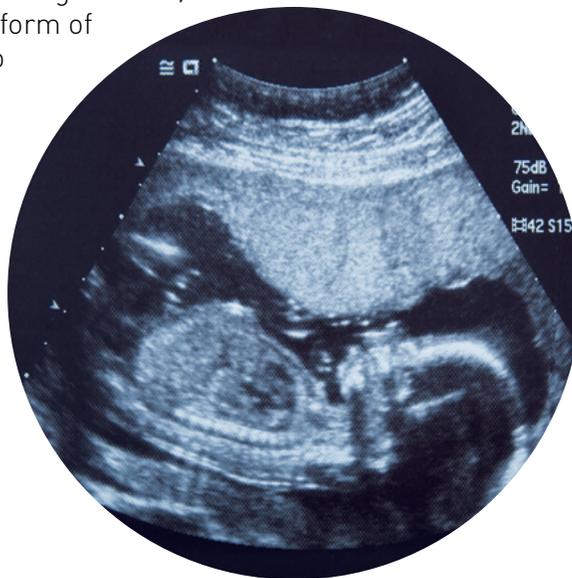


Non-Invasive Prenatal Testing

Prenatal diagnosis involves the use of tests during pregnancy to determine whether a fetus is affected with a particular disorder. These tests have been a part of prenatal medicine for over 30 years. Testing methods vary both in level of invasiveness to the fetus as well as the degree of accuracy. Generally, a set of non-invasive screening methods — such as maternal serum analysis or ultrasound — are initially performed. Suspicious results are followed up with more invasive diagnostic testing e.g. amniocentesis or chorionic villus sampling (CVS). These invasive approaches obtain amniotic fluid and/or fetal cells that are then biochemically or genetically analyzed. Genetic tests may be genome wide — such as karyotyping or array comparative genome hybridization [see pg. 36] — or more narrow in scope, such as testing a single gene. Both amniocentesis and CVS carry a small but significant risk of miscarriage.

Scientists have recently developed a novel, non-invasive testing method. In the 1990s, it was discovered that fetal DNA crosses the placenta into the maternal bloodstream. Today, relatively straightforward techniques can isolate and analyze this DNA, beginning as early as seven weeks gestation. This test can be performed several weeks earlier than conventional techniques and carries no risk to the health of the fetus. As a result, a larger number of pregnant women may choose to undergo prenatal testing. In 2012, three companies introduced this form of non-invasive prenatal testing into the clinic.

Non-invasive prenatal testing is currently classified as a screening, rather than a diagnostic test. It signals whether further, often more invasive forms of testing should be considered.



Prenatal diagnosis is a standard part of discussion around egg and sperm formation and abnormalities that can occur during meiosis. The advent of non-invasive techniques is an exciting addition for Biology (COS standards 12, 12a, 11, 3c). The application of this new technology to health and society links to classroom conversation in AP Biology (Big Idea: Information) and Health (COS objectives 5 and 6). Clearly there are a number of ethical concerns related to non-invasive prenatal testing. Depending on the context of the conversation and the maturity of the class, these questions may be appropriate for exploration and detailed discussion.

Whether this will ultimately replace CVS and amniocentesis as a diagnostic test will depend upon improvements in the sensitivity and specificity of the testing. However a number of significant ethical issues are associated with safer, earlier prenatal diagnosis. For example, by offering early non-invasive diagnosis, will there be increased social pressure to have the test and terminate an “abnormal” pregnancy? What or who decides the definition of “abnormal”? As the genetic components of many disorders become better understood, would non-invasive diagnostic testing allow parents — with only a blood test — to identify mild, adult-onset disorders as well as nonmedical traits such as eye color?