US doctors' group says patients should have option not to know their DNA

New genome-sequencing recommendation will enable patients to opt out of testing.

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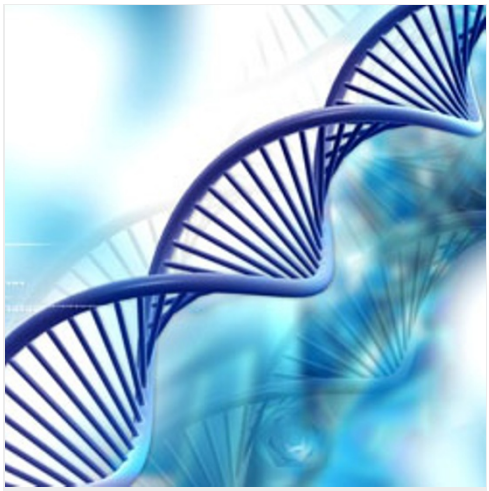

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In 2030, Joe Johnson goes to the doctor with vision problems. The doctor does what doctors in 2030 do for almost any ailment: she runs a full scan of the patient's genome. The test gives the doctor crucial information she needs to understand Joe's troubles — and then some. When Joe gets his lab report back his doctor discovers Joe has about an 80% chance of developing colorectal cancer. She advises him to receive regular colonoscopies, since the screenings could catch polyps that would be the precursor to the cancer. But Joe does not relish facing years of colonoscopies, nor is it a sure thing that his genetic mutation will lead to cancer.

In 2014, of course, this type of genetic analysis is not routinely offered to most patients, but in the coming years it may be. Many patients will not want to know about their genetic predisposition to diseases, especially those with no treatment, such as Alzheimer's disease. But it remains murkier whether they would want to know about results that suggest they are at a heightened risk for certain treatable illnesses. Today, geneticists took an important step to give patients more choice in the matter. The national standard-setting group, the American College of Medical Genetics and Genomics (ACMG), rolled out an updated policy that will allow patients to completely opt out of recommended testing for genetic mutations that could indicate specific disease risks.

The spectre of medical testing dredging up unexpected findings is not new; it currently exists with CT scans that could reveal tumors. But the advent of cheap genome-scale sequencing has upped the ante. Sequencing has vast power to uncover a wide range of underlying genetic mutations, but in many cases we are still not sure exactly what these mutations mean. When doctors test for one reason, clues to an entirely different disease may emerge. Sometimes those genetic clues provide an early warning that will save a life, but they can also be wrong, leading to unnecessary stress and risky treatments.

The new recommendation amends earlier guidelines that called for doctors to screen for genes linked to 24 medical conditions, even if they are irrelevant to the ailment the patient is being tested for. The ACMG chose those maladies because researchers know enough about them to say that these particular mutations would result in a heightened risk for specific conditions that have available treatment or management options.

The earlier decision was controversial, however, partly because it raised uncomfortable questions about patient choice. Before ordering the test a patient and her clinician would discuss what the findings could mean and decide together if the patient would want that information returned. The bar for refusal, however, was pretty high since most clinicians would push for the patient to have that full list. Even if the patient decided against it, the laboratory would usually still gather the data and give it to the clinician.

Since the information would still wind up in the patient's medical record even if her current physician agreed not to share the information, it could conceivably come up in later encounters with other medical professionals. "It could easily leak back to you and undermine your effort to opt out," says James Evans, director of Adult Genetics Services at the University of North Carolina and a board member of ACMG. The arrangement also put doctors in a "sticky position" he says, since a finding could come back that the doctor felt the patient should know about, while the doctor's hands were tied from revealing it, he says.

The new decision from the ACMG recommends giving patients the option to not have the information gathered at all. The move has widespread support among genetic experts. At their annual conference in Nashville, Tenn., last week an informal poll of more than 400

experts working in the field revealed that 88% were in favor of such an opt-out option.

Still, the situation remains contentious. Some doctors oppose the decision since they feel it will make it too easy to refuse potentially life-saving testing. “I worry that allowing this opt out will disproportionately impact the disenfranchised. It will become too easy for a lazy doctor just to say ‘ok, we’ll skip [this important testing],” says Robert Green, an associate professor of medicine at Harvard Medical School and a lead writer of the original ACMG recommendations. “Frightened patients, those less well-educated patients or perhaps those distracted by a child’s disease will be the ones that opt out,” he says.

The issue of genetic sequencing raises thorny issues of ethics and patient-doctor communication. If a patient chooses to opt out of testing for that recommended list of mutations does she or he really understand what that decision means? Was the physician able to make the significance of the mutations clear in a relatively short appointment? But patients are currently afforded the opportunity to opt out of life-saving procedures, so why should opting out of information about possible genetic mutations be any different? The ACMG board, which put forth this new decision, is implicitly stating that it isn’t.

Some laboratories that currently offer genetic sequencing already offer opt-out options so it is unlikely that it will be a challenge for laboratories to comply with the new recommendations, says Gail Herman, president of ACMG. But it remains unclear how future opt-out choices will play out on an individual gene level.

Although the new ACMG recommendations suggest a patient could opt out of — or go forward with — the list as a whole, geneticists and bioethicists are already discussing scenarios where patients may approach such decisions more like a menu, saying they want to know about increased risk of heart disease but not cancer, for example. “At this point we are not recommending selective opting out,” says Herman. “Of course the physician and the patient can decide what is in the patient’s best interest, but we are not [recommending] that at this level at this point.” That short list of recommended genes to examine for underlying mutations is expected to evolve. A new ACMG committee that met for the first time last week will develop criteria for how a gene would get added or removed from future iterations of the list.

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