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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FIG 1 The fetus showing cleft lip and palate and digital anomalies.

FIG 2 Interstitial deletion of chromosome 4.

Discussion

Deletion of a segment of the long arm of chromosome 4 is relatively rare. There have been 14 reports describing deletion of the terminal segment of chromosome 4 (4q31→qter).¹⁻¹¹ Mitchell *et al*⁸ also described two cases with similar deletions to the present case, del(4)(q21·3→q26) and del(4)(q21·1→q25) respectively, but these cases did not have cleft palate which features prominently in the specific and recognisable phenotype, the '4q deletion syndrome'.⁶

The characteristic features of this syndrome are outlined in the table. The Robin malformation combination of cleft palate, posteriorly displaced tongue, and micrognathia appears as a highly consistent manifestation. The association with cleft lip seems to be less marked than that with palatal

cleft. Some form of mid-facial asymmetry or hypoplasia was also present in all but one of the cases reviewed. Abnormalities of the auricles have been variously described as 'elfin like', 'low set', and 'satyr'. Cardiac anomalies are common and are often the cause of death. Frequently reported anomalies of the hands and feet are displaced toes, fifth finger clinodactyly, and abnormal palmar creases. The present case showed most of the salient features, with a complete bilateral cleft lip and palate, a preductal coarctation of the aorta, double vena cava, and digital flexion deformities.

Survival is inversely proportional to the size of genetic deletion. As with deletion of chromosome 4(q31→qter), deletion of 4(q21→q27) accounts for 1·2% of the haploid genome and so about half the subjects with this would not survive. Of the 14 cases of deletion 4(q31→qter) reported, only four survived beyond 2 years of age, with physical and mental retardation a universal finding.

There does not appear to be a parental age effect. Only one recorded case was born to relatively old parents, the mother being 38 and the father 49 years of age. Apart from one case reported which resulted from a paternal 4;20 translocation,⁶ the aberration seemed to arise de novo.

It is interesting to note that this chromosomal abnormality was detected as a result of amniocentesis, prompted by a high serum AFP level. This is a recognised association in trisomy 18, Turner's syndrome, and other chromosomal anomalies, and we have experience of four other cases associated with

high AFP levels, one trisomy 18, one Klinefelter's syndrome, and two Down's syndrome, although this conflicts with recent publications on Down's syndrome.¹²

This brings into question whether all analyses of amniotic fluid for raised serum AFP should also have chromosomal analysis done. It is the practice in this region, because of the risk involved in amniocentesis, to perform both.

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