Q&A Summary
Pharmacogenomics: Providing Personalized Medicine
April 28, 2020

Presenter: Russ B. Altman, PhD, MD, Kenneth Fong Professor of Bioengineering, Genetics, Medicine, Biomedical Data Science and (by courtesy) Computer Science, Stanford University, Stanford, CA

Available On Demand:
optumhealtheducation.com/pharmacogenomics-2020

1) Which practitioner/health care professional (HCP) is responsible for initiating pharmacogenetic testing?

At Stanford, we have a consult service, so this comes from mostly primary care physicians of individuals who have particularly difficult drug-management situations. In other places, this can be part of a clinical pharmacology consulting unit. Eventually, I hope this will be in the hands of primary care physicians who will use it at the point of decision making.

2) What is the availability of pharmacogenetic testing?

There are several companies that offer testing; typically, a sample must be mailed, and the report takes about a week or two to return.

3) Is there a national directory for centers that do genetic testing?

Not to my knowledge, no. These tests are primarily done in academic centers, but some specialty practices have also emerged.

4) What are the primary barriers to broad adoption of pharmacogenomics in health care today?

First, testing and interpretation will need to be integrated into the routine electronic health record. Second, HCPs must be aware of when it can be useful. Third, and most importantly, there needs to be reimbursement for it.

5) How "mainstream" is genetic testing for things such as chemotherapy, since this impacts the likelihood of it being covered by Medicare and commercial payers?

Some cancer pharmacogenetics, like the testing done before administering Herceptin (trastuzumab), is quite routine. My talk was about non-cancer applications where I would say that pharmacogenomics is not yet mainstream.

6) How do we discuss this topic with individuals who are nervous about having their "genetic data out there" and concerns about future denial of health care or life insurance coverage?

This is a legitimate concern. The Genetic Information Nondiscrimination Act (GINA) gives some protections but not complete protection. Medical records are secure, but t not absolutely guaranteed, we hear about security breaches in the news regularly. Fortunately, current tests only test a small fraction of the genome, so these results may not be identifiable. If we move to whole-genome sequencing, these issues (including incidental findings and others) would become more pressing. However, the entire medical record is already very identifying (even without genetic data), and so this is part of that same issue.

7) What recommendations would you give psychiatrists who are trying to determine when/if testing would be beneficial for an individual?

Many selective serotonin reuptake inhibitor (SSRI) drugs have variations in metabolism related to genetics. Thus, the approach of using pharmacogenomics for the choice of first antidepressant is reasonable to me, as a physician. Certainly, if an individual has failed first-line therapy, it could also be useful for focused choice of a second-line agent. Since depression it is an urgent prescribing decision anything to find success without experimenting with multiple drugs would be valuable.

8) With the opioid epidemic, will pharmacogenetic testing help delineate the success of rehabilitation?

This is an intriguing idea, and there are studies in this direction, but I am not aware of any definitive trials or guidelines in this area yet.

9) Can you comment on the status of pharmacogenetics related to treating hypertension?

There currently no Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for hypertensive medications, so it is not a major target currently for pharmacogenomics implementation.

10) How well (or not) do you see regulatory agencies such as the US Food and Drug Administration (FDA) trying to address these areas of opportunity?

The FDA are aware of pharmacogenomics technology but are still trying to find the right balance of regulatory oversight policies. Part of the challenge comes because pharmacogenetic tests are regulated by the Center for Diagnostics and Radiological Health (CDRH) but considerable pharmacogenetic expertise also lies in the Center for Drug Evaluation & Research (CDER).

11) Can these practice guidelines be extrapolated to pediatrics, or are they only for adult patients?

Pediatrics is an area of great need for more pharmacogenetic research. There are a few examples that show the great importance of genetic variation in children, but it simply has not been sufficiently studied. The metabolizing capabilities of the liver in children changes from birth to adulthood, and this must be fully understood to implement pharmacogenetics appropriately for children.

If you have questions regarding this document or the content herein, please contact: moreinfo@optumhealtheducation.com.