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Three Translocations Involving C- or G-group Chromosomes

Summary. Three translocations each involving C or G chromosomes are reported. A familial translocation t(Cq+; Eq-) was identified to be rcp(6;18)(q2;q1) and two malformed children were then found to have a 46,XY(or XX),-6,+der(6) constitution. One of the carrier's pregnancy in this family was monitored by amniocentesis and a fetus was identified as being a male translocation carrier (balanced). Two other translocations were identified as rcp(11;14)(q12 or 13;q32?) and t(17;22)(p12 or 13?;q11?), respectively.

Individual chromosomes in the C- or G-group cannot be distinguished with certainty either by conventional karyotype analysis or autoradiography. However, various Giemsa-banding and quinacrine fluorescence techniques have made it possible to recognize every individual chromosome pair by their characteristic patterns of bands (Caspersson, Lomakka, and Zech, 1971; Evans, Buckton, and Sumner, 1971).

In the present report the chromosomes involved in three different structural rearrangements, including two Cs and one G, were identified by the techniques of both Giemsa-banding and quinacrine fluorescence. Further, the points of exchanges were determined within narrow limits.

Materials and Methods

Metaphase spreads were obtained by conventional leucocyte culture technique. Slides were flooded with ice chilled trypsin solution (freshly prepared, Difco 1:250 trypsin, 0.05% in Ca⁺⁺ and Mg⁺⁺ free Hanks' solution) for 10 to 30 seconds, rinsed in physiologic

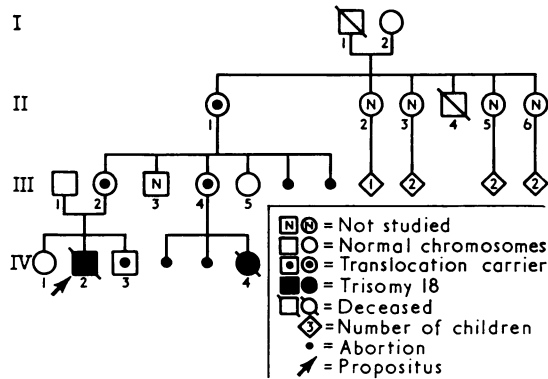


FIG. 1. Pedigree of the family with $t(6q+;18q-)$.

saline and in tap water, and then stained with Giemsa (Merck, diluted to 1:50 in 1/15 M phosphate buffer, pH 6.8) for 30 minutes. Some of the slides were stained with 0.005% solution of quinacrine mustard dihydrochloride (Polysciences Inc) in McIlvaine's buffer, pH 7.0, and observed under an Olympus FLM fluorescence microscope.

Case Reports

Case A was referred to us because of multiple congenital malformations. The propositus was 14 days of age and had typical features of trisomy 18 syndrome. A maternal cousin (case IV. 4, Fig. 1) showed similar clinical features. Both of them died later and necropsy revealed ventricular septal defects and kidney cysts. In case IV.4 there were a few additional abnormalities including Meckel's diverticulum and eventration of the diaphragm.

Chromosome analysis revealed that both case A (IV.2) and IV.4 had a $46,XY$ (or XX), $Cq+$ karyotype. Their mothers (III.2 and III.4) and maternal grandmother (II.1) appeared to have a translocation $t(Cq+;18q-)$. Amniocentesis was carried out on case III.2 in the 18th week of gestation and a fetus (IV.3) was found to be a male carrier of the same translocation (Fig. 2). Based on both Giemsa banding (G-banding) and quinacrine fluorescence (Q-banding) patterns the translocation was identified as $rcp(6;18)(q2;q1)$ according to the Paris Conference (1971) nomenclature (Fig. 2).

Case B, a 20-month-old girl, was examined because of multiple minor defects. They included webbed neck, downward slanting of eyes, flat nasal bridge, and hypoplastic nails. She was sex chromatin (buccal smear) positive and had a $46,XX,t(Cq-;Dq+)$ karyotype. G- and Q-banding studies revealed that the chromo-

somes involved were numbers 11 and 14, the propositus having a $46,XX,rcp(11;14)(q12\text{ or }13;q32?)$ karyotype (Fig. 3). Both of her parents showed a normal karyotype.

