Case reports

Journal of Medical Genetics, 1980, **17**, 389–402

A liveborn case of 49,XXXY,+18

SUMMARY The first case of a liveborn male infant trisomic for both the X and the No 18 chromosome is presented. The patient had multiple congenital anomalies many of which were similar in appearance to patients with trisomy 18. The proband died after 2 days. Both maternal and paternal karyotypes were normal.

Double aneuploidy has been described earlier.[1] Most reported cases involve trisomy for a sex chromosome as well as a classical autosomal trisomy syndrome.[2–4] To our knowledge this is the first case of a liveborn male infant with a chromosomal complement of 49,XXXY,+18.

Case report

The proband, a male, was born to a 19-year-old primagravida mother and a 24-year-old father. Pregnancy was normal except for a urinary tract infection during the 5th month of gestation. Labour occurred spontaneously at 36 weeks and delivery was complicated by placenta abruptio. Polyhydramnios was noted. Apgar scores were 3 at 1 minute and 4 at 5 minutes.

Physical examination showed a small premature male infant with several congenital anomalies. The birthweight was 1130 g and the length 40 cm. The head appeared elongated with a circumference of 28 cm. There was hypertelorism. The ears were low set in position and only a rudimentary auricular appendage was present on the left (fig 1). The mouth was very small and micrognathia was apparent. Examination of the chest revealed a 2/6 holosystolic murmur along the left sternal border. The penis was small and the testes were palpable in the inguinal canal. Rockerbottom feet were present bilaterally. The hands had flexion contractures with overlapping of the second and third fingers and clinodactyly was present. The palmar ridge creases were normal, but the dermal ridge patterns were not recorded.

The infant's hospital course was brief. Marked respiratory distress persisted despite vigorous resuscitation efforts and he died at 48 hours of age. Permission for a necropsy examination was not granted.

Metaphase preparations from short term lymphocyte cultures and skin fibroblast cultures showed a consistent karyotype (fig 2) of 49,XXXY,+18 (GTG). Fifty cells were analysed from cultures of each tissue. Normal karyotypes were found in short term lymphocyte cultures from both parents.

Discussion

Double aneuploidy involving trisomy for an autosome and a sex chromosome has been described by

Received for publication 17 May 1979

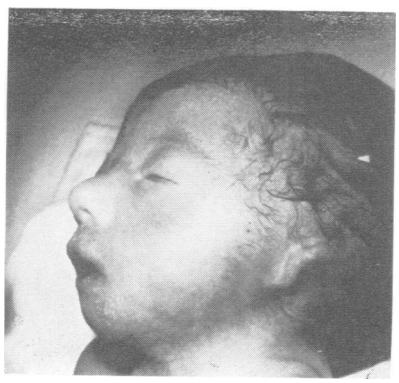

FIG 1 *Proband at 24 hours of age. Note unilateral microtia.*

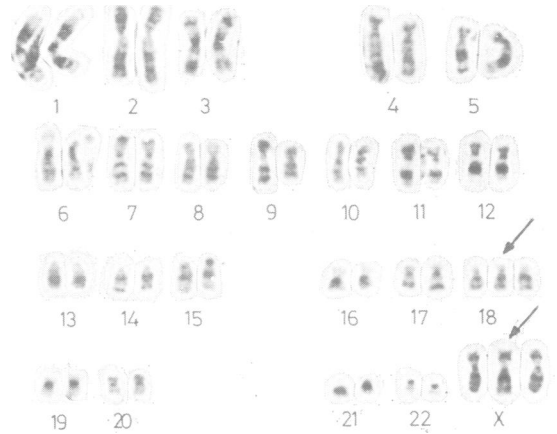

FIG 2 *Karyotype of the proband: 49,XXXY,+18 (GTG). (Original magnification × 1600).*

several investigators.[1–3] The modal chromosomal number reported in all cases has been 48. This is the first case in which 49 chromosomes were found in a patient with double aneuploidy.

