

Age, Pregnancy and Genetic Testing

By Karen Stebner, CGC, and Philip Buchanan, PhD

In 1994, there were twice as many women having babies after age 35 compared to 1984. The medical significance is that as a couple's ages increase, so does the chance for having a baby with a chromosome abnormality, such as Down syndrome. Advanced technology has allowed for prenatal testing for the age-related risk of chromosome abnormalities.

Sandra and Bill (pregnant couple 35 and 41 years of age): *We know at our ages, we are at a higher risk for having a child with a chromosome abnormality, but what is the risk?*

Genetic Counselor: *Your age-related risk for having a baby with any type of chromosome abnormality is 2.1 percent (1 in 48 chances). Although any pregnancy can be affected by a chromosome abnormality by chance alone, this chance increases with age. Between 34 and 35 is the age at which doctors begin offering diagnostic testing. Three diagnostic procedures are available during pregnancy to test for chromosome problems.*

Chorionic Villus Sampling (CVS), performed between 10 and 13 weeks gestation, involves sampling a tiny piece of the placenta to analyze the chromosomal make-up of the fetus. Amniocentesis performed after 14 weeks samples the amniotic fluid surrounding the baby. Early amniocentesis is performed between 11-14 weeks. The amniotic fluid contains fetal cells, which can be analyzed for chromosome abnormalities. A protein in amniotic fluid, alpha-fetoprotein (AFP), is measured to detect 98% of pregnancies affected with open spine and skull defects. Amniocentesis detects 99.9 percent and CVS detects 99.5 percent of chromosomal abnormalities and are safe when performed by experienced physicians. These procedures are unable to detect all causes of birth defects and mental retardation and do not guarantee a perfect baby.

When comparing the various testing options, genetic counselors provide non-directive counseling to educate couples about the risks, benefits, and limitations of each procedure.

Sandra: *What are the benefits of one test over another? Is one procedure safer than the others?*

Genetic Counselor: *CVS and Early Amniocentesis are performed early, which allows for earlier decision-making if an abnormality is detected. Amniocentesis testing is also beneficial for providing information about spinal defects and some other disorders. The risk for limb defects is increased when CVS is performed before 10 weeks gestation. Also, there is a small increased risk for complications with CVS.*

Bill: *What is the benefit of having prenatal testing?*

Genetic Counselor: *Testing provides knowledge about the baby's chromosomal make-up so a decision can be made about whether or not to continue an affected pregnancy. For couples choosing to continue affected pregnancies, the information helps them prepare for the births of their babies.*

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