Perspectives

CCMG statement on gene patents


Conflict of interest

All authors declare that they have no conflict of interest.

Statement development

This statement was developed by the CCMG Ethics and Public Policy committee with input from the membership-at-large. It reflects the general opinion of the CCMG membership and reflects the values of the CCMG and has been approved by the CCMG Board of Directors.

Summary

This document enunciates the position of the Canadian College of Medical Geneticists (CCMG) on the patenting of isolated sequences of DNA and RNA. It is the opinion of the CCMG that the information encoded by the human genome, whether found in vivo or artificially reproduced in vitro, should not be patentable.

The granting of patents on diverse aspects of the genome has raised considerable international attention in the last two decades. This is particularly true of Canada’s closest neighbor, where a legal battle is still ongoing. Its outcome, and the conclusions of the related international debate, will have long-lasting effects on the practice of medical genetics worldwide.

In order for Canadians to continue to benefit from the advances of basic and clinical research in health and medicine resulting from the study of the human genome, cost effective health care and optimal
interpretation of clinical investigations are needed. The patenting of DNA and RNA sequences has, and will further, greatly limit the ability of our patients to benefit from progress in the area of human genetics, especially as knowledge continues to rapidly expand with the development of high throughput genomic technologies. The intention of this document is to highlight the need for a detailed legislative revision of patent laws as they apply to the human genome.

For the purpose of this statement, it is understood that reference to human gene sequences includes any and all components and variations of the sequences of DNA and RNA that make up the total human genome, whether found in vivo or artificially reproduced in vitro. DNA and RNA sequences are products of nature that undergo breakage of chemical bonds (including hydrogen and covalent bonds) and, as applicable, replication, amplification, and modification. While we agree that the novel methodologies developed to visualize the sequence of DNA and RNA are patentable, the sequences produced in vitro, which reflect the in vivo sequences, do not constitute inventions and should not be patentable.

Background

With the advent of high-throughput genomic technologies, our understanding of the human genome is rapidly increasing. New genes, and ways of regulating genes, are being identified at the same time that the biological functions of previously known genes are elucidated. These advances have led to the development of an already unprecedented number of genetic diagnostic tests and the pace of development is accelerating. Moreover, the technical possibilities afforded by high-throughput genomic technologies combined with new knowledge of gene function has allowed the development of testing platforms that simultaneously interrogate multiple genes and/or loci. Such platforms include, but are not limited to, chromosomal microarrays, resequencing microarrays (i.e. targeted sequencing of multiple genes), and whole exome/genome sequencing. These platforms allow the study of multiple genes in the same pathway, multiple genes that give rise to similar phenotypes, or assessment of the entire exome or genome. It is our contention that while new testing platforms themselves are often inventions subject to patents, the growing body of knowledge about the sequences of the human genome generated by the use of such platforms should not be patentable, and in particular, DNA and RNA molecules corresponding to human genomic sequences should not be patentable.

Patenting components of the human genome, including isolated DNA and RNA sequences, leads to suboptimal standard of medical care, especially within public healthcare systems. While some patented genetic tests are of high quality, some are not. Yet when test quality is low or reporting is inadequate, there is no alternative to a patented test because there is no competition for quality or price, and no motivation for patent holders to improve their test or to grant access to others through licensing. Moreover, such patents pose barriers to the use of data obtained from currently available untargeted genetic testing technologies. For example, recent examination of whole exome sequencing data revealed pathogenic mutations in high penetrance cancer susceptibility genes in slightly over 1% of participants in a research project on atherosclerosis (Am J Hum Genet. 2012 Jul 13;91(1):97–108). Disclosure of such information is of utmost importance for the future health of patients; however, patents exist for many of these disease associated DNA sequences. A literal reading of the Canadian patent claims suggests they would be infringed. Courts might decide otherwise, but finding out the opinion of the courts would entail litigation – an expensive and drawn-out process incompatible with getting this information to the patients in time for them to make life-changing medical decisions. If the patents on the DNA sequences comprising specific genes are upheld, this will significantly penalize the patients undergoing whole exome/genome sequencing by impairing the ability of the clinical laboratory to comprehensively report on incidental clinical findings. Moreover, the interpretation of sequence variations identified in patented genes may be incorrect if the data needed to infer clinical significance are held in proprietary databases that are not shared publicly. These patents also pose significant barriers to the development and exploitation of the full potential of testing platforms designed to simultaneously assess multiple genes responsible for similar phenotypes, thereby adversely affecting the cost and time-to-diagnosis efficiencies recognized with such platforms. Patent protection has unfortunately been extended to substances it should not cover, the DNA and RNA corresponding to sequences found in the human genome. It would be far better to engender respect for patent law by restricting it to valuable new inventions, rather than including substances found in nature that were discovered and not invented. In this context, we believe that patents on DNA and RNA sequences (whether isolated in vitro or not) pose a significant threat to the ability of Canadian citizens to access appropriate and cost effective genetic care.

The discoveries that have resulted in patents on isolated DNA sequences, and will continue to do so under the current law, are often the product of massive public investments including funding from government and charities, as well as decades of collaborative research involving innumerable participants, including researchers, students, clinicians and patients from around the world. Of particular importance is the fact that patients and other research participants usually contribute their time and biological samples altruistically with a motivation to promote better care for others. If one ignores the issue of whether or not human genes should be patented, attributing the discovery to a specific individual or entity fails to recognize the essential public investment in this process of collaboration and discovery; further, it fails to respect the wishes of patients who generously contribute with the hope of helping others. And finally, failure to respect the core values of patients who altruistically contribute to
research may have tremendously damaging effects on the future of genetic medicine by limiting the willingness of our patients to participate in future research endeavors.

The attained knowledge is the fruitful result of decades of research, significant public investments and tissue donations from patients. We firmly believe that Canadian citizens should be able to benefit from any knowledge to which they actively contributed and continue to contribute. Patents on DNA and RNA sequences can be used to restrict the potential benefits of genomic discoveries; unreasonable exploitation of the entitlements of a patent holder will be unfair to Canadians and detrimental to their present and future health and well-being. While some may argue that fair access could still be achieved through equitable licensing, we believe that this only creates secondary issues of defining and enforcing what is fair and equitable licensing to remediate a situation that would not exist in the absence of what we judge to be unfair and unwarranted patents. That is, while some problems might be solved by more enlightened licensing policies, we believe patents on DNA molecules corresponding to human DNA sequences should not be granted at all, thus avoiding the problems entirely.

In order to: (i) improve the quality of life of people, most particularly patients who have diseases associated with genetic sequences, or a high genetic risk of developing such diseases, (ii) promote fair access to new technologies for the Canadian population, (iii) respect the values of Canadians altruistically participating in research, and (iv) help to realize the potential benefits of knowledge of the human genome, we believe patents claiming DNA and RNA sequences comprising the human genome must be reconsidered. The Canadian College of Medical Geneticists is of the opinion that any existing patents on human genetic sequences must not be used to (i) enforce commercial monopoly on any use of the protected knowledge, (ii) build genetic information banks subject to restricted access despite preferences of individual patients, particularly where that information is relevant to the health and well-being of Canadians, and (iii) restrict access to medical advances in the context of our publicly funded health care system.

We call upon the Government of Canada to promptly enact the legislative and regulatory changes necessary to ensure that all Canadians can benefit from the advances in genetics to which they are active contributors both as research participants and taxpayers.

Acknowledgements

We would like to acknowledge the CCMG Board of Directors and membership for their helpful comments. We would also like to thank Dr Alessandra Duncan for her tremendous editorial help.