

No faulty-gene carrier need apply

Consistent laws against genetic discrimination are needed to allow the development of genomic medicine and ensure individual participation in genetic research.

Six years after the publication of the Human Genome sequence, genome-wide association studies have started uncovering gene variants indicating a predisposition to one disease after another. Yet a clear legal framework for protection against genetic discrimination is still largely missing. But wheels are in motion.

The Genetic Information Nondiscrimination Act of 2007 (GINA 2007) is awaiting a vote in the US Senate. More than ten years in the making, and after several favorable votes by one chamber not followed through by the other, the bill finally stands a good chance of being implemented into law this year. It was passed by an overwhelming majority in the House of Representatives in April and comes to the Senate with support from the White House. If enacted, this legislation will make it illegal for health insurance companies to increase premiums or refuse coverage for individuals with a genetic predisposition to disease. It will also prohibit employers from using genetic information in hiring, firing, as well as deciding job assignments and promotions.

To date, there has been little evidence of discrimination by health insurance providers and employers. Yet there is widespread fear of discrimination. In a 1997 US survey, 63% of participants indicated that they would not take genetic tests if health insurers or employers could get access to the results. In a 2000 survey of genetic counselors, 68% of the participants indicated that for fear of discrimination they would not bill their insurance for a genetic predisposition test, and 26% would even go so far as to use an alias.

If not addressed, such fear of discrimination will discourage people from using genetic tests to make better preventive healthcare decisions and also risks limiting participation in genetic research. The apprehension is understandable in the absence of a clear legislative framework. Most US states have enacted laws to make up for the lack of federal regulation, but these laws differ in their scope and implementation schemes. For example, the type of health insurance regulated and the definition of 'genetic information' vary from state to state, and penalties range from a small fine to the revocation of the insurer's license (see the National Conference of State Legislatures website for a current update).

Europe has a similar legal patchwork. Some countries like Austria, Belgium and France have enacted legislation to prohibit insurance companies from using results

of genetic tests. In other countries, such as the United Kingdom, the government has agreed on a moratorium with the national insurers' association. But even when legislation is in place, definitions are not perfect. The Belgian law, for example, leaves the door open for discrimination based on family history.

The Charter of Fundamental Rights of the European Union prohibits discrimination on the basis of genetic features. It draws on the 1997 Convention on Human Rights and Biomedicine, Europe's most central international legal document in the health field. These prescriptions, however, are not legally binding until they are enacted as national laws by the individual countries. In practice, many countries either have not signed the Convention, or have signed but not ratified it (see the Council of Europe website for an update).

To effectively reassure citizens, a consistent and comprehensive legal framework must replace this patchwork. In that regard, the American bill hits a lot of important points that are inconsistent in the state laws.

For starters, GINA 2007 makes clear that the proposed legislation will not interfere with the delivery of healthcare. By no means should it prevent physicians from requesting genetic tests for patients or their relatives. Along the same lines, it will be acceptable to use genetic testing to justify reimbursement by the insurer for preventive interventions or intensive monitoring, which while appropriate for someone at high risk, could not realistically be offered to the general population. Appropriately, the proposed legislation prohibits discrimination based on the mere request of genetic services and family medical history.

Interestingly, the definition of genetic testing under GINA 2007 includes "analysis of human DNA, RNA, chromosomes, proteins or metabolites that detects genotypes, mutations, chromosomal changes." But this definition excludes analyses of proteins or metabolites that are directly related to a manifested disease, thus making a clear distinction between risk and pre-existing condition.

Hopefully GINA 2007 will receive the priority it deserves on the US Senate's agenda and will inspire law makers in Europe. Genomic technology is moving fast. The legal system must catch up quickly to provide a framework allowing research progress and its implementation for better healthcare.