NEWS BLOG

Geneticists debate what to tell patients about clinical genome sequences

29 Mar 2012 | 20:01 BST | Posted by Erika Check | Category: Biology & Biotechnology, Health and medicine, Policy

Should patients undergoing genome sequencing be screened for a minimum set of disease-causing mutations, and should adults and children receive different types of genetic results?

Last night, geneticists debated these issues at the annual meeting of the American College of Medical Genetics (ACMG) in Charlotte, North Carolina. In an open forum at the meeting, the ACMG released a new policy statement on whole-genome sequencing and presented a report from a work group that is drawing up guidelines about what information should be given to patients about 'secondary findings' that turn up during the course of sequencing tests. Secondary findings are genetic mutations that predispose a patient to a disease but are unrelated to the initial reason for the patient's decision to undergo sequencing.

The draft recommendations, which will not be finalized until this summer, are part of a larger debate over what geneticists should do about the 'return-of-results' issue, which focuses on how much information patients and research subjects should learn about their genomes. A project funded by the US National Institutes of Health recommended on 21 March that researchers who find disease-causing mutations in archived data should consider notifying research participants of the mutations. But the ACMG's recommendations will focus specifically on patients being sequenced for clinical, rather than research, purposes.

Robert Green of Brigham and Women's Hospital in Boston, who co-chairs the ACMG work group on secondary findings, says that the field must develop standards for informing patients about them.

"We don't think it's going to be a sustainable strategy for the evolving practice of genomic medicine to ignore secondary findings of medical importance," he says.

These findings could arise in several ways. A child undergoing sequencing to diagnose the cause of a developmental delay might find out that he also has a genetic predisposition to certain cancers, or a cancer patient undergoing sequencing to guide personalized therapy might find out that she has a mutation linked to a treatable syndrome, called familiar hypercholesterolaemia, marked by high cholesterol.

The ACMG is considering recommending that clinical laboratories test patients' genomes for a minimum set of mutations such as these that meet a checklist of criteria and are not detected by newborn screening programmes (see slides from the work group's presentation here). High-penetrance mutations — those very likely to lead to disease — that cause treatable

conditions would be high on the work group's list, whereas genetic variants that only sometimes cause disease or are linked to untreatable conditions wouldn't make the cut.

For instance, familial hypercholesterolaemia variants would be reported to patients. But variants of the gene that encodes apolipoprotein E, which is linked to an increased risk of developing Alzheimer's disease, wouldn't be reported, because these variants don't actually predict that a patient will develop the disease, and because no early intervention has been shown to prevent Alzheimer's disease.

Green said that some geneticists at last night's forum were concerned that testing a standard set of genes would violate patients' rights not to know about their genetic predispositions to disease.

And, although the current working group guidelines don't distinguish between information given to children or adults, some geneticists argued at the meeting that it is inappropriate to tell children about predispositions to disease that will not affect them until they grow up.

Green says, however, that a patient's right not to know about certain mutations could be protected, for instance, if a patient tells her doctor that she doesn't want to know about them. And Green points out that a child's genetic information will also be relevant to his or her parents. For instance, a child carrying a mutation that predisposes him or her to a certain cancer inherited it from at least one parent, who may not know that he or she is also likely to develop that cancer.

"That information sitting in our hands could save a parent's life," Green says.

The work group, co-chaired by Leslie Biesecker of the US National Human Genome Research Institute, is also proposing that clinical labs disclose their policies on reporting secondary findings to patients and doctors. The AMCG is still soliciting input on the draft guidelines and aims to deliver the finished recommendations in June.

Follow Erika on Twitter at @Erika_Check.

Previous post
Gaining control of our circadian rhythms

Next post
Welcome to the biobank

Comments

Comments are closed.