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Women and health providers at odds on risks/benefits of noninvasive prenatal tests

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The day when a simple maternal blood draw can be used to diagnose chromosomal abnormalities in a fetus has arrived. But many issues remain to be sorted out as this technology is applied. The detection of Down syndrome is its first widely used application. In the United Kingdom and Australia, widespread use of noninvasive prenatal diagnosis (NIPD) for Down syndrome brings questions of how to present such tests to prospective parents. To understand how women view such tests as compared with health-care providers, Melissa Hill et al. conducted a National Health Service–sponsored study in England that used a series of questions designed to rank the importance of four attributes: test accuracy, time to get a result, risk of miscarriage, and completeness of information provided. The results showed that women ranked the noninvasive test as more desirable than the current invasive procedure even when told the NIPD test is less accurate. The findings demonstrate that women hold safety of the fetus as the highest consideration, with one-third of the women making choices based solely on the fact that there was no risk of miscarriage. By contrast, health-care providers were more concerned about test accuracy. The study's results point to the need to develop effective prenatal counseling tools that help women make informed choices about new NIPD tests. —*Karyn Hede, News Editor*



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Imaging a useful indicator of potential cardiac disease in Marfan syndrome patients

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Individuals with Marfan syndrome commonly experience cardiac disease, due to aortic dilatation and stiffness of the vascular system. Such individuals are prone to aortic aneurysm and dissection. Disease of the ascending aorta also correlates with dilatation of the trunk of the pulmonary artery. Rigmor Lundby, of Oslo University Hospital, conducted a magnetic resonance imaging (MRI) study of 87 patients as part of a Norwegian national study of Marfan syndrome. A multivariate analysis of the results suggested that dilatation of the pulmonary artery may occur even with no visible aortic disease. The findings also demonstrated that previous surgery on the ascending aorta correlated with dilatation of the trunk of the pulmonary artery but not of the root. The majority of the patients with dilation of the trunk of the pulmonary artery also had aortic disease (40 of 47), but the remainder (7 of 47) showed no immediate sign of aortic disease. Echocardiography showed that none of the study participants had high pressure in the pulmonary artery. The author therefore suggests that dilation of trunk of the pulmonary artery as imaged by MRI (or computed tomography) may be an early marker of vascular disease in Marfan syndrome patients. —*Karyn Hede, News Editor*



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NEWS BRIEFS

Did you vote? Thank your dopamine gene

Political participation is at least partially under the purview of our genetic predisposition, according to the burgeoning field that might be dubbed politicomics. The quest to understand why individuals come to hold particular political views has prompted some investigators to drill down into the genetic contribution to partisan behaviors. Peter Hatemi, of Pennsylvania State University—who may be the only academic to hold joint appointments in departments of political science, microbiology, and biochemistry—along with Rose McDermott, of Brown University, reviewed the genetics of politics in the August 2012 issue of *Trends in Genetics*.

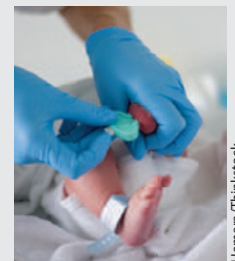


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The concept that people have innate tendencies when it comes to political ideology is relatively novel, as social scientists have tended to view political views as the product of family, environment, and social interactions. Geneticists have added to the conversation by revealing that, for example, variants of dopamine (*DRD2* and *DRD4*) and serotonin (*5HTT*) genes can influence whether people participate in political causes and turn out to vote. Unsurprisingly, no genetic studies have revealed a single significant variant that can account for political leanings. Rather, most researchers understand that political traits are influenced by the complex interplay of multiple genes and life experience. But the methods developed to integrate genetic research with complex human behavior should benefit a wide range of research endeavors, and perhaps even our political discourse, as we continue to wrestle with such contentious issues as allocation of health-care resources. —*Karyn Hede, News Editor*

Heel-stick DNA offers potential epigenetic bonanza, invites controversy

Researchers who study epigenetic changes over time and try to associate them with disease state have run into a classic chicken/egg conundrum: how to distinguish epigenetic changes that drive disease risk from incidental changes. Guthrie cards—those archived newborn blood spots—have now been shown in principle to be a source of epigenetic fingerprints documenting



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NEWS BRIEFS

DNA methylation status acquired in the womb as well as a potential window into how epigenetic patterns change over time. In an article published online in *Genome Research* in August 2012, a team of European researchers describe their method for generating a complete map of DNA methylation patterns derived from Guthrie cards. The group then compared that “methylome” with one for the same individuals three years later and found that the pattern was stable for each individual and variable between individuals. The authors hint at the re-

search potential offered by use of the cards, but that potential is tempered by recent controversy over ownership and research use of this potential archival DNA resource. Although newborn screening for genetic disorders is nearly universal in developed nations, informed consent from the newborn’s parent or guardian for research use has not been routinely obtained. Privacy concerns will probably lead to opt-in consent for research going forward, but the extent to which archival material can be used remains unclear. —*Karyn Hede, News Editor*

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- 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:

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- 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.