**Introduction**

At the request of Delegate Shane Pendergrass, Vice Chair of the Health and Government Operations Committee, the Department of Health and Mental Hygiene conducted a review of the laws and policies of other states and conducted a literature review on the potential harms and benefits of allowing direct-to-consumer genetic testing (DTC) in Maryland. Since that time, the Department has completed a review of the literature on the harms and benefits as well as the clinical utility of services offered by DTC companies. Additionally, the Department has included a review of an expanded set of state policies and laws that have been used to protect consumers in their interaction with DTC genetic testing companies.

**Review of the clinical utility of services offered by DTC genetic testing companies:**

Direct-to-Consumer genetic testing companies offer a wide range of tests, from those with little clinical utility, such as a genes determining eye color, to those that evaluate serious medical conditions, such as the BRCA breast cancer genes (1).

Validity of testing relies on two components: (a) analytic validity, whether the lab accurately analyzes the biologic sample and (b) clinical validity, whether the result from the lab is interpreted in a clinically meaningful way. In the United States, the Clinical Laboratory Improvement Amendments of 1988 (CLIA) regulates laboratories that provide testing services and helps to ensure the analytic validity of services. After some initial concerns, now most DTC companies in the U.S. are regulated by CLIA (1).

In a 2013 study by Kalf and colleagues examined how the results of genetic testing were interpreted by 3 companies. Investigators created genotypes for a hypothetical population of 100,000 individuals and then calculated the predicted risks of disease using the methods published on the websites of 23andMe, deCODEme and Navigenics. The companies used different sets of single nucleotide polymorphisms (SNPs) and different average population risks for many of the diseases tested, resulting in substantially different predictions for the risk of the disease in individuals. For example, 27.1% of the hypothetical individuals would have received opposite responses from the different companies regarding their risk of Crohn's disease (2).

Of note, the company 23andMe now offers BRCA testing, a test that is characteristically different than many of tests that were evaluated in these studies. BRCA testing evaluates genomes for a specific gene, rather than SNPs, which give less specific information about disease risk. BRCA carriers have a greatly increased risk of breast cancer and ovarian and carrying this gene has potential profound health implications, with many people recommended to undergo a prophylactic mastectomy (3). This and other high risk testing on the market, have high clinical utility, but present their own potential risks to patients.

**Literature Review:**

In our previous communication to Delegate Pendergrass, the Department reviewed several studies of actual and hypothetical users of Direct-to-Consumer genetic testing. This review demonstrated mixed

findings on the impact of DTC genetic testing on patient anxiety and healthcare utilization. Since that time, a few studies have been published that further address these issues.

*Effects on Health Status and Behaviors:*

A large long-term evaluation of users of DTC genetic testing was recently published. In this study by Bloss and colleagues, 3,416 study participants initially purchased a genomic test and 1,325 had long-term follow-up over 1 year. The authors investigated the impacts of testing participation on patient anxiety, fat intake and exercise. In the 3 month follow-up study, published in the New England Journal of Medicine, and the year-long follow-up, participation in the DTC genetic testing service had little impact on these patient outcomes. Ninety seven percent of the sample had no test related distress. Of note, the genetic testing involved in this study did not involve more specific testing such as BRCA tests (4,5)

A small exploratory study by Wasson and colleagues followed 20 patients recruited from an urban primary care clinic. These patients were offered free DTC genetic testing from 23andme and were interviewed periodically over the course of one year. The panel of testing included a broad range of test results including testing for susceptibility to diabetes and varicose veins as well as BRCA testing. Many reported no significant impact from the testing, with several unable to recall the specific results of the study after 1 year. Most patients reported feeling relieved or pleased by their results. The authors did not report what specific results patients received and in such a small sample it would unlikely for these patients to have positive results for any of the rare, serious genetic diseases that were screened (6).

*Effects on Health Utilization:*

Bloss and colleagues also evaluated participants' use of health services, in particular their increased use of screening tests. Across the entire study population, the researchers did not find an increased rate of screening test completion according to patient self report. Thirty six percent of individuals in the overall study shared their test results with their physician and the sharing test results was associated with increased screening test completion. It is unknown if this increased screening represented increased compliance with screening recommendations or unnecessary, over- testing (4). In a 2012 study by Reid colleagues, 1,599 participants were offered genetic testing (that did not include BRCA testing), with 217 (13.6%) choosing to complete the testing. Utilization of health services was determined by evaluating health records. There were no significant differences in physicians visits or utilization of medical tests or procedures in the post-test period between those who did and did not choose testing (7).

