quent pseudocyst formation at 29 days and 10 weeks of age. The obstetric history was normal in 18 (72%) of the infants with cyst formation in this study. It is unlikely that any noxious cellular agent caused destruction of subependymal cell population in utero in all of these. An hypothetical explanation is that concealed subependymal haemorrhage caused by intrauterine hypoxia or another unknown factor may occur several weeks before delivery and result in cyst formation.

Neurological and physical examination of these neonates were within normal limits during the neonatal period as well as during their monthly follow up. Although the cysts were reabsorbed in all cases followed, it takes time to predict the integrity of the infant's high cortical function and the normality of psychomotor development. In summary, subependymal cyst in normal neonates may suggest intrauterine subependymal haemorrhage. Its aetiology needs further investigation. Various races and communities with different qualities of prenatal care may have a different incidence of cyst formation. We look forward to further reports for comparison.

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Marfan’s syndrome presenting as an intrapartum death

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SUMMARY A rare case of Marfan’s syndrome presenting as an intrapartum death is described. Recognisable mitral valve lesions were present.

Marfan’s syndrome is a generalised connective tissue disorder, and cardiovascular symptoms result from a loss in tensile strength of the supporting tissue of the aorta and cardiac valves. With more sophisticated diagnostic procedures it is realised that cardiovascular defects occur in virtually all patients with this syndrome, but they do not usually become manifest before the second decade. They are rare in childhood but have been described in infants, and very rarely, recognisable defects are present at birth.

Case presentation

This baby was the second child of a 25 year old mother. The antenatal period was uneventful apart from the lie, which was a breech. The mother’s pelvis was adequate. She was admitted, in labour, at 39 weeks and under a pudendal block an assisted breech delivery was performed. Low forceps were used and the head was delivered with no undue delay or force.

The fetal heart had been heard during labour but on delivery the apex beat was not felt, although the baby gasped once. Intensive resuscitation was unsuccessful.

The infant was a girl weighing 2800 g. Her length crown to rump was 34 cm, and crown to heel 50 cm. Her head circumference was 35 cm, and foot length 8.5 cm. The feet were long and narrow and the hands were long with tapering fingers (Fig. 1). The palate was not high arched, there were no flexion deformities of the limbs, and no signs of injury to the head or scalp were seen.

A small pericardial effusion was present. The heart was enlarged due to right and left ventricular dilatation. The great vessels were macroscopically normal but the tricuspid and mitral valve rings were dilated, the cusps of both valves were voluminous and pale (Fig. 2).

The pulmonary and aortic valves were normal. There was free blood over the surface of the brain,
Marfan's syndrome presenting as an intrapartum death

Discussion

The basic defect in Marfan's syndrome is unknown. Some patients, but not all, have increased hydroxyproline in the urine, an indication that increased collagen turnover and abnormalities in the cross linkage of collagen have been found. Other tissue components, however, may be involved. Histologically, in the media of the aorta, elastic fibres show fragmentation and deflection around pools of mucopolysaccharide. This weakening of the wall leads to aneurysmal dilatation and aortic dissection. Other cardiovascular abnormalities include dilatation of the mitral and aortic valve rings and 'floppy' valves in which large, lax leaflets lead to prolapse or insufficiency.

In a review of 36 infants and children with Marfan's syndrome, 17 were found to have mitral regurgitation, one aortic valve regurgitation, and three a combination of the two. Four died in childhood from cardiovascular disease. Necropsy, performed on one who had had severe mitral insufficiency, showed thickened, redundant cusps in the mitral and tricuspid valves.

Twin girls with skeletal features of Marfan's syndrome were noted to have heart murmurs at 3 months of age, and severe mitral regurgitation by 5 months. One died at 8 months and the other at 2 years. Necropsy examination in both showed large redundant mitral valve cusps and medial degeneration of the aorta.

