where the vertebral defects do not fall into either of the preceding categories.

Whereas type I patients are obviously abnormal because of their appearance—that is a short neck, the limitation of rotation, and a low hairline—type III patients are not necessarily recognizable for their anomalies. Because of their normal appearance, type II patients are usually not recognized until the complications caused by their malformation are investigated or their anomaly is discovered incidentally (Gunderson *et al*, 1967; Poznanski, 1974).

Our patient represented a problem in classification, for her abnormal appearance was like that of a type I patient, but her cervical anomalies were like those in type II with variable fusion. Because she did not have blocked vertebrae but because of her anomalies in at least three sites—C1, C2–3, and C3–4—we considered her defects most likely to be classed as type II with variable fusion.

Hearing deficiency. Deafness has been said to be the second most common anomaly associated with the syndrome (Palant and Carter, 1972; Stark and Borton, 1973; McLay and Maran, 1969). There is histological and *x*-ray evidence that the structure of the inner ear is abnormal (Palant and Carter, 1972). Because there was no other deafness in the family and also because our patient seemed less severely affected than often occurs in the hereditary forms of deafness (Fraser, 1964), we concluded that her hearing deficiency was most probably associated with the syndrome and not independently determined.

Genetic determination and mode of inheritance. The patient appeared in the pedigree as a sporadic occurrence of her congenital malformations. Because there was no suspicion of any environmental agent, we concluded that genetic determination was probable. After rejection of the possibility of a phenocopy—an environmentally caused mimic of a genetically determined disorder—there remained three considerations: spontaneous mutation, reduced penetrance, and recessive homozygosity. The normality of the cervical vertebrae of the parents plus the phenotypic normality of the parents and the grandparents argued against the likelihood of reduced penetrance. Parental consanguinity favoured an interpretation of determination by a single autosomal recessive gene. The probability of spontaneous mutation can be further considered when there are more progeny.

The implication of the consanguinity to an understanding of the aetiology of this instance of the cervical vertebral fusion syndrome is the same as that when there are consanguineous parents of any offspring who has a rare disorder, namely, that the disorder is genetically determined, probably by a recessive gene, which most likely came to be homozygous in the proposita as a result of descent from an ancestor common to both parents. Of course, this does not mean that one or both genes could not have risen independently.

Gunderson *et al* (1967) suggested that type II with variable cervical fusion is caused by a single dominant gene with considerable variation in both penetrance and expression. If our case fits into type II with variable cervical fusion and if our conclusion of a single recessive gene is correct, then there is evidence of genetic heterogeneity for this clinical class, there apparently being both a dominant and a recessive gene producing the phenotype.

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Pterygium syndrome

Summary. The pterygium syndrome consists of webbing of the neck, the antecubital fossae and the popliteal regions together with flexion deformities of the limb joints and anomalies of the vertebrae. A family, three offspring of which appear to be affected with the same disorder, is presented. All three are female; there is also a normal female child of the same union.

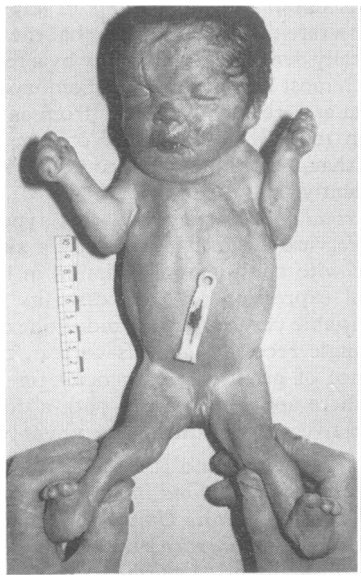

FIG. 1. Case 2: abnormal facies, small chest, and flexion deformities of upper and lower limbs.

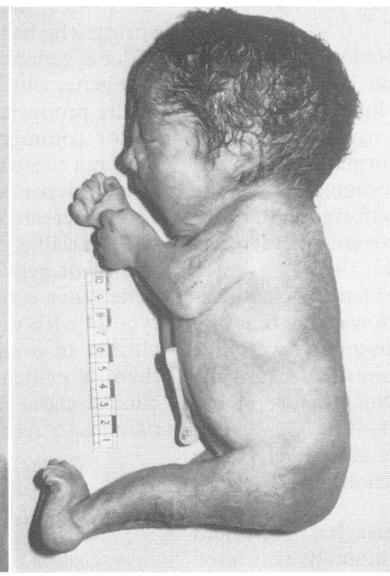

FIG. 2. Case 2: lateral view showing pterygia and flexion deformities.

