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# COMMITTEE OPINION

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## Committee on Genetics

*This Committee Opinion was developed by the American College of Obstetricians and Gynecologists' Committee on Genetics in collaboration with committee members Joseph R. Biggio Jr, MD, MSHQS, and Steven J. Ralston, MD, MPH.*

*This document reflects emerging clinical and scientific advances as of the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed.*

## Counseling About Genetic Testing and Communication of Genetic Test Results

**ABSTRACT:** Given the increasing availability and complexity of genetic testing, it is imperative that the practicing obstetrician–gynecologist or other health care provider has a firm comprehension of the benefits, limitations, and risks of offering a specific genetic test, as well as the importance of appropriate pretest and posttest counseling. The purpose of this Committee Opinion is to provide an outline of how obstetrician–gynecologists and other health care providers can best incorporate these tests into their current practices and provide appropriate pretest and posttest counseling to patients. Obstetrician–gynecologists and other health care providers should determine which tests will be offered as the standard in their practices so that similar testing strategies are made available to all patients. Practices should have procedures in place that ensure timely disclosure of test results to patients. As with any medical test, expectations regarding the performance of a genetic test should be discussed with the patient before the test is ordered. After counseling, patients should have the option to decline any or all testing. Pretest and posttest counseling should be done in a clear, objective, and nondirective fashion, which allows patients sufficient time to understand information and make informed decisions regarding testing and further evaluation or treatment. In addition to counseling each patient about her own personal risk, obstetrician–gynecologists and other health care providers should counsel patients regarding the risk for family members, including their potential to have affected offspring.

### Recommendations

- Pretest counseling that includes information on the types of potential results as well as the risks, limitations, and benefits of testing should be provided to all patients before performing any form of genetic test. After counseling, patients should have the option to decline any or all testing.
- Pretest and posttest counseling should be done in a clear, objective, and nondirective fashion, which allows patients sufficient time to understand information and make informed decisions regarding testing and further evaluation or treatment.
- Obstetrician–gynecologists and other health care providers should determine which tests will be offered as the standard in their practices so that similar testing strategies are made available to all patients.
- In order to optimize options for evaluation and medical care, results should be communicated in a timely manner.
- Patients should be informed that genetic testing could affect insurance premiums or eligibility for life or long-term care insurance.
- If genetic testing reveals clinically significant mutations with heritable potential, patients should be strongly encouraged to share the results with affected or at-risk family members.
- If an obstetrician–gynecologist or other health care provider does not have the necessary knowledge or expertise in genetics to counsel a patient appropriately, referral to a genetic counselor, medical or gynecologic oncologist, maternal–fetal medicine specialist, or other genetics specialist should be considered, as appropriate for the condition being examined.

## Introduction

Over the past decade, there has been a profound increase in the number of genetic tests available and in the scope of information that these tests make available. In addition, direct-to-consumer marketing has increased patient awareness of genetic tests and the desire for these tests (1, 2). Although prenatal **screening tests** for chromosome abnormalities and prenatal **diagnostic tests** have been routine components of prenatal care for several decades, new screening tests and diagnostic tests are being introduced into clinical practice, each with the potential to provide information that may be useful, confusing, or, in some circumstances, even harmful. In addition, although the American College of Obstetricians and Gynecologists has recommended cystic fibrosis **carrier screening** for more than a decade, new genetic technologies are making **expanded carrier screening** attractive to some obstetrician–gynecologists and other health care providers and patients. Similarly, in gynecologic practice, testing for genetic susceptibility to cancer is becoming increasingly commonplace for patients who have received a diagnosis of cancer or who have a family history of malignancy (3, 4). Given the increasing availability and complexity of genetic testing, it is imperative that the practicing obstetrician–gynecologist or other health care provider has a firm comprehension of the benefits, limitations, and risks of offering a specific genetic test, as well as the importance of appropriate pretest and posttest counseling.

The American College of Obstetricians and Gynecologists' Committee on Genetics recognizes that these are complex issues that require expertise and knowledge on the part of the obstetrician–gynecologist or other health care provider who offers and administers these tests, as well as time and resources for necessary counseling. The purpose of this Committee Opinion is to provide an outline of how obstetrician–gynecologists and other health care providers can best incorporate these tests into their current practices and provide appropriate pretest and posttest counseling to patients. If an obstetrician–gynecologist or other health care provider does not have the necessary knowledge or expertise in genetics to counsel a patient appropriately, referral to a genetic counselor, medical or gynecologic oncologist, maternal–fetal medicine specialist, or other genetics specialist should be considered, as appropriate for the condition being examined.

