Stephen Scherer, head of Applied Genomics at Toronto's Hospital for Sick Children and project leader for one of Genome Canada's Genomics and Personalized Health projects, says that "Many countries are looking to us." Photo: Robert Teteruck, The Hospital for Sick Children

Canada's Advantage in Genomic Medicine

Stephen Scherer

In the early 1990s, Canada was not considered to be in the top 10 countries in genome science. Today, we are recognized as in the top five, arguably the top three. Canada's health care system offers unique opportunities for the development of genomic science that have already delivered results, with breakthroughs in autism, cardiac disease and drug reaction research. With those successes come great expectations, which Canada is poised to meet.

There is an emerging technology that promises to change health care for Canadian patients. It may be the most powerful and comprehensive diagnostic tool in the history of medicine. It is fueling exciting new scientific discoveries. Canada is emerging as a world leader in the field. As a society, we need to ensure that our science investments, health policy and ethical frameworks keep pace with genomic science.

Genomic sequencing is becoming faster, cheaper and better—a trend that

can deliver significant health benefits to Canadians. I have been a part of this transformation throughout my career, and yet, there are still moments when I am awestruck. This past September, The Hospital for Sick Children in Toronto recognized the 25th anniversary of one of its major research achievements. In 1989, Lap-Chee Tsui's research laboratory discovered the gene that causes cystic fibrosis. As a graduate student on that team, I observed the long hours of making small, steady incremental contributions towards this discovery. Talking with my former mentor, we marveled at how, back then, identifying a cystic fibrosis gene took more than five years of painstaking work. Using today's technology, it would take us a week.

This acceleration in science is about more than boosting the ability of researchers to uncover the nature of inherited diseases. Today, we can know our own personal genetic inheritance. Put another way: If you came to my laboratory on a Monday and spit in a cup, I could show you your full genomic sequence by Friday. That knowledge could have profound health implications for you, your siblings and your children. Increasingly, Canadians want to know such detailed information about themselves, as do their physicians. There are important ethical considerations, but by proceeding carefully and confidently, Canadian researchers can deliver scientific gains and help strengthen health care.

Genetic sequencing can often confirm a medical diagnosis, saving critical time. It is helping to tailor treatments and therapies for some types of cancer. It is used to find rare diseases in patients. At the McLaughlin Centre at the University of Toronto, one personalized medicine project isolated genetic variants that cause congenital heart defects, a condition found in 1 per cent of children. As a scientist, one of my own research interests has been to identify the formula for diagnosing autism spectrum disorder at an earlier age. This will help create more advanced genetic diagnostic tests enabling earlier detection and behavioral interventions helping children. In Canada, we are uniquely positioned to reap such benefits in genome science and personal medicine. Our universal health care system creates both opportunity and incentive to continue investing in this field. Such investments often also seed other international investments with a notable example being my own laboratory's leadership role in the international effort to sequence the genomes of 10,000 families with autism.

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There is an opportunity because we have a relatively stable patient population. People and families do not move around among health providers as much as they do in the United States. Everyone has access in one of the world's most ethnically diverse, and therefore genetically diverse, countries. A universal health care system is also well-suited to collecting and utilizing the vast amounts of data that are essential to genetic discoveries in health care.

As for incentive, genome science can make the system work better for patients and doctors, while helping to sustain the universal system itself. Right now, a physician orders a test to look for genetic evidence of a specific inherited condition that could be causing the symptoms they have observed in the patient. If they fail to confirm a diagnosis, they order more tests.

However, the cost of full genome sequencing is also decreasing, and we anticipate in 2015 it will be in the $1,000 range. Sequencing a patient's full genome could often remove the need for multiple tests. There is less trial and error. Plus, it would likely contribute to improved care. Sequencing the full genome often uncovers genetic evidence for inherited conditions that had not yet been identified by the clinician. In many cases, these conditions were contributing to the patient's symptoms, and so treatment can be improved.

To fully realize the benefits of genomics in health care, important policy conversations need to happen. For one, personal health information of patients who undergo full genome sequencing need to be protected. Canada has not yet enacted genetic non-discrimination laws, as have many other countries. With Senate hearings ongoing there does look to be good progress towards this end.

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will need to consider impacts on the patient relationship.

