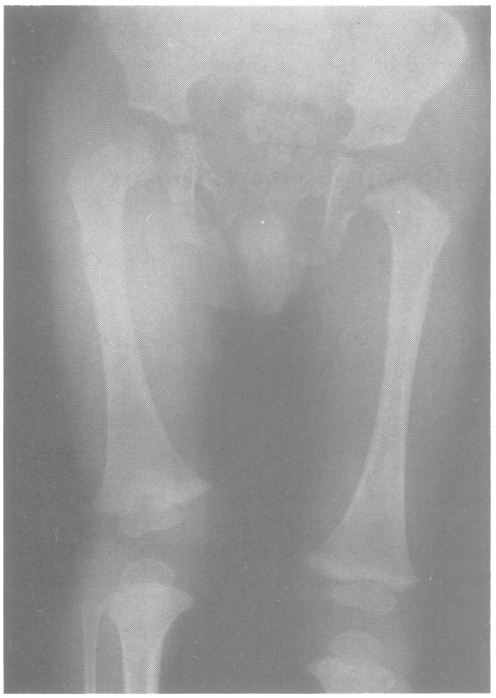

FIG 2 *Twin 2 aged 17 months.*

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. Ring[2] described a pair of concordant twins with congenital short femur in 1959, but no comment was made about their zygosity. Kelly[3] noted that a third degree relative had a curved femur, and in his comprehensive family study Hamanishi[4] 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 zygosity 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.

We wish to thank the Wellcome Trust and the National Fund for Research into Crippling Diseases for financial support.

References

1 Rogala EJ, Wynne-Davies R, Littlejohn A, Gormley J. Congenital limb anomalies: frequency and actiological factors. Data from the Edinburgh Register of the newborn (1964–1968). *J Med Genet* 1974;**11**:221–33.

2 Ring PA. Congenital short femur. Simple femoral hypoplasia. *J Bone Joint Surg (Br)* 1959;**41**:73–9.

3 Kelly TE. Proximal focal femoral deficiency (familial). *Birth Defects* 1974;**10**(12):508–9.

4 Hamanishi C. Congenital short femur. Clinical, genetic and epidemiological comparison of the naturally occurring condition with that caused by thalidomide. *J Bone Joint Surg (Br)* 1980;**62**:307–20.

5 Asch AJ, Myers GJ. Benign familial macrocephaly. Report of a family and review of the literature. *Pediatrics* 1976;**57**:535–40.

6 Westin GW, Gunderson FO. Proximal femoral focal deficiency: a review of treatment experiences. In: Aitken GT, ed. *Proximal femoral focal deficiency. A congenital anomaly.* Symposium held in Washington, June 13, 1968. Washington: National Academy of Sciences, 1969:1–22.

Correspondence and requests for reprints to Dr J M Connor, Duncan Guthrie Institute of Medical Genetics, Yorkhill, Glasgow G3 8SJ.

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

D T PIŠTELJIĆ*, D VRANJEŠEVIĆ*, S APOSTOLSKI†, AND D D PIŠTELJIĆ*

**Institute for Neurology and Psychiatry of Childhood and Adolescence, and †Clinic for Neurology, 11000 Belgrade, Yugoslavia.*

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.

Received for publication 2 March 1985.
Revised version accepted for publication 4 July 1985.

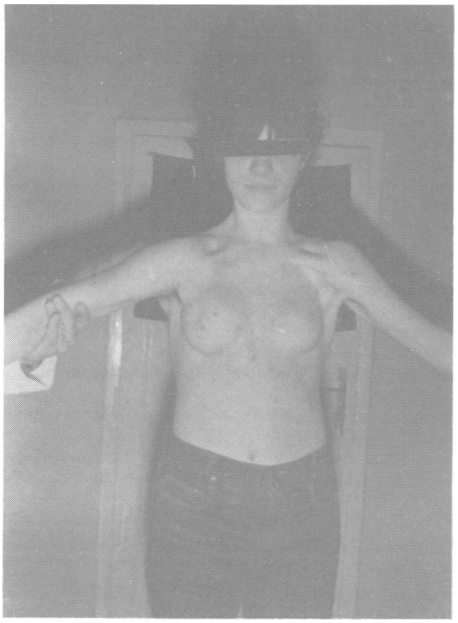

FIG 1 *Absence of the sternal portion of the left pectoralis major muscle with ipsilateral breast and nipple hypoplasia.*

