Genetic Counselors and Testing

Pennsylvania is currently the only state with genetic counselor licensure to explicitly disallow their ability to order genetic testing. This is not in the interest of patients and leads to inefficient delivery of genetic services as patients get referred back and forth between physicians and genetic counselors needlessly. Allowing genetic counselors to order genetic testing will enable patients to receive convenient high quality genetic services as evidence based research demonstrates that genetic counselors order the right test, at the right time, for the right patient.

**Why should genetic counselors be allowed to order genetic testing?**

*Research Demonstrates Genetic Counselors Order Appropriate Tests*

There are a considerable number of studies in the literature that show that incorporating genetic counselors into the testing process leads to cost-efficient risk identification and more appropriate genetic test orders. This results in significant savings in healthcare spending for patients with cancer, neurological disorders, cardiac disorders, and other genetic diseases. Several payers and providers have calculated the savings within their programs once genetic counselors were included in the process of ordering genetic testing for their members. An increasing number of publications now exist in the literature that demonstrate the value proposition and potential savings that can be achieved by incorporating genetic counselors into this process, before tests are ordered or in a pre-authorization framework. Ensuring patients receive the most appropriate test leads to better health outcomes.

*Genetic Counselors have the Unique Training to Order the Correct Test*

Genetic counselors make appropriate test selection whereas many non-genetics specialists do not. This includes better identification and utilization of genetic tests that have adequate supporting evidence (e.g., targeted mutation testing when appropriate vs. comprehensive full-gene sequencing).

More recently, primary care physicians (PCPs), and providers such as advanced practice nurses have been called upon to provide these highly specialized services without appropriate training, and often in a setting with significant time constraints. Although many PCPs are well-equipped to screen and refer individuals who may be at an increased risk for hereditary conditions, the vast majority of PCPs are not adequately trained to perform genetic counseling and risk assessment (1-4). Physicians’ self-reported levels of knowledge in basic genetics and cancer genetics indicate that they have not received adequate training to deliver these services (3, 4) and the vast majority (>75 percent) felt that they needed more training on when to order the test, how to interpret the test, and how to counsel the patient (3).

In a survey-based study of family physicians, 54.4 percent of respondents felt that they were not knowledgeable about available genetic tests (5). Furthermore, one-third to one-half of PCPs and naturopaths reported that they do not refer to a genetics professional even when they suspect that their patient has an increased risk for a serious hereditary cancer syndrome (4). Uninformed and inefficient practice may detract from the promise of genetics as a powerful tool to promote health and reduce suffering from disease. Appropriately trained genetic counselors are ideally suited to provide these increasingly essential services in the most cost-effective and efficient manner, and therefore should have the ability to order tests in Pennsylvania.

*Misuse of Genetic Testing is Occurring*

A survey of practitioners who self-identified as providing cancer genetic services in their practices assessed knowledge and clinical practices around genetic counseling and testing for Hereditary Breast and Ovarian Cancer. Results of the study showed that many non-genetics providers are not ordering the appropriate genetic test for their patients. Less than 40 percent recognized the need for rearrangement testing after standard BRCA testing was negative in a woman at 30-percent risk, which could result in missing a mutation in a high-risk family. Less than 15 percent recognized appropriate follow-up testing for a BRCA variant of uncertain significance – 43 percent of non-genetics providers stated that they would order a $3,340 comprehensive test on an unaffected family member, when in fact no testing is warranted in this scenario (9).

A case-based survey study conducted on 225 non-genetics practitioners in Texas showed similar results, with the majority of providers indicating that they would order more expensive and often unnecessary tests. The total cost of testing if the National Comprehensive Cancer Network guidelines had been followed for each of these cases was $950/case. The average total cost of testing based on respondents’ answers was $ 10,000 per case, a nine-fold difference in cost (10).