## Pretest Considerations

Determining the appropriate patient population that should be offered testing is important. Although some national organizations provide recommendations about offering testing in certain circumstances, the exact type of testing often is not specified (eg, Practice Bulletin No. 163, **Screening for Fetal Aneuploidy**, recommends that all women be offered prenatal screening for aneuploidy early in pregnancy but does not specify which

test to use) (5). Obstetrician–gynecologists and other health care providers generally have latitude in selecting the test that is most appropriate for their practice setting. For scenarios in which different testing options are acceptable alternatives, obstetrician–gynecologists and other health care providers should determine which tests will be offered as the standard in their practices so that, in accordance with the ethical principle of justice, similar testing strategies are made available to all patients. In the context of pregnancy, obstetrician–gynecologists and other health care providers should develop protocols that standardize their practice of offering prenatal screening tests for aneuploidy and carrier screening for genetic conditions. Although the validity of a wrongful birth or wrongful life claim varies by state, use of a standardized protocol may help minimize the potential for omission and assist in defending any claims of physician negligence in the event of the birth of an affected child (6, 7).

Obstetrician–gynecologists and other health care providers should consider whether a patient with a family history of cancer meets criteria for genetic testing regardless of whether cancer **susceptibility testing** is offered in the health care provider's practice. In some instances, when patients were not offered genetic testing and when risk-reducing treatments were not made available, claims for failure to diagnose have been successful (8, 9). A hereditary cancer risk assessment is the key to identifying patients and families who may be at increased risk of developing certain types of cancer. This assessment should be performed by an obstetrician–gynecologist or other health care provider and should be updated regularly. If a hereditary cancer risk assessment suggests an increased risk of a hereditary cancer syndrome, referral to a specialist in cancer genetics or another health care provider with expertise in genetics is recommended for expanded gathering of family history information, risk assessment, education, and counseling, which may lead to genetic testing (10).

Obstetrician–gynecologists and other health care providers should use standard protocols to identify patients for whom testing or referral for further genetic counseling is indicated in order to minimize the likelihood of missed opportunities to offer testing (8). Protocols have been developed to identify individuals at risk of hereditary cancer syndromes that can be adopted for use in a general obstetric–gynecologic practice (10).

As with any medical test, expectations regarding the performance of a genetic test should be discussed with the patient before the test is ordered. Pretest counseling that includes information on the types of potential results as well as the risks, limitations, and benefits of testing should be provided to all patients before performing any form of genetic test. After counseling, patients should have the option to decline any or all testing. Pretest and posttest counseling should be done in a clear, objective, and non-directive fashion, which allows patients sufficient time to

understand information and make informed decisions regarding testing and further evaluation or treatment.

A discussion of the *sensitivity* and *specificity* of the test for each of the disorders being tested is important to ensure patient understanding. For example, in the case of expanded carrier screening, patients should be informed of the overall range of the carrier detection rate and the range of *residual risk* of the disorders examined. With reference to each patient's specific *a priori risk*, the patient should be informed of the meaning and significance of positive, negative, or indeterminate test results, as well as results that are normal but may have variable phenotypes. This discussion of the *positive predictive value* and *negative predictive value* of the test result facilitates a discussion of the potential need for follow-up diagnostic testing. For example, discussion of cell-free DNA screening allows the obstetrician-gynecologist or other health care provider the opportunity to discuss the high sensitivity and specificity of this testing while also stressing the positive predictive value and the need for follow-up diagnostic testing. The potential for the discovery of *variants of uncertain significance* should be addressed with patients; such variants may prompt further testing or collecting of additional data from other family members or even necessitate long-term follow-up. Although additional discussion will be needed during posttest counseling, addressing these issues during pretest counseling conveys the message that an abnormal test result may not mean there is a specific problem or diagnosis.

## Potential Risks

A discussion about the potential risks and pitfalls of testing is particularly pertinent in the setting of genetic testing, especially with some of the newer prenatal genetic testing modalities. Although cost should not be a driving factor in whether a medically indicated test is offered to a patient, many genetic tests are expensive and may not be covered by a patient's insurance. Therefore, patients should be encouraged to discuss their eligibility for coverage with their insurance providers.

Patients may be concerned about the possibility of discrimination based on their genetic testing results. The 2008 federal Genetic Information Nondiscrimination Act aims to protect Americans against discrimination by health insurance providers and employers based on their genetic information. In general, Title I makes it illegal for health insurance providers to use or require genetic information to make decisions about an individual's health insurance eligibility or coverage. The protections of the Genetic Information Nondiscrimination Act do not extend to other forms of insurance such as life, disability, or long-term care insurance; thus, patients should be informed that genetic testing could affect insurance premiums or eligibility for life or long-term care insurance (11). Title II makes it illegal for employers to use an individual's genetic information when making deci-

sions about hiring, promotion, and several other terms of employment.

