Inherited Translocations in Two Families 
(t(14q+;10q--) and t(13q--;21q+))

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Summary. Two families with reciprocal translocations (t(14q+;10q--) and t(13q--;21q+)) are described. In both families the proband had multiple congenital anomalies and an unbalanced karyotype, 46,XY,14q− and 46,XX,21q− respectively. Routine, autoradiographic and fluorescence techniques were used for analysis of karyotype of probands and their relatives. The probands' phenotypes and the results of their family members' dermatoglyphic analysis are presented in detail.

Zusammenfassung. Zwei Familien mit reziproker Translokation (t(14q+;10q−) und t(13q--;21q+)) werden beschrieben. In beiden Familien weist der Proband multiple angeborene Mißbildungen und einen unbalancierten Karyotyp (46,XY,14q− bzw. 46,XX,21q−) auf. Für die Analyse aller untersuchten Personen wurden neben der Routine-Methode autoradiographische und Fluoreszenz-Methoden verwendet. Die Phänotypen der Probanden sowie die Ergebnisse einer Analyse der Dermatoglyphen bei ihren Familienangehörigen werden genau beschrieben.

Introduction

In two non-related families with reciprocal translocations 46,t(14q+;10q−) (Family F1) and 46,t(13q--;21q+) (Family F2) the transmission of translocated chromosomes was observed in five and three generations respectively. The present paper presents the results of clinical investigation of two probands with unbalanced karyotypes, and the results of chromosomal, autoradiographic, fluorescence and dermatoglyphic studies of these probands and their family members.

Family F1

Proband, a male infant, was born in August 1968 after a normal pregnancy lasting 43 weeks with a birth weight of 2700 g and a length of 48 cm. He cried at once, but his cry was weak and high-pitched. The proband's inviability was evident shortly after the birth. Normal contact with the child was never obtained, he never even started to smile and he was unable to sit. In the 5th month he was admitted to the hospital because of a high temperature and congenital malformations. He died in the 11th month from cardiovascular insufficiency during a bout of fever.

Family History

At the time of the proband's birth the mother was 27 and father 30 years old. Both parents were phenotypically normal. The first pregnancy ended in the birth of a phenotypically normal
girl (see Fig. 1, IV 22) in 1965. The second pregnancy had been disrupted by a legal abortion in 1966 and the third one in 1967 by a spontaneous abortion in the 12th—14th week of pregnancy. The proband was born of the 4th pregnancy. In 1970 the 5th pregnancy, which had been complicated by bleeding in the 16th—17th week, terminated in the birth of a phenotypically normal boy (IV 26). The proband’s grandmother (II 1) had had one miscarriage, and the father’s elder sister one. Proband’s father’s sister’s son (IV 11) has one miscarriage, one stillborn baby and a daughter with congenital malformations.

Proband was seen several times, the last in July 1969 when the proband was 11 months old. His weight was 7100 g (−2σ), length 68 cm (−2σ), head circumference 41 cm (−3.6σ) and circumference of thorax 43.5 cm (−1.5σ). The examination revealed severe physical and psychomotor retardation with microcephaly, microphthalmia, narrow eye-slits with antimongoloid slant, hypertelorism and broad nasal bridge, prominent glabella, high-arched palate, micrognathia, low-set ears, short thick neck, a brown band on the distal part of the nails, abnormal position of the fingers, wide I—II interdigital space on the feet, bilateral cryptorchismus and slightly marbled skin on the limbs (see Fig. 2).

Neurological examination revealed a weak pupillary reaction to light; optic papillae were light pink with dimmish borders. There was moderate muscle hypertony. Therapeutic examination revealed a 4-grade precordial systolic murmur.

Electrocardiograms revealed bioelectrical changes showing the predominance of the right ventricle.

A congenital heart defect was diagnosed roentgenographically and bone age was retarded (without symptoms of rickets). The spinal roentgenograms showed a congenital defect of the 2nd and 3rd lumbar vertebrae (lateral cuneiform vertebrae, left scoliosis).

Intravenous urography revealed diminished kidney function.

Routine analysis of the blood (E.S.R., hemoglobin, numbers of erythrocytes, blood platelets, and leukocytes, leukocyte formula, cholesterol, bilirubin, lipids, phosphorus, sodium, potassium, calcium, residual nitrogen, and saccharum contents) yielded normal values. In serum

1 This child will be described elsewhere.