FOLLOW-UP INVESTIGATION OF A FAMILIAL TRANSLOCATION 14/18

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A follow-up investigation of a family 14/18 translocation has shown the first affected child to be followed by a healthy boy and by a similarly affected girl.

In 1967, a chromosome analysis was carried out for a child born with multiple malformations. It showed an unbalanced D/E translocation that was later identified as a 14/18 translocation (Meyer-Robisch M. and Schwanitz G. 1967. Familiäre D/E Translokation. Acta Genet. Med. Gemellol. (Roma), 16: 365-375).

The father was carrier of a balanced translocation: karyotype 46,XY,t(14;18) (p or q 11; p or q 11) (Figure).

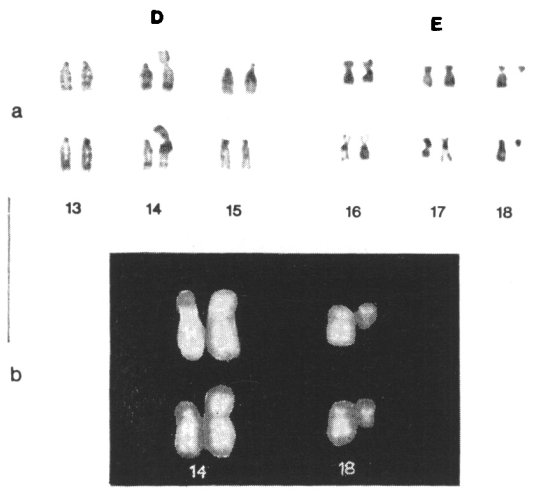

Figure. Chromosomes 14 and 18 showing Q-banding D and E groups of father showing Giemsa bands.

The proband, a girl, died at 10 weeks of bronchial pneumonia and circulatory failure.

The couple was informed of the increased genetic risk in further pregnancies. As the child with the unbalanced translocation had, in the view of the pediatrician, a short life expectancy because of its serious malformations, the couple decided to take the risk of a further pregnancy. The possibility of prenatal chromosome analysis was at that time not available.

The next child, a boy, was phenotypically healthy, and a chromosome analysis carried out in 1975 showed a normal male karyotype (46,XY).

The third child, a girl, was born in 1972 in Spain. This child again showed congenital malformations and died after six days. A chromosome analysis was not performed, but the clinical symptoms showed similarities to those of the first child, i.e., omphalocele, deep-seated ears, and hypertelorism. Further information was unfortunately not available.

In 1975, the family returned to Germany and received further genetic counseling. We informed them of the possibility of prenatal chromosome analysis and advised that an amniocentesis should be performed for any further pregnancy.

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