Case report

The female infant was the child of non-consanguineous Caucasian parents. The mother had had one previous early miscarriage and there was one normal female sib. Delivery was by caesarian section at 38½ weeks because of polyhydramnios and a large head. At birth the baby weighed 3.5 kg, length 44 cm (<3rd centile), and head circumference 38 cm (>98 centile). The limbs were markedly short with an arm span of 27 cm and a pubis to heel length of 13 cm. There was midline cleft upper lip and cleft palate with a very narrow thorax (fig 1). The digits were short, but there was no polydactyly. Radiographs showed very short, horizontal ribs, highly placed clavicles, small scapulae, small ilia, short tubular bones with bowing of the radius and ulna, and minimal metaphyseal irregularity and spiking (fig 2). Chromosomal analysis showed a normal female karyotype. Unfortunately cartilage histology was not examined and necropsy was not carried out.

Discussion

Although a diagnosis of one of the short rib-polydactyly syndromes manifesting without polydactyly must be considered here, this case is very similar to two unrelated cases, one born to a consanguineous couple, described by Beemer *et al.*¹ These authors did consider the possibility of their cases being examples of a type of short rib-polydactyly, particularly type II (Majewski); however the radiological features were not consistent with this condition. In short rib-polydactyly syndromes I (Saldino-Noonan) and III (Verma-Naumoff), metaphyseal irregularity and spiking is a marked feature.² It was not present in the case described here, apart from possible mild irregularity of the distal end of the femora. Short rib-polydactyly syndrome type II (Majewski) characteristically manifests with hypoplastic, oval shaped

TABLE Common features in three reported cases.

	Present case	Beemer <i>et al.</i> ¹	
		Case 1	Case 2
Macrocephaly	+	+	+
Flat face	+	+	+
Midline cleft lip	+	+	+
Cleft palate	+	+	—
Ascites	—	+	+
Omphalocele	—	+	+
Narrow thorax	+	+	+
Short ribs	+	+	+
Highly placed clavicles	+	+	+
Small scapulae	+	+	+
Small ilia	+	+	+
Short tubular bones	+	+	+
Bowing radius/ulna	+	+	—
Metaphyseal irregularity/spiking	+/-	—	—
Family history	Isolated case	Parents consanguineous, affected sib diagnosed prenatally by ultrasound	Isolated case

tibiae,³ quite unlike the well formed tibiae seen in the present case. Thus, it seems possible that a separate short rib syndrome is being described. The common features are shown in the table. Inheritance is assumed to be autosomal recessive, because of the sib pair reported by Beemer *et al.*¹

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Intrauterine death in megacystis-microcolon-intestinal hypoperistalsis syndrome

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SUMMARY Megacystis-microcolon-intestinal hypoperistalsis syndrome is an uncommon condition, possibly inherited as an autosomal recessive trait. This report describes an affected sib pair with intrauterine death of one of the sibs.

Winter and Knowles¹ recently described the occurrence of megacystis-microcolon-intestinal hypoperistalsis syndrome in two female sibs who were the offspring of first cousin parents, suggesting the possibility that this disorder has an autosomal recessive aetiology. Few cases of this condition, characterised by hypoperistalsis of the bowel and dysfunctional bladder contractility, have been reported and only three other possibly affected sib pairs have been noted.²⁻⁴ This report describes

Case reports

another affected sib pair, with intrauterine death of one of the sibs, a finding not previously noted.

Case reports

The parents were a young, unrelated white couple with no history of bladder or bowel dysfunction. They have had two pregnancies.

CASE 1

This female infant was born at term by caesarean section for failure to progress and weighed 4040 g. Apgar scores were 8 at one minute and 9 at five minutes. Abdominal distension was noted shortly after delivery. Ultrasonography showed bilateral hydronephrosis, bilateral hydroureter, and a grossly distended urinary bladder. Initial renal function was moderately impaired but returned to normal after catheterisation of the bladder. Permanent bladder drainage was required and a draining vesicostomy was performed. A barium enema revealed a non-rotated microcolon, and at laparotomy at two months to fix the position of the colon, the mesentery was short. She could not be fed due to the absence of peristalsis and a gastrostomy for gastric drainage was needed. Based on these features, the diagnosis of megacystis-microcolon-intestinal hypoperistalsis syndrome was made. Total parenteral nutrition was initiated shortly after birth, but at 10 months of age complications were noted, including portal fibrosis. Progressive liver damage became evident. She died of sepsis at 14 months of age. Although a formal developmental assessment was not conducted, her major motor milestones were noted to be delayed before her death. At 10½ months she was able to sit independently but did not roll over.

Histological examination of the colon biopsied at laparotomy at two months of age showed a marked thinning of the external longitudinal smooth muscle layer and the presence of normal appearing ganglion cells.

CASE 2

In the second pregnancy, detailed fetal ultrasonography revealed distension of the urinary bladder and some dilatation of the renal pelvises in the 19th week. At 30 weeks of gestation, the fetal heartbeat was no longer audible and at ultrasonography no fetal heart action could be seen. A large cystic structure filled the abdomen and was thought to be the fetal urinary bladder. A female fetus was delivered by repeat caesarean section. Although detailed necropsy was not undertaken, hydroureter, hydronephrosis, and a massively dilated bladder were noted.

Discussion

The occurrence of affected sib pairs, especially with consanguineous parents,¹ suggests the possibility of an autosomal recessive pattern of inheritance for this syndrome. Genetic heterogeneity is possible since these features have been considered to be a marked form of hereditary hollow visceral myopathy,⁵ previously described as having an autosomal dominant mode of inheritance in some families. In the absence of a positive family history of bowel and bladder dysfunction, a recurrence risk of at least 25% seems appropriate for families with one affected infant.

Based on the clinical features, the diagnosis of megacystis-microcolon-intestinal hypoperistalsis syndrome was made shortly after the birth of the first infant in this report. Although a detailed necropsy was not performed on the second infant, the prenatal ultrasonographical findings strongly suggest she also had this condition. The detection of the distended urinary bladder and renal pelvic anomalies at 19 weeks of gestation shows the feasibility of second trimester prenatal diagnosis for this syndrome, which previously has not been noted in a fetus at risk. Intrauterine death has not been described in megacystis-microcolon-intestinal hypoperistalsis syndrome. Although the cause of the intrauterine death was not clear, other affected fetuses may be at risk of early death and pregnancies must be monitored for this event.

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