

## A t(2q; 15q) TRANSLOCATION IN A FAMILY WITH HABITUAL ABORTION AND PRENATAL DIAGNOSIS IN A RECENT PREGNANCY\*

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**SUMMARY :** We here present a family with t(2q; 15q) balanced translocation. The family has experienced 3 consecutive abortions. The mother displayed normal 46, XX chromosome complement while her husband was 46, XY, t(2q; 15q) (q37; q15). The spouses were advised to use the estimated 4/24 chances for a novel pregnancy and performed chorion biopsy sampling at the 12 th week of gestation. The fetus was found to be 46, XX. Pregnancy resulted in a normal female child which is getting very well.

**Key Words :** Chromosome Human 2, Chromosome Human 15, Prenatal Diagnosis, Habitual Abortion.

### INTRODUCTION

Translocations involving heterologues are of special interest, since they may lead to the formation of balanced as well as unbalanced zygotes. Unbalanced zygotes are either lost early or late in pregnancy or result in birth of a child with different malformations (1, 3, 4, 9). The common denominator is that abnormal karyotypes formed through translocations involving all of the members of the human complement are associated with fetal wastage with no discrimination or a phenotype that might be related to a specific pair (2, 10).

### FAMILY PRESENTATION

D. family came to our attention in early 1989. They are from a village of Nevşehir, a central Anatolian city, famous with its ancient Roman History and fairy chimneys. The man, Y.D., was 27 year - old. He was an unqualified worker in a municipal office. He had no health problem.

His wife, Z.D., 21, is from the same village. She

too is normal and healthy. They did not describe consanguinity. D. family had marriage for about 4 years. The first pregnancy occurred in the first months of union. However, it ended up in a spontaneous abortion at two - and - half months, following vaginal bleeding of about two weeks. and the story repeated 2 more times.

All tests so far applied to both couples were found within normal limits, including histerosalpingography. The family story was considered not contributory.

Cytogenetic analysis of the female was found to be 46, XX with a normal G-banding. That of the man, however, showed a balanced translocation involving Nos 2 and 15 with breakpoints (q37; q15) (Fig 1 and 2).

The family was advised for a pregnancy, for they may have a zygote with a balanced karyotype, the estimated chance being 4/24. Fetal karyotype revealed normal 46, XX complement after chorio-



Fig - 1 : Metaphase plaque (GT - banding).

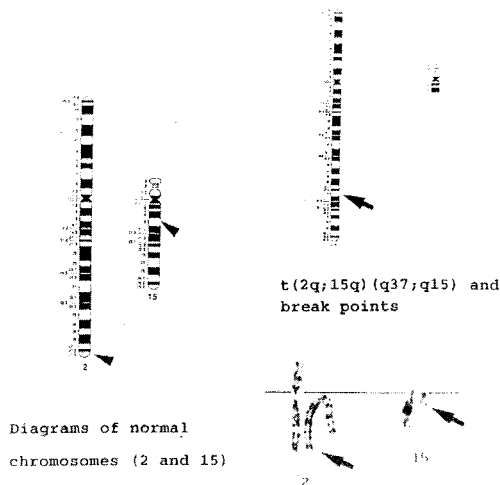


Fig - 2 : Diagrammatic representation and partial karyotype of husband.

nic villus sampling. Couples did not accept amniocentesis for control. They later wrote us that pregnancy resulted in a birth of a female child who appeared normal. They did not either accept our invitation for karyotyping the child, but declared she has been getting very well.

## DISCUSSION

Even though no fetal chromosomal analyses have been carried out, one would say that exchanged segment (s) as (were) the same or similar in all 4 previous pregnancies. We found break points (q37; q15) and it appears not to be reported hitherto. On the other hand, since individuals from the previous generation were not available for the study, the ori-

gin of the translocation could not have been traced back. However, since there is no another subject with a similar story and / or an abnormality we would conclude that it is of de novo occurrence.

Instances of t(2q; 15q) seem to be relatively fewer among reports associated with fetal wastage than, for example 13/14 translocations. We here present another case of t(2q; 15q) with habitual abortion (6, 7, 11). Yet it appears our case is somehow different from others.

There are 24 possibilities of chromosome constitution a fetus would carry of these 4/24 are expected to be normal with 46, XX (XY) complements, 2/24 balanced complements, and the remaining 16 unbalanced ones. Of these latter 4/24 are trisomy - 2 and disomy - 15; 4/24 disomy - 2 and trisomy - 15; 4/24 disomy - 2, monosomy - 15 and 4/24 monosomy - 2 and disomy - 15.

As mentioned above, we were able to diagnose fetus prenatally and the karyotype was 46, XX.

The mechanisms involved in such cases are not understood completely. But it appears that balanced chromosomal translocations are relatively infrequent in individuals having repeated abortions. Dissimilarity between reports are probably due to the differences in break points and / or segments exchanged. Alternatively, one would argue that there are separate loci in different pairs to be accounted for (5, 8, 10).

*\* This family has been presented under the same name by the some authors in 24th Annual meeting of the European Society of Human Genetics, 29 May - 1 June 1992, Elsinore, Denmark.*

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