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Angelina Jolie turns heads—toward genetic information

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It's no secret that celebrities' medical conditions draw attention. Interest in spinal injuries rose after actor Christopher Reeve fell off his horse and became paralyzed. Apple CEO Steve Jobs raised the profile of pancreatic cancer. Certainly actress Angelina Jolie's public announcement of her genetic predisposition and decision to pursue risk-reducing bilateral mastectomy got lots of people talking about genetic testing for the *BRCA1* and *BRCA2* genes. Now, Juthe and colleagues document the impact of that increased public interest on Internet traffic to online cancer genetics resources available at the National Cancer Institute (NCI). The authors used digital media analytics to calculate page views for available fact sheets and Physician Data Query (PDQ) cancer genetics information summaries. On the date that Jolie's announcement appeared in the *New York Times*, page views of the NCI's preventive mastectomy fact sheet increased 795-fold compared with views one week previously. Use of other cancer genetics resources, including fact sheets for skin and prostate cancer, also had large increases in the days following Jolie's announcement. In addition, resources intended for health professionals experienced a similar surge in Internet traffic. The authors suggest that the "Jolie effect" extended to health-care providers attempting to learn more about *BRCA1* and *BRCA2* by accessing PDQ information. Such instances offer genetics professionals an opportunity to educate patients about credible sources of online health information and highlight the opportunity for celebrity announcements to serve as teachable moments for both the public and medical professionals. —Karyn Hede, News Editor

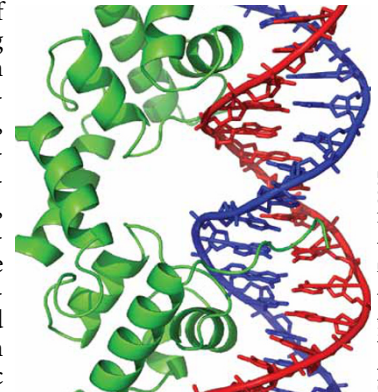


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Utility of gene-expression profiling in breast cancer still unproven

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Despite widespread use of gene-expression profiling to assist clinical decision making in women diagnosed with breast cancer, significant questions remain concerning its prognostic usefulness. In 2009, the Evaluation of Genomic Applications in Practice and Prevention Working Group (EWG) found evidence of an association between the prognostic ability of two gene-profiling systems—MammaPrint (Agendia) and Oncotype DX—and actual disease recurrence. Only MammaPrint is approved by the US Food and Drug Administration for determining the risk of distant recurrence in women less than 61 years old with stage I or II lymph node-negative early breast cancer. The two products measure gene expression in entirely separate sets of genes. Marrone et al. report that a new compilation of systematic reviews evaluating the clinical utility of the two products reveals no direct evidence that use of the tests improved outcomes of women with breast cancer. The reviews do show that use of the tests often led to a change in treatment. Six studies reported that 13–34% fewer patients received chemotherapy, and one study reported that 27% of patients changed their own treatment decisions after gene-expression testing. Two ongoing clinical trials, TAILORx and MINDACT, are currently evaluating whether using Oncotype DX and MammaPrint to guide treatment decisions for women with early-stage breast cancer changes patient outcomes. These findings are intended to provide input toward an updated recommendation from the EWG. —Karyn Hede, News Editor

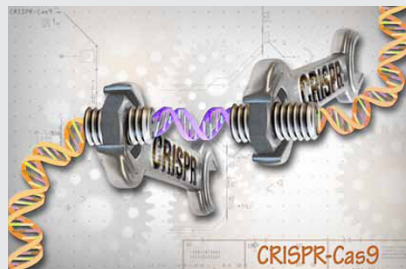


Richard Wheeler (Zephyris) 2007

NEWS BRIEFS

Gene editing of human embryos in China prompts federal ban in US

The recent announcement by scientists in China that they had conducted gene-editing experiments on human embryos prompted a swift response by the National Institutes of Health (NIH), which banned federal funding on any similar human embryo research. NIH Director Francis Collins cited the "serious and unquantifiable safety issues, ethical issues presented by altering the germline in a way that affects the next genera-



Ernesto del Aquila, NHGRI

tion without their consent," in his 29 April 2015 statement. The response addressed the use of CRISPR/Cas9, a novel technique that allows scientists to insert

small pieces of DNA at precise locations. Adapted from a bacterial adaptive immunity response to foreign DNA, CRISPR/Cas9 has rapidly reduced the time it takes to produce mouse models of disease since its introduction only two years ago. The Chinese scientists reported in an 18 April 2015 research report published in the journal *Protein & Cell* that they had attempted to use the CRISPR/Cas9 system to edit DNA in embryos donated by an in vitro fertilization clinic. In their report, they state that the human homologous recombination-directed repair system required to complete the editing

NEWS BRIEFS *(continued)*

process was weak in the embryos and repair was incomplete. They also reported the cutting mechanism snipped embryonic DNA in places that the scientists did not intend, further damaging them. The report divided scientists worldwide, with some advocating nonreproductive uses of the gene editing system in humans and others saying any human research should be off limits. For now, the NIH has reiterated that the ban for federally funded researchers remains firmly in place. —*Karyn Hede, News Editor*

Genetic testing for wellness at work?

Health insurer Aetna has a new twist on wellness by offering employees of its largest corporate customers genetic testing to identify risk factors for obesity as part of a larger exercise and weight-loss program. It is the first insurer to venture into using genetic tests as part of a preventive wellness program. Aetna announced earlier



this year that it had signed a commercial agreement with the Canada-based company Newtopia, which offers the service. Specifically, the program targets metabolic syndrome, a loosely defined condition that may include obesity, poorly controlled blood sugar, and high cholesterol among other markers that increase diabetes risk. The test looks at genes encoding three proteins: fat mass and obesity-associated protein (FTO), melanocortin receptor 4 protein (MC4R) and dopamine receptor

D₂ (DRD2), variants of which have been suggested to increase the risk of obesity. The remainder of the wellness package looks a lot like other weight-loss programs, with a personalized diet and exercise plan and a “coach” available to provide information on nutrition and behavior management. The company tested the program on its own employees and on employees of The Jackson Laboratory, the nonprofit mammalian genetics research organization based in Bar Harbor, Maine. Now it is seeking to sign up corporate clients willing to bet that motivating employees to reduce their health risks by losing weight will pay off in reduced health-care costs. The scientific and clinical use of this program is questionable, at best. Moreover, whether thinking your genes explain why you pack on more pounds than your co-worker in the next cubicle is motivating or crushing remains an open question.

—*James P. Evans, Editor-in-Chief, and Karyn Hede, News Editor*

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.