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 zygosity. 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 parenthood ages were found and only one set of parents was consanguineous. Furthermore, three sets of discordant twins of uncertain zygosity have been reported.5 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

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.
Poland syndrome consists of absence of the sternal portion of the pectoralis major muscle and symbrachydactyly. 'Morning glory' syndrome 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 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 HSV\textsubscript{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,\textsuperscript{3} preaxial polydactyly type I, absence of extensor tendons of the hand and hypoplastic thenar muscles,\textsuperscript{4} dextrocardia,\textsuperscript{5} skeletal and genitourinary tract abnormalities.\textsuperscript{6} It has also been associated with Möbius\textsuperscript{7} and Pierre-Robin\textsuperscript{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 encephalocoele\textsuperscript{9} \textsuperscript{10} and Duane's retraction syndrome.\textsuperscript{11}

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

**INTERSTITIAL DELETION OF CHROMOSOME 4q DIAGNOSED PRENATALLY**

**J M CAMPBELL\textsuperscript{*}, J WILLIAMS\textsuperscript{†}, AND G BATCUP\textsuperscript{‡}

*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 examination was carried out at 20 weeks gestation.

**TABLE Characteristics of 4q deletion syndrome.**

<table>
<thead>
<tr>
<th>特征</th>
<th>前期情况</th>
</tr>
</thead>
<tbody>
<tr>
<td>精神发育迟缓</td>
<td>10 (100%)*</td>
</tr>
</tbody>
</table>
|颅面异常 | 13 (92.8%)
|面中线不对称或发育不良 | 5 (35.7%)
|唇裂 | 13 (92.8%)
|鼻裂 | 12 (85.7%)
|先天性小颌 | 14 (100%)
|异常耳孔 | 13 (92.8%)
|手指/脚趾异常 | 11 (78.6%)

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

REFERENCES


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

**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 tissues obtained after termination. The karyotypes of both parents were normal.