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PCA3 testing discouraged until better evidence obtained

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The unacceptably high rate of false-positive and false-negative testing results in prostate cancer screening has led to a search for more reliable screening tests. One such assay—the prostate cancer antigen 3 (PCA3) nucleic acid amplification test—has been proposed as a tool to assist physicians in deciding whether to perform or repeat a biopsy in at-risk men. PCA3 messenger RNA (mRNA) is highly elevated in prostate cancer cells as compared with normal prostate cells. A clinical test that measures the ratio of PCA3 mRNA to prostate-specific antigen (PSA) mRNA has been approved by the US Food and Drug Administration and is commercially available from a single vendor. But the clinical effectiveness of the test remains an open question. After reviewing the available published scientific evidence, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group reports in this issue that at this time there is insufficient evidence to recommend PCA3 testing in men. The group looked specifically at evidence to support testing for patients who were deemed at risk because of an elevated PSA level or suspicious physical examination or who had previously had a negative biopsy but were being considered for re-biopsy. They found no outcome studies showing that PCA3 scores can predict the behavior of a particular tumor or can yield information related to diagnostic accuracy, prognosis, or quality of life. The authors call for studies designed to compare PCA3 testing with two other commonly used add-on tests that measure free PSA and total PSA doubling time. —Karyn Hede, News Editor



Ted Foxx/Alamy

Regulatory loopholes raise concern about noninvasive prenatal tests

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The use of noninvasive prenatal testing (NIPT) has become attractive because of its high sensitivity and specificity as well as the potential to avoid invasive procedures and their associated risks. These tests evaluate circulating cell-free DNA to determine the risk of fetal aneuploidy. But many such tests being marketed to health-care providers and patients lack clinical validation and regulatory oversight, according to the review by Lutgendorf et al., who fear that neither clinicians nor the public fully appreciate the limitations and shortcomings of NIPT. The authors describe regulatory loopholes that allow companies to avoid US Food and Drug Administration oversight of lab-developed tests and suggest that patients and providers may be unwittingly participating in an unregulated public experiment in which adverse events are not being tracked. They emphasize the importance of prenatal counseling that clearly communicates the strengths and limitations of NIPT. In addition, they recommend that counselors clearly communicate with patients the possibility of false-positive and false-negative results as well as of results that don't correlate with fetal findings. Along these lines, the American College of Medical Genetics and Genomics recently advocated changing the terminology from NIPT to NIPS (noninvasive prenatal screening) to emphasize its limitations. There remain many questions surrounding the best use of these tests and how they should be integrated into prenatal screening programs. —Karyn Hede, News Editor



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NEWS BRIEFS

Artificial intelligence to help interpret genomic data

Watson—IBM's intelligent computer that can speak and learn—is now being harnessed to break down the barriers between genomic data and personal health. Best known for beating human game-show champions, Watson will now be spending time scanning genomic publications and incorporating those findings into a proprietary infrastructure developed by Coriell Life Sciences in Camden, NJ, which won IBM's Global Entrepreneur of the Year award for 2014. As a prize, Coriell is using Watson technol-



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ogy to speed the process of incorporating data from relevant genomic studies into its genetic risk-assessment system.

Coriell plans to offer genomic reports in language meaningful to physicians. For example, the report might flag a patient with an increased risk of an adverse drug reaction and identify an alternative treatment. The company plans to securely store genomic data and to continually update its databases as new genomic discoveries are made, using artificial intelligence to modify and enhance the advice it offers to physicians and patients. As described by Coriell, the aim is to bridge the gap between genomic information and actionable data so as to change how patients receive treatment, ultimately improving their health. —Karyn Hede, News Editor

NEWS BRIEFS *(continued)*

Researchers high on potential of pot genome

First it was *Arabidopsis* (rock cress), then crop plants such as rice, corn and wheat. Next up...pot? Few plant species have had their full genomes sequenced—so why choose cannabis, a plant that until very recently could not be grown legally in the United States? Nolan Kane, assistant professor of ecology and evolutionary biology at the University of Colorado, Boulder, thinks now is the time to sniff out the pot genome. His Cannabis Genomic Research Initiative will attempt to sequence 150 cultivars of cannabis,



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ranging from those intended to provide hemp to various medicinal varieties. For a species that has captivated humans

for centuries, relatively little is known about the plant, and little to no scientific research has been possible, at least in the United States. Even state legalization didn't really clear the way for research. But a provision in the most recent farm bill permits universities in states that permit hemp cultivation to conduct research without losing federal funding. That clears the way for genomics research that could provide hemp farmers and marijuana growers worldwide with genomic information to assist breeding of desired traits and that may provide information valuable to the plant genomics community. —Karyn Hede, *News Editor*

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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.