

The Benefits of Preconception Genetic Counseling, Indications for Referral

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Any patient who is planning a pregnancy may benefit from preconception genetic counseling, regardless of his or her age or method of conception. Patients who use assisted reproductive technologies (ART) are particularly suited for preconception counseling, because they plan the details of their reproductive treatments and interact with health-care professionals before conception. Therefore, they can be informed about and referred for genetic counseling before initiating a pregnancy. However, too few patients who are using ART are informed about or take advantage of preconception genetic counseling. OB/GYN and infertility nurses can support their patients' reproductive goals by knowing the indications for referral and by informing their patients of the benefits of preconception genetic consultations.

Preconception Genetic Counseling

Certified genetic counselors have master's degrees in medical genetics or genetic counseling and have achieved certification through the American Board of Genetic Counseling. Genetic counselors work in diverse clinical settings, including prenatal, oncology, and neurology clinics. Members of the ART special interest group (SIG) of the National Society of Genetic Counseling (NSGC) and the genetic counseling SIG of the American Society of Reproductive Medicine specialize in working with patients who use reproductive technologies.

A genetic counseling appointment includes collection of a patient's family medical history; a risk assessment to identify potential health risks to offspring; and discussion of the inheritance patterns of those risks, preventive measures, and available genetic testing options. Genetic consultations also include support with decision-making about which genetic tests, if any, may be right for that patient's individual reproductive plans.

Any patient who is planning a pregnancy may consider preconception genetic counseling for genetic carrier screening, a family history risk assessment, and discussion of the options available for the management of any concerns identified. For example, if a risk for a specific medical problem is identified during the genetic risk assessment, or if both partners test positive as carriers of mutations for the

same genetic condition, such as cystic fibrosis, they can be informed about preimplantation genetic diagnosis (PGD) as an option for their reproductive plans.

This is particularly beneficial to patients who are already planning to use IVF. If these health risks are identified postconception, the options for managing them may be limited to taking no action, or using invasive prenatal diagnosis procedures, such as chorionic villus sampling or amniocentesis, and considering possible termination of a much-wanted pregnancy.

Patients who plan to use ART are especially likely to benefit from preconception genetic consultations.

Too often, patients are left to wonder why no one offered this genetic screening and information before they became pregnant, so that they could have avoided dealing with these stressful and unexpected situations during their pregnancies.

Patients who plan to use ART are especially likely to benefit from preconception genetic consultations, because they can be informed about the risks, benefits, and limitations of PGD and other ART procedures. They can also discuss and compare these procedures to prenatal screening and diagnostic testing options. In one study, patients were found to have limited knowledge of PGD even though they had used it to conceive a pregnancy. They often did not understand which types of health issues were screened using this tool, and assumed that they did not need to consider prenatal screening or diagnostic testing during pregnancy if they used PGD. It is not clear if this misinformation may be a result of poor patient comprehension or if physicians and nurses lack the time to provide sufficient education.

When patients are not aware that prenatal diagnosis is recommended after using PGD, they can be greatly distressed by this information during pregnancy. The anxiety arises from the realization of unscreened medical risks in their offspring, and the limited time available to make decisions about these procedures. Therefore, precon-

ception genetic consultations are ideal, because they provide patients time to carefully consider their options, ask questions, and acquire support in developing their fertility and prenatal management plans.

Indications for Referral

Patients who are at advanced parental age at the time of delivery—aged ≥ 35 years for women and ≥ 45 years for men—or who have a family history of recurrent pregnancy loss, infant death, mental retardation, birth defects, or specific genetic disorders, should be strongly encouraged to schedule a consultation.

Patients using fertility treatments as a result of known and unknown causes of infertility are also prime candidates for preconception genetic counseling. It is important for them to be informed that they may pass on inherited forms of infertility to their offspring by using certain reproductive technologies. In addition, certain causes of infertility, such as structural chromosome abnormalities, can lead to miscarriage or the birth of a child with major birth defects and/or mental retardation. Preconception genetic counseling can provide education about alternatives, such as gamete donation, if patients are concerned about transmitting these issues to their offspring.

Individuals involved in third-party reproduction, including donors, recipients, surrogates, and intended parents, may all benefit from preconception genetic counseling. Genetic screening of surrogates and egg and sperm donors is variable in the United States. The guidelines for screening these populations are not specific, and each egg and sperm bank and surrogate agency independently determines its own procedures for screening applicants.

Genetic screening of a third-party individual may not be sufficient for a specific recipient or intended parent, depending on his or her own medical history. Additional genetic screening of the third party, when appropriate, should be discussed before a pregnancy is initiated, so that all parties have time to agree as to how to proceed.

Preconception genetic counseling may allow a surrogate to be informed about the health risks to a child, as elicited from the biological parents' risk assessment. The biological parents' history may indicate a need for invasive testing or a risk for medical com-

plications during pregnancy, for which the surrogate may need to be prepared. Even when a legal agreement has been established among all parties before conception, difficult situations can arise during pregnancy if a thorough discussion of health risks and their meanings has not occurred. In one situation, a surrogate who previously agreed to prenatal diagnosis later declined testing after being informed about what the procedure was testing, and that the results may lead to decisions about pregnancy termination or fetal reduction.

Barriers to Preconception Genetic Consultations

Patients often believe that they are familiar with their own family histories and that they are aware of any risks to their offspring. However, many patients do not truly understand the risks posed by their medical histories and are surprised to learn about possible risks and management options that they may wish to consider. Patients frequently leave a genetic consultation feeling more informed and in control of their plans than they had been when they arrived, because the consultations empowered them with information and support in making decisions that are optimal for their individual circumstances.

Individuals who use fertility treatments often feel burdened by multiple medical appointments. In addition, genetic counseling services are available most often in major metropolitan areas. At California Cryobank Genetic Counseling Services, we attempt to address some of these issues by offering appointments outside of traditional clinic hours and by providing in-person and over-the-phone genetic consultations. In-person consultations are preferable whenever possible; however, over-the-phone consultations provide significant benefit compared with no consultation.

OB/GYN and infertility nurses can collaborate with genetic counselors to provide excellent patient care by establishing a strong network of relationships for case management and establishing the appropriateness of referrals. Nurses can provide guidance on the benefit of preconception genetic counseling to encourage patients to utilize these services that will enhance their reproductive experiences. To locate a genetic counselor in your area, visit www.nsgc.org. ■