

Amniotic Fluid Analysis in Diagnosing Fetal Abnormalities Some Clinical and Experimental Observations

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Although the genesis and mechanisms leading to formation of amniotic fluid have not yet been completely clarified, it is obvious that the amniotic sac and its fluid constitute a "fetal compartment" surrounded by the chorion and the placenta, both of which are products of conception. Although water, electrolytes and other solutes contained in the amniotic fluid may originate in part from the mother, the cells and macromolecules contained in this fluid are of fetal origin, and hence would logically reflect fetal properties. Therefore, sampling the amniotic fluid by amniocentesis would appear to be a useful means of detecting genetic disorders of the intrauterine fetus.

Clinical Applications of Amniocentesis

The first systematic attempt to sample amniotic fluid for the purpose of obtaining information about the condition of the fetus was conducted in 1952 by Bevis.¹ This researcher determined the content of bile pigment in cases of erythroblastosis fetalis due to Rh isoimmunization of the mother. Then, in 1960, Riis and Fuchs² first applied the examination of amniotic fluid in their investigation of sex-linked hereditary diseases.

During the past few years, amniocentesis (ie, removal of a sample of amniotic fluid by percutaneous transabdominal puncture) has been gaining importance as a means of detecting genetic disorders and congenital abnormalities *in utero*.³⁻⁸ Table 1 lists the indications and tests used in diagnosing various fetal conditions.

Prenatal detection of chromosomal aberrations.

The cytologic and cytogenetic study of cultured or uncultured amniotic-fluid cells has become established as a practical diagnostic technique. Interpretation of results helps in providing adequate genetic counsel to mothers having a high risk of bearing a child with chromosomal abnormalities. Down's syndrome (mongolism) is one of the many

genetic diseases which may be diagnosed by cytogenetic techniques. Among the other diagnosable chromosomal abnormalities is 13-trisomy, a condition characterized by multiple congenital anomalies.

Enzymes in amniotic fluid. A new and challenging approach to the prenatal diagnosis of hereditary disease is the analysis of enzymes contained in cultivated or noncultivated cells collected from amniotic fluid. Reviews of enzymatic studies describe at least 15 enzymes isolated from cultivated cells. Numerous metabolic disorders have recently been diagnosed in the fetus by enzyme analysis.

Sex-linked hereditary disease. The antenatal determination of sex is useful in preventing sex-linked hereditary disorders. Although this approach does not permit one to make a direct diagnosis of all sex-linked diseases, it is highly useful since only the male suffers from these diseases and, thus, selective preventive measures can be taken. The technique should only be used as a medical indication for therapeutic abortion when sex-linked disease is a distinct possibility. Otherwise, the test could be exploited by those having a strong preference for a child of a particular sex.

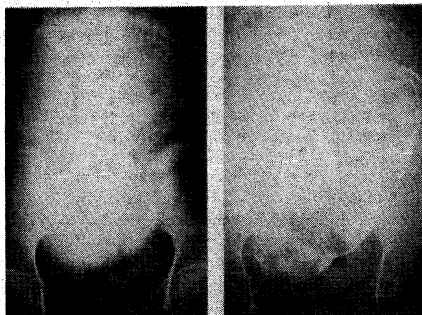


Fig. 1.—Normal human fetus as shown by fetography.

Fig. 2.—Fetograph reveals anencephalic human fetus.

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