Double Autosomal Trisomy (Trisomy D+G) with Mosaicism

IAN H. PORTER, WILLIAM PETERSEN, and CHARLES D. BROWN

From New York State Department of Health, and Department of Pediatrics, Albany Medical College, Albany, New York, U.S.A.

Reports of patients with double autosomal trisomy are rare (Table). In contrast, patients with double aneuploidy involving autosomal and sex chromosomes are slightly more frequent (Zellweger and Abbo, 1967). The reason for this is not known.

The case we wish to report is a double autosomal trisomy with mosaicism involving group D and G chromosomes.

Case Report

The patient, a male, the seventh child of a 37-year-old father and a 34-year-old Caucasian mother, was the product of a pregnancy complicated by massive polyhydramnios. The mother previously had had two miscarriages. During the present pregnancy she had not had any infections or X-irradiation. There was no family history of congenital malformations, mental retardation, leukaemia, autoimmune disease, or diabetes.

Three days after birth the patient was transferred to the Albany Medical Center Hospital because of persistent vomiting, absent bowel movements, and x-rays showing massive distension of the stomach and proximal duodenum, with no gas in the distal intestine. In addition, the child had the features of Down's syndrome: flat facial profile, epicanthic folds, oblique palpebral fissures, Brushfield spots, broad hands and short fingers,

incurving little fingers, and bilateral simian creases. He was, however, hypertonic, and held his back in opisthotonos suggesting the presence of other abnormalities (Fig.). The cerebrospinal fluid was normal.

At operation an annular pancreas was found compressing the duodenum at the junction between the first and second portions. A duodenostomy was performed but in spite of this the child died shortly after the operation.

Cytogenetics. Chromosomal analyses were made from preparations obtained from cultures of peripheral blood lymphocytes. The chromosomes from 43 cells were karyotyped. Forty-one cells had 47 chromosomes of which 10 had 47,XY,G+ and 31 had 47,XY,D+. The remaining two cells had 48,XY,D+,G+.

Discussion

The only previously reported case of 47,XX,D+/47,XX,G+ mosaicism also had some of the characteristic signs of Down's syndrome, and also was the daughter of an older mother (39 years). She did, however, have some of the features of the D₁ trisomy syndrome, such as micrognathia, spasticity, and rockerbottom feet (Smith, Tips, and Howard, 1965) (Table).

Of the cases with 47,XX,E+/47,XX,G+ mosaicism, one showed predominantly the features of the trisomy-18 syndrome (Hsu *et al.*, 1965), the others the features of Down's syndrome (Marks,

Received October 14, 1968.

TABLE
CASES OF MOSAICISM FOR DOUBLE AUTOSOMAL TRISOMIES

Author	Cell Lines	Clinical Features
Hsu <i>et al.</i> (1965)	46,XX	Trisomy-18 syndrome
Smith <i>et al.</i> (1965)	46,XX 47,XX,G+ 48,XX,D+,G+	Down's syndrome with micrognathia, rockerbottom feet, and spasticity
Baikie <i>et al.</i> (1965)	47,XX,D+ 48,XX,D+,E+	Mixture of D ₁ trisomy and trisomy-18 syndromes
Marks <i>et al.</i> (1967)	47,XX,E+ 47,XY,E+ 47,XY,G+	Down's syndrome
Zellweger and Abbo (1967)	46,XY 47,XY,E+ 48,XY,G+,E+	Down's syndrome with duodenal atresia and hypertonia
Zellweger and Abbo (1967)	47,XY,G+ 47,XY,D+ 47,XY,E+	D ₁ trisomy
This report	47,XY,D+ 48,XY,D+,G+	Down's syndrome with annular pancreas and hypertonia

FIG. Note the opisthotonos, mongoloid features, and simian hand, with incurving little finger.

Wiggins, and Spector, 1967; Zellweger and Abbo, 1967). It is interesting that the latter case also presented with duodenal atresia and hypertonia.

The two cases with 47,XX,D+/47,XX,E+ mosaicism had a mixture of the characteristic clinical signs of these syndromes (Baikie, Garson, and Birrell, 1965; Zellweger and Abbo, 1967).

The simplest explanation of the way the mosaicism arose in our case seems to be that the aneuploidy arose by nondisjunction during meiosis in gametogenesis followed by nondisjunction or anaphase lag during mitosis in the zygote.

Two other cases of double autosomal trisomies have been reported (Gagnon *et al.*, 1961; Becker, Burke, and Albert, 1963). These were not, however, mosaics.

Summary

A boy with multiple congenital malformations and three chromosomal stem lines, 47,XY,D+,

47,XY,G+, and 48,XY,D+,G+, has been described, and the other reported cases of double autosomal trisomy have been reviewed briefly.

REFERENCES

- Baikie, A. G., Garson, O. M., and Birrell, R. G. (1965). Mosaicism for trisomy 17-18 and trisomy 13-15 in man. *Nature (Lond.)*, **207**, 1419.
- Becker, K. L., Burke, E. C., and Albert, A. (1963). Double autosomal trisomy (D trisomy plus mongolism). *Proc. Mayo Clin.*, **38**, 242.
- Gagnon, J., Katyk-Lontin, N., DeGroot, J. A., and Barbeau, A. (1961). Double trisomie autosomique à 48 chromosomes (21 + 18). *Un. méd. Can.*, **90**, 1220.
- Hsu, L. Y-F., Schwager, A. J., Nemhauser, I., and Sobel, E. H. (1965). A case of double autosomal trisomy with mosaicism: 48/XX (trisomy 18 + 21) and 46/XX. *J. Pediat.*, **66**, 1055.
- Marks, J. F., Wiggins, K. M., and Spector, B. J. (1967). Trisomy 21-trisomy 18 mosaicism in a boy with clinical Down's syndrome. *ibid.*, **71**, 126.
- Smith, G. S., Tips, R. L., and Howard, H. (1965). Autosomal mosaicism occurring in conjunction with Down's syndrome. *Amer. J. ment. Defic.*, **70**, 218.
- Zellweger, H., and Abbo, G. (1967). Double trisomy and double trisomic mosaicism. *Amer. J. Dis. Child.*, **113**, 329.