

GENOMICS & HUMAN HEALTH

THE RIGHT TREATMENT AT THE RIGHT
TIME FOR THE RIGHT PATIENT



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GENOMICS & HUMAN HEALTH

Genomics is probably most commonly associated with human health. With the mapping of the human genome, we now know of the roughly 20,000 genes that make us who we are. And each day, as the costs of sequencing goes down, we understand a little more about what each of those genes do to control the proteins and ultimately the cells in our bodies.

Genomics has tremendous potential to help us manage our health better. From understanding our genetic predisposition to a disease, to providing earlier and more accurate diagnosis, to pinpointing the drugs that are best suited to the genetic variation of disease, genomics is playing a vital role in moving us from a 'one size fits all' approach to healthcare, to 'personalized medicine' where our prevention, care and treatment are informed by our personal genome.

PATIENT STRATIFICATION

Genomics can inform how we assign patients for care. It can create efficiencies and reduce unnecessary, expensive and time-consuming treatments.



For example, there has been a lot of talk about Herceptin, a costly drug for cancer treatment that is highly effective on the roughly 20% of breast cancers where women possess a mutation of a protein called HER2. National guidelines now require HER2 testing upon diagnosis of breast



cancer to determine whether treatment with Herceptin is warranted, saving both the patients and the health care system wasted time and money.



DIAGNOSTICS

Early and accurate diagnosis of disease is pivotal to effective interventions.



Genomics can help to validate the presence of disease, as well as pinpointing what variation of that disease a patient has. For instance, a genetic mutation is responsible for a deadly heart condition in families in Newfoundland and Labrador. This information has led to a diagnostic test that helps doctors definitively determine who has the condition, and proactively implant a defibrillator that will restart their heart when it stops.

ADVERSE DRUG REACTIONS

Adverse drug reactions cost the Canadian system \$15B/year in medical treatments and hospital visits. But genomics can help us assign the right drug to the right person at the right time.

For example, through genomics we know that some post partum mothers carry a genetic mutation that makes them process the painkiller codeine into morphine much faster than normal. When that is transmitted to their breastfeeding newborns, the effect is lethal. When this new genetic information was revealed, guidelines around the use of painkillers for new moms were revised, and countless babies are alive today because of it.



WHAT IS PERSONALIZED MEDICINE?

Personalized medicine is an emerging practice of medicine that uses an individual's genetic profile to guide decisions made in regard to the prevention, diagnosis, and treatment of disease.

With the many advances in our understanding of the human genome since the Human Genome Project completed in the first part of this century, we now know much more about the genetic make-up of our bodies in health and in illness.

Knowledge of a patient's genetic profile can help doctors select the proper medication or therapy and administer it using the proper dose or regimen. Although in early stages, the development of so-called 'designer drugs' based on this knowledge could also prove to be a game-changer for the health care industry.

EXAMPLE: LUNG CANCER

Lung cancer remains the leading cause of cancer-related death worldwide accounting for over a quarter (27%) of all cancer deaths each year. In Canada, there are roughly 25,300 new cases of lung cancer diagnosed each year, with over 20,000 resulting in death.

Although surgery is one of the most effective treatments, most lung cancer patients are diagnosed too late for surgical intervention. For these patients platinum-based chemotherapy and radiation therapy, alone or in combination, become the treatment of choice. Unfortunately, the five-year survival rate is only about 15%, and has not significantly changed in the past several decades.

Thankfully, lung cancer shows great promise for gene-specific therapy. Through a pilot clinical diagnostic service in Halifax, genomics is helping us find the genetic markers that identify subgroups of patients that are more likely to benefit from targeted therapies. Early results show that those whose treatment is correlated with the genetic mutation behind their specific type of lung cancer are showing marked improvements in surviving this terrible disease.