In 2011, Shah et al. showed that BRCA 1/2 tests were frequently inappropriately requested by physicians. Twenty-five percent of individuals with a personal cancer diagnosis did not meet coverage criteria, had a clinical history that suggested a different cancer syndrome, or had a relative who would be more appropriate to initiate testing. For those unaffected patients in which testing was based on family history alone, the proportion of inappropriate test orders was much higher – greater than 40 percent (6).

*Cost Effectiveness of Genetic Counselors*

Many studies present quantified savings from: 1) reducing inappropriate tests that are not clinically indicated, 2) reviewing genetic test orders to ensure that they are in accordance with evidence-based medicine, and 3) reducing unnecessary treatment. A study performed by a large genetic testing reference lab compared tests ordered by non-genetics and genetics providers. After review of the test request forms by a certified genetic counselor and follow-up with the ordering physician, the study revealed that up to one-third of tests ordered by non-genetics providers were inappropriate. Modification or cancellation of the tests ordered resulted in savings of $402,357 over a 10-month period. Reasons for inappropriate testing included ordering the wrong test (e.g. ordering Hereditary Hemorrhagic Telangiectasia sequencing when the provider intended to order the Hemochromatosis Mutation Panel) and ordering full-DNA sequencing when a less expensive, targeted mutation test was needed (8).

Some argue that misuse of testing would be largely avoided if payers developed rigorous genetic testing policies. Even in this scenario, however, misuse and overuse of testing can occur. Data that Priority Health (a Michigan-based health maintenance organization) presented in 2009 showed that despite stringent genetic testing criteria, increased utilization of BRCA testing occurred after a genetic testing company launched a direct-to-physician marketing campaign in 2002. When the policy was accompanied by clinical genetic counseling and risk assessment by a certified genetic counselor, 25-33 percent of the genetic tests that initially seemed appropriate according to the documentation of the ordering provider would have been avoided. Based on Priority Health’s authorization form, approximately 10,000 beneficiaries initially met the criteria for BRCA genetic testing. The plan’s mandatory independent genetic counseling more accurately assessed these criteria and prevented over $10,000,000 worth of inappropriate tests. The cost of genetic counseling for these 10,000 people was estimated to be $2,800,000, resulting in an overall savings of $7,220,000 (Priority Health data).

The aforementioned evidence demonstrates that licensed genetic counselors should have the ability to order genetic tests. Many states allow this practice. Such a policy would not preclude other healthcare providers who have the ability to order tests from doing so, but would allow uniquely trained healthcare practitioners, genetic counselors, to do so as well. Adopting this policy would protect the public from unnecessary medical expense and ensure that they receive appropriate testing and follow-up care.

1. United Healthcare Working Paper 7, March 2012; www.unitedhealthgroup.com/reform.

2. Secretary’s Advisory Committee on Genetics, Health, and Society’s Executive Summary on the Oversight of Genetic Testing 2008; http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS\_oversight\_report.pdf

3. Klitzman, R et al J Genet Counsel 2012; epub 15 May 2012:1-11.

4. Cox, et al J Cancer Epidemiol. 2012;2012:294730. Epub 2012 Oct 24.

5. Mainous AG, et al Fam Med. 2013 Apr; 45(4): 257-62.

6. Mayo Medical Laboratories. (2012) Communique: Improving Patient Care through Esoteric Laboratory Testing. May/June: Volume 37(3):6.

7. Shah A, Harris H, Brown T, et al. (2011) Analysis of insurance preauthorization requests for BRCA1 and BRCA2 genetic testing: experience of the Humana Genetic Guidance Program. Personalized Medicine, Vol. 8(5): 563-569.

8. Arup Laboratories: Value of Genetic Counselors in the Laboratory. (2011)

9. Pal T, Cragun D, Lewis C, Doty, et al (2013). A statewide survey of practitioners to assess knowledge and clinical practices regarding hereditary breast and ovarian cancer. Genet Test Mol Biomarkers. 17(5):367‐75.

10. Plon SE, Cooper HP, Parks B, et al (2011) Genetic testing and cancer risk management recommendations by physicians for at‐risk relatives. Genetics in Medicine; 13(2): 148‐154.