Tackling the Root Cause of Disease

By Rob Abbott

The American philosopher and scholar, Henry David Thoreau, once famously observed that “there are a thousand hacking at the branches of evil to one who is striking at the root”. I think of this whenever I meet a Canadian geneticist toiling in the shadows of the health care system he or she wants to improve. Let me explain.

While many Canadians may not know it, there is a revolution underway in medicine and geneticists are leading the charge. Medical genetics, once a tool for diagnosing a handful of relatively rare diseases inherited from a parent, has rapidly expanded into new territories – the prediction of a healthy person’s risk of developing even common diseases such as cancer and cardiovascular disease; the analysis of patterns of gene expression as a complement to conventional diagnostic methods; and the evaluation of multigenic diseases and responses to environmental agents and drugs. Further, knowledge about of the genomes of microbes is rapidly expanding the opportunities for diagnosing, preventing, and treating infectious diseases.

The most revolutionary development, however, has been the work of the Human Genome Project (HGP). Knowledge gleaned from the HGP is making it possible to identify individuals at risk for disease, to diagnose and treat disease in ways that until recently were not possible, and to improve health through early diagnosis, health promotion activities, more targeted treatments and increased understanding of prevention. It is fair to say that biology in the post-HGP age has become an information science with the potential to transform human understanding in ways that were heretofore unimagined.

All of this means better health outcomes for Canadians, allowing more of us to live lives of access and opportunity unburdened by disease; and new forms of economic activity as knowledge is translated from the university lab into biopharmaceutical companies. Oh yes, the medical genetics revolution is also a novel approach to the containment of health care costs – upwards of $100 billion annually in Canada. So, you’d think medical geneticists would be feted like celebrities. Sadly, the vast majority struggle for recognition and funding. The Canadian Gene Cure Foundation (CGCF) wants to address this; the CGCF

wants to help these extraordinary people do their best work. More important, CGCF wants to help the millions of Canadians who currently live with a genetic disease. At least 3 million Canadians suffer from one or more rare or orphan diseases – ailments that individually affect 1 in 10,000. A much larger number of Canadians, estimated by the eminent scholar Charles Scriver at 60% of the population, will be affected by more common genetic diseases at some point in their life. This is a national challenge that should unite us all, from sea to sea to sea. We can find cures, we can improve people's lives.

The CGCF (www.jeansforgenes.ca) has long recognized the importance of supporting genetic disease research. Through its annual "Jeans for Genes Day" campaign, the foundation raises money to fund research into both rare diseases such as Purine Nucleoside Phosphorylase deficiency, a disease that affects the immune systems of children; and Downs Syndrome, a more common genetic disorder which results in mental disability, heart defects, hearing loss, obesity, diabetes, and Alzheimer's disease.

This year's Jeans for Genes Day will be held on October 12. On that day, I encourage all Canadians to buy the distinctive pin, wear their jeans, and show they care enough to contribute to the search for the root cause of disease.

Rob Abbott is the CEO of the Canadian Gene Cure Foundation