Case C was a 4-month-old girl with failure to thrive,

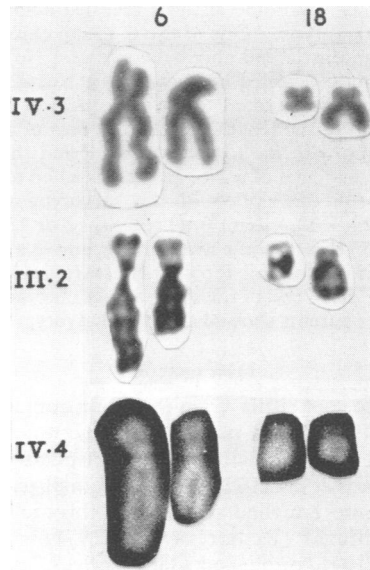


FIG. 2. Partial karyotypes showing $t(6q+;18q-)$ or $rcp(6;18)(q2;q1)$. Top: regular karyotype obtained by amniocentesis. Middle: G-banding karyotype in a carrier. Bottom: Q-banding karyotype in an 18 trisomy. Case numbers correspond to Fig. 1.

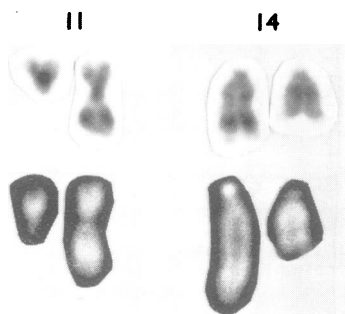


FIG. 3. G- and Q-banding karyotypes showing translocation $rcp(11;14)(q12 \text{ or } 13;q32?)$ in case B.

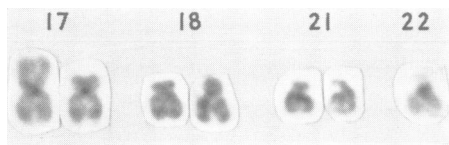


FIG. 4. Partial karyotype (G-banding) of case C. A No. 22 is missing and the short arm of No. 17 is enlarged.

broad nasal bridge, malformed low set ears, high arched palate, small mandible, short and broad neck, patent ductus arteriosus, small pelvis, and unusually long big toes. Her dermal ridges were hypoplastic.

Routine chromosome analysis revealed a $45,XX,+C,-E,-G$ karyotype. One of the C-group chromosomes was mediancentric and about the size of a No. 12. G-banding study revealed that one of the No. 22 pair was missing and translocated to the short arm of a No. 17. Comparison of the length of the short arm of the 17p+ chromosome with its homologue suggested that almost the entire long arm of a No. 22 was attached to 17 (Fig. 4). The full description of the karyotype would be $45,XX,-17,-22,+der(17),t(17;22)(p12 \text{ or } 13;q11?)$. The $der(22)$ chromosome was missing, and as a result the patient was monosomic for the pericentric region of No. 22 and the distal end of the short arm of chromosome 17. Both of her parents showed a normal karyotype.

Discussion

By the use of either Giemsa (G) or quinacrine (Q) banding, individual pairs of chromosomes can be identified without ambiguity (Caspersson *et al*, 1971; Evans *et al*, 1971). Even a small part of one chromosome attached to another chromosome has been identified (Rethoré *et al*, 1972). Recently Francke (1972) published a large series of quinacrine banding studies, in which chromosomes or parts of them involved in various translocations were determined.

In the present study, two C-group chromosomes and one G-group chromosome in three different translocations were identified; points of exchanges were also located within narrow limits.

Translocations in either case A or B seemed to be reciprocal and balanced presumably without any phenotypic effects. On the other hand, a small part of chromosomes No. 22 and 17 seemed to be missing in case C. The correlation between this seemingly minor chromosome loss and clinical abnormalities is not known.

There are various techniques of G-banding. The ASG method (Sumner, Evans, and Buckland, 1971) results in beautiful bands; however, it usually works only on freshly prepared slides. The technique used in the present report was found satisfactory when it was applied on slides that were kept in a vacuum desiccator for up to 6 months. In certain circumstances, where freshly prepared slides cannot be obtained, this technique may prove to be useful.

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