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Genomic Screening for Improved Public Health

March 7, 2013 –Bethesda, MD| In ten years time, routine preventive health care for adults may include genetic testing alongside the now familiar tests for cholesterol levels, mammography and colonoscopy. As genomic testing prepares to enter the realm of general medical care, an interdisciplinary team of researchers is suggesting in a commentary in the May 2013 issue of *Genetics in Medicine*, the peer-reviewed journal of the American College of Medical Genetics and Genomics (ACMG), that now is the time to explore genetic testing to identify people at high risk for carefully selected, preventable disease.

The technology is available, and the price is coming down so rapidly that it will soon be possible and practical to offer a carefully selected panel of genetic tests that could avert disastrous health consequences in people at high risk for serious life-threatening diseases, according to James P. Evans, MD, Ph.D, Bryson Distinguished Professor of Genetics & Medicine, at the University of North Carolina – Chapel Hill and Editor-in-Chief of *Genetics in Medicine*. The commentary authors, experts in both genetics and public health, believe it is time to start looking at genetic testing through the lens of disease prevention. There are enough genetic conditions that are both preventable and strongly predispose people to specific cancers or to a catastrophic vascular event that it is sensible to try to identify those people early so they can seek preventive care, the researchers argue.

"Added together, the number of people who are walking around, who unknown to them, have mutations that greatly predispose to serious but eminently preventable disease, comes out to roughly 1 percent of the population," says Evans.

For example, about 1 in 400 people in the United States carries an inherited genetic predisposition to develop colon cancer at an early age. Currently, people carrying this genetic risk would be unaware of it unless enough close family members develop colon cancer prompting doctor to suggest genetic testing or it is identified by analysis of their own or a family member's tumor. If, instead, at-risk people could be identified before cancer has occurred, a program of early, regular colonoscopy screening would help prevent the disease in the first place the researchers argue.

Using a preventive approach, the general public could be offered a panel of tests for this and other selected genes for which preventive care or early treatment is available, the researchers say. But discussion about which genes to include in such a panel will take time, as will the necessary research into the cost-effectiveness of such testing, as well as ethical, legal, and social implications. Now is the time to begin the discussion, they say, and to form a new partnership between geneticists and members of the public health community to help realize the full promise of public health genomics.

"Investigating the possible benefits of such screening, as well as the challenges and potential pitfalls that might exist, should involve a partnership between the genomics community and the public health community," said Evans. "There would be many important issues to sort out, including which genes should be screened and how individuals respond to screening."

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