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THE PIERRE ROBIN SYNDROME ASSOCIATED WITH FEMORAL DYSGENESIS

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In recent years there have been several reports of children born with clefts of the secondary palate in association with multiple skeletal abnormalities, notably aplasia or hypoplasia of the femur. Three further cases of this syndrome form the basis of this paper.

CASE REPORTS

Case 1. This boy was the first of 3 children, the other two being normal. One of his grandparents was a diabetic. He was found to have multiple abnormalities. Extreme shortness of both legs (Fig. 1), was shown on X-ray examination to be due to total absence of both femora apart from the lower epiphyses and a short fragment of the shaft on the right (Fig. 2). This would be classified according to Aitken (1969) as a Type V proximal focal deficiency. There was aplasia of the hip joints, the fibulae were short and there were bilateral club feet. Only 3 lumbar vertebrae were present with a spina bifida of L3 and the sacrum (Fig. 3). He also had hypoplasia of the lateral condyles of the humerus with antero-lateral dislocation of the head of the radius and abnormalities of both shoulder joints. There was an incomplete cleft of the secondary palate, tongue-tie and a small mandible which caused some difficulty with feeding in the first few weeks of life.

Both testes were undescended and he was noted to have slight webbing of the neck and elbows.

Treatment has included repair of the cleft palate and release of the tongue-tie, at the age of 14 months. The club feet corrected well with splintage and although he stood at 14 months, walking was slow due to the unstable hip joints. Helped by special footwear and calipers he walked independently at the age of two.

When last seen at the age of eight, his speech was normal and he was walking remarkably well with special prostheses.

Case 2. This boy, who has 4 normal siblings and no family history of diabetes, was noticed to be short at birth with hypoplastic femora. Overlying the femoral head on the right side was an obvious skin dimple with two further dimples in the corresponding position on the left side (Fig. 4). X-rays taken on the 5th day confirmed the presence of bilateral short femora of Type 1 with a fracture of the left femur which was thought to be an intra-uterine lesion because of the presence of callus (Fig. 5). He too had minor abnormalities of his hips and shoulders and arthrogryphosis multiplex congenita.

A cleft of the secondary palate in association with marked retrognathia and glossoptosis required nursing on the special Burston frame (Burston, 1978). The cleft palate was repaired at the age of 17 months, together with release of a tongue-tie. Other abnormalities included undescended testes and a systolic murmur at the left sternal edge. An accurate diagnosis of the cardiac abnormality was not made but he developed signs of congestive cardiac failure for which he was digitalised and given a diuretic. By the age of 3 months this treatment was discontinued. There has been no recurrence of any symptoms of heart failure and no difficulties with subsequent operations or anaesthetics.

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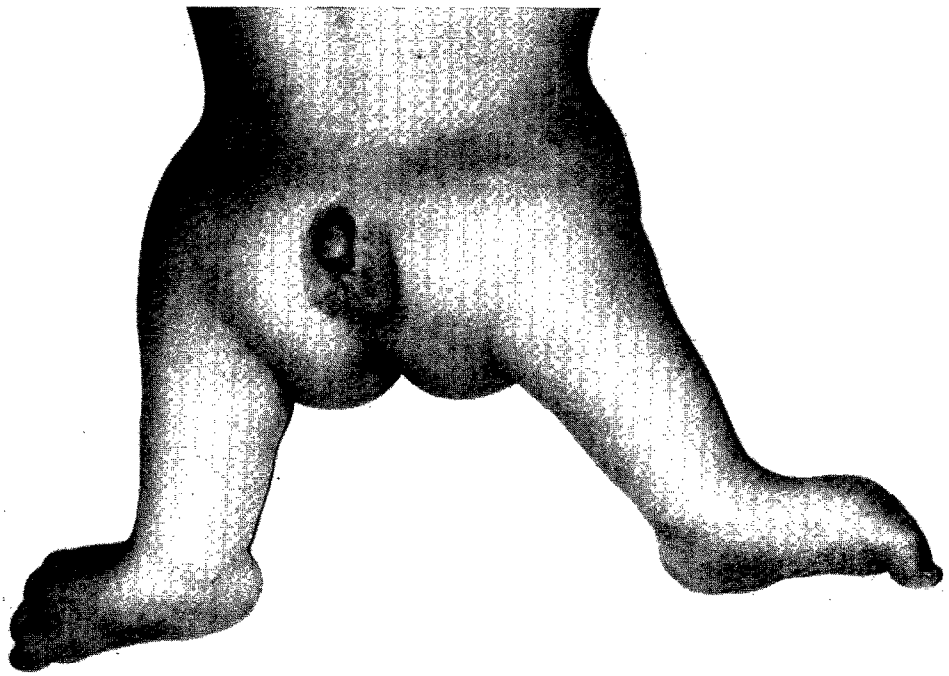

