J Genet Counsel (2010) 19:419–421
DOI 10.1007/s10897-010-9292-2

COMMENTARY

Uhlmann Rejoinder to: Taubman's "Letter to the Editor"

Wendy R. Uhlmann

Received: 16 February 2010 / Accepted: 24 February 2010 / Published online: 1 April 2010

Keywords Genetic testing · Genetic counseling · Insurance coverage · DNA banking · Genomic medicine

The challenges of integrating new genetic tests into healthcare, using the example of hereditary breast cancer, were recently raised by Resta (2009) in his Commentary "Unprepared, Understaffed and Unplanned: Thoughts on the Practical Implications of Discovering New Breast and Ovarian Cancer Causing Genes" and in Commentary responses by Hampel (2009) and Uhlmann (2009). I appreciate the thoughtful points that Dr. Edward Taubman makes in his Letter to the Editor (2010) about these Commentaries. While I agree with Dr. Taubman that genetic testing companies and insurers are key players, I respectfully disagree about the roles he proposes for each of them in informing patients about genetic testing advances.

Dr. Taubman accurately notes that insurers' policies limit who can be covered for testing but I disagree that they have a responsibility "to help inform their clients when something more informative comes along." It is impractical for insurance companies to keep on top of genetic testing advances and inform patients, especially given the large number of genetic conditions and the fact that laboratories may offer different tests for the same condition. There are over 6,800 genetic conditions (Online Mendelian Inheritance in Man [OMIM]) and currently there are over 1,600 genetic tests with a couple hundred more in the research pipeline (GeneTests). Testing decisions can be complex because depending on the genetic condition, laboratories can differ in genes and mutations tested, methodologies, test sensitivities and costs; therefore, determining whether genetic testing is indicated, which test should be ordered and appropriate laboratory selection are critical. Determining whether genetic testing is now more informative for a patient will require genetics/specialists' expertise, especially since few genetic tests currently have practice guidelines and/or are considered standard of care. Insurance companies have a limited number of physicians to review coverage requests for testing and genetics background of most physicians is also limited.

What is practical is to work with insurance companies for better coverage of genetic testing. As noted in the Secretary's Advisory Committee on Genetics, Health and Society 2006 report on Coverage and Reimbursement of Genetic Tests and Services, problems with coverage and reimbursement of genetic tests and services are limiting their accessibility and integration into the health care system. I propose that genetic tests should be ordered by primary care physicians if consistent with practice guidelines and/or considered standard of care and other genetic tests should be ordered by geneticists/specialists, given the expertise needed to make genetic testing determinations, select the laboratory and interpret test results. To increase efficiency in making insurance coverage decisions about genetic testing, I propose the following:

- If genetic testing is ordered consistent with practice guidelines and/or is considered standard of care for a condition, it should be covered.
- If a geneticist or other specialist is requesting a genetic test, it should be covered. Sure, some tests may be inappropriately ordered but in general, paperwork and time to review testing requests and insurance

W. R. Uhlmann (✉)
Department of Internal Medicine,
Division of Molecular Medicine and Genetics and Department of Human Genetics, University of Michigan,
Ann Arbor, MI, USA
e-mail: wuhlmann@umich.edu

claims could be cut significantly if it was presumed that the specialist physician ordering the test used their expertise to make this determination. The geneticist/specialist in making the decision to offer the patient a genetic test should give careful consideration as to how good is the test/how likely is it that a mutation(s) will be identified, whether test results would impact their patient's medical care, life decisions and/or have implications for other atrisk family members.

- When a patient with a genetic condition has at-risk family members, cover the genetic test. For many genetic conditions, gene mutation(s) need to be identified in an affected family member first before at-risk family members can be tested. While other at-risk family members may not be insured by the same insurer, insurers' agreement to cover genetic testing on these affected individuals would globally enable at-risk individuals in their and other insurance plans to be tested. For a number of genetic conditions, ruling out familial gene mutation(s) in at-risk family members can save healthcare dollars by eliminating the need for costly tests and evaluations that would otherwise regularly be needed for surveillance. For at-risk family members who have positive genetic test results, timely surveillance/medical care can be initiated, which can potentially reduce complications and also globally lower healthcare costs.
- Additional genetic testing by different methodologies and retesting in the future may be indicated and this should be covered if a geneticist/specialist makes the testing request.
- Cover DNA banking so cost is not a barrier for patients.