The sharing of tests results with primary care physicians may present new counseling burdens on already busy practices. In 2012 nationally representative survey of primary care physicians by Bernhardt and colleagues, 58% of respondents felt confident interpreting genetic testing and with 40% feeling that results would be helpful in disease management (8). In most surveys, few physicians encountered DTC genetic testing frequently and no studies estimate the amount of time counseling may take (9).

Overall, there appears to be little impact of genomic testing on healthcare utilization. However, no well designed studies address the impact of DTC access to testing of higher risk studies, such as BRCA tests.

**Legal Action:**

DTC genetic testing has been challenging to regulate in part because no single agency clearly oversees all aspects of regulation (10). In our last communication, we noted that 37 states and the District of Columbia allow direct to consumer genetic testing. In 23 of the states that allow this type of testing, laws are silent on the issue, rather than explictly allowing and regulating DTC genetic testing. A handful of states allow limited direct to consumer testing for a select number of specific tests. There has been varied federal and state action taken to regulate DTC testing.

*Federal Action:*

The Food and Drug Administration (FDA) has the regulatory ability to oversee genetic testing, but has not to our knowledge clarified its regulatory approach (1,11). In 2010, the FDA sent several letters to DTC genetic testing companies indicating its intention to assert regulatory authority over them and held several public meetings to discuss possible regulatory approaches (1). Further regulatory action by the FDA is expected soon (12) .

The Federal Trade Commission (FTC) has authority to regulate the marketing of DTC genetic testing. On its website, it warns consumers to take at-home genetic testing with a "healthy dose of skepticism" (13). Thus far they have limited their action to the most flagrant false claims (1).

The Secretary of Health and Human Services' Advisory Committee on Genetics, Health and Society also weighed in on the regulation of DTC genetic testing. In its April 2010 report, it recommended a stratified approach such that higher-risk results would require more oversight than lower-risk results (19). This approach has been supported by other thought leaders as well (1).

*State Action:*

In addition to regulating access to DTC genetic testing, states have also enacted legislation to address misleading advertisement. According to a 2009 survey by the Genetics and Public Policy Center, 48 states have more general provisions addressing the false representation of the benefits of services provided (17). 4 states have more specific laws that could apply to DTC genetic testing. California and Nevada specifically outlaw the presentation of false or misleading scientific or medical claims and Nebraska and Pennsylvania explicitly outlaw false or misleading claims in privacy policies(18).

**Other policies and recommendations for regulation in the literature:**

Several medical associations have weighed in on approaches to regulating DTC genetic testing. The American College of Medical Genetics and the American Medical Association recommend that testing should only be performed with the guidance of a licensed healthcare provider (1). U.K. Human Genetics Commission and the European Society of Human Genetics, allow DTC but only with the involvement of

genetic counselor (1). American College of Medical Genetics suggests that consumers be specifically informed about the benefits and risks of genetic testing (14).

Many experts have raised concerns about the privacy of the potentially sensitive data held by DTC genetic testing companies. These companies are not required to meet HIPAA requirements (1). Special concerns have arisen when companies are sold or go bankrupt, which has occurred frequently in the DTC marketplace. It is often unclear what happens to the sensitive genetic information held by the defunct companies.

The U.S. Preventive Services Task Force recommends BRCA genetic testing only for patients with previous family history of breast cancer due to the likelihood of unnecessary stress and additional testing and treatment (15).

Secretary's Advisory Committee on Genetics, Health and Society raised concerns about the use of the resources of the DTC companies for research (16).

Finally, ethicists and policy makers have urged the prohibition of using DTC testing to evaluate third parties, minors and prenatal testing of fetuses(17) .

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**Questions for Public Comment:**

The Department would like to receive comments from impacted stakeholders and the public on whether DTC testing should be allowed in Maryland and if so, what, if any consumer protections should be put into place. Specifically, the Department would like comments on:

**1. Whether consumers should have direct access to genetic testing.**

**2. If direct access to genetic testing is authorized, what types of consumer protections (if any) should be included, such as:**

- **Quality controls;**
- **Disclosure requirements;**
- **Requirements for access to genetic counselors as part of the testing package; and**
- **Privacy requirements for companies conducting the testing.**

**3. If direct access to genetic testing is authorized, whether and what types of testing should be prohibited (such as testing for the BRCA gene or other high risk tests with clinical significance, or for prenatal testing of fetuses).**