FIG. 1. Case 1. Showing the extreme shortness of both lower limbs.

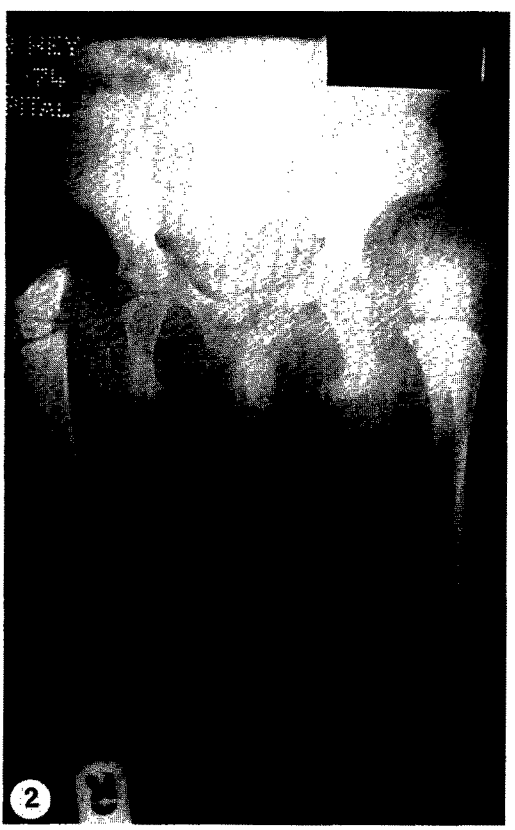

FIG. 2. Case 1. X-ray to show absent femora apart from a short fragment of the shaft and lower epiphysis on the right side.

FIG. 3. Case 1. X-ray to show the presence of only three lumbar vertebrae and spina bifida affecting L3 and the sacrum.

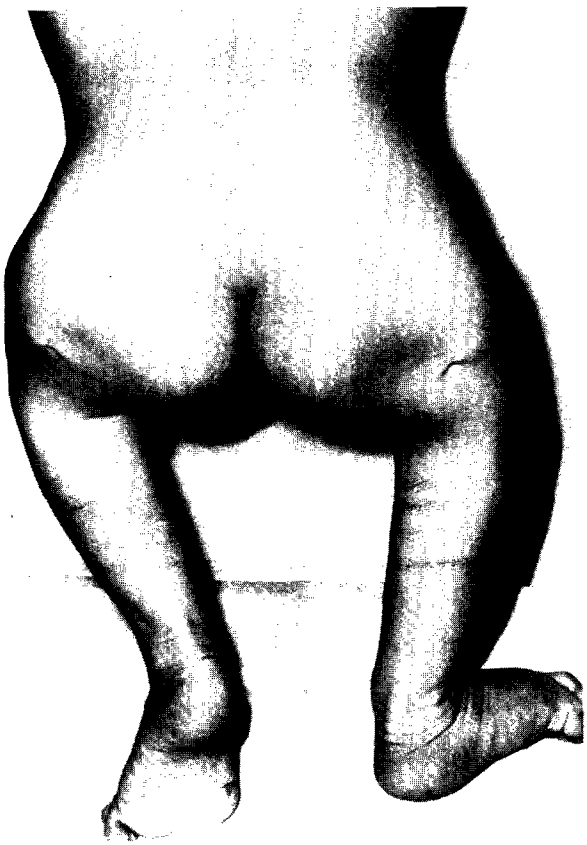

FIG. 4. Case 2. Skin dimples overlying the femoral head on each side. Both lower limbs are short.

