ABSTRACT: With the increased emphasis on patient-driven health care and readily available access to patients through the internet and media, many genetic testing companies have begun to market directly to consumers. Direct-to-consumer genetic testing raises unique concerns and considerations, including limited knowledge among patients and health care providers of available genetic tests, difficulty in interpretation of genetic test results, lack of oversight of companies that offer genetic testing, and issues of privacy and confidentiality. When undergoing any direct-to-consumer genetic testing, the consumer should be apprised of risk from screening or susceptibility test results that can neither prove nor eliminate disease potential. Because ordering an appropriate genetic test and interpretation of genetic test results are complex, an obstetrician–gynecologist or other health care provider with knowledge of genetics should be involved in ordering and interpreting the results of any genetic test with medical implications. When an individual presents test results to a health care provider from a direct-to-consumer genomic test that putatively assesses the risk of specific diseases, the patient should be referred to an obstetrician–gynecologist or other health care provider who is skilled in risk assessment for the diseases of interest and interpretation of genetic testing results in the context of the individual’s relevant medical and family history. Because of these considerations and in view of the fact that the interpretation of the results requires specific training and medical knowledge, direct-to-consumer genetic testing should be discouraged because of the potential harm of a misinterpreted or inaccurate result.

Recommendations
The American College of Obstetricians and Gynecologists makes the following recommendations:

- Direct-to-consumer genetic testing should be discouraged because of the potential harm of a misinterpreted or inaccurate result.
- Given the insufficient data to support the use of single nucleotide polymorphisms (SNP) testing for medical purposes, SNP testing to provide individual risk assessment for a variety of diseases or to tailor drug therapy outside of an institutional review board-approved research protocol is not recommended.
- Because ordering an appropriate genetic test and interpretation of genetic test results are complex, an obstetrician–gynecologist or other health care provider with knowledge of genetics should be involved in ordering and interpreting the results of any genetic test with medical implications.
- When undergoing any direct-to-consumer genetic testing, the consumer should be apprised of risk from screening or susceptibility test results that can neither prove nor eliminate disease potential. The consumer also should be made aware of the potential for unanticipated information or information that may have implications for other family members.
- Consumers should be aware of privacy concerns before undergoing direct-to-consumer commercial genetic testing, including who will have access to the results; what systems are in place to provide protection of confidential health information; how the sample will be handled after testing is complete; whether the test results will have an effect on issues...
related to life, long-term care, or disability insurability; and how genetic information will be handled if the company closes.

- When an individual presents test results to a health care provider from a direct-to-consumer genomic test that putatively assesses the risk of specific diseases, the patient should be referred to an obstetrician–gynecologist or other health care provider who is skilled in risk assessment for the diseases of interest and interpretation of genetic testing results in the context of the individual’s relevant medical and family history.

- There currently are no standard clinical indications for the use of pharmacogenetic testing in the routine practice of obstetrics and gynecology.

**Precision Medicine**

*Precision medicine* is defined by the National Academy of Sciences as the use of genomic, epigenomic, exposure, and other data to define individual patterns of disease, potentially leading to better treatment. More recently, the concept of “precision medicine” also has been defined as “the tailoring of medical treatment to the individual characteristics of each patient,” which incorporates not only genetics, but also other aspects of biology and prognosis (1). The National Academy of Sciences uses the term “precision medicine” rather than “personalized medicine” to clarify that it “does not literally mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease, in the biology and/or prognosis of those diseases they may develop, or in their response to a specific treatment” (1). Precision medicine is also sometimes referred to as “precision health.”

Predictive testing (which determines whether an individual carries a genetic variation associated with later development of a genetic condition) and pharmacogenetics (which attempts to identify DNA or RNA variations and their relationship to drug response) are examples of precision medicine. Variations in genes related to drug metabolism and other human disease have been increasingly identified through advancing genetic technologies, including the completion of the sequencing of the human genome in 2001. Predictive testing and pharmacogenetics recently have been marketed directly to consumers.

**Predictive Testing**

A single nucleotide polymorphism, frequently called SNP, is a variation at a single position in the DNA sequence. Single nucleotide polymorphisms occur throughout the human genome, once in every 300 nucleotides on average. Most SNPs are not causative of disease or impaired development but may be used to predict risk of a variety of complex diseases (such as heart disease or diabetes), predict an individual’s response to certain drugs, or estimate susceptibility to environmental toxins. Predictive testing in precision medicine involves SNP testing to provide individual risk assessment for a variety of diseases or to tailor drug therapy. This should be done under the care of a health care provider in a traditional office setting, although it is increasingly offered directly to the patient through at-home testing.

