FIG 2 Twin 2 aged 17 months.

circumference of 58.5 cm and the absence of neurological findings.⁵

Discussion

With the present report there are now four instances of familial recurrence of congenital short femur. Ring² described a pair of concordant twins with congenital short femur in 1959, but no comment was made about their zygoty. Kelly³ noted that a third degree relative had a curved femur, and in his

comprehensive family study Hamanishi⁴ found only one familial recurrence in first cousins among 56 families studied. In this study,⁴ no raised parental ages were found and only one set of parents was consanguineous. Furthermore, three sets of discordant twins of uncertain zygoty have been reported.^{4, 6}

With the exception of thalidomide, which is usually associated with more severe femoral defects, no consistent environmental factors have been identified. Unifactorial inheritance is most unlikely and the observed patterns of familial recurrence and twin concordance favour multifactorial inheritance for simple femoral hypoplasia.

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Poland syndrome associated with 'morning glory' syndrome (coloboma of the optic disc)

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SUMMARY A 12 year old girl with the Poland syndrome and the 'morning glory' syndrome is described. The patient presented with absence

of the left pectoralis major muscle, hypoplasia of the left arm, symbrachydactyly, and ipsilateral coloboma of the optic disc. This is the first report of the association of these two congenital anomalies.

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FIG 1 Absence of the sternal portion of the left pectoralis major muscle with ipsilateral breast and nipple hypoplasia.

FIG 2 Symbrachydactyly of the left hand.

Poland syndrome consists of absence of the sternal portion of the pectoralis major muscle and symbrachydactyly.¹ 'Morning glory' syndrome² or coloboma of the optic disc is thought to result from disturbances in closure of the embryonal optic fissure. Congenital malformations of the optic disc are seldom specifically distinguishable without histological examination, but they are usually represented by specific clinical findings. We report a patient with a combination of these abnormalities involving the same side.

Case report

The patient was a 12 year old girl with signs of Poland syndrome on the left side and coloboma of the left optic disc. The family history was negative for both anomalies.

Aplasia of the sternal portion of the left pectoralis major muscle was associated with hypoplasia of the breast and nipple and reduced axillary hair (fig 1). The left arm was thinner and 2.5 cm shorter than the right. The middle phalanges of the fingers of the left hand were missing (symbrachydactyly) (fig 2).

The right eye was myopic with a visual acuity of 6/6 –10.0. The left eye was blind, divergent, and raised. The visual field and colour perception were normal in the right eye and intraocular pressures

FIG 3 Coloboma of the optic disc.

were normal. The appearance of the right fundus was normal but the left fundus showed features of the 'morning glory' syndrome (fig 3). Fluorescein angiography indicated normal duration of retinal blood vessel filling on both sides. ERG was normal on the right but subnormal for the left eye.

Neurological, neuroradiological, neuroendocrinological, and EEG examinations revealed no abnormalities. Antibody titre measurement showed no antibody production against CMV, rubella, HSV₁,

and HSV₂ viruses. The karyotype was 46,XX (in five cells).

Discussion

Poland syndrome is often associated with other congenital abnormalities: hypoplasia of the breast and nipple, reduced axillary hair, anomalies of the bony thorax, syndactyly, cleft hand deformities,³ preaxial polydactyly type 1, absence of extensor tendons of the hand and hypoplastic thenar muscles,⁴ dextrocardia,⁵ skeletal and genitourinary tract abnormalities.⁶ It has also been associated with Möbius⁷ and Pierre-Robin⁸ syndromes.

'Morning glory' syndrome is usually a unilateral congenital defect that occurs as a consequence of developmental disturbance of the optic disc in the course of the first six weeks of gestation and is accompanied by severely impaired or completely absent vision. This syndrome has been associated with basal encephalocele⁹⁻¹⁰ and Duane's retraction syndrome.¹¹

The aetiology of 'morning glory' syndrome and the nature of its association with Poland syndrome remain unclear.

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Interstitial deletion of chromosome 4q diagnosed prenatally

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SUMMARY The prenatal diagnosis of 4q deletion was made as a result of amniocentesis for high serum alphafetoprotein

Case report

A 27 year old primigravida (husband 28 years old), a non-smoker with no history of illness, x-ray ex-

posure, or medication in early pregnancy, was seen after 13 weeks' amenorrhoea. In the 15th week of gestation the serum alphafetoprotein was found to be high on two occasions and therefore amniocentesis was performed in the 17th week. The amniotic fluid AFP level was normal. However, the fetal karyotype showed a deletion of the long arm of chromosome 4. The parents chose to terminate the pregnancy.

The fetus (weighing 360 g) had a complete bilateral cleft lip and palate and there were deformities of the fourth finger on the right hand and the second toe on the left foot (fig 1a and b). Necropsy showed a preductal coarctation of the aorta and a double superior vena cava.

CYTOGENETIC STUDIES

G banding showed an interstitial deletion in all cells analysed. The karyotype was interpreted as: 46,XY,del(4)(pter→q21::q27→qter) (fig 2). This finding was confirmed in fetal skin and lung tissue obtained after termination. The karyotypes of both parents were normal.

TABLE Characteristics of 4q deletion syndrome.

	Previous cases
Mental retardation	10 (100%)*
Craniofacial anomalies	
Mid-facial asymmetry or hypoplasia	13 (92.8%)
Cleft lip	5 (35.7%)
Cleft palate	13 (92.8%)
Micrognathia	12 (85.7%)
Abnormal auricles	14 (100%)
Abnormalities of fingers and/or toes	13 (92.8%)
Cardiac anomalies	11 (78.6%)

*Cases of Townes *et al*⁶ (died at birth), Mitchell *et al*⁶ (one died at 1 hour, one died at 23 days), and Chudley *et al*¹¹ (reported at age of 1 day with no further information given) are not included.

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