Prevalence of common aneuploidies and sex chromosome abnormalities in second trimester amniocentesis

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Background: Aneuploidy is the most common chromosome abnormality in humans, and is the leading genetic cause of miscarriage and congenital birth defects. The aim of this study is to assess the frequency of pregnancies with common aneuploidies (trisomy 13, 18, and 21) and sex chromosome abnormalities between 2 subgroups of age, the age >=35 years vs. the age < 35 years.

Methods: A total of 2,697 prenatal samples of amniotic fluids (AF) at Ramathibodi Hospital between 2011 and 2014 were analyzed for aneuploidy of chromosomes 13, 18, 21, X, and Y using conventional G-banding.

Results: A total of 78 cases (2.89 %) with common aneuploidies and sex chromosome abnormalities were identified by fetal karyotyping. In advanced maternal age group (>= 35 years), 60 abnormal cases were identified. Down's syndrome was the most commonly diagnosed (15 cases, 25.00%) and others including 3 cases (5.00%) of Patau syndrome, 11 cases (18.33%) of Edward's syndrome and 10 cases (16.67%) of X and Y chromosomal abnormalities were detected. However, sex chromosome abnormalities was the most commonly found in maternal age < 35 years (6/18 cases, 33.33%). Common aneuploidies were detected only 6 cases in this subgroup including 1 cases (5.56%) of Patau syndrome, 3 cases (16.67%) of Edward's syndrome and 2 cases (11.11%) of Down's syndrome.

Conclusions: In this study, advanced maternal age was the most common indication for second trimester amniocentesis. Down's syndrome was the most commonly diagnosed in advanced maternal age group but sex chromosome abnormalities was the most commonly found in maternal age < 35 years.