Case reports

This case was complicated in that 9% of the non-polyplodid cells had 18 trisomy, so that it was unclear which chromosome abnormality might be responsible for which anomalies.

The cases most similar to ours are those of Kohn et al (1967) and Kelly and Rary (1974). The former was a tetraploid/diploid mosaic infant with 69% tetraploid leucocytes and 1% tetraploid marrow cells but no demonstrable tetraploid in skin, muscle, or lung tissues. The other was a 2-year-old girl with 29% tetraploid cells in different tissues. Both infants had low birthweight, small size, decreased in utero movement, hypotonia, and a simian line as did our patient. Kohn's patient, like ours, had eye abnormalities, failed to gain weight or develop, and died suddenly before 1 year of age.

A unique facet of our case is that there was no mosaic diploid cell line. Many ‘broken’ metaphase spreads with a chromosome number of 80 to 90 were seen but this was felt to be an artefact of slide preparation. The two counts of 45 chromosomes (identified as ‘diploid in Table 1) most probably represent such ‘broken’ spreads and not a true diploid cell line. The cell nuclei on the necropsy tissue slides were all of one size and did not fall into two size classes as seen in the mosaic case of Kohn et al (1967). The relation between the maternal use of spray adhesive and the chromosome abnormality is probably spurious. The original claim of karyotype irregularities secondary to use of spray adhesives has since been refuted (Seely, 1973; Cervenka and Thorn, 1974). We, as well as others, have performed chromosome studies on pregnant women exposed to spray adhesives but no increase in chromosome breaks or other abnormalities was noted (F. Conte, M. Golbus, and B. Hall, unpublished observations).

The most significant aspect of this case is not that certain anomalies were associated with the tetraploidy, but that the anomalies were not more severe. Our patient, with 46 extra chromosomes, had abnormalities comparable to those seen in patients with the single extra chromosome in trisomy 13 or trisomy 18. This suggests that the balance between chromosomes and/or the ratio between different portions of the chromatin is more important than the absolute number of chromosomes present, but does not negate the fact that even ‘balanced’ polyploidy is developmentally deleterious.

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REFERENCES

46,XY/46,XY,21q– mosaicism in an infant with neutropenia and properdin deficiency*

Summary. An infant with neutropenia, properdin deficiency, and a 46,XY/46,XY,21q– mosaicism is described. It is not known whether these two findings are related to the missing 21q material.

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The propositus is normal in appearance, and has none of the phenotypic features associated with the G-group deletion syndromes.

Lejeune and co-workers (1964) described an infant lacking G-group chromosomal material, and referred to the phenotype of their patient as 'le contre type' of Down's syndrome. The term 'antimongolism' was later applied to this syndrome by Reisman et al (1966).

Subsequently, other patients with total or partial G-monosomy have been reported, and two distinct syndromes have emerged (Warren and Rimoin, 1970; Kelch et al 1971). Banding studies have shown that monosomy for part of the number 21 chromosome is responsible for the clinical entity called antimongolism (Warren et al, 1973; Richmond, MacArthur, and Hunter, 1973), and perhaps this syndrome should be referred to in the future as the 21 deletion syndrome.

The clinical features of individuals with the 21 deletion syndrome include downward slanting palpebral fissures, protruding nose, blepharochalasia, micrognathia, large low-set ears, and other anomalies. The case presented here is that of an infant with 46,XY/46,XY,21q- mosaicism; he has none of the clinical features seen in the 21 deletion syndrome, and is quite normal in appearance.

Case report

The propositus is the first child of 20-year-old parents, and was born at term after a normal pregnancy. He was referred to the State University Hospital at 1 week of age because of pneumonia and neutropenia. Physical examination at this time revealed a phenotypically normal infant with the presence of râles in the lower lobe of the right lung. A complete blood count showed an absolute neutropenia with neutrophil counts ranging from 100 to 180/mm³. A bone marrow aspiration was performed, and the aspirate was a normocellular specimen with normal myeloid maturation. No rise in neutrophil counts occurred with the presence of any infection, and the neutropenia resolved spontaneously at 6 months of age. During this interval the subject experienced other episodes of infection, including pneumonia (from which no bacterial pathogen could be isolated), cellulitis, and numerous episodes of oral moniliasis. Candida and streptokinase-streptodornase skin tests were repeatedly negative. Serum immunoglobulins were normal for his age. Beginning at 6 months of age no further unusual infections were noted. A physical examination of the propositus at 1 year of age was within normal limits; his height and weight were at the 25th centile; his psychomotor development was within normal limits.

Laboratory studies

A few drops of the bone marrow aspirate taken at 1 week of age were sent to the cytogenetics laboratory for chromosome analysis. The modal chromosome number was 46, but 9 of the 20 metaphases examined were found to have a Gq-chromosome (Fig. 1). A chromosome analysis of 100 metaphases from a peripheral blood culture revealed that 49 had a 46,XY,Gq- complement and 51 a 46,XY complement. One hundred metaphases were also examined from a skin fibroblast culture; 43 were 46,XY,Gq- and 57 were 46,XY. Subsequent G-banded chromosome preparations made using the technique of Frey et al (1972) showed the deleted chromosome to be a number 21 (Fig. 2). There was no
Evidence that the deleted 21q material was translocated onto another chromosome.

The chromosome complements of both parents were normal, which is consistent with the post-fertilization development of mosaicism.

Studies were also made of the host defence mechanisms because of the patient’s recurrent infections. The remarkable finding was a persistent decrease in the level of serum properdin as quantified by radial immuno-diffusion (Mancini et al., 1965). Serial determinations of the patient’s serum properdin for an 11-month period (made by Dr. Spitzer) varied from 29 to 44%, of normal age-matched controls. Serum properdin levels vary with age during the first 6 to 9 months of life; thus the above values are expressed as a percentage of the mean normal value for that age. All of the patient’s values were lower than 3 SDs below that normal mean.

In addition, it was shown that the properdin deficiency contributed to, or was largely responsible for, a failure to promote phagocytosis.

Another defect which was interesting but transient in nature was an initial deficit in cell-mediated immunity. Whether this was secondary to his infectious process or indicated a delay in maturation or temporary involution of cellular function could not be determined.

**Discussion**

The patient has no phenotypic features of the 21 deletion syndrome. This may be attributable to the presence of the normal cell line, an undetectable translocation of the deleted material to another chromosome in the apparent 46,XY,21q- cells, or to the fact that the short arms of the 21q- chromosome appear to be intact. The previous cases of this syndrome are monosomic for some of the short arm as well as long arm material of chromosome 21.

The immunological abnormalities represent an interesting finding in relation to this patient’s chromosomal defect. Whether the properdin deficiency can be directly related to the chromosomal abnormality is entirely speculative; further data pertaining to this relation would be of great value.

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**References**