Despite laboratories offering genetic testing for disease-associated SNPs, there remains a paucity of evidence in most clinical scenarios to support SNP testing as a method to provide medically actionable results. Few SNP testing protocols have been clinically validated (2, 3). Given the insufficient data to support the use of SNP testing for medical purposes, SNP testing to provide individual risk assessment for a variety of diseases or to tailor drug therapy outside of an institutional review board-approved research protocol is not recommended. Further investigation is necessary to prospectively assess the validity and use of incorporating disease-associated SNPs into management recommendations. Therefore, the American College of Obstetricians and Gynecologists’ Committee on Genetics recommends that the use of these technologies be viewed as investigational at this time.

**Pharmacogenetic Testing**

Pharmacogenetics refers to the interactions of multiple genetic differences and environmental influence on drug responses. Currently, the U.S. Food and Drug Administration includes pharmacogenetic testing in the labeling of a select few drugs (4). In specific situations, pharmacogenetic testing is done to determine if an individual is likely to benefit from the specific therapeutic agent. Prospective clinical trials are needed to develop the appropriate algorithms to introduce pharmacogenetic testing into routine clinical practice in ways that are demonstrated to improve health outcomes. There currently are no standard clinical indications for the use of pharmacogenetic testing in the routine practice of obstetrics and gynecology. However, given its potential applications to women’s health care, obstetricians and gynecologists and other health care providers should be aware of this rapidly developing field.

**Direct-to-Consumer Genetic Testing**

With the increased emphasis on patient-driven health care and readily available access to patients as consumers through the internet and media, many genetic testing companies have begun to market directly to consumers. This includes testing for medically significant conditions, carrier status for genetic diseases such as cystic fibrosis or hemochromatosis, or both. Additionally, some companies offer genetic testing for nonmedical information, including ethnicity, athletic performance, behavior, aging, or metabolism.

Direct-to-consumer genetic testing raises unique concerns and considerations, including limited knowledge
among patients and health care providers of available genetic tests, difficulty in interpretation of genetic test results, lack of oversight of companies that offer genetic testing, and issues of privacy and confidentiality. When undergoing any direct-to-consumer genetic testing, appropriate pretest and posttest counseling should be provided, including a discussion of the risks, benefits, and limitations of the testing. Because direct-to-consumer testing typically is provided without input from a health care provider, it is the responsibility of the entity offering this testing to ensure that appropriate and accurate information is conveyed to the consumer. When undergoing any direct-to-consumer genetic testing, the consumer should be apprised of risk from screening or susceptibility test results that can neither prove nor eliminate disease potential. The consumer also should be made aware of the potential for unanticipated information or information that may have implications for other family members. Consumers should be aware of privacy concerns before undergoing direct-to-consumer commercial genetic testing, including who will have access to the results; what systems are in place to provide protection of confidential health information; how the sample will be handled after testing is complete; whether the test results will have an effect on issues related to life, long-term care, or disability insurability; and how genetic information will be handled if the company closes. Direct-to-consumer testing should be held to the same standard as other medical services. Informed consent, risks and benefits, accuracy and privacy concerns should all be addressed.

Because ordering an appropriate genetic test and interpretation of genetic test results are complex, an obstetrician–gynecologist or other health care provider with knowledge of genetics should be involved in ordering and interpreting the results of any genetic test with medical implications. It must be recognized that direct-to-consumer genetic testing will create downstream needs for counseling, support, and care for those identified as carriers of genes associated with deleterious medical conditions. When an individual presents test results to a health care provider from a direct-to-consumer genomic test that putatively assesses the risk of specific diseases, the patient should be referred to an obstetrician–gynecologist or other health care provider who is skilled in risk assessment for the diseases of interest and interpretation of genetic testing results in the context of the individual’s relevant medical and family history. A vignette on direct-to-consumer testing for BRCA mutations can be accessed through the Washington State Department of Health Genetics Services Policy Project (5).

Recognizing the concerns associated with direct-to-consumer genetic testing, the U.S. Federal Trade Commission, U.S. Food and Drug Administration, and the Centers for Disease Control and Prevention advise consumers to be skeptical of these tests’ claims and encourage consumers to seek guidance from a knowledgeable health care provider before pursuing this type of genetic testing (6).

**Conclusion**

All genetic testing, including pharmacogenetics and direct-to-consumer testing, should be considered medical testing because results might have an effect on future medical care and clinical decision making. Although some tests have been marketed as nonmedical by commercial entities, any test that provides information or risk estimates for medical conditions should be considered a medical test. Because of these considerations and in view of the fact that the interpretation of the results requires specific training and medical knowledge, direct-to-consumer genetic testing should be discouraged because of the potential harm of a misinterpreted or inaccurate result. Genetic testing should be performed only under the supervision of an appropriate obstetrician–gynecologist or other health care provider who is skilled in risk assessment for the diseases of interest and interpretation of genetic testing.

**References**


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