

Should advanced maternal age be a reasonable indication for invasive diagnostic testing?

Advanced maternal age (AMA) is usually defined as a mother who is 35 years of age or older at delivery. AMA is related to aneuploidies by nondisjunction of the chromosomes during maternal oogenesis (meiosis) and is therefore an important risk factor for fetuses with chromosomal abnormalities. AMA has also been used as a sole indicator for undergoing cytogenetic amniocentesis. As such, pregnant women with AMA have been offered invasive diagnostic testing, such as amniocentesis or chorionic villus sampling (CVS), as well as genetic counseling.

In Korea, the proportion of pregnant women with AMA is continuously increasing due to increases in the average age of marriage and the social participation of females, and has more than doubled over the past 10 years according to source of the National Statistical Office (from 7.4% in 2001 to 18.0% in 2011). It is important to monitor various indicators in pregnant women with AMA in order to predict the risk of fetal chromosomal abnormalities that depend on the mother's age. However, the validity of AMA as a potential indicator relating to the risk of fetal chromosomal abnormalities is still being studied and its usefulness is still under debate.

In 2007, the American Congress of Obstetricians and Gynecologists (ACOG) recommended that patients should have the option to take a diagnostic test such as amniocentesis or CVS regardless of maternal age [1]. In previous studies, Forabosco et al. [2] and Boyd et al. [3] reported that the decision to undergo amniocentesis should not be based solely on maternal age. These changes in the guidelines of genetic counseling for pregnant women with AMA appear to be based on the high detection rate of prenatal screening tool such as integrated test. Due to improvements in the diagnostic accuracy of prenatal screening, doctors' individual risk assessments show a tendency to prefer the easier screening algorithms rather than offering amniocentesis to all pregnant women with AMA. Recently, Kwon et al. [4] reported that the quadruple screening test is a better choice than direct amniocentesis for pregnancies complicated by AMA under conditions in which first trimester screening test is not available.

However, quite a few obstetricians still use a maternal age of 35 years as a cutoff for offering diagnostic testing. Park et al. [5] determined that AMA was a strong risk factor for chromosomal abnormalities. Bornstein et al. [6] suggested that the benefit of undergoing genetic amniocentesis based only on AMA far outweighs the potential amniocentesis-related fetal loss rate and Henry et al. [7] reported that the rate of Down syndrome births for women with AMA increased considerably, while the same rate for pregnant women <35 years of age remained stable. This demonstrates that especially for women who do not undergo combined screening and who want definitive information on the chromosomal status of their fetus, AMA is a reasonable indicator for invasive prenatal diagnosis.

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In this controversial situation regarding the use of AMA as a sole indicator for invasive diagnostic testing, all pregnant women should have the option to choose invasive diagnostic testing such as amniocentesis or CVS, regardless of maternal age, and women should be counseled regarding the differences between prenatal screening and invasive diagnostic testing, according to ACOG guidelines [1]. Obstetricians should guide pregnant women through genetic counseling that is focused on the importance of individual choice, based on the risk that the fetus will have a chromosomal abnormality and the risk of pregnancy loss from an invasive procedure, as well as the morals and viewpoints of the individual, couple, and/or family.

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