It is also impractical for genetic testing laboratories to have the responsibility of helping to notify patients though Dr. Taubman makes the excellent point that they have databases with patients' test results and could easily identify who needs retesting. Some laboratories do send clinicians announcements about test changes and new genetic tests and may even send updated reports of results when testing advances change the interpretation of previous test results. While commercial laboratories may have more resources to inform patients about testing advances, many genetic tests are performed in academic laboratories that generally lack resources to mount media campaigns and recontact patients. There are other logistical challenges in recontacting patients discussed in the Commentaries by Resta (2009), Hampel (2009) and Uhlmann (2009) including time required, patients relocating and for insurers and genetic testing laboratories, the fact that some may cease to exist. Globally in medicine, we do not expect testing laboratories and insurers to notify patients about the availability of more informative testing—why should this be different for genetic testing?

What genetic testing laboratories can do is 1) include information on their website on test parameters (e.g. sensitivity) to make it easy to ascertain when advances have been made for genetic tests that they offer 2) note in reports if other genetic testing methodologies and/or retesting in the future should be considered 3) note that a clinical genetics evaluation may be indicated (a genetic test does not take the place of a genetics evaluation and other conditions and genetic tests may be diagnostic considerations) 4) include in their test reports that DNA banking should be considered when no mutation(s) are identified in an affected individual, particularly if the condition is life-limiting, to preserve the option for future testing. If a genetic testing laboratory does not offer DNA banking, consideration should be given to contracting with a laboratory that does in order to make this service readily available to their patients. As Dr. Taubman notes, unfamiliarity with DNA banking is "limiting how often it is done."

Dr. Taubman proposes just having a box on genetic testing forms that patients simply could check to have their DNA banked when having genetic testing done. However, consenting patients for DNA banking is quite different than consenting patients for a genetic test. For DNA banking, one must specify to whom the sample can be released for future genetic testing. Family dynamics may complicate who the family member is willing to release the sample to and family members who may need access to the sample in the future may be denied access or may not yet be born. In addition, there will be potential ethical issues including a family member accessing a banked DNA sample when other at-risk relatives in the family line have decided not to learn their carrier status; testing could potentially reveal the carrier/future affected status of family members who have decided not to learn this information. Relatives' knowledge that DNA has been banked on a family member and where it has been banked are an issue. A DNA bank in each state or a national centralized DNA bank could solve the issue but this would then raise more issues that would need to be resolved including use, privacy and access. It is important to globally address DNA banking issues including length of storage; family members, clinicians and researchers sample access; and release of samples for future testing and to educate patients, physicians and other allied healthcare providers on the availability of DNA banking.

Patients' personal medical and family histories are not static and neither are genetic testing and other aspects of medical care. The onus needs to be on patients to seek regular healthcare and inquire whether advances have been made in testing that will benefit their care (Uhlmann 2009). Determining when a test is ready to be a test (particularly a challenge for rare genetic conditions) and communicating genetic testing advances to patients and healthcare providers will take collaboration with the genetics community, laboratories,

groups involved in genetic testing oversight, insurers, disease advocacy groups and other key stakeholders.

Incorporating advances in genetic testing and genomic medicine in healthcare will take a multi-pronged approach and involve many stakeholders. As Dr. Taubman notes, genetic testing companies and insurers are two key team members that need to be involved. The time to engage in discussion is now and we need to work on hearing from diverse stakeholders, figuring out how to best inform patients and healthcare providers about advances so that genetic testing and genomic medicine are successfully integrated into healthcare.

References

GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1993–2010. Available at http://www.genetests.org. Accessed February 9, 2010.

Hampel, H. (2009). Recontacting patients who have tested negative for BRCA1 and BRCA2 mutations: how, who and why? *J Genet Counsel, 18*, 527–529.

Online Mendelian Inheritance in Man, OMIM (TM). McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD), Accessed February 9, 2010. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/.

Resta, R. G. (2009). Unprepared, understaffed and unplanned: Thoughts on the practical implications of discovering new breast and ovarian cancer causing genes. *J Genet Counsel, 18*, 521–523.

Secretary's Advisory Committee on Genetics, Health and Society. (2006). Report on coverage and reimbursement of genetic tests and services. http://oba.od.nih.gov/oba/sacghs/reports/CR_report.pdf.

Taubman, E. (2010). Letter to the Editor: Re: Unprepared, Understaffed, and Unplanned. *J Genet Counsel*. doi:10.1007/s10897-010-9290-4.

Uhlmann, W. R. (2009). Response to Robert G. Resta Commentary (Unprepared, understaffed and unplanned: Thoughts on the practical implications of discovering new breast and ovarian cancer causing genes). *J Genet Counsel, 18*, 524–526.