Topic Page: Amniocentesis

Definition: **Amniocentesis** from *Black's Medical Dictionary, 43rd Edition*

A diagnostic procedure for detecting abnormalities of the FETUS. Usually carried out between the 16th and 18th week of pregnancy, amniocentesis is performed by piercing the amniotic sac in the pregnant UTERUS with a hollow needle, under ULTRASOUND guidance and withdrawing a sample of AMNIOTIC FLUID for laboratory analysis. As well as checking for the presence of abnormal fetal cells, the procedure can show the sex of the fetus. The risk of early rupture of the fetal membranes or of miscarriage is low (around 1 per cent). Currently it is offered (in the UK) to women over age 35, those who have previously had a baby with SPINA BIFIDA, CHROMOSOME abnormality and those known to carry a serious genetic disorder. It is also offered to women who have had an abnormal result on fetal ultrasound screening or blood testing for DOWN'S SYNDROME.

Summary Article: **Amniocentesis**
From *Encyclopedia of Special Education: A Reference for the Education of Children, Adolescents, and Adults with Disabilities and Other Exceptional Individuals*

Amniocentesis is the sampling of amniotic fluid surrounding a fetus. A physician anesthetizes a small area of the pregnant woman's abdomen, inserts a small needle through the abdominal wall, and, with the aid of ultrasonography, enters the amniotic sac and removes 20 ml (approximately 1 oz) of fluid. It is performed most frequently between 15 and 18 weeks gestation to detect hereditary disease or congenital defects in the fetus. One disadvantage is that analysis of the fluid takes 2 to 4 weeks. Damage to the fetus also may occur, but the risk is small—.06% or 1 in 1600 (Eddleman, Malone, & Sullivan,; March of Dimes,).

Midtrimester amniocentesis plays an important role in genetic and other prenatal counseling by providing potential parents with reproductive options. It should be considered when the pregnant woman is over 35, or a family history of genetic or congenital disorders is apparent (Kaback,; March of Dimes,). Cytogenetic analysis of fetal fluid leads to prevention of birth of approximately 15,000 chromosomally abnormal infants each year in the United States alone (Pritchard, MacDonald, & Gant,).

Amniocentesis allows identification of about 300 chromosomal, single-gene, and other congenital abnormalities (Pritchard et al.,). The list grows with the discovery of new markers. Chromosomally based disorders are identified through karyotyping and resultant abnormal appearance of one or more chromosomes; other disorders are identified through elevated or reduced levels of particular substances. Among the disorders that can be reliably diagnosed are (a) all chromosomally based disorders such as Down syndrome and cri du chat; (b) about 75 inborn errors of metabolism, including galactosemia, Tay-Sachs disease, and Lesch-Nyhan syndrome (X-linked), but not phenylketonuria; (c) some central nervous system defects including meningocele (a form of spina bifida) and anencephaly; (d) some fetal infections (cytomegalovirus, herpes simplex, and rubella); (e) and some hematologic disorders (e.g., sickle-cell anemia; Pritchard et al.,).

The widespread availability of amniocentesis forces many women to confront the decision to terminate an advanced pregnancy. Attachment grows throughout pregnancy, and confronting the decision of choosing termination at a late stage can be emotionally painful (Brewster,). Many women are unprepared for the anxiety associated with both waiting several weeks for results of their

amniocentesis and choosing between life and quality of life. Optimally, women in high-risk groups should weigh this decision and discuss other reproductive options with a genetic counselor prior to conception. Some counselors suggest that health caregivers be sensitive to pregnant women's emotional reactions and not use measures such as a doppler to hear the fetus's heartbeat or ultrasonography to take pictures of the fetus, that promote maternal attachment prior to amniocentesis (Brewster,).

A new diagnostic technique, chorion-villus biopsy, usable as early as 8 weeks gestation, may be preferable in some cases, but risks include 1 in 100 pregnancies being at risk for miscarriage.

See also Chronic Villus Sampling; Genetic Counseling; Inborn Errors of Metabolism

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