The phenotypic characteristics of this patient most closely resemble those of trisomy 18. This observation is consistent with those described in other cases of double aneuploidy involving chromosome 18 and sex chromosomes.[4–6] One explanation for this is that there are no specific phenotypic findings in the sex chromosome syndromes in the newborn period.[4]

It is of interest to note the presence of unilateral microtia in our patient. This is not a characteristic finding in trisomy 18. Unilateral malformed ears have been reported in other cases of double aneuploidy with karyotypes of 48,XXY,+13[2] and 48,XXY,+18.[3 5] This might be a reflection of the major chromosomal imbalance present since it is not syndrome specific.

NATALINE B KARDON,* ANN L BERGER,* MICHAEL ELICE,* JESSICA G DAVIS,* AND EDMUND C JENKINS†
*Child Development Center/Genetics, Department of Pediatrics, North Shore University Hospital; the Department of Pediatrics Cornell University Medical College; and the †Department of Genetics, New York State Institute for Basic Research in Mental Retardation, 1050 Forest Hill Road, Staten Island, New York 10314, USA

References

1 Hamerton JL. *Human cytogenetics.* vol 1. New York: Academic Press, 1971:254–5, 279–80.
2 Mailhes JB, Moore CM, Gershanik JJ. A case of double trisomy in a liveborn infant: 48,XXY,+13. *Clin Genet* 1977;**11**:147–50.
3 Madahar DP, Dosik H, Wexler I. 48,XXX,+18 double trisomy. *J Med Genet* 1974;**11**:309–11.
4 Nielsen, J, Vetner M, Holm S, Askjaer AA, Reske-Nielsen E, Zizken J. Klinefelter's syndrome and trisomy 18 in a newborn boy. *Clin Genet* 1978;**13**:259–64.
5 Cohen MM, Bumbalo TS. Double aneuploidy. Trisomy 18 and Klinefelter's syndrome. *Am J Dis Child* 1967;**113**: 483–6.
6 Bach C, Toublanc JE, Gantier M. Une observation de double aneuploidie chromosomique trisomie 18 et XXY. *Ann Genet (Paris)* 1973;**16**:61–6.

Requests for reprints to Dr N B Kardon, Child Development Center/Genetics, North Shore University Hospital, 300 Community Drive, Manhasset, New York 11030, USA.

Proximal femoral focal deficiency associated with the Robin anomalad

SUMMARY A case of unilateral proximal femoral focal deficiency (PFFD) and the Robin anomalad is reported. Since bilateral PFFD and unusual facies have been reported before, we suggest an association between the Robin anomalad and PFFD.

The purpose of this report is to describe a patient with unilateral proximal femoral focal deficiency (PFFD) and coexisting Robin anomalad. Daentl and associates reported the occurrence of PFFD with unusual facies.[1] In each of their cases the femoral defects were bilateral. The unusual facies described by Daentl, including micrognathia and cleft palate, would seem to be representative of the Robin anomalad.[2 3]

Case report

A male term infant was born to a 22-year-old gravida 1 para 1 aborta 0 female after an uncomplicated gestation. The patient was delivered by caesarian section because of breech presentation. The Apgar scores at 1 and 5 minutes were 8 and 8,

Received for publication 7 November 1979

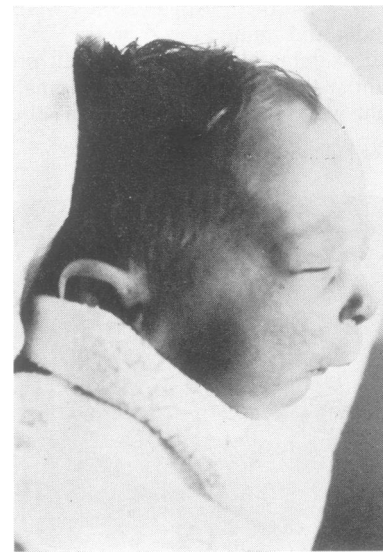

FIG 1 *Profile of proband showing the pronounced mandibular hypoplasia that is seen with the Robin anomalad.*