Ring chromosome 15 in a patient with features of Fryns’ syndrome

SUMMARY A stillborn male infant with a ring chromosome 15 and some features compatible with Fryns’ syndrome is presented. Neither diagnosis is common and the overlap may be of significance.

History
Prenatal. No drugs or infections in pregnancy. Mother had no antenatal care.

Birth. Normal vertex delivery at 38 weeks after sudden antepartum haemorrhage and cessation of fetal movements. Birth weight 2000 g (<3rd centile), head circumference 30 cm (<3rd centile), and length 44.3 cm (<3rd centile). The stillborn male infant was meconium stained and the cord was wound tightly around the neck. The amount of amniotic fluid was not recorded.

Family history. Normal, healthy, non-consanguineous ‘Cape Coloured’ parents; mother and father 37 and 38 years old, respectively. Normal older brother, but the younger brother had unilateral Perthes’ disease of the hip and haemophilia B. All were dissimilar in appearance to the patient.

Clinical features
He had brachycephaly, a large posterior fontanelle, coarse looking facies, low set, large ears with poorly shaped auricles, short palpebral fissures, short philtrum, large, carp shaped mouth, and micrognathia (fig 1). Widely spaced nipples gave the appearance of a broad chest. The limbs showed brachydactyly, abnormal palmar creases, and rockerbottom feet, but no finger nail hypoplasia. He had cryptorchidism, but a well developed scrotum and short penis (0.5 cm).

Investigations
Necropsy. Left diaphragmatic hernia with lung hypoplasia (weight of right lung=4.7 g, left=1.2 g) and small dysplastic kidneys.

Chromosome analysis. 46,XY,r(15)(p11→q26) (fig 2). Chromosome complements of mother and both brothers normal. Father unavailable for chromosome studies. The ring appeared stable in all nine cells analysed from blood lymphocytes. No other tissue was available.

Discussion
A specific phenotype, namely slight to moderate mental retardation, pre- and postnatal growth failure, triangular
Dysmorphology reports

kidneys, and dysmorphism are the result of the ring 15 and whether this patient has Fryns’ syndrome. The latter is a rare syndrome, not yet reported from this country. If the patient has Fryns’ syndrome, could this be the result of monosomy of the recessive gene on the morphologically normal chromosome?

The authors wish to thank the Cape Provincial Administration, the South African Medical Research Council, and the University of Stellenbosch for their support. The secretarial assistance of Mrs M Page is gratefully acknowledged. We would like to thank Dr M Nelson for reviewing the manuscript and for helpful suggestions.

G de Jong, R A Rossouw, and A E Retief
MRC Research Unit for Cytogenetics, Medical School, University of Stellenbosch, Tygerberg, Republic of South Africa.

References


Correspondence to Dr G de Jong, MRC Research Unit for Cytogenetics, Medical School, University of Stellenbosch, PO Box 63, Tygerberg, Republic of South Africa.

Unknown syndrome:
Noonan-like craniofacial features, digital anomalies, and premature birth

SUMMARY. We report a mother and two of her children, one female and the other male, who have ptosis, hypertelorism, epicantthic folds, downward slanting palpebral fissures, broad nasal bridge, and minor digital anomalies (fig 1); the children had delayed closure of a large anterior fontanelle. All three affected persons were born prematurely.

History

The normally intelligent mother was born six weeks prematurely when her father was aged 33 years, and was...