

The Idea Behind Prenatal Genetic Screening

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Jackie is a 38 year old mother of two who is pregnant for the third time. Her obstetrician has referred Jackie and her husband, Albert, to a genetic counselor to discuss her screening test which shows their baby is at an increased risk for having Down syndrome. Understandably, Jackie and Albert have trouble thinking about much else besides their baby before their meeting with the counselor.

The genetic counselor explains that chromosomal abnormalities, like Down syndrome, occur more often as the ages of the parents increase. Jackie says "My friend had this blood test which showed her baby had Down syndrome but it was wrong because the baby was fine."

The counselor clarifies for Jackie that her friend may indeed have had an increased risk screening test for Down syndrome, but her friend's screening test was not designed to make a definite diagnosis. The purpose of the maternal blood test is to screen for chromosomal or open spinal abnormalities in patients who are less than 35 years of age and have a low risk for having a baby with health problems. A small number of patients having this screening test will receive an increased risk report, but most of these patients will still have a normal baby. Because screening tests do not make a diagnosis, they should be reported as "increased" or "not increased risk", never as "abnormal" or "positive".

Jackie asked the counselor "Should we have had an amniocentesis to have a diagnosis rather than the screening test?" The counselor responded that "Amniocentesis very safely provides a diagnosis for 99.9% chromosomal abnormalities and 99.9% of open spine and skull defects. Screening on the other hand is only meant to provide a screening risk for Down syndrome and Trisomy 18, another type of chromosomal abnormality, but not a risk for all the problems that can be diagnosed by amniocentesis testing". Jackie and Albert continue the discussion with the genetic counselor and understand that the screening test they had cannot diagnose an abnormality in their baby, just suggest a possible increased risk for which follow-up diagnostic testing might be helpful. The counselor discusses the benefit of knowing early whether their baby has a problem.

The medical societies involved with prenatal diagnosis and screening recommend that women 35 years of age and older have amniocentesis to diagnosis 99.9% of all chromosomal abnormalities rather than have maternal blood or ultrasound screening. Maternal blood screening provides a risk for only two chromosomal abnormalities. Similarly, an ultrasound exam cannot diagnose chromosome abnormalities, only adjust the possible risk for several of the hundreds of types of chromosomal abnormalities. While ultrasound provides an image of the unborn baby, ultrasound cannot diagnosis mental retardation, chromosomal and many types of birth defects because the problems associated with those disorders are usually not seen on ultrasound in mid-pregnancy. As a result, ultrasound can only detect certain changes in appearance of the fetus, but cannot provide the same information that an amniocentesis or a screening test can. Also parents should be aware that the normal appearance of a baby on ultrasound may be misleading and provide false reassurance that their baby does not have any problem when in fact not all problems are detectable by ultrasound. Couples who are at an increased risk of having a baby with a chromosomal abnormality due to age or family history but decline amniocentesis, should be aware that screening cannot be a diagnostic substitute for amniocentesis. Additionally, women 38 years of age and older rarely benefit from screening because most will have an increased risk associated with their age which screening cannot significantly reduce.

Jackie and Albert elect to have an amniocentesis following their discussion with the genetic counselor. In their case, as it happens in the majority of pregnancies at increased risk for birth defects due to age or screening risk, the counselor reported the amniocentesis test shows normal chromosomal and open spine test results.

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