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Knowledge of carrier status low among relatives of CF patients

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Diagnosis of a newborn with cystic fibrosis (CF) has consequences not only for the parents but also for the extended family. With a carrier frequency of 1 in 25, the CF mutation is the most common serious recessive condition among northern Europeans. After diagnosis, parents face issues with regard to informing family members about their increased risk of being a carrier. Little is known about how these conversations affect the subsequent decision by relatives as to whether to be tested for carrier status. McClaren et al. studied this communication process and decisions regarding testing among a population of patients at Royal Children's Hospital in Melbourne, Australia. Parents of newly diagnosed children were counseled about the implications for immediate-family members and were provided with a sample letter to assist in communicating the information. Despite these efforts, only 11.8% of relatives accessed the free carrier testing available at the hospital. Follow-up interviews with parents and extended family of 30 newly diagnosed children revealed that notification was uneven and depended on personal relationships among family members. Parents reported that they were comfortable with their own understanding of the genetic cause and inheritance of CF but that they felt the concepts were difficult for their relatives to understand. The study's results indicated that relatives outside the immediate family were less likely to have been tested. The authors suggest that, after a period of adjustment, additional contact by health-care providers with parents to discuss implications for relatives might be useful in increasing family members' knowledge. *—Karyn Hede, News Editor*

KRAS testing in metastatic colorectal cancer patients

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Colorectal cancer tumors that harbor specific mutations affecting epidermal growth factor receptor (EGFR) pathway signaling have been shown to be generally unresponsive to therapy with anti-EGFR antibodies such as cetuximab and panitumumab. The Evaluation of Genomic Applications in Practice and Prevention Working Group recently reviewed the evidence in order to determine the implications of these observations and concluded that clinical use of *KRAS* mutation analysis is warranted for metastatic colorectal cancer patients (the recommendation statement appears in full in this issue). The group concluded that analyses to identify those who are *KRAS*-mutation-positive would benefit patients by avoiding potentially

NEWS BRIEFS

DNA collection for tracking human-rights victims

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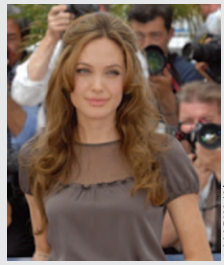
The exquisite specificity of DNA-sequencing technology to identify individuals makes it an attractive approach for helping to curb human trafficking by identifying its victims. One such program, DNA-PROKIDS, is an international collaboration initiated by the University of Granada Genetic Identification Laboratory and involving the University of North Texas Center for Human Identification. The group seeks to establish international DNA registries to identify trafficked children and reunite them with their families. Similar programs are already up and running. However, government collection of DNA from children raises questions regarding civil rights and justice as well as numerous ethical concerns. In a commentary published online by *Trends in Genetics* (15 May 2013), Joyce Kim and Sara Katsanis of the Duke Institute for Genome Sciences & Policy, Durham, NC, discuss the issues arising from use of DNA to track individuals not accused of crimes. The authors point out that, although applying DNA technologies to identify victims of human-rights violations is a "noble cause," collection of genetic information itself can also lead to human-rights violations. They address the social concerns facing efforts to implement DNA programs for the identification of victims and outline how to define best practices for the establishment of international DNA registries. *—Karyn Hede, News Editor*

UK aims to make cancer gene screening common practice



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The announcement by actress Angelina Jolie that she had undergone a double mastectomy in an effort to stave off breast cancer increased public awareness of genetic testing for cancer predisposition. In Jolie's case, the presence of a *BRCA1* mutation known to greatly increase her risk of breast or ovarian cancer, along with having a first-degree relative who died of breast cancer, made the decision clear-cut for her. Now, more women may be faced with making such choices in the United Kingdom, as the National Health Service (NHS) pilots a

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ineffective and harmful treatment. Another conclusion is that, whereas there are convincing data to support *KRAS*-mutation testing, there is inadequate evidence that testing for the *BRAF* V600E mutation or loss of PTEN expression improves health outcomes. Nor was evidence found of improved health outcomes among those tested for *NRAS* or *PIK3CA* variants or AKT protein expression levels in this patient group. The working group's recommendations include consideration of the fact that eliminating anti-EGFR therapy as an option following a positive test for *KRAS* mutation may promote quicker access to potentially more effective treatment. *—Karyn Hede, News Editor*

NEWS BRIEFS

model program to test women with breast or ovarian cancer using a panel of nearly 100 cancer-predisposition genes. The three-year program, called Mainstreaming Cancer Genetics, is led by Britain's Institute of Cancer Research and the Royal Marsden NHS Foundation Trust, with funding by the Wellcome Trust. It will be focused on breast and ovarian cancer patients at first, but the plan is to expand it to many more types of cancer, eventually making cancer genetic testing a routine part of cancer care. As part of the initial study, organizers also plan to consider the ethical and social impact of genetic testing in cancer patients and to evaluate outcomes—as well as cost, a controversial issue highlighted by the high price of the patent-protected test that revealed Jolie's cancer susceptibility. *—Karyn Hede, News Editor*