John Hickner, MD, MSc
Editor-in-Chief

Will screening open Pandora's box?

If it ain't broke, don't fix it" or "A stitch in time saves nine"—which do you prefer?

When I taught epidemiology at the University of Chicago, I asked first-year medical students that question before discussing the science of screening for early detection of disease. Each year, the class was about evenly divided. Their split response reinforced to me the need for shared decision making when we offer screening tests to our patients.

Shared decision making is especially important in light of new evidence about the effectiveness (or lack thereof) of some screening tests. Several bread-and-butter screening procedures and tests promoted for years have been debunked as having no value (routine testicular exam and monthly self-breast exam), having harms that might outweigh the benefits (PSA for prostate cancer), or having marginal benefit for those in certain age groups (mammography in women younger than 50). And, as treatments for cancer get better and better, screening will have less and less value.

What would a 30-year-old do if he found out he has a gene that makes him susceptible to Alzheimer's disease?

The biggest screening test challenge, however—genome screening—is still to come. Genomic sequencing analysis is already useful for the diagnosis of certain genetic disorders and for treatment decisions in certain cancers. Genomic sequencing to screen for disease, however, is fraught with ethical challenges and the absolute need for shared decision making.

What if gene analysis uncovers "incidental findings" about risk faced by asymptomatic patients, like the "incidentalomas" described in "When to worry about incidental renal and adrenal masses" on page 476? The debate about what to do with incidental findings from genetic analysis is heating up because of the American College of Medical Genetics and Genomics' recent recommendations[1] to automatically screen for 56 genes that may contain "potentially important" findings when genome sequencing is done for any reason.

Talk about Pandora's box! Suppose a 30-year-old finds he carries a gene that makes him susceptible to Alzheimer's disease. What would he do with that information, other than get depressed when he realizes there are not yet any effective early interventions?

Family physicians are likely to be asked more and more questions about genome analysis.* Be prepared. You can start by asking patients whether they adhere to an "If it ain't broke…" " or "A stitch in time…" approach.

1. Green RC, Berg JS, Grody WW, et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genet Med*. 2013;15:565-574. Available at: https://www.acmg.net/docs/IF_Statement_Final_7.24.13.pdf. Accessed August 20, 2013.

jfp.eic@gmail.com

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