

Potential Regulation of DTC Genetic Testing

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As direct-to-consumer (DTC) genetic testing services rise in popularity and prevalence, greater federal oversight of the system has become necessary in order to protect consumers. Proponents of DTC testing argue that removing the necessity of passing through a medical professional allows consumers increased autonomy, freer access to their results, and enhanced privacy. However, critics argue that the government should monitor the system because without regulation, consumers might receive results without adequate context. Consumers may base pivotal life decisions on the information received from genetic tests, including whether to have children, take certain types of medications, or take part in treatments, so ensuring that the information received by consumers is accurate should be a system subject to extensive federal regulation. The FTC and certain divisions of Health and Human Services such as the CMS, CDC, and FDA are responsible for regulating the system, but at this point, the federal government only formally monitors the quality of genetics testing facilities. In order to protect consumers, the federal regulatory system must exercise increased oversight in areas of lab quality control, test validity and reliability, availability of adequate genetic counseling, and accuracy of advertising.

The federal government must regulate the quality of genetic testing labs to ensure the analytic validity of tests. Currently, the 1988 Clinical Laboratory Improvement Act (CLIA) ensures the accuracy, reliability and timeliness of patient test results: “CLIA, which is administered by the Centers for Medicare & Medicaid Services (CMS), imposes basic requirements that address personnel qualifications, quality-control standards, and documentation and validation of tests and procedures” (Javitt 59). To some extent, CLIA helps ensure the analytic validity of tests, and the act ensures that tests deliver accurate

information as to whether a mutation is present or not. However, no special standards or specific procedures apply to genetic testing facilities, except for labs which conduct cytogenetic tests for chromosomal abnormalities such as Down's syndrome. In the future, many organizations propose that regulations should be modified to create specific standards and ensure total quality control. In 2006, three organizations, including the Genetics and Public Policy Center (GPPC) petitioned CMS to issue updated standards for genetic testing labs, but CMS denied the petition on the basis of cost concerns in 2007 (Huang "Who...?"). For most high complexity tests, CLIA requires facilities to undergo specialized proficiency testing, but even though genetic testing is considered high complexity, CMS has not created special requirements for genetic testing. Although genetic testing facilities are already monitored by CMS under CLIA, many organizations have argued that creating requirements specific to genetic testing labs will ensure higher quality control.

Beyond ensuring the analytic validity of tests through regulating the quality of genetic testing facilities, the clinical validity and reliability of tests should also be regulated by the federal government. The Federal Food, Drug, and Cosmetic Act grants the FDA the authority to regulate medical devices, which are defined as articles "intended for use in the diagnosis of disease or other conditions, or in the cure, mitigation, treatment, or prevention of disease" (Huang "FDA"). However, at the moment, the FDA distinguishes between lab-developed genetic tests and genetic test kits, and the organization only regulates genetic test kits. External manufacturers create and distribute test kits as a single unit, and these are considered medical devices and therefore regulated by the FDA. Lab-developed genetic tests, on the other hand, are not subject to regulation

because they are made within labs and considered in-house tests. The American Society of Human Genetics proposes that the “FDA should be involved in the regulation of genetic tests, whether they are packaged and sold as kits or provided as a laboratory service” (Hudson 1). Because the majority of DTC tests are considered lab-developed tests, the FDA should expand regulations to cover lab-developed tests as a way of ensuring the clinical validity of DTC tests. As it is, the FDA believes that lab-developed tests fall outside of its jurisdiction and therefore does not monitor whether the results of DTC genetic tests correlate with the presence, absence, or heightened risk of a disease. Assessing clinical validity is crucial because this determines whether genetic tests are accurate predictors of disease or other conditions.