FIG. 5. Case 2. X-ray on the 5th day of life showing a fracture of the left femur, possibly sustained in utero.

FIG. 6. Case 3. Congenital short right femur.

Case 3. This boy was the fourth child with three normal brothers. There was no history of diabetes in the family. He had a short right femur (Type 1) but no other significant skeletal abnormalities (Fig. 6). Walking was not delayed and at the age of $6\frac{1}{2}$ is normal apart from a slight drag of the right foot when running. A grid film taken at the age of 6, shows 2 cm of shortening of the femur.

A typical severe Pierre Robin Syndrome, that is a cleft of the secondary palate with marked retrognathia and glossoptosis, required treatment on the special frame for 3 months to deal with the airway obstruction. At the age of 11 months the cleft palate was repaired but because of poor speech a pharyngoplasty was carried out when he was $5\frac{1}{2}$ years old.

Bilateral inguinal herniae were repaired when he was 5 months old, at which time he was noted to have undescended testes.

Discussion

Daentl *et al.* (1975) gave the name "femoral hypoplasia—unusual facies syndrome" to a complex group of congenital abnormalities that included short or absent femora, a typical facies, cleft palate and a number of other anomalies which are listed in their paper. They reported 4 cases and could only trace 2 others in the literature. However, we have been able to find a large number of papers describing a bewildering collection of abnormalities associated with femoral aplasia or hypoplasia and a cleft palate (Frantz and O'Rahilly, 1961; Kučera *et al.*, 1965; Passarge, 1965; Bailey and Beighton 1970; Gorlin *et al.*, 1976). It is interesting that in series of 38 cases of short femora (Ring, 1961) no mention is made of cleft palate among the associated defects noted which were all of a skeletal nature. It has been thought that the presence of maternal diabetes could be a possible contributory factor. Indeed Bailey and Beighton (1970) drew a distinction between two types of femoral dysgenesis depending on the presence or absence of such a diabetic history. It may therefore be worth recording that our first patient had a distant family history of diabetes.

What is of particular interest to us is the fact that, almost without exception, in those children with a short femur associated with a cleft palate, the cleft has only involved the secondary palate. Furthermore in a strikingly high proportion of cases a degree of retrognathia often with glossoptosis has been reported. This suggests to us that there is a specific clinical entity in which the significant features are short femora and the Pierre Robin Syndrome. Walden *et al.* (1971) described such a case believing it to be the first in the literature. Clearly this is not so although it does substantiate our views.

We cannot accept the concept of Daentl and his colleagues that there exists a "Femoral hypoplasia—unusual facies syndrome". The unusual facies which they describe is in our view typical of the Pierre Robin Syndrome of which, with one exception, their cases were examples.

Summary

Three cases are described of the Pierre Robin Syndrome in association with femoral dysgenesis. This would appear to be a definite clinical entity.

Addendum

Since this paper was submitted for publication, a fourth child, a girl, has been born in Liverpool with an underdeveloped lower jaw, a cleft of the soft palate and bilateral short humeri and femora. Both parents were of normal build, but on the mother's side there is a family history of short stature apparently due to shortness of length of the tibiae. Further investigations are being made.

Our thanks are due to all those who have been involved in the treatment of these children's complex deformities at Alder Hey Children's Hospital, Liverpool and the Royal Liverpool Children's Hospital, Heswall. These include Dr W. R. Burston and Mr P. G. Bush, Consultant Orthodontists who supervised their care on the special nursing frame and Mr J. Monk, Mr R. Owen and Mr G. Shatwell, who were responsible for their orthopaedic care.

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