

## **Analysis of Reasons for and Outcome of Amniocentesis at Danbury Hospital Over a Five-Year Period**

JACQUELINE P. BURNS, PH.D. AND MICHAEL DI FILIPPANTONIO  
Department of Laboratory Medicine

During the early part of the second trimester of pregnancy, analysis of amniotic fluid is usually done to rule out a chromosome abnormality or a neural tube defect. Among the most frequent reasons given for performing an amniocentesis are: advanced maternal age (AMA), maternal serum alfafetoprotein (MSAFP) levels which are increased or decreased, history of a familial chromosome aberration, or maternal anxiety.

Analysis of 1,689 consecutive amniotic fluid specimens processed at Danbury Hospital during the past 64 months revealed the following results: More than 80% were done for reasons of AMA (56%), or decreased MSAFP (24.6%), or a combination of both of these together with a family history of chromosome problems or to rule out trisomy 21 (1.2%). The remainder were done to rule out: 1) the presence of a neural tube defect because of increased MSAFP only (8.2%), or in conjunction with AMA (0.3%), or to rule out trisomy 21 or other familial chromosome aberration (0.1%); 2) other chromosome aberrations because of family history (5.2%); 3) Down's syndrome (2.5%) whether or not there was a previous family history; 4) abnormal ultrasound or maternal anxiety.

The results showed 34 cytogenic abnormal findings (2.0%), the majority (1.3%) in specimens from women 35 years of age or older. There were five abnormalities found from specimens which were ordered for decreased MSAFP (0.3%) and three because of increased MSAFP (0.2%). There was one which was ordered for both AMA and a decreased MSAFP (0.06%). If the abnormalities are categorized according to the reason for doing the prenatal test, the findings become: AMA (22/948) 2.3%; increased MSAFP (3/139) 2.2%; decreased MSAFP (5/415) 1.2%; AMA and decreased MSAFP (1/14) 7.1%.

There was a total of nine cases of trisomy 21. Of these, seven were found in women with AMA, one with abnormal ultrasound, and one with both AMA and decreased MSAFP. Therefore, trisomy 21 was found only once with a decreased MSAFP and this was in association with AMA. The low number of specimens with a decreased MSAFP may be a reason for finding only one case of trisomy 21 associated with it. Our findings reaffirm what other investigators have found, namely that patients with a low serum AFP should be counseled not only for the risk of trisomy 21, but for other chromosomal aberrations as well.