Pioneer Spotlight

Intermountain Healthcare

A precision health approach has the potential to drive better value and quality in health care. Precision health is the intersection of disease prediction and prevention with personalized treatment. By amplifying the role of big data in disease prevention and detection, precision health zeroes in on the various factors that help maintain health throughout life. At Illumina, we strongly believe that genomics-powered precision health has the potential to improve patient care, deliver better healthcare outcomes, and open new venues for health care discoveries and insights.

Lincoln Nadauld, MD, PhD Vice President and Chief of Precision Health & Academics at Intermountain Healthcare



We believe that precision medicine has the ability to improve clinical outcomes while simultaneously reducing costs

To dive into details of their clinical implementation of precision health approaches, we spoke with Lincoln Nadauld, MD, PhD, a practicing oncologist who serves as Vice President and Chief of Precision Health & Academics at Intermountain Healthcare. Though there is no one-size-fits-all approach to implementing precision health, his insights can inform and guide others embarking on their own journey.

"We believe that precision medicine has the ability to improve clinical outcomes while simultaneously reducing costs," says Dr. Nadauld. He sees precision medicine with genomics as a population health management tool something that can help teams of healthcare professionals improve outcomes for groups of individuals by designing proactive care strategies.

Getting Started with Precision Medicine -**Precision Oncology with Genomics**

Intermountain began their precision health journey by applying precision medicine in Intermountain's Oncology Clinical Program. After observing individual successes, the team at Intermountain ran studies to systematically compare precision oncology to standard chemotherapy or best supportive care. After several years, they found that genomic testing and targeted therapy doubled patients' average survival to 52 weeks while decreasing healthcare costs per patient by an average of \$700 per month.^{1,2}



Our findings of improved survival rates and decreased overall healthcare costs ultimately led to changes in our health plan's coverage policies," says Dr. Nadauld. "Any patients with advanced cancer who are covered by our system's health plan can now receive comprehensive genomic profiling.

From there, Intermountain continued expanding their applications of precision health. The next step was focused on inherited cancer conditions. After reviewing genomic data from nearly 300 cancer patients within the health system, they found that nearly 15 percent—regardless of tumor type or stage—have an inherited cancer-related gene mutation, in line with findings by other institutions.^{3,4}

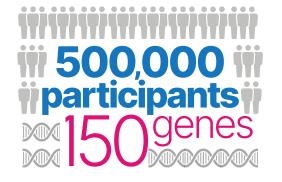


15% of patients have inherited cancer-related gene mutation. Opportunity to discover additional germline variants in the other 85% of patients.

"There are probably additional germline variants in the other 85 percent of patients; we just haven't defined them yet," notes Dr. Nadauld. Based on these findings, all newly diagnosed cancer patients are also getting germline testing, which according to Intermountain's soon-to-be-published data does not increase the overall cost of care.

Moving beyond oncology, Intermountain has also expanded pharmacogenomic testing in primary care, implemented rapid whole-genome sequencing in the neonatal intensive care unit, offered non-invasive prenatal testing (NIPT) to detect chromosomal abnormalities during pregnancy, and recently started exploring polygenic risk score as part of managing care in their cardiovascular clinic. Even more clinical services throughout the Intermountain health system will soon incorporate genomics as a standard component of delivering the highest possible quality of care.

HerediGene®: **Population Study**



Whole-Genome Sequencing for Everyone

Throughout the precision health journey, Dr. Nadauld says he used to joke that "everyone should have their whole genome sequenced." But he's not joking anymore. Intermountain recognized that they had reached an inflection point where a population-scale genomics implementation would serve both the patients and the health system. Recently, Intermountain announced the HerediGene®: Population Study, which ultimately aims to enroll 500,000 individuals to undergo genome mapping. Patients who participate, along with their care teams, will receive information on about 150 genes with known clinical impact.

As of October 2021, The HerediGene®: Population Study enrolled more than 81,700 individuals, performed genetic screening on most enrollees and mapped the genomes for ~28,000 already. Through this initial review, the team identified 255 individuals who carry pathogenic variants contributing to CDC Tier 1 conditions—a proportion in line with other large-scale studies.^{5,6} As these results have immediate clinical implications for family members, they also anticipate 1,020 cascade tests. By start of 2022, they ultimately expect more than 1,000 Tier 1 results and another 4,000 cascade tests that will identify individuals in their catchment area who need a personalized-approach to standard of care at Intermountain Healthcare.



Among ~28,000 genomes reviewed, 255 individuals were found to carry pathogenic variants.

Reaping the Benefits of Precision Genomics

Dr. Nadauld says the implementation of precision medicine at Intermountain has created a sort of overarching "halo effect" that is driving growth and sustainability throughout the health system. Intermountain has increased its market share amongst newly diagnosed patients, and providers within the system are enjoying a heightened atmosphere of innovation, which in turn has improved recruitment amongst new physicians and scientists.

With successes like these on the leading edge of genomics-powered precision health, there is great promise for other health systems applying these strategies and lessons to meet the unique needs of their own patients and populations.

For more insights from Dr. Nadauld, check out this webinar.

Explore our website to discover how Illumina can support your precision medicine journey.

¹ Haslem, D. S. et al. Precision oncology in advanced cancer patients improves overall survival with lower weekly healthcare costs. Oncotarget 9, 12316-12322 (2018).

² Haslem, D. S. et al. A retrospective analysis of precision medicine outcomes in patients with advanced cancer reveals improved progression-free survival without increased health care costs. J. Oncol. Pract. 13, e108-e119 (2017).

³ Bertucci, F. et al. Prospective high-throughput genome profiling of advanced cancers: results of the PERMED-01 clinical trial. 1-20 (2021).

⁴ Cobain, E. F. et al. Assessment of Clinical Benefit of Integrative Genomic Profiling in Advanced Solid Tumors. 48109, 1–9 (2021).

⁵ Dewey, F. E. et al. Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science (80-.). 354, (2016).

⁶ Grzymski, J. J. et al. Population genetic screening efficiently identifies carriers of autosomal dominant diseases. Nat. Med. (2020) doi:10.1038/ s41591-020-0982-5.