Another area in which increased federal oversight might benefit consumers is in ensuring that adequate genetic counseling is associated with DTC testing. The nature of DTC testing is to bypass physicians, and only several companies provide clients with genetic counselors to help them interpret their results, including Navigenics. In a service provided by AtlasGene described in the New York Times, parents subject their toddlers to ACTN3 testing in order to figure out whether their children will excel at endurance or fast-twitch sports. However, experts say that without the help of a genetic counselor, parents may not realize that “athletic performance has been found to be affected by at least 200 genes” (Macur 1). Because consumers may not necessarily understand how to interpret their test results, many organizations strongly suggest implementing regulations mandating that adequate genetic counseling be made available to DTC genetic testing customers. The American Society of Human Genetics proposes that the CDC might conduct a study on the impact of DTC testing on consumers to assess to what extent

consumers are benefiting or being harmed from a mode of delivery without the advice of a medical professional (Hudson 1). In addition, the organization suggests that the FTC should require companies to disclose all risks associated with testing, including psychological risks to individuals and family members. Genetic counseling should be made available because many consumers may base life-defining decisions on the information received from DTC tests, such as whether to have children or take part in treatment for a medical condition. In order to make sure that consumers are making informed decisions when they act on their genetic test results, many organizations propose that the federal government ought to ensure that adequate genetic counseling is made available.

In addition to genetic counseling, a final area of concern in which federal regulation may be implemented over DTC genetic testing is the accuracy of advertising and claims made by companies providing DTC genetic tests. The American Society of Human Genetics believes that regulating the claims made by companies about the predictive power of genetic tests is highly important:

“Claims made regarding DTC genetic tests may in some cases be exaggerated or unsupported by scientific evidence. Exaggerated or unsupported claims may lead consumers to get tested inappropriately or to have false expectations regarding the benefits of testing. Further, consumers may make unwarranted, and even irrevocable, decisions on the basis of test results and associated information, such as the decision to terminate a pregnancy, to forgo needed treatment, or to pursue unproven therapies” (Hudson 1).

As stated in the concerns of the American Society of Human Genetics, consumers base weighty life decisions on the information supplied by DTC genetic tests, and because companies are unregulated, they may exaggerate the predictive power of genetic tests. Consumers then forget that genes and environment interact and define whether a person

ultimately develops a disease or any other condition—genetics are not the only influence on whether a person develops a disease. The Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) was developed in order to advise the secretary of Health and Human Services on genetic issues. In 2004, SACGHS proposed that greater federal oversight of the advertising of genetic tests ought to be implemented, and the committee recommended “enhanced collaboration between the FTC and FDA and other appropriate HHS agencies on advertising for genetic tests, clarification of FDA's role in monitoring the advertising of laboratory developed genetic tests, and an analysis of the public health impact of DTC advertising and direct access to genetic tests” (FTC 1). Since 2004, several study committees have been convened to assess the impact of inaccurate claims made by genetic testing companies, but no formal regulations have been instituted in order to police the claims made by DTC genetics companies. So far, no regulations have been instituted to ensure a correlation between the claims made by DTC genetic testing advertising and the scientific evidence available to support these claims.

Essentially, the current federal regulatory system surrounding DTC genetic testing is insufficient to ensure that companies provide accurate, valid, and reliable information to consumers receiving services. To some extent, analytic validity of genetic tests is ensured by CLIA, which regulates the quality control and personnel selection of laboratories where genetics tests are analyzed. However, the issues of clinical validity, adequate genetic counseling, and accuracy of advertising are not reinforced by federal regulations in order to ensure that consumers are able to understand and utilize their test results. The FDA currently does not regulate the clinical validity of DTC genetic tests because they do not fall under the category of medical devices—rather than being

considered test kits, they are labeled “lab-developed tests.” In order to protect consumers, the FDA must regulate both categories of tests. Another area where federal regulations might benefit consumers is in requiring that adequate genetic counseling be made available for people who utilize DTC genetic testing. In order to confirm that consumers can make informed decisions about their genetic information, regulations might ensure that professionals are available to help consumers interpret their test results. Additionally, federal regulations ought to ensure that the claims made in advertisements of DTC services are accurate and can be verified by scientific evidence. While DTC genetic testing gives consumers a greater degree of autonomy, empowerment, and control over their genetic information, the associated risks necessitate federal regulation of DTC companies.

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