Nonpaternity has been estimated to be found in 3–10% of pregnancies (12). Genetic tests that examine fetal DNA in comparison with parental DNA (eg, carrier testing followed by diagnostic testing of the fetus) have the potential to discover nonpaternity. The possibility of nonpaternity should be discussed with the patient without her partner present so that she can fully consider her options if there is a chance of nonpaternity. Genetic technologies that rely on sequence analysis may reveal parental *consanguinity*, which is a risk that should be disclosed to patients before such testing is done. In the case of suspected consanguinity in the pregnancy of a minor patient, obstetrician-gynecologists and other health care providers are encouraged to discuss reporting requirements with qualified legal professionals.

Genetic tests are different from many other medical tests because the results can have potential medical and psychological consequences for the patient as well as the patient's family members. Multiple studies have demonstrated that abnormal results, such as detected mutations or variants of uncertain significance, can have profound psychosocial effects on patients and their families. Even in the setting of negative test results, patients can experience considerable "survivor guilt" or alterations in family dynamics, especially in the setting of cancer susceptibility testing (8).

Although many genetic tests can be performed using a routine blood sample, such tests should not be adopted into routine practice without patient consent. The ethical principles of autonomy, beneficence, and nonmaleficence require that the unique circumstances of each patient be considered before performing this type of testing. In some circumstances, this type of pretest counseling may require the assistance of an obstetrician-gynecologist or other health care provider with expertise in genetics. Patients also should be given a reasonable time frame within which they can expect to be informed about their test results, and they should be encouraged to call if they have not received their results at the end of that period.

## Posttest Considerations

As with the results of all medical tests, timely communication of genetic test results is a benchmark by which all obstetrician-gynecologists and other health care providers will be judged by patients and payers in the changing landscape of medical practice. More importantly, delays in communicating test results in obstetric practice have the potential to limit diagnostic and management options. Practices should have procedures in place that ensure timely disclosure of test results to patients (13). The method and schedule of communication should be appropriate to the type of testing performed and the urgency of the timeline in which any further testing may be needed. A policy of "no news is good news" is not

consistent with high-quality care. In order to optimize options for evaluation and medical care, results should be communicated in a timely manner, especially with prenatal testing. Although the data are limited with regard to the best schedule of disclosure, there does not seem to be any difference in patient anxiety when results are disclosed as soon as they become available compared with when they are disclosed at the time of a prespecified, prescheduled appointment.

The increased use of electronic health records adds further urgency for timely communication of test results to patients. In a growing number of medical practices, test results are made available to patients through a web portal shortly after they are received by the obstetrician-gynecologist or other health care provider. Therefore, obstetrician-gynecologists and other health care providers need to be able to communicate results to patients in the context of counseling. Discovery of results without concomitant counseling has the potential to cause considerable patient anxiety.

There are no data to determine whether delivering results in person or over the telephone produces less patient anxiety. Regardless of the method chosen for disclosure, results should be delivered clearly, objectively, and in a nondirective manner. The significance of the test result, a description of the diagnosis and its prognosis in the context of contemporary care and practice, and the options for further evaluation and management should be discussed. In the setting of a negative screening test result, discussion of the result and the residual risk is appropriate. For example, in a non-Hispanic white woman who has cystic fibrosis carrier screening performed with a panel that detects 92% of mutations, a negative screening test result reduces her risk of being a carrier from 1 in 25 to a residual risk of 1 in 200. An example of residual risk in carrier screening for cystic fibrosis is shown in [Table 1](#). With a screening test indicative of increased risk or an indeterminate result, discussion of available

diagnostic tests and the likelihood of an abnormal diagnostic test result are vital. Such discussions should enable patients to understand their likelihood of having the particular condition and empower them to make decisions about whether to proceed with further testing with full respect for patient autonomy.

These discussions can be augmented by the provision of additional resources that a patient and her family may use for further information, such as publications or websites of national organizations or advocacy groups. In the case of prenatal testing, obstetrician-gynecologists and other health care providers should be prepared to discuss all reproductive management options with patients, including (where appropriate) termination of pregnancy, adoption, neonatal palliative care, or prenatal surgery.

