89. A Preliminary Note on a Partial C/D Translocation Related to Repeated Abortions

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Recent cytogenetical studies have revealed that partial translocations such as those represented by B/C (Makino et al. 1965), D/D (Jacobsen et al. 1965), and E/C (Punnett et al. 1966) are a cause of repeated abortions.

The present paper reports some familial cytogenetic data involving a partial C/D translocation associated with repeated abortions.

Clinical notes and family history: The pedigree involving the family of the proband and his relatives is given in Fig. 1. The proband (Fig. 1: II, 4) is a 34 years old and clinically normal male. His wife (Fig. 1: II, 5), in a duration of her six years' marriage life, experienced five consecutive spontaneous abortions (Fig. 1: III, 8–12) at three-month-gestation in every case, and had a fullterm healthy live-birth (Fig. 1: III, 13). He has three brothers (Fig. 1: II, 2, 3, 8) and three sisters (Fig. 1: II, 1, 6, 7); they are all physically as well as mentally normal. They had all no miscarriage and no clinically abnormal child (Fig. 1: III, 1–7, 14–19). The wife of the proband (Fig. 1: II, 5) had five consecutive spontaneous abortions (Fig. 1: III, 8–12) at three-month-gestation in every case, and had a fullterm healthy live-birth (Fig. 1: III, 13). The present paper reports some familial cytogenetic data involving a partial C/D translocation associated with repeated abortions.

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Fig. 1. Pedigree involving the family of the proband with a C/D translocation.

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II, 5) is 30 years of age, being clinically normal.

Cytogenetical findings: Leucocyte cultures were set up with blood samples derived from the proband, his wife, his son and his youngest brother. Chromosome slides were made according to the Moorhead method (1960) with a minor modification.

The chromosomes of the proband showed a consistent count of 46 in 50 cells examined. Karyotype analyses in 20 cells made it possible to detect two abnormal chromosomes in C and D groups; the one corresponds most likely to no. 3 in its size, while the other is acrocentric chromosome slightly larger in size than the members of groups G (indicated by arrows in Fig. 2). The most plausible interpretation for the origin of these two abnormal chromosomes is that a translocation might have occurred between the short arm of one of the C group chromosomes and the long arm of one of the D group chromosomes. Two abnormal chromosomes are tentatively placed as heteromorphic partners of no. 10 and 15 chromosomes, respectively (Fig. 2).

![Karyotype of the proband, a partial C/D reciprocal translocation (arrows).](image)

Chromosome-counts in 50 cells and karyotype-analyses in 10 cells revealed that the wife (Fig. 1: II, 5), the son (Fig. 1: II, 13), and the youngest brother (Fig. 1: II, 8) of the proband had a normal karyotype in each.

Warkany et al. 1964, Yunis et al. 1964), it is most probable that translocation of the chromosomes is an important factor as a cause of repeated abortions and/or of malformations. Individuals with balanced translocation have been known to be phenotypically normal. But there is a possibility that, in the course of gametogenesis, genetically unbalanced gametes may be formed with a predictable frequency.

Recent literature indicates that monosomy and partial disomy of autosomes are extremely rare in livebirth, even in spontaneous abortuses. This suggests that such abnormal individuals are incompatible with livebirth and abort in early pregnancy.

Evidence presented in this study indicates that the husband had a C/D translocation, and that his wife experienced five repeated spontaneous abortions, irrespective of the fact that she is karyotypically normal having a 46, XX constitution. A possible cause for her repeated abortions is the occurrence of spermatozoa with abnormal combinations of chromosomes in the husband.

Cytogenetical features presented in this paper will provide complementary data relating chromosome abnormality to spontaneous abortion.

Summary. The wife experienced five spontaneous abortions at three months of gestation in every case. The proband showed a chromosomally balanced C/D partial translocation, while his wife and son had a normal chromosome complement.

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References