Case Reports

dehydrogenase determination and Drs Kenneth Adams, Steven Myers, and Phillip Rennick (Lafayette Clinic, Detroit, Michigan) for the neuropsychological studies. Hyland Laboratory, California, kindly provided the Xg® anti-serum.

WALDEMAR A. PALUTKE, YINGSAN CHEN, and HAROLD CHEN

Departments of Pathology and Medicine, Detroit General Hospital and Wayne State University, and the Growth and Development Center, Children's Hospital of Michigan, Detroit, Michigan, USA

REFERENCES


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 Ca2+ and Mg2+ free Hanks' solution) for 10 to 30 seconds, rinsed in physiologic

Received 18 September 1972.
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 chromosomes involved were numbers 11 and 14, the propositus having a 46,XX,rcp(11;14) (q12 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,

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 chromosomes involved were numbers 11 and 14, the propositus having a 46,XX,rcp(11;14) (q12 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,
Case Reports

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.

This study was supported in part by grant 87557 from the Ministry of Education, Japan. The authors would like to thank Dr E. Matsunaga for his encouragement and to Dr F. A. Lilienfeld for reading the manuscript. Thanks are due to Miss H. Kamegai for her superb technical assistance.

YASUO NAKAGOME, KAZUSO IINUMA, and
ICHIRO MATSUI

Department of Human Genetics, National Institute of
Genetics, Mishima
and the Division of Medical Genetics,
Kanagawa Children’s Medical Centre,
Yokohama, Japan

References

fluorescence patterns of the human metaphase chromosomes—
distinguishing characters and variability. Hereditas, Genetiskt

Evans, H. J., Buckton, K. E., and Sumner, A. T. (1971). Cyto-
logical mapping of human chromosomes. Results obtained with
quinacrine fluorescence and the acetocarmine-Giemsa techniques.
Chromosoma, 35, 310–325.

Francke, U. (1972). Quinacrine mustard fluorescence of human
chromosomes. Characterization of unusual translocations.
American Journal of Human Genetics, 24, 199–213.

Foundation—March of Dimes, New York.

Rethoré, M. C., Dutrillaux, B., Baheux, G., Gerveaux, J., and
Lejeune, J. (1972). Monosomie pour les régions ixexactcroto-
mérés d’un chromosome 21. Experimental Cell Research, 76,
455–456.

technique for distinguishing between human chromosomes.

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 deter-
mined.

Fig. 3. G- and Q-banding karyotypes showing translocation
rcp(11;14)(q12 or 13;q32 ?) in case B.

Fig. 4. Partial karyotype (G-banding) of case C. A No. 22 is miss-
ing 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 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.
Three Translocations Involving C- or G-group Chromosomes

Yasuo Nakagome, Kazuso Inuma and Ichiro Matsui

doi: 10.1136/jmg.10.2.174

Updated information and services can be found at:
http://jmg.bmj.com/content/10/2/174

These include:

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/