Table 2 Radiological findings

Condition	Schinzel and Giedion (1978)		Present case
	Case 1	Case 2	
Short and sclerotic base of skull	+	+	-
Steep base of skull	+	+	+
Wide occipital 'synchronosis'	+	+	+
Multiple Wormian bones	+	+	+
Orbital hypertelorism	+	+	+
Wide cranial sutures and fontanelles	+	+	+
Broad cortex and increased density of long tubular bones and vertebrae	+	+	-
Broad ribs	+	+	+
Hypoplastic or aplastic pubic bones	-	+	-
Moderate mesomelic brachymelia	-	+	+
Congenital hydronephrosis and hydroureter	+	+	?
Short metacarpals of thumbs	+	+	-
Hypoplasia of distal phalanges in hands and feet	+	+	+

we compare the clinical and radiological findings of our patient with those of Schinzel and Giedion. With the exception of an atrial septal defect, for which there is no clinical evidence at present, our patient has all the clinical findings and most of the radiological features found in both of Schinzel and Giedion's patients (Table 1), and his clinical course has been similar to that of their first patient who survived long enough to permit comparison. Schinzel and Giedion concluded that autosomal recessive inheritance was likely in view of the involvement of sibs of different sex born to normal parents. This is of great importance in determining what genetic counselling should be given, but further reports of affected sibs and increased frequency of parental consanguinity are necessary to support the conclusion that this form of inheritance is involved. Our report helps to establish the existence of this syndrome as a separate defined entity, but since only one sib is affected and the parents are not consanguineous, it does not help to elucidate the mode of inheritance.

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Reference

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Roberts's syndrome and clonidine

SUMMARY A child is described with features of Roberts's syndrome. The mother took clonidine during pregnancy. The question of a relationship between the child's abnormalities and clonidine is raised.

In 1966, Appelt *et al.* defined a syndrome as Roberts syndrome. The most prominent symptoms of Roberts's syndrome (Roberts, 1919) are low birth weight, tetraphocomelia, cleft lip and palate, dysmorphism, failure to thrive, vitium cordis, and low life expectancy. A similar condition has been described under the names of pseudothalidomide or Roberts syndrome by Hermann and Opitz (1977).

Roberts's syndrome is a rare condition and only a few papers on it have been published since 1966 (for review see Freeman *et al.*, 1974; Hermann and Opitz, 1977). We observed a boy recently with Roberts's syndrome. Our observations add another case to previously reported publications and we consider the question of the relationship between our patient's abnormalities and clonidine.

Case report

The father, 37 years of age, is healthy. The mother had hypertension before pregnancy. Therefore, she took a hypotensive drug, clonidine 0.3 mg per day from the beginning of pregnancy. From the sixth month of pregnancy, diazepam (10 mg per day) was added to the clonidine. During pregnancy the mother took no other drugs, she was not irradiated and except for hypertension she was not unwell. The parents are not related. An older sister was stillborn but had no malformations. The family history was normal.

The course of the pregnancy, apart from the hypertension, was normal. The child was born by caesarean section at 36 weeks' gestation on 2 July 1977. Weight at birth was 1300 g, length 33 cm. At physical examination the following abnormalities

were seen (Fig.): tetrphocomelia, three fingers on the right hand and a very small fourth high-placed finger on the cubital side of the hand, four fingers on the left side, cleft lip and palate, marked prominence of the eyes, ocular hypertelorism, protrusion of the intermaxillary portion of the upper jaw, epicanthus, large frontal bones, blond but not silvery hair, low positioned ears, midfrontal haemangioma, bilateral contracture of the knees, syndactyly of the 4th and 5th right toes, six toes on the left foot, club foot, and large penis and testes. A systolic murmur (grade 4/6) was present.

At x-ray examination there was bilateral absence of the radius and cubitus, four metacarpal bones, three fingers on the right side and four on the left with phalanges. The vertebral column was normal. The femur was present but only one bone was found in the foreleg. There were five metatarsal bones, with only the proximal phalanges present.

The karyotype using conventional and RHG-banding was normal. Fundoscopy was normal. Electrocardiogram showed right ventricular hypertrophy. At birth, thrombocytes were $104\,000/\text{mm}^3$.

The clinical course was characterised by failure to thrive. At 16 months of age, his weight was 4500 g (-5 SD), length was 52 cm (-8 SD), and head circumference was 42 cm. The child smiled at 3 months, held his head up at 4 months, and sat alone at 12 months.

At 7 months, for the first time, glucose was found in his urine. Proteinuria was 0.50 g/l and urea 0.49 to 0.65 g/l. IPG showed a duplicity of the left calices.

Discussion

A review of Roberts's syndrome was recently undertaken by Freeman *et al.* (1974). Our patient has all

the abnormalities found in the reports of patients with tetrphocomelia and cleft lip or palate or both.

Roberts's syndrome differs from the SC phocomelia or 'pseudothalidomide' syndrome (Hermann and Opitz, 1977) by the lack of silvery-blond hair and blue sclerae, and the presence of ulnar rather than radial abnormalities of the upper extremities. Features common to both syndromes are intra- and extrauterine retardation of growth. Mental retardation is probably common to both syndromes but is difficult to evaluate in Roberts's syndrome. Our patient, at 8 months of age, has only mild mental retardation.

If we refer to the R-score or S-score established by Hermann and Opitz (1977) for distinguishing between Roberts's syndrome and the SC syndrome, our patient has Roberts's syndrome, as birth length was under 37 cm, he had a cleft palate and short arms, but his nasal alae were not hypoplastic. Klein (1977, personal communication) stressed the point that patients with the SC syndrome have ocular abnormalities like cataracts. Our patient was free of such features.

Several observations suggest that Roberts's syndrome is a genetic disorder transmitted in an autosomal recessive manner (Freeman *et al.*, 1974; Zergollern and Hitrec, 1976; Hermann and Opitz, 1977). All the malformations associated with Roberts's syndrome could have resulted from a disturbance in development before the seventh week of gestation (Smith, 1976). The mother of our case had taken clonidine from the beginning of gestation. The question arises as to whether clonidine could be involved in the occurrence of the syndrome in our patient. We were unable to find a report saying that such a causal relation could exist. Clonidine is a drug which is sometimes given to pregnant women with hypertension in our country, and the occurrence of malformations in the fetus has not been reported, but usually clonidine is not taken after the first month of pregnancy.

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Fig. Probant at one week of age.

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