Familial D/E Translocation

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Summary. A familial D/E translocation is described. The proposita, a girl with features of the trisomy-E1 syndrome, had 47 chromosomes. The extra chromosome was a small acrocentric one. Her mother and little brother had 46 chromosomes, and showed a missing chromosome in the groups D and E, and an extra chromosome in the groups C and G. The former had a sub-terminal centromere. The latter could not be distinguished morphologically from the other small acrocentrics.

The morphology and the autoradiographic analysis of the chromosomes concerned in the translocation, indicated that it was a (17q+; 14q−) translocation. It could also be proved that the extra chromosome of the proposita represented mainly a partial trisomy 14. The father and little sister of the patient had a normal karyotype.

A comparison of the karyotypes, found in the children of the present family and in cases of D/E-translocation reported in the literature, pointed to a high frequency of non-disjunction in D/E-translocation carriers. As a possible explanation, a convergent orientation of a trivalent at metaphase I of meiosis is proposed.

Familial D/E-translocation has been described by VISLIE et al. in 1962 [9]. In two mentally retarded children an extra acrocentric chromosome was found in the G-group. In the mother there was a missing chromosome in the groups D and E and an extra chromosome was present in the groups C and G respectively. The first extra chromosome (T1), with a sub-terminal centromere, had the short arms of an E-chromosome. The small acrocentric extra chromosome (T2) in the mother as well as in both children was morphologically indistinguishable from the other G chromosomes. These findings were considered to be the result of a balanced reciprocal D/E-translocation in the mother, which gave rise to a partial trisomy D and E in the children. BREIBART et al. [1] observed a morphologically identical translocation chromosome T1 in a retarded and spastic child. In this infant the karyotype showed only 45 chromosomes and one chromosome was missing in the groups D and E. The same karyotype was described by TOWNES et al. [8] in a four-year-old child. No balanced D/E-translocation, however, was found in the parents of these two children.

A family will be described here in which an identical translocation was found. In the propositus an extra acrocentric chromosome was observed in the G-group.

Clinical Description

The propositus (Fig. 1) was born on June 26th, 1965, and was the child of healthy parents. The father was 32 and the mother 29 years old. The two other children, a girl of 6 and a boy of 2½ years old, apparently were normal.
A full term pregnancy had been uneventful and delivery occurred without complications. Birth-weight was 2800 g. Immediately after birth pronounced cyanosis and dyspnea were observed. It was noted that the child cried without making any noise. The child was placed in an incubator for oxygen therapy. The general condition, however, did not improve, and cyanosis and dyspnea persisted. Feeding was difficult. Because of the progressive deterioration of his condition, the patient was transferred to our clinic on August 9th, 1965.

On admission an atrophic baby was seen with severe respiratory difficulties and who cried without making any noise. His body-weight was 2800 g and his head-circumference 35 cm. The following abnormalities were noted. The eyes were protruding and had a slightly mongoloid position, the nose was small, there were low-set and deformed ears, a hairy forehead and micro- and retrognathia. The neck was short and showed dilated veins. On the thorax the shortness of the sternum and the lateral displacement of the nipples was obvious. Except for an umbilical hernia, the abdominal examination was normal. The fingers of both hands could not be extended fully and the index overlapped the third finger. The big toe was shorter and more bent than the second toe. On auscultation of the heart, a systolic murmur was noted, extending over all the ostia but with a maximal intensity at the apex. On ECG a right hypertrophy was seen, and X-ray pictures revealed a global enlargement of the heart. Auscultation of the lung was completely normal. Blood examinations showed a slight, normochromic anaemia, urinalysis was normal. Because of the patient's poor condition, no further serological investigations could be performed.

Digitalisation was started immediately. On August 10th, 1965, crepitations were heard on both lungs and a treatment with penicillin was given. During the following days, the patient’s general condition deteriorated, the cyanosis and dyspnea increased and on radiography progressive enlargement of the heart was seen. On August 13th, 1965, heart decompensation occurred and the patient died. Autopsy was refused by the parents.