Personal Genome Project Canada study results show promise for health care in Canada

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First results from the Personal Genome Project Canada, which sequenced the entire personal genomes of 56 healthy participants, suggest whole genome sequencing can benefit health care in Canada, according to results published in *CMAJ (Canadian Medical Association Journal)*.

"The technology is here now to give us a $1000 genome, we will be at a $100 genome very soon, and everyone may have this done," says Dr. Stephen Scherer, Director of the University of Toronto's McLaughlin Centre and The Centre for Applied Genomics (TCAG) at The Hospital for Sick Children (SickKids). "Family physicians will be involved in interpreting and communicating genomic information to patients in the context of personal and family histories. This requires awareness of the range of potential findings and the strengths and limitations of the tests."

One-quarter (14) of the 56 adult participants who had their entire personal genomes sequenced had genetic variants associated with diseases such as cancer, cardiovascular disease and neurological disease. Thirteen participants (23%) were at risk of potentially life-threatening adverse drug reactions. All had medically relevant findings, including genes for drug reactions, copy number variation of genes involved in mental health conditions, and other genetic alterations that could affect the health of future generations of an affected individual's family.

Individual findings with health implications:

- A cancer-linked BRAC1 genetic variant was identified in a 65-year-old man whose father had died of prostate cancer.
- Gene sequencing found a variant in a 49-year-old man that is associated with a manageable condition causing kidney stones or crystals.
- Chromosome loss in blood cells linked to Turner syndrome was found in a 54-year-old woman who had no clinical manifestations; screening for cardiac or endocrine disorders is recommended in this situation.

Participants were highly engaged in the research study, providing full consent to publicly communicate results. They were offered sessions with genetic counsellors, who are specialists in delivering complex, often nuanced, genetic information, to receive their results in context.

The CMAJ study is the first research paper published since the project's inception in 2007 and has implications for government.

"Canada as a nation does not yet have a national sequencing project for precision medicine like England, the US, Australia and many other countries," says Dr. Scherer. "Our Personal Genome Project could be scaled up, and there are other ways to leverage it as has been done in the US, England and others."

He adds, "We selected CMAJ because we wanted to tell the 'genome sequence' story in the manner most relevant to Canadian medicine, since by definition genetics is personal, and each health care jurisdiction needs to deal with their citizens' data in its own way."

The Globe and Mail has been an active follower of the Personal Genome Project Canada, covering topics on personal genomic screening research.

A 2012 Globe series helped to attract and survey volunteer recruits to the Personal Genome Project Canada and raise awareness of Bill S-201, Canada's Genetic Non-Discrimination Act, which is now law.

In a related commentary, Dr. Robert Hegele, Schulich School of Medicine and Dentistry, Western University, London, Ontario, writes that "the study highlights the potential of widespread implementation of whole genome sequencing, including early diagnosis and perhaps an opportunity to prevent or delay disease-associated outcomes or complications, screening of family members and implications for family planning."

Misdiagnosis of disease because of misclassified DNA variants, risk of incidental findings, diagnosis of diseases that have no treatments, and increased resource utilization triggered by a positive diagnosis are all potential pitfalls with this technology.

However, "the potential benefits deserve further careful and intensive evaluation, particularly within the Canadian context," he concludes.

More information: The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants, *CMAJ*, February 05, 2018 190 (5) E126-E136; DOI: doi.org/10.1503/cmaj.171151

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