A FETAL CASE OF PARTIAL TRISOMY 6q(q21-qter) WITH RENAL DYSPLASIA

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Summary A fetal case of partial trisomy 6q(q21-qter) was reported. Renal dysplasia and coarctation of the aorta with persistent left superior vena cava were detected in the fetus in addition to the typical external phenotype, which has been described in children and adolescents with partial trisomy 6q. The significance of associated abnormalities was briefly discussed.

INTRODUCTION

In a collaborative study of fetuses aborted after prenatal diagnosis (Miyabara et al., 1985), a case of partial trisomy 6q was encountered. This type of partial trisomy has not been fully documented. This report describes some morphological features of partial trisomy 6q in the fetal stage and discuss their developmental significance.

CASE REPORT

The fetus in question was the fifth product of a 33-year-old gravida 5, ab 4 mother and a 33-year-old father, her second husband. This woman's first three pregnancies, all with her former husband, terminated in spontaneous abortions at about two or three months. Her fourth pregnancy, with her present husband, also terminated in a spontaneous abortion at four months. The cytogenetic examination of the fourth fetus revealed a karyotype 46,XY,der(7),t(6;7)(q21;p22). It was further observed that the mother was a balanced translocation carrier. She had a reciprocal translocation between the long arm of chromosome No. 6 and the short arm of chromosome No. 7. The break points occurred at bands 6q21 and 7p22, karyotype being t(6;7)(q21;p22) (Figs. 1A and 1B).

Received October 29, 1985/revised version received February 13, 1986
Fig. 1. A. Schematic illustrations of normal and derivative chromosomes 6 and 7. Arrows indicate breakpoints. B. G-banded chromosomes 6 and 7 from the mother and the fetus: t(6;7) (q21;p22).

In the conception under discussion, the cytogenetic examination by amniocentesis at 17 weeks of pregnancy showed a partial trisomy 6q having an identical karyotype 46,XY,der(7),t(6;7)(q21;p22)mat (Figs. 1A and 1B) with that of the fourth fetus. The fetus was artificially aborted at 19 weeks.

*Jpn. J. Human Genet.*