The power to help, hurt and confuse: Direct-to-consumer whole genome testing

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Two UNC experts write in a *JAMA* commentary that whole genome and whole exome sequencing technology "will routinely uncover both trivial and important medical results, both welcome and unwelcome … and presents the medical community with new challenges." Credit: National Institute of General Medical Sciences

The era of widely available next generation personal genomic testing has arrived and with it the ability to quickly and relatively affordably learn the sequence of your entire genome. This would include what is referred to as the "exome," your complete set of protein-coding sequences.

But as University of North Carolina at Chapel Hill medical geneticists point out, this avalanche of information also includes the totality of one's genetic mutations and as such arrives with both promise and threats associated with its use.

James P. Evans, MD, PhD is the Bryson Distinguished Professor of Genetics and Medicine at UNC and is a member of the Lineberger Comprehensive Cancer Center. He is also editor-in-chief of *Genetics in Medicine*, the journal of the American College of Medical Genetics. "What you're now dealing with is a real medical test, one that has the power to help, hurt and to confuse. I believe we need to think carefully about how to best use it and how that use should be regulated in order to maximize benefit and minimize harm," he said.

In a commentary published in JAMA on Wednesday, Dec. 7, 2011, Evans and UNC co-author Jonathan S. Berg, MD, PhD, Lineberger member and assistant professor of genetics and medicine, argue that whole genome and whole exome sequencing technology "will routinely uncover both trivial and important medical results, both welcome and unwelcome … and presents the medical community with new challenges."

"What we have had up until this point with direct-to-consumer genetic testing, despite all the hoopla, was arguably rather trivial from the standpoint of either benefits or threats. It was a fairly worthless technology because it really didn't give people medically significant findings," Evans said.

"Now we are entering an entirely different era due to the advent of robust sequencing technology. We have now the potential to tell people very real and important things about their genomes. Some of those things can be very useful and very welcome if acted upon in the right way, but some of that information may not be very welcome to people: being at high risk for an untreatable disease such as dementia, for example."

As to regulation, Evans and Berg suggest that it need not be draconian but must be nuanced. "Basically, what we call for is that this new generation of medical testing be treated like other medical tests that

involve complex medical information – and that there should be a reasonable expectation that an individual who gets it done has some relationship with a qualified care provider."

That person doesn't need to be a physician, Evans adds. "There are genetic counselors capable of dealing with this. But it must be a person not employed by the company or laboratory doing the testing since that invites egregious conflict of interest."

As physicians pledged to avoid causing harm, the authors acknowledge the inevitable tension that exists between paternalism and the reasonable protection of people. They point to three compelling arbiters of whether the acquisition of medical information should require a relationship with a healthcare professional: the information's complexity, ability to mislead and potential for harm.

"The advent of next generation sequencing technology marks a threshold at which genomic testing easily meets these bars," they state.

Provided by University of North Carolina School of Medicine