Trisomy of Chromosome 16 in a Neonate, 47,XY,?16+

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M.W., date of birth 27 November 1969, reference PRU 5621/8114 was the third child of healthy unrelated parents, mother and father both aged 30 years. He was born at term by Caesarean section for fetal distress after an otherwise uneventful pregnancy. His birthweight was 2660 g., the placenta weighed 481 g., and there were three vessels in the chord. At birth he was noted to have major somatic anomalies and his condition was very poor. Respiration was established with difficulty and he died at 8 hours of age. He had an abnormally shaped head with microcephaly and marked flattening of the occipital region. Ocular hypertelorism was marked and epicanthic folds present. He had bilateral microphthalmos. There was a severe double harelip and a cleft palate. The ears were unusual, being cup-shaped and low set to a degree usually seen in Potter's syndrome (Fig. 1). There was brachydactyly of hands and feet, and there was a single transverse palmar crease on the right. The genitalia were underdeveloped with a penis only 2-3 mm. long and undescended testes.

At necropsy, there was ventricular septal defect and the ductus arteriosus was patent. The spleen was large, the large intestine malrotated, with an ascending mesocolon and a large pelvic colon. The adrenals and urinary

tract were normal. There was a small right inguinal testis otherwise macroscopically normal. The left testis, also inguinal, was represented by an amorphous blob of tissue. The skull was abnormally thickened posteriorly, but the brain though small was macroscopically normal. Optic and olfactory nerves were present.

Histological examination showed that the right gonad was completely replaced by a tumour resembling a seminoma or embryonic teratoma. There was no testicular structure and the tumour filled some of the blood vessels and invaded the capsule (Fig. 2). The left gonad consisted of connective tissue (Fig. 3).

Dermatoglyphs. Inspection of the patterns revealed simple arches on all 10 digits and normally situated *a*, *b*, *c*, *d*, and *t* triradii on the palms.

Chromosome studies on peripheral blood, taken *post mortem*, showed a modal number of 47 in all 29 cells counted, with an extra submetacentric chromosome resembling number 16 in arm ratio, size, and position of the secondary constriction in the long arm (Fig. 4).

Oral smears and fibroblast nuclei from right and left gonads were chromatin negative. The tumour tissue was also chromatin negative though most cells were too anaplastic to be assessed (Fig. 2). The absence of sex

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FIG. 1. Front and side view of patient showing extreme hypertelorism, harelip, low set cup-shaped ears, and brachycephaly.

FIG. 2a. Low power views of right 'gonad' showing replacement by tumour, infiltration, and invasion of the capsule by round cells.

FIG. 2b. High power view of right 'gonad' showing cellular anaplasia and mitotic figures.

FIG. 3. Low power view of left 'gonad' showing connective tissue.

chromatin supports the conclusion that the extra chromosome is an autosome. Both parents have normal chromosomes. Apparently the infant is a primary 16 trisomic male, 47,XY,?16+. However, it cannot be excluded that the extra chromosome is a structurally abnormal autosome which arose during gametogenesis and which is present in an unbalanced state following non-disjunctional segregation.

Comment

In the literature there are three reports of trisomy 16, by Lewis *et al.* (1963), Melnyk, Thompson, and Hecht (1967), and Gilgenkrantz *et al.* (1967). In the first two cases the affected subjects are adults with severe mental retardation and relatively minor somatic anomalies. Gilgenkrantz *et al.* report an infant with multiple malformations including microcephaly, arrhinencephaly, and congenital heart disease who died at the age of 7 months. She did not resemble the present case. A further case of interest is that of van Wijck *et al.* (1961) with multiple malformations of the midline type including marked hypertelorism, low set ears, a double harelip and a cleft palate. There was a large meningocele which malformation was also present in a sib. She had a modal number of 47 with an extra chromosome 'in group 16-18 of the Denver system' which the authors regarded as 18 but which, on inspection of their published karyotype, could equally be regarded as chromosome No. 16. This case of van Wijck resembles the present one.

FIG. 4. Karyotype showing 47,XY,?16+.

Trisomy 16, well known in spontaneous abortions, is, thus it seems, exceedingly rare in live births. On the basis of 5 cases only 2 of whom resemble one another, and with the uncertainty of chromosome identification, it would be premature to talk of a trisomy 16 syndrome in live births.

Summary

An infant born at term with major somatic anomalies and a gonadal tumour lived only for 8 hours. Cytogenetic studies showed apparent trisomy of chromosome 16, 47,XY,?16+. Four other possible cases of 16 trisomy are discussed.

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