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?

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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
thought to resemble neither her parents nor her four healthy sibs who were born at term. As a teenager spontaneous pneumothorax was treated by pleurodesis. Her first pregnancy was uncomplicated until premature rupture of membranes and spontaneous onset of labour at 28 weeks' gestation. The female proband's birth weight was on the 50th centile. Respiratory distress syndrome was treated by artificial ventilation and patent ductus arteriosus closed by indomethacin. Osteitis of the femur at one month of age was treated with antibiotics. In her second pregnancy spontaneous onset of labour occurred at 31 weeks and the male infant's birth weight was on the 50th centile. He had mild respiratory distress. Her third pregnancy went to term when a normal male infant was born.

**Clinical examination**

**Proband:** at 12 weeks chronological age, downward slanting palpebral fissures, hypertelorism, large anterior fontanelle. At four years three months, height 50th centile, weight 25th centile, OFC 75th centile, prominent forehead, ptosis, epicanthic folds, hypertelorism (IPD=60 mm, >97th centile), hypermetropic astigmatism, strabismus, entropion, persistent hyaloid artery, unilateral hearing loss, broad nasal bridge, fifth finger clinodactyly, and dysplastic toenails. Joint laxity was present at the knee and finger joints. Psychomotor and speech delay present in the first three years resolved so that detailed psychological assessments at four and a half years placed her within normal ranges. Normal Giemsa banded female karyotype in blood lymphocytes.

**Affected male sib:** at three years, height 3rd centile, weight 3rd centile, OFC 3rd centile, prominent forehead, two strawberry naevi, unilateral ptosis, downward slanting palpebral fissures, epicanthic folds, hypertelorism (IPD=55 mm, >97th centile), broad nasal bridge, short nose, long philtrum (75th centile), fifth finger clinodactyly, left inguinal hernia (repaired), and dysplastic toenails. Joint laxity detected at the knee and finger joints. Developmental progress similar to that of his sister with diminishing evidence of psychomotor delay as he gets older.

**Proband's mother:** facial features in childhood (fig 2) similar to her affected children; she was also 'loose jointed' as a child but lost this trait in adulthood. Normal intelligence, height 25th centile, weight 25th centile, OFC...
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3rd centile, slight left ptosis, downward slanting palpebral fissures, slender fingers, clinodactyly and camptodactyly of the fifth digit with unilateral postaxial remnant (fig 3), dysplastic toenails.

Discussion

Ptosis, hypertelorism, downward slanting palpebral fissures, and digital anomalies may be present together in Aarskog’s syndrome and Noonan’s syndrome. However, other features of these conditions (cardiac defects, short stature, ear and genital anomalies) were absent in this family and thus it seems likely that this craniofacial phenotype is non-specific; for example, there are reports of autosomal dominant\(^1\) and autosomal recessive\(^2\) conditions mimicking Aarskog’s syndrome. The occurrence of premature birth and a degree of joint laxity in each affected subject is interesting and may indicate an underlying connective tissue disorder even though the skin was normal. It would seem worthwhile to investigate collagen biosynthesis in this family.

References


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