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## A STUDY ON 85 PREGNANCIES WITH CHROMOSOME ABNORMALITY DIAGNOSED BY PRENATAL CYTOGENETIC ANALYSIS

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There is no doubt that, in the last decades, conventional cytogenetics has been a powerful tool for early prenatal diagnosis (PD). The present report describes chromosomal aberrations in 85 cases referred to our private laboratory for PD in a period between 1999-2002.

Amniocentesis (AC) and chorionic villus sampling (CVS) were two methods employed to obtain samples for cytogenetic analysis. Of 85 samples with abnormal karyotypes, 84 came from AC, while only 1 was from CVS. Of those, 41 were numerical (35 trisomies, 3 sex chromosome aberrations and 3 triploidy) and 44 were struçtural aberrations. The distribution of the results is given: Down syndrome (DS) (total= 33, trisomy 21 n=30, mosaic n=1, t(14;21) type n=1, t(21;21) type n=1), trisomy 18 (n=3), mosaic trisomy 20 (n=1), 69,XXX (n=3), Turner syndrome (n=2), 47,XXY (n=1), +mar (n=2), balanced reciprocal translocation (n= 13), pericentric and paracentric inversions (n=6), and pericentric inversion 9 (n=18).

The major indications were advanced maternal age (42.5 %), positive triple test (TT) (37.0%), ultrasound abnormalities (11.1%), recurrent miscarriages (5.5 %), parental structural rearrangement(1.8%), and previous chromosomal abnormality (1.8%). Maternal age ranged between 18-47 (mean=34). Of those who had fetus with Down syndrome, 12 were below 35 year of age, and 14 were above 35. Indications for the diagnosis of DS were advanced maternal age (48.3%), positive triple test (TT) (34.5%), ultrasound abnormalities (10.3%), and recurrent miscarriages (6.9%). Of those with ultrasound abnormalities, nuchal thickness was the major finding indicating DS (66%). Pregnancies with abnormal karyotypes (n=41) were terminated following a written consent of the families. On the other hand, those with balanced chromosome changes were not terminated.

The study clearly demonstrates that cytogenetic analysis should be indicated in a situation that there is even little suspicion for high risk pregnancy. We can conclude that:

1. Average maternal age for DS child is 34.
2. Advanced maternal age was the most frequent indication for PD (42.5%).
3. Maternal age and TT still remain as significant indicators for chromosome analysis.
4. Approximately 50% of the chromosome abnormalities are structural abnormalities. This means that DS itself is not a single indication for prenatal cytogenetic analysis.
5. Prenatal ultrasound is not a method of first choice for the diagnosis of DS. Because majority of DS cases revealed by cytogenetic analysis were normal at ultrasound controls.
6. Nuchal thicness is an important finding indicating prenatal cytogenetic diagnosis

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## THE RESULTS OF 863 AMNIOCENTESIS OF ZEKAİ TAHİR BURAK HOSPITAL

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Aim: This study performed to present and discuss the results and indications of amniocentesis performed and analyzed in Zekai Tahir Burak Women's Hospital.

Material-Methods: This retrospective study covers 863 genetic amniocentesis performed cases between 1998-2001 in our hospital. These cases had firstly referred from Antenatal outpatient clinics to High Risk Pregnancy Outpatient clinic. After obstetric evaluation, all cases had referred to the Genetics outpatient clinic for genetic counseling. Amniocentesis had been performed in High Risk Pregnancy Department and amniotic fluids had been evaluated in Genetics Laboratory.

Results: 863-amniotic fluid materials had been analyzed. Indications of these cases were listed in Table 1. 798 of them presented normal fetal karyotype (373 cases were 46,XX and 439 cases were 46,XY) while 46 of presented abnormal karyotype (%5,3) (Table 2). 21 Down syndrome cases, 5 trisomy 18, 4 trisomy 13, 6 translocations, 2 pericentric inversions, 1 triploidy, 3 sex chromosome anöploidy, 1 deletion,

1 duplication, 1 Angleman syndrome and 1 46,X,+21/47,X,+21,+mar (Y chromosome) were detected. 19 cases could not reported at the first time and reamniocentesis was performed 14 of them and reported at the second study. 6 out of 19 cases were presented slow culture and 13 cases were contaminated.
Conclusions: The greatest group of indication was maternal age (35 and up) and there were 396 cases in this group while 20 had chromosomal abnormalities (% 5.05). Abnormal karyotypes were detected nearly in all groups but the most effective indications were ultrasound abnormalities, parental translocations and poor obstetric history. There are so few cases in the group of elevated AFP that, the detection rate of this group is worthless.

**Table 1: Distribution and detection rate of indications**

| | Karyotype | | | |
|---|---|---|---|---|
| | Normal | | Abnormal | |
| | No | % of total | No | % of group |
| AFP (0,75 MoM Ø) | 130 | 15,06 | 5 | 3,84 |
| AFP (2,5 MoM ≠) (NTD risk) | 3 | 0,03 | 1 | 33,34 |
| UE3 (0,75 MoM Ø) | 122 | 14,14 | 8 | 6,55 |
| HCG (0,50 MoM Ø) | 30 | 3,5 | - | - |
| HCG (2,0 MoM ≠) | 138 | 15,99 | 9 | 6,52 |
| Triple test Down syndrome (1/250) | 300 | 24,76 | 15 | 5,00 |
| Triple test Trisomy 18 (1/250) | 10 | 1,15 | 3 | 30 |
| USG abnormality | 81 | 9,38 | 14 | 17,28 |
| Chromosomaly abnormal offspring | 5 | 0,06 | - | - |
| Offspring had Down Synd. | 45 | 5,21 | 1 | 2,22 |
| Parental translocation | 7 | 0,08 | 2 | 28,57 |
| Poor obst. history | 87 | 10,08 | 8 | 9,19 |
| Offspring had cong. Anomaly | 66 | 7,64 | 2 | 3,03 |
| Maternal age over 35 | 396 | 45,89 | 20 | 5,05 |
| Colchicine uptake during pregnancy | 4 | 0,05 | - | |
| Maternal stress | 7 | 0,08 | - | |
| Papp-A/ Free B hCG | 1 | 0,01 | 1 | |

**Table 2: Distribution of abnormal karyotypes**

| Abnormal karyotype | No | Abnormal karyotype | No |
|---|---|---|---|
| 47,XY,+21 | 12 | 46,XX/ 47,XX,+13 | 1 |
| 47,XX,+21 | 7 | 47,XXY | 1 |
| 47,XY,+18 | 2 | 47,XYY | 1 |
| 47,XX,+18 | 3 | 46,XX/ 47,XXX | 1 |
| 47,XY,+13 | 1 | 46,XY, t(1;10) | 1 |
| 45,XY, t(14;21)(q10;q10) | 1 | 46,XX, t(11;16) | 1 |
| 45,XX, t(14;21)(q10;q10) | 2 | 46,X,+21/ 47,X,+21,+mar (male fetus) | 1 |
| 45,XY, t(13;14)(q10;q10) | 1 | 47,XX,+mar Par.Trisomy 9 | 1 |
| 46,XY, t(13;14)(q10;q10) | 1 | 69,XXY | 1 |
| 46,XY, t(14;21)(q10;q10) | 1 | Angelman syndr | 1 |
| 46,XY, t(13;13)(q10;q10) | 1 | 46,XY,add 13 | 1 |
| 46,XY / 47,XY,+21 | 1 | 46,XY,inv (9)(q12;q13) | 2 |