

## IN THIS ISSUE

### Family cancer history doesn't necessarily lead to more healthful living

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Public health messages about the ways people can reduce their risk of cancer may not be making an impact on those with a family history of the disease, according to a new population-based study reported in this issue. Researchers from the Centers for Disease Control and Prevention used data from the 2005 California Health Interview

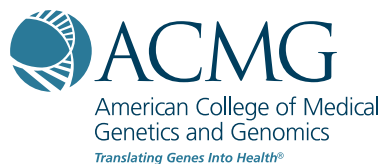


Survey to examine the relationship between having a first-degree relative with breast or colorectal cancer and receiving recommended screenings as well as following recommended guidelines regarding modifiable lifestyle factors. Among the findings, both men and women with family history of colorectal cancer were actually less likely than those without a family history to maintain a healthy weight and consume the recommended amount of fruits and vegetables. However, those with family history of cancer were more likely to follow guidelines for periodic mammogram or colonoscopy, although many of those with such a history did not receive screenings. The investigators could not assess the reasons behind these behaviors, which would seem to put some individuals at an additional increased risk of cancer, but they suggest that health-care providers counsel patients with family histories of cancer about what they can do to reduce their risk. —Karyn Hede, News Editor

### New ACMG Practice Guideline: genetic testing for *MTHFR* variants rarely warranted

see page 153 (February 2013)

Carrying common genetic polymorphisms in the gene encoding 5,10-methylenetetrahydrofolate reductase (*MTHFR*) is no longer considered a significant risk factor for abnormal blood clotting, making genetic testing for their



presence unnecessary, according to the ACMG. Previous research had suggested that genetic polymorphisms in *MTHFR*, which are associated with marginal elevations in plasma levels of homocysteine, might increase the risk of venous thromboembolism, coronary heart disease, and recurrent pregnancy loss. But recent meta-analyses have disproven these suspected associations, making *MTHFR* polymorphism testing minimally useful. Therefore, the ACMG issued a new Practice Guideline, detailed in the February issue, that recommends discontinuation of *MTHFR* testing as part of a routine evaluation for patients suspected of increased risk of blood clots. Patients who test homozygous for the more common “thermolabile” variant, and who also have normal plasma homocysteine, can now be reassured that they are not at increased risk for venous thromboembolism or recurrent pregnancy loss related to their *MTHFR* status. In addition, fortification of grain products with folic acid has resulted in higher serum folate concentrations and lower serum total homocysteine levels in general, possibly reducing some of the already low risk of carrying such an *MTHFR* polymorphism. —Karyn Hede, News Editor

## NEWS BRIEFS

### Study questions the security of genomic information

Public genome databases, while providing a rich source of information for research purposes, also expose participants to the possibility of being identifiable, despite measures designed to provide anonymity. A recent informatics study published in *Science* showed just how easy it is to use publicly available online databases to track and identify study subjects. The team of researchers pinpointed (although didn't publish) the identity of nearly 50 individuals who participated in genomic studies that became part of the 1000 Genomes Project. To track individuals, the authors relied on a combination of data submitted as part of research on genetic patterns of the Y chromosome, paternal transmission, and cultural transmission of a family's surname through male family members. The finding implies that genetic data could be used to identify distant relatives who might have no idea that their genetic heritage could be made public. An accompanying commentary suggests that, rather than stirring fear, the study should provoke a robust debate about the risks and benefits of broadly sharing genomic data. Social norms, attitudes, and expectations of privacy are rapidly changing. Indeed, individuals participating in the study in question consented twice and were told it was possible, although not likely, that they could be identified through their genomic information. Yaniv Erlich of the Whitehead Institute, the study's lead author, says, “We also hope that this study will eventually result in better security algorithms, better policy guidelines, and better legislation to help mitigate some of the risks described.” —Karyn Hede, News Editor



### Brain's reward system linked to longer life

A gene variant previously associated with “novelty seeking” and possibly selected for during the human population expansion out of Africa has now been suggested as a contributor to longevity as well. The dopamine receptor gene *DRD4*, which has been shown to play a major role in the neural network responsible for the brain's internal reward system, has been extensively studied for its role in personality variation. The new research, published in the *Journal of Neuroscience*, combined human and mouse studies to identify the *DRD4* 7R allele as a



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contributor to long life. Mice in which the gene was knocked out had a 7–10% shorter life. Conversely, mice with the *DRD4* 7R allele lived significantly longer than control mice when raised in an activity-rich environment. Similarly, in human studies of people more than 90 years old, the allele was found more often and was associated with higher levels of physical activity. People who carry the gene variant seem to be more motivated to pursue social,

intellectual, and physical activities, according to lead researcher Robert Moyzis of the University of California, Irvine. Although the variant may not directly control longevity, the research team suggests that it contributes to developing personality traits such as sociability, intellectual curiosity, and pursuit of physical activity that have been shown to be important for living a longer, healthier life. —*Karyn Hede, News Editor*

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- Original reports which enhance the knowledge and practice of medical genetics
- Strategies and innovative approaches to the education of medical providers at all levels in the realm of genetics

As the official journal of the American College of Medical Genetics and Genomics (ACMG), the journal will:

- Provide a forum for discussion, debate and innovation concerning the changing and expanding role of medical genetics within the broader context of medicine
- Fulfill our responsibility to the College membership through the publication of guidelines, policy statements and other information that enhances the practice and understanding of medical genetics

Finally, as genetics becomes increasingly important in the wider medical arena, we will be an accessible and authoritative resource for the dissemination of medical genetic knowledge to providers outside of the genetics community through appropriate reviews, discussions, recommendations and guidelines.