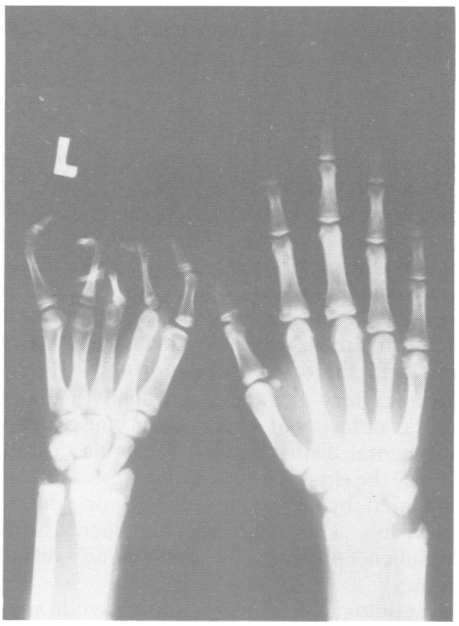

FIG 2 *Symbrachydactyly of the left hand.*

Poland syndrome consists of absence of the sternal portion of the pectoralis major muscle and symbrachydactyly.[1] 'Morning glory' syndrome[2] 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

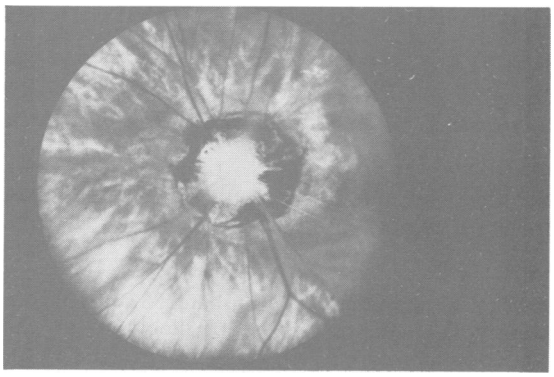

FIG 3 *Coloboma of the optic disc.*

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, HSV_1,

and HSV_2 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 Möbius[7] and Pierre-Robin[8] 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 encephalocele[9] [10] and Duane's retraction syndrome.[11]

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

References

1 Poland A. Deficiency of the pectoralis major muscles. *Guy's Hosp Rep* 1841;**6**:191–5.
2 Kindler F. Morning glory syndrome: unusual congenital optic disk anomaly. *Am J Ophthalmol* 1970;**69**:376–83.
3 Engberg WD. Cleft hand and pectoral aplasia. *J Hand Surg* 1981;**6**:574–7.
4 Senrui H, Egawa T, Horiki A. Anatomical findings in the hands of patients with Poland's syndrome. Report of four cases. *J Bone Joint Surg (Am)* 1982;**64**:1079–82.
5 Hanka S. Dextrocardia associated with Poland's syndrome. *J Pediatr* 1975;**86**:312.
6 Oppolzer A, Sacher M. Poland syndrom. *Klin Paedr* 1983;**195**:135–7.
7 Sugarman GI, Stark HH. Möbius syndrome with Poland's anomaly. *J Med Genet* 1973;**10**:192–6.
8 Wood VE, Sandin C. The hand in the Pierre Robin syndrome. *J Hand Surg* 1983;**8**:273–6.
9 Caprioli J, Lesser RL. Basal encephalocoele and morning glory syndrome. *Br J Ophthalmol* 1983;**67**:349–51.
10 Koenig SB, Naidich TP, Lissner G. The morning glory syndrome associated with sphenoidal encephalocoele. *Ophthalmology* 1982;**89**:1368–73.
11 Kawano K, Fujita S. Duane's retraction syndrome associated with morning glory syndrome. *J Pediatr Ophthalmol* 1981;**18**:51–4.

Correspondence and requests for reprints to Dr D. Pišteljić, Institute for Neurology and Psychiatry of Childhood and Adolescence, Dr Subotića 6a, 11000 Belgrade, Yugoslavia.

Interstitial deletion of chromosome 4q diagnosed prenatally

J M CAMPBELL*, J WILLIAMS†, AND G BATCUP‡

Departments of Obstetrics and Gynaecology and Pathology‡, University of Leeds; and †Department of Cytogenetics, St James's University Hospital, Leeds.*

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 exposure, 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.

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*[8] (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.

Received for publication 12 April 1985.
Revised version accepted for publication 16 September 1985.

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.