Trisomy D/Trisomy E Mosaicism in an Infant Male

RICHARD J. WARREN* and JOHN I. KEITH

From Edward Mallinckrodt Department of Pediatrics, Departments of Medicine and Pathology, Washington and the University School of Medicine, Division of Genetics, St Louis Children's Hospital, St Louis, Missouri, USA

The rarity of double autosomal trisomy has been pointed out by Porter, Petersen and Brown (1969). The patient reported here is the third case of double autosomal trisomy as a mosaic for the group D and E chromosomes (47,XY,D+/47,XY,E+).

Case Report

The patient, a male, was the third child of a 41-year-old mother and 45-year-old father. The pregnancy was terminated after a 39-week gestation by a Caesarean section for elective sterilization.

Apart from the development of polyhydramnios (estimated 5 l), the pregnancy was uneventful. There was no history of viral infection, drug ingestion, or radiation exposure. The mother had previously given birth to two normal children (both by Caesarean section) when she was 19 and 40 years old, respectively. Family history was negative for congenital malformations.

The birthweight was 2000 g, and body length was 44 cm. The child was cyanotic and in extreme respiratory distress. No breath sounds were heard over the left thorax or over the right anterior thorax. He lapsed into periodic apnoeic spells and died within one hour.

A complete necropsy was performed, excepting the eyes, 3 hours after expiration. Externally, there were multiple anomalies. Both auricles were low-set and slanted posteriorly. The antihelix was absent (Fig. 1). There was a right cleft lip and an ipsilateral posterior cleft palate (Fig. 2). Both hands showed marked radial deviation and there was a simian crease across each widened palm (Fig. 3). The thumbs were relatively hypoplastic, and all fingers were flexed with deviation of the index and fifth fingers towards the longitudinal axis of the hand. The nails of the fingers and the toes were narrow and hyperconvex. There were rocker-bottom feet with prominent heels (Fig. 4).

Exposure of the thoracic and peritoneal cavities revealed that most of the anterior abdominal cavity was occupied by a large symmetrical liver (Fig. 5). Most of the small intestine had herniated through a large posterolateral (Bochdalek) defect of the left hemidiaphragm. The mediastinum was deviated to the right, and the heart occupied most of the anterior right thoracic cavity.

Received 26 November 1970.
* Present address: Mailman Center for Child Development, PO Box 6, Biscayne Annex, Miami, Florida 33152, USA.

Fig. 1. The ear; note the absent antihelix.

Fig. 2. The face; note the cleft lip and palate.
Posterior to the herniated small bowel was a compressed hypoplastic left lung (2 g in weight and 2 cm in greatest dimension). The right lung weighed 12 g, was atelectatic and compressed behind the displaced heart. There were a patent foramen ovale and patent ductus arteriosus. There was a small accessory spleen adjacent to the splenic artery near the splenic hilum. There was no Meckel's diverticulum in the small bowel, but due to malrotation of the colon, the caecum was attached by the mesocolon to the left posterior abdominal wall. The only urogenital anomaly was an intra-abdominal left testicle.

Extensive cerebral subarachnoid hemorrhages were found, but the central nervous system was morphologically normal, both on gross examination and on microscopy.

**Cytogenetics.** Peripheral lymphocytes were cultured for chromosomes. Seventy-six cells were analysed and all contained 47 chromosomes (Table I). Karyotype analyses revealed a mosaic pattern (47,XY,D+ / 47,XY,E+) (Fig. 6).

**Discussion**

Two cases of double autosomal trisomy for the group D and E autosomes have been reported. The first had 3 cell lines (47,XX,D+/47,XX,E+/48,XX,D+,E+) and showed a mixture of the clinical features of trisomy D and trisomy E (Baikie, Garson, and Birrell, 1965; Garson et al, 1969). The other karyotyped like our patient (47,XY,D+/
More complicated schemes have been devised (Zellweger and Abbo, 1967) and showed the clinical features of trisomy D (Zellweger and Abbo, 1967). Our patient, like the first, showed a mixture of the phenotypes.

The singular notable complication in the prenatal course of our patient was polyhydramnios. Polyhydramnios was reported in another instance of double autosomal trisomy, a case of 47,XY,D+/
47,XY,G+/48,XY,D+,G+ (Porter et al, 1969). Advanced maternal age was present in our case and has been a factor twice before with double autosomal trisomies (Hsu et al, 1965; Smith, Tips, and Howard, 1965). The relationship between advanced maternal age and trisomy G, as well as other aneuploid abnormalities, has long been acknowledged.

Double autosomal mosaicism could arise as the result of either meiotic or mitotic packaging errors. This type of mosaicism can most simply be explained by two nondisjunctional events at the two-cell stage of the zygote. One would have to hypothesize that one cell disjoined in the E group; the other in the D group, and the resulting monosomic cells died. More complicated schemes have been devised (Zellweger and Abbo, 1967), but this is the simplest and involves the fewest karyokinetic mistakes. Aberrant karyokinetic events later during the formation of the zygote would result in the existence of normal cells, but none were found in the blood of our patient. The early demise of the patient precluded our culturing other tissue for the presence of normal cells.

**Summary**

A patient is described who had two trisomic cell lines, 47,XY,D+/
47,XY,E+ . The necropsy findings are given in detail. A possible mechanism for double autosomal trisomy is discussed.

We gratefully acknowledge the expert technical assistance of Mrs S. Yates and Mr Ronald E. Durbin. We are indebted to Drs Philip Dodge, Robert Kaufman and William Sly for reviewing the manuscript and to Drs John Karpel and David Raimon for referring the patient.

**References**


Trisomy D-trisomy E mosaicism in an infant male.

R J Warren and J I Keith

*J Med Genet* 1971 8: 384-386
doi: 10.1136/jmg.8.3.384

Updated information and services can be found at:
[http://jmg.bmj.com/content/8/3/384.citation](http://jmg.bmj.com/content/8/3/384.citation)

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](http://group.bmj.com/group/rights-licensing/permissions)

To order reprints go to:
[http://journals.bmj.com/cgi/reprintform](http://journals.bmj.com/cgi/reprintform)

To subscribe to BMJ go to:
[http://group.bmj.com/subscribe/](http://group.bmj.com/subscribe/)