Genomics: Lots of press, but how much progress?

Despite the mapping of the human genome, the hundreds of millions of dollars invested in genetic research, and the hype about the transformation of basic science, physicians can be forgiven for questioning when—or whether—the genomics revolution will lead to improvements in patient care. At press time, a glimmer of an answer appeared.

In January, the federal entity charged with evaluating the clinical usefulness of genetic applications recommended that patients with newly diagnosed colorectal cancer should be offered testing for Lynch syndrome, a rare inherited condition associated with an elevated risk for this type of cancer.

EGAPP’s first definitive recommendation
Aimed at reducing morbidity and mortality in relatives of colorectal cancer patients, testing for Lynch syndrome is the first definitive recommendation to come out of Evaluation of Genomic Applications in Practice and Prevention (EGAPP). The group, which is made up of clinicians and researchers steeped in evidence-based medicine, was formed in 2004. But until this January, EGAPP had issued only one report, in the fall of 2007.

In the 2007 report, EGAPP examined the usefulness of genetic testing for cytchrome P450 (CYP450) before prescribing SSRIs. Testing for this marker, EGAPP reported, was not ready for prime time. Actually, EGAPP said “I”—there was insufficient evidence to recommend for or against testing for this genetic marker.

In January, EGAPP issued 2 additional “I” ratings. The group found insufficient evidence to make a recommendation for or against both tumor gene expression profiling (eg, Oncotype DX) in women with breast cancer and UGT1A1 genotyping for patients with metastatic colorectal cancer being treated with irinotecan.

Physicians are no strangers to the frustrating “I,” which leaves us pretty much where we were before the extensive evidence review and deliberation process began. We also tend to be a skeptical bunch. I suspect that most physicians take an “I” to mean a vote of no confidence, early adopters notwithstanding.

But “I” is an accurate reflection of the state of the evidence for the vast majority of genetic tests available today. Genomics should not get a free pass in today’s health care system, no matter how much money and marketing are devoted to it.

More than 1000 genetic tests, much uncertainty
The biggest problem facing EGAPP is the genetic testing deluge. There are already more than 1000 genetic tests on the market. And the excitement about genome-wide association studies (GWAS)—an
approach that involves rapidly scanning for markers across the entire human genome to identify genetic variants associated with particular traits and diseases—promises to yield even more questionable clinical applications.

So what are physicians to do, faced with all of this uncertainty? The same thing we’ve always done. Use good clinical judgment, review the available evidence, cut through the hype, and practice medicine for the good of our patients.

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References

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