

47,XX,13+ with Snodgrass Phenotype II* Are Different Chromosomes Associated with Two Clinical Varieties of D-trisomy?

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Snodgrass *et al* (1966) divided the D trisomy syndrome into two phenotypic categories based on facial appearance. Patients in their first category had facies associated with defects of prosencephalic cleavage. A striking finding was median cleft lip and palate. In addition there was orbital hypotelorism with severe ocular anomalies, hypoplasia or aplasia of the crista galli, of the median philtrum, and of other parts of the ethmoid bone and the nasal septum. According to these authors, this malformation complex is due to the aberrant development of the embryonic median fronto-nasal process.

In the second category of Snodgrass *et al*, the facies did not exhibit the prosencephalic type of defect. These patients closely resemble each other and bear little resemblance to category I patients. They show mild microcephaly and micrognathia. The nose is large, with a broad ridge and bulbous tip. There are redundant skin folds in the mandibular and periorbital regions. The mouth may be large and turned down. There are no severe optic defects; cleft lip and palate are not usually observed.

We have had the opportunity to do an autoradiographic study on an infant girl with category II D trisomy (Fig. 1). The extra chromosome in this case appears to be a D₁ (Fig. 2).

Comment

Yunis, Hook, and Mayer (1964) proposed that the D group chromosome with late replication in the distal portion of the long arm be designated as D₁. The D₂ chromosomes in the metaphase were more heavily labelled in the proximal portion, and the D₃ chromosomes were the most lightly labelled in the D



FIG. 1. The patient's face at age 1 week, showing lack of prosencephalic defects (phenotypic category II).

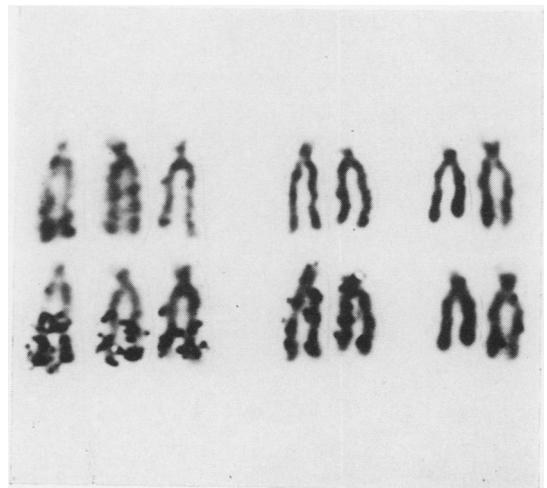


FIG. 2. Labelled D group chromosomes from the patient in Fig. 1 showing the extra chromosome to be a D₁.

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group. Autoradiographic evidence was also presented by these authors that the extra chromosome in a case of phenotypic category II D-trisomy was a D_1 chromosome.

Other studies have also indicated that a D_1 chromosome is the extra one in D trisomy (Büchner, Pfeiffer, and Stupperich, 1965; Giannelli, 1965; Giannelli and Howlett, 1966). Allderdice and Miller (1968), however, reported a D trisomy patient whose extra D chromosome labelled more heavily in the proximal portion, indicating it was a D_2 . Clinical descriptions of the patients were not given in the last four references, so it is not clear whether these patients fit into phenotype category I or II.

Based on these findings it occurred to us that the two clinical categories of D trisomy might involve two different D group chromosomes, D_1 and D_2 . Further autoradiographic studies on D trisomy patients from both phenotypic categories I and II would help resolve this question.

Summary

Autoradiographic studies have been made on an infant with D trisomy of the Snodgrass category II

phenotype. The results indicated that the trisomy was associated with a D_1 chromosome. This suggests the possibility that D trisomy of the phenotype I category may involve other chromosomes of the D group.

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