

SHOULD YOU BE SCREENED? What to ask each other before seeking a gene test

There's not much couples can do to prevent the misreading of a prenatal test. But even when the process goes smoothly, difficult questions can arise. Partners can prepare themselves by discussing these questions in advance, including the toughest of all. Get further guidance from the National Society of Genetic Counselors at NSGC.org.

How early can we get screened? Even before pregnancy, you and your partner can have blood tests to pinpoint gene mutations that are known to cause inherited diseases.

How high is our risk for a genetic defect? Tay-Sachs and Canavan disease, a fatal, degenerative brain disorder, are most common among Ashkenazi Jews; African-American and Hispanic couples are more likely to pass on sickle-cell anemia. The American College of Obstetricians and Gynecologists in Washington, D.C., now recommends that all couples seeking prenatal care be screened for cystic fibrosis, which one in 29 Caucasians and Ashkenazi Jews carry.

Is IVF an option? Couples who discover they are both carriers may decide to undergo in vitro fertilization so their doctor can prescreen their embryos for defects.

What about amniocentesis? This test allows doctors to look for defects by drawing amniotic fluid from the sac surrounding the fetus. But it carries a chance of miscarriage: Talk to your doctor about the rate in her practice.

How late would we wait? Doctors must hold out until the second trimester to detect some problems; testing for spina bifida, for instance, is done between 15 and 18 weeks.

Would we end the pregnancy? "Of course, what you think you will do and what you will actually do sometimes differ," says Kathy Hudson, Ph.D., director of the Genetics and Public Policy Center at Johns Hopkins University in Washington, D.C. "But talking about it is an important exercise before you jump in." —Erin O'Donnell