Poland 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.

References


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

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.

FIG 3 Coloboma of the optic disc.

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, HSV,
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, preaxial polydactyly type I, absence of extensor tendons of the hand and hypoplastic tenar muscles, dextrocardia, skeletal and genitourinary tract abnormalities. It has also been associated with Möbius and Pierre Robin 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 and Duane's retraction syndrome.

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

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-

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<th>TABLE Characteristics of 4q deletion syndrome.</th>
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<tr>
<td>Mental retardation</td>
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<tr>
<td>Craniofacial anomalies</td>
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<td>Mid-facial asymmetry or hypoplasia</td>
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<td>Cleft lip</td>
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<td>Cleft palate</td>
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<tr>
<td>Micrognathia</td>
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<td>Abnormal auricles</td>
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<td>Abnormalities of fingers and/or toes</td>
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<tr>
<td>Cardiac anomalies</td>
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<td>Previous cases</td>
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<td>10 (100%)*</td>
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<td>13 (92.8%)</td>
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<td>5 (35.7%)</td>
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<td>13 (92.8%)</td>
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<td>12 (85.7%)</td>
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<td>14 (100%)</td>
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<tr>
<td>13 (92.8%)</td>
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<tr>
<td>11 (78.6%)</td>
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</tbody>
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*Cases of Townes et al* (died at birth), Mitchell et al* (one died at 1 hour, one died at 23 days), and Chudley et al† (reported at age of 1 day with no further information given) are not included.

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References


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