Of course, policy is not up to me, but as a scientist and leader of an organization dedicated to advancing biomedicine, I can say this: Canada cannot easily import the genomic science and solutions it needs. We have to do it ourselves because our health care system and the population we support are unique to us. The truth is, there are few countries Canada can to look to for an example. We're already one of the leaders. Many countries are looking to us.

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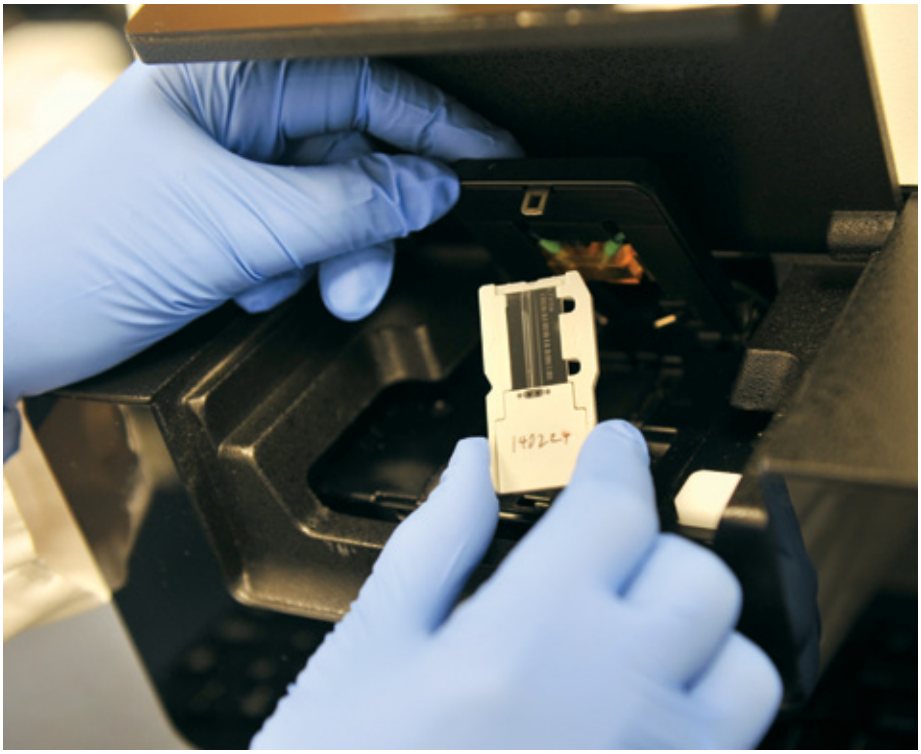

A researcher loads a sample onto a next-generation genome sequencer at the Genomics Innovation Centre at the BC Cancer Genome Sciences Centre. Photo courtesy of Genome Canada

Canadian governments have made solid commitments to boosting national genomics research capacity. Led by Genome Canada and six regional Genome Centres, Canada has made a significant and sustained investments. In the early 1990s, Canada was not considered to be in the top 10 countries in genome science. Today, we are recognized as in the top five, and I would argue top three. When I attend international conferences, everyone is talking about what is happening across Canada.

Such leadership comes with important obligations. Expectations are rising. Soon after identifying a new disease-causing genetic mutation, people start asking about a cure, especially those affected by that condition. It is understandable, but these types of advances are, unfortunately, coming more slowly than the diagnostic advances. There have been notable successes in treatment. In one Canadian example, defibrillators were implanted in people who tested positive for a fatal gene that causes sudden cardiac death. Furthermore, genetic testing is already leading to a reduction in adverse drug reactions, which cost billions of dollars to Canadian governments annually.

There is also encouraging progress on drug development. One of my colleagues at the University of Toronto, Aled Edwards, is chief executive of the Structural Genomics Consortium. They bring together scientists in academia and the pharmaceutical industry to discover and validate targets for new medicines.

Genomics technology is already allowing health providers to do things in medicine that have never been done before. As these new tools improve wellbeing, it can unleash human potential in ways never imagined before. That is the true beauty of health research breakthroughs. They yield exponential returns outside the hospital and the laboratory. People of all ages, living longer and healthier lives, contribute to our communities in the arts, science and the economy. They are parents, friends and mentors who enrich the lives of others. It is impossible to measure, but we know from our own life experience that such value exists. It is the reason Canadians treasure their universal health care system. Genomics can help sustain and strengthen that system.

We are on the right track. Canada needs to keep moving forward with confidence; advancing science, adding to humanity's storehouse of knowledge and continually improving care for Canadians and people throughout the world. P

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