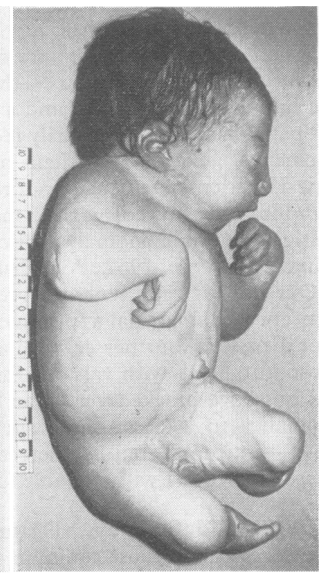

FIG. 3. Case 3: abnormal facies, flexion deformities, and kyphoscoliosis.

This syndrome was described in 1969. Scott described one patient and Norum, James, and Rabry reported another 3 patients who were cousins. Pterygium is Greek for a 'wing' and describes a major feature of these cases. Another three cases are presented below.

Case reports

Mother is English, aged 30 years, married in 1965 and in good health. There was no history of any congenital abnormality in her own or her husband's family, nor any history of consanguinity.

In 1969 she was delivered of a normal female infant at 38 weeks' gestation; this child is alive and well.

Case 1. In 1972 an intrauterine death occurred at 32 weeks' gestation, with delivery at 35 weeks of a macerated stillborn female, weighing 960 g.

The main findings were webbing of the neck and axillae, flexion deformities of the wrists with claw-like hands, and dorsiflexion of the ankles with prominent heels. The chest was small with hypoplastic lungs.

Case 2 (Fig. 1 and 2). In 1973 an abnormal female infant, weighing 1560 g, was delivered at 33 weeks' gestation. Death occurred at the age of 1 hour. The head circumference was 32.5 cm, which was large relative to body weight and crown heel length of 37.5 cm, using the standards of Usher and McLean (1969). The face showed epicanthic folds, hypertelorism, flattened nose, haemangioma of mid-forehead, and micrognathia. The neck was short, with a pterygium stretching from chin to sternum. The chest appeared small with a prominent sternum. Webbing of the neck, axillae, elbows, and hips was present. There were also obvious flexion deformities of the hips and elbows, dorsiflexion of the ankles with prominent heels ('rocker bottom'), and clawing of the fingers and toes.

Case 3 (Fig. 3). During her fourth pregnancy in 1974 an amniocentesis was performed at 15 weeks. Cell culture showed a normal female karyotype and alpha-feto protein estimations on maternal serum (42 ng/ml) and on liquor (26.5 μg/ml) were considered normal. Both parents were found to have normal karyotypes. Ultrasound cephalometry suggested normal fetal growth up to 36 weeks' gestation. Labour was induced at 39 weeks and an abnormal female infant was delivered who died at 7 hours of age. Birthweight was 2.375 kg (less than 2 standard deviations (SD) below the mean for 39 weeks' gestation), crown heel length was 47.0 cm (at 2 SD below mean), and head circumference was 33 cm (at 2 SD below mean) (Usher and McLean, 1969). The facial appearance resembled those of Cases 1 and 2 with epicanthic folds, hypertelorism, flattened nose, haemangioma of mid-forehead, micrognathia, and a short neck with skin stretching from the chin to the sternum. There was also webbing of the neck, axillae, and hips. The chest was small, with kyphoscoliosis and hypoplastic lungs. Flexion deformities involved the elbows, hips, and ankles (left 'rocker bottom' foot

and right equinovarus deformity), with clawing of the fingers and toes.

Discussion

Our three cases and the four previously described cases appear to have several major findings in common.

(1) Pterygia of neck, antecubital, hip, and popliteal regions were present in all cases.

(2) Flexion deformities involved the elbows, hips, ankles, hands, and feet to a varying degree in all cases.

(3) There were abnormalities of chest development in 6 of the 7 cases.

(4) A capillary haemangioma of mid-forehead was noted in 4 cases.

Of those described, our cases appear to be the most severely affected. One of the patients described by Norum *et al* (1969) died of pneumonia at 12 months. At the time of reporting, the other cases were aged from 3 to 17 years.

In previously reported cases there was no attempt to define the underlying abnormality. We examined sections of the skin, muscle, spinal cord, and brain from the postmortem tissues of Cases 2 and 3 in an attempt to find any neuromuscular or connective tissue abnormality. Anterior and posterior thigh muscles, blocked transversely and longitudinally, were stained. No abnormality of skeletal muscle fibres in size or appearance was seen. There was no cellular infiltrate present and the nerves in these sections appeared normal. Anterior horn cells from cervical, thoracic, and lumbar spinal cord were normal in number and appearance, showing no degeneration.

Skin from the abdominal wall was examined and no abnormality of connective tissue or elastic tissue was found. Unfortunately skin from the pterygia was not examined. We assume the inheritance to be autosomal recessive in type.

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