Patients receiving results of susceptibility genetic tests (such as those for cancer susceptibility) or *predictive tests* (such as those for adult-onset genetic conditions) need thorough counseling on the significance of the results based on their particular risk status. Patients found to have a mutation associated with an increased risk of malignancy should be counseled regarding their age-related and lifetime risks of particular types of cancer and the surveillance and management strategies available for risk reduction and health maintenance. The discovery of a cancer susceptibility mutation can have a considerable negative psychologic effect on patients, especially those with limited support systems and coping mechanisms. Providing a patient with additional resources for information and support may help alleviate anxiety by providing examples of how the result can be managed successfully. A patient from a high-risk family who receives results indicating that she does not carry a cancer susceptibility mutation may experience “survivor guilt,” so obstetrician-gynecologists and other health care providers should be prepared to provide support and resources for these patients. Importantly, in the setting of a high-risk family without a known familial mutation,

**Table 1.** Cystic Fibrosis Detection and Carrier Rates Before and After Testing ↵

Racial or Ethnic Group	Detection Rate* (%)	Individual Carrier Risk Before Testing	Approximate Individual Carrier Risk After Negative Test Result†
Ashkenazi Jewish	94	1/24	1/380
Non-Hispanic white	88	1/25	1/200
Hispanic white	72	1/58	1/200
African American	64	1/61	1/170
Asian American	49	1/94	1/180

\*Detection rate data based on use of a 23-mutation panel.

†Bayesian statistics used to calculate approximate carrier risk after a negative test result.

Modified from the American College of Medical Genetics. Technical standards and guidelines for CFTR mutation testing. American College of Medical Genetics Standards and Guidelines for Clinical Genetics Laboratories. Bethesda (MD): ACMG; 2011. Available at: <http://www.acmg.net/StaticContent/SGs/CFTR%20Mutation%20Testing.pdf>. Retrieved September 12, 2016.

a patient needs to be counseled that receiving a negative result does not return her risk to baseline general population levels.

Family members of any patient found to be a carrier of a genetic disease—whether it be cancer susceptibility or a single-gene disorder such as cystic fibrosis—are at risk of carrying the same mutation. In addition to counseling each patient about her own personal risk, obstetrician–gynecologists and other health care providers should counsel patients regarding the risk for family members, including their potential to have affected offspring. If genetic testing reveals clinically significant mutations with heritable potential, patients should be strongly encouraged to share the results with affected or at-risk family members. Although the ethical issues surrounding disclosure of test results to family members is complex, tested patients generally should be informed that they have a duty to notify relatives of the familial risk, though it is neither required nor appropriate for an obstetrician–gynecologist or other health care provider to contact and notify at-risk family members (8, 14, 15). If there is any uncertainty regarding local legal disclosure parameters, physicians are encouraged to consult with qualified legal professionals.

## Glossary

**A Priori Risk:** The best assessment of risk before testing results are known. ↩

**Carrier Screening:** Genetic testing performed on an asymptomatic individual to determine whether that person has a mutation or abnormal allele within a gene associated with a particular disorder. Carrier screening can be performed for one specific condition or for multiple disorders. ↩

**Consanguinity:** A union between two individuals who are second cousins or closer in family relationship. ↩

**Diagnostic Test:** Determines whether a specific condition is present in an individual. ↩

**Expanded Carrier Screening:** Disease screening that evaluates an individual's carrier state for multiple conditions at once and regardless of ethnicity. ↩

**Negative Predictive Value:** The chance that a negative test result is a true negative. ↩

**Positive Predictive Value:** The chance that a positive test result is a true positive. ↩

**Predictive Test:** Determines whether an individual carries a genetic variation associated with later development of a genetic condition. An example of predictive testing is Huntington disease testing—every individual who carries the mutation will eventually develop the disorder. Most predictive tests cannot determine the severity of a condition or the timing of its onset. ↩

**Residual Risk:** The risk that an individual is a carrier for or is affected by a disorder despite a negative screening test. ↩

**Screening Test:** Determines whether an individual is at increased risk of either being a carrier for or being affected by a specific disorder. ↩

**Sensitivity:** The proportion of individuals with a condition correctly identified as positive. ↩

**Specificity:** The proportion of individuals without a condition correctly identified as negative. ↩

**Susceptibility Testing:** Determines whether an individual carries a genetic variation that increases the potential for developing a specific condition. Not every person with a genetic variant will develop the condition, but they are at increased risk. An example of susceptibility testing is *BRCA* testing. ↩

**Variants of Uncertain Significance:** An identified DNA change that either cannot be characterized reliably as benign or pathogenic at the time of the study because of limited data describing outcomes in association with the changes or is associated with a variable phenotype (because of incomplete penetrance or variable expressivity). ↩

## For More Information

The American College of Obstetricians and Gynecologists has identified additional resources on topics related to this document that may be helpful for ob-gyns, other health care providers, and patients. You may view these resources at [www.acog.org/GeneticCounseling](http://www.acog.org/GeneticCounseling).

These resources are for information only and are not meant to be comprehensive. Referral to these resources does not imply the American College of Obstetricians and Gynecologists' endorsement of the organization, the organization's website, or the content of the resource. The resources may change without notice.

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