventions and optimize health guidance through genomic screening, not only for the patients in front of us, but also for their families,” says Dr. Hulick. Many CDC Tier 1 Conditions, such as hereditary breast cancer and familial hypercholesterolemia, are currently not screened for adequately. Proactively identifying the genetic risk factors for these otherwise healthy patients can help them and their families make informed health decisions and seek life-changing preventative care.

“We felt that this initiative could create a differentiated and elevated experience for our patients and for the health system,” says Dr. Hulick. To realize this potential, they made sure to gain insight and buy-in from every stakeholder, from administrative leaders and clinical providers to patients and community members. They focused on education efforts to boost understanding of novel genomic technologies' potential and made sure to build agreement around promising application areas, goals, milestones, and areas for improvement and reinvestment.

Promising Results from the Initial Genomic Screening Program

NorthShore's initial population screening pilot efforts revealed the importance of eliminating traditional barriers such as cost and family history that have historically limited eligibility for these types of programs. Physicians and administrators also learned best practices for obtaining consent, developing cost-sharing models, and integrating results into patients’ records.

Within the NorthShore healthcare system, 99 percent of primary care physicians ordered genomic testing for at least one patient, with one third of eligible patients enrolling in the screening program at no cost. To assess the initial genomic screening program in more detail and glean lessons to apply in the future, Dr. Hulick and his colleagues systematically surveyed physicians and patients who participated. Physicians reported high satisfaction with the program, which they saw as a differentiator for their practice that showcased their commitment to providing cutting-edge healthcare for their patients. 77 percent of physicians agree that genetic testing is useful in determining patient management, while nearly half indicated that they are more likely to follow screening recommendations after participating in the program. Meanwhile, 87 percent of patients reported overall satisfaction with their experience of genomic screening and any resulting clinical actions.
Building on Success

Based on the outcomes of their pilot screening programs, NorthShore decided scale up and deploy genomic screening offerings to their entire healthcare system. “We now have over 20,000 patients who have pharmacogenomic information in their Electronic Health Record in a structured way,” says Dr. Hulick. “We’ve done a lot in a relatively short amount of time, but our journey is certainly not over.”

NorthShore is further expanding their efforts to bring genomic medicine to the masses through their Advanced Primary Care initiative, which aims to address disparities and break down inequities that currently exist in personalized medicine. This program will go beyond traditional single-gene conditions and incorporate new information, such as polygenic risk scores, into screening results. The ultimate mission is not just to perform individual tests but also aggregate and analyze data on a larger scale as part of a Learning Health System model.7

“Genomics transcends and crosses all departments and divisions across our organization,” says Dr. Hulick. “It has been an exciting challenge to scale the complexity of genomics in a cost-effective, seamless, and sustainable way across our mid-sized health system.”

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