

IN THIS ISSUE

No health benefit of current genetic tests for type 2 diabetes

see page 612

An evaluation of several commercial tests for type 2 diabetes published in this issue found little to no benefit to patients. An accompanying systematic review and recommendation statement conclude there currently is no advantage to such testing and therefore discourage its use at this point. After reviewing the published literature and information made available on company websites, the Evaluation of Genomic Applications in Practice and Prevention Working Group (EWG) concluded that there is no evidence of analytical validity (reproducible test performance) or clinical utility for currently available tests. Several companies offer genomic testing for type 2 diabetes, either through a health-care provider or direct to consumers. Evaluation of test performance proved difficult because the EWG found that it could not even identify the testing platform or methodology used in some cases. Looking at testing both for the general population and for high-risk groups, they detected little, if any, ability to evaluate or classify individual risk. Specifically, they found insufficient evidence to recommend testing for the transcription factor 7-like 2 (*TCF7L2*) gene marker. Although the gene has been demonstrably linked to a higher risk of developing type 2 diabetes, there is no evidence that knowledge of the presence of the gene variation could improve health outcomes. The EWG states that, to be useful, genomic testing must be shown to have predictive value superior to that of traditional risk factors. Therefore, until advances in the clinical validity of these genomic panels can be demonstrated, their use is discouraged. —Karyn Hede, News Editor



©istockphoto/Thinkstock

Genomic literacy conference yields research agenda

see page 658

In an era in which advances in genetic knowledge are bound to touch many, lack of public understanding of genetics and genomics poses a potentially confounding knowledge gap. Closing this gap with effective educational messages about genetics and genomics was the goal of a national workshop on genomic literacy held in November 2011 and sponsored by the National Institutes of Health's National Human Genome Research Institute. In this issue, a workshop report details the challenges to educating the public and provides a research agenda designed to produce effective educational mes-



©istockphoto/Thinkstock

NEWS BRIEFS

Genomics solves mysterious disappearance of leprosy

No one wants to be labeled a leper. So powerful was the fear of leprosy in medieval Europe that its specter still exerts a hold



University of Winchester

Skeletons at the St. Mary Magdalen leprosarium in Winchester, UK.

on our collective psyche. Yet the disease itself is nearly unknown in developed countries today. To understand why the scourge of leprosy virtually disappeared, an international team of geneticists and archeologists systematically compared the genomes of five strains of *Mycobacterium leprae*, the bacterium responsible for leprosy, found in human remains buried in medieval graves. The findings, published in the 14 June 2013 issue of *Science*, indicate that the bacterium's genome is nearly identical in the medieval and modern forms. For example, a medieval strain of *M. leprae* found in Sweden and the United Kingdom was nearly identical to the strain currently found in the Middle East. The research team suggests that host resistance may have been the result of social isolation and widespread infection rather than of bacterial mutation—people simply ceased becoming as susceptible to infection. The findings add to our understanding of the natural history of epidemics. In addition, the difficulty of isolating the bacterial DNA from human DNA fostered development of exquisitely sensitive sequencing methods that the authors report are among the most precise ever developed. The methods will assist isolation and sequencing of other difficult mixed and ancient DNA samples. In addition, the state of bacterial DNA preservation suggests that additional studies might be able to trace the path of leprosy even further back in time. —Karyn Hede, News Editor

Patently obvious

What has been self-evident to much of the genetics community for more than two decades has finally, at least in part, been affirmed by the highest court in the land.

As most readers of this journal know, on 13 June 2013, the US Supreme Court ruled that Myriad Genetics' patent claims covering the genomic versions of the *BRCA1* and *BRCA2* genes were invalid, on the grounds that genes are naturally occurring substances and that merely isolating them is insufficient to warrant patent protection. Although the court made clear that manipulated



©istockphoto/Thinkstock

IN THIS ISSUE

sages to promote public understanding of the role of genomics in human health. Workshop attendees discussed what it means to have a working knowledge of genomic science and its role in society. Given the public's lack of genomic-health literacy, participants agreed that determining and evaluating culturally appropriate terminology, with minimal jargon, are important research priorities. As individuals will increasingly be asked

to make personal and family health decisions based on genetic and genomic information, providing useful educational tools becomes imperative and will require new partnerships among health-care systems, government, community advocacy organizations, and others. To keep the conversation going, participants also recommended establishing a national conference on genomics education. —*Karyn Hede, News Editor*

NEWS BRIEFS

genes whose material properties have been changed (e.g., cDNA) may be patented, the overwhelming "take" on this decision is that it is a win for those who object to the patenting of human genes. Indeed, the impact of the decision was clear within hours of its announcement, when at least two large commercial laboratories announced plans to start offering sequence analysis of *BRCA1* and *BRCA2*, either individually or as part of expanded panels.

It appears, then, that the long battle of those who have fought the patenting of human genes has largely been won, and the

result will be expanded access and healthy competition among purveyors of genetic tests—a happy outcome for patients and those who care for them.

But the next battle looms. Properly interpreting the medical implications of genetic variants relies on access to data. However, Myriad Genetics has kept private the data obtained from their patent-protected analyses of countless women who have suffered from breast and ovarian cancer. The field (and patients) will suffer if such data remain sequestered for the perceived financial advantage of a single company. —*James P. Evans, Editor-in-Chief*

***Genetics in Medicine* | Mission Statement**

Genetics in Medicine is a monthly journal committed to the timely publication of:

- 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.