A previous case of congenital Marfan's syndrome was found in the English published reports, in which the baby died at birth with similar changes in the aortic and mitral valves. In addition, there was haemorrhage into the media of an umbilical vessel, and evidence of an intrauterine infection, possibly the result of prolonged rupture of the membranes. It was postulated that both these features were an expression of the connective tissue disorder. In the patient we have described, death resulted from a tear in the falx. As there had been no problem over the delivery of the head, it seems likely that this tear was a manifestation of the basic collagen defect of Marfan's syndrome, and these infants may be at an increased risk from shearing injuries to the supporting tissues of the brain.

No family history of Marfan's syndrome was obtained and the condition presumably arose as a spontaneous mutation, which occurs not infrequently. The paternal age, however, was not high as is often found in dominant mutations.

We thank Mr C Gale for his permission to publish this case.

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Neonatal inferior vena cava and renal venous thrombosis treated by thrombectomy and nephrectomy

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SUMMARY Neonatal inferior vena cava and renal venous thrombosis with obstruction was diagnosed clinically and confirmed by ultrasonography. Successful thrombectomy and nephrectomy were performed at 40 hours of age. Thrombus with obstruction occluding the inferior vena cava favours immediate surgery.

Renal venous thrombosis occurring in the neonate causes haematuria, oliguria, acute renal failure, and hypertension. Diagnosis is usually made on clinical grounds and supported by intravenous urography. Recent advances in ultrasonography have improved the ease of diagnosis, in most cases giving precise information on the location and extent of the thrombus. This directly influences management as shown by the case below.

Case report

Clinical details. This boy was born to a mother who had previously had two successful pregnancies producing live infants and one miscarriage at 10 weeks' gestation. After a normal pregnancy, spontaneous labour occurred at 39 weeks' gestation. During the first and second stages, meconium stained liquor was present but no other evidence of fetal distress was noted. The infant was born by vaginal delivery and his Apgar scores were 6 at one minute, 8 at five minutes, and 9 at 10 minutes. His birthweight was 3.55 kg, and there was a true knot in the umbilical cord.

At 22 hours of age he was noted to have macroscopic haematuria, and by 30 hours, oedematous lower limbs, grunting respirations, a fine generalised purpuric rash, and bilaterally palpable kidneys. The systolic blood pressure was 150 to 140 mm Hg. Investigations showed a haemoglobin concentration of 15 g/dl, the total white cell count was 27.2 x 10^9/l, platelets 25.0 x 10^9/l, prothrombin time 17 seconds (control 13), partial thromboplastin time 62 seconds (control 35), thrombin time 25 seconds (control 10), and fibrin degradation products were greater than 10 but less than 40. The plasma sodium concentration was 134 mmol/l, potassium 4.5 mmol/l, bicarbonate 15 mmol/l, urea 6-6 mmol/l, and creatinine 230 muol/l. Urinary electrolytes values were sodium 109 mmol/l and urea 22 mmol/l.

After transfer to our unit at 37 hours of age, an abdominal ultrasound scan showed two large echogenic kidneys with no evidence of obstruction and an empty bladder. A thrombus 3 cm in length was seen in the inferior vena cava totally occluding the lumen of the vessel at the level of the renal veins but the latter were not visualised (Figure).

Management. The metabolic acidosis was corrected by intravenous administration of 4 mmol of sodium bicarbonate. A laparotomy was performed at 40 hours of age. Operative findings were a large black right kidney and a large dusky left kidney. Both renal veins and the vena cava from below the renal vein orifice to the hepatic vein orifice were occluded by thrombus. A longitudinal venotomy allowed complete removal of the organising clot from the vena cava, and thrombectomy of the left renal vein was successful, with improvement in kidney size and colour. A similar procedure on the right renal vein failed to produce any improvement in the kidney's appearance, so a right nephrectomy was performed. A Tenckhoff peritoneal dialysis catheter was left in situ.

After operation a bolus of 150 U of heparin followed by an infusion of 10 U/kg per hr intravenously was given for 22 days. A 99m-Tc