

Roberts Syndrome

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Roberts Syndrome was first described in 1919¹ in 2 siblings, (a sister and a brother) who had severe limb deformities (tetraphocomelia) with bilateral cleft lip and palate as major features but it was first defined and recognized as a syndrome in 1966². Previously only three families have been reported with affected siblings including that by Roberts. Out of these, two families had consanguinity established between parents^{1,2}. Characteristically a child with this syndrome has profound growth deficiency of prenatal onset. Usually birth weight ranges between 1.5-12kg and birth length is less than 40 cm. There is cleft lip with or without cleft palate, prominent premaxillae, hypertelorism, mid-facial hemangioma, thin nares and shallow orbits with prominent eyes. Also there is hypomelia ranging from phocomelia (seal limb) to lesser degrees of limb reduction, often including radial aplasia, syndactyly, clinodactyly of fifth finger and talipes. Encephalocoele or hydrocephalus, corneal clouding, cataract, cardiac anomalies, renal anomaly (polycystic or horse shoe kidney) bicornuate uterus, thrombocytopenia and hypospadias may be present¹⁻³. We report two sibs of the same family having features similar to this congenital malformation syndrome. Parents of the patients were first cousins. Father was forty years and mother was thirty three years old when first affected child was born. They have five Children, four males and one female who are alive and healthy.

Case 1

The girl presented at the age of five years. She was a product of an uneventful full-term vaginal home delivery. There was no history of drug use in pregnancy. She had multiple congenital defects at birth, but was never taken for professional opinion. At five years she was seen by us when her younger brother was admitted in hospital. Her weight was 8.0 kg, height 76.0 cm and head circumference was 43 cm, all being less than third percentile for her age. She had hypertelorism, narrow palpebral fissure, irregular cornea with clouding and central cataract, alae nasi were hypoplastic with thin nares. She had high arched palate but did not have a cleft palate. Her right elbow was flexed at 90 degrees and immobile. Left forearm was short, hypoplastic with radial angulation of left hand. Fifth digits had clinodactyly with abducted thumbs in both hands. Lower limbs were very short. Knees though mobile, showed flexion contracture causing deformity. Lateral angulation at knees was present (Figure 1).



Figure 1. Short stature compared with a normal child of the same age. The case has ankylosed left elbow joint and flexion contracture of both knees.

Little toes of both feet were overriding. Child was able to walk slowly, was intellectually normal and had nasal tone in speech. Complete blood picture was normal, platelet were ,000 per cmm. Radiological examination showed absent right elbowjoint with ankylosis of radial head with humeral condyles. In the left fore- arm radius was absent and only one ossification centre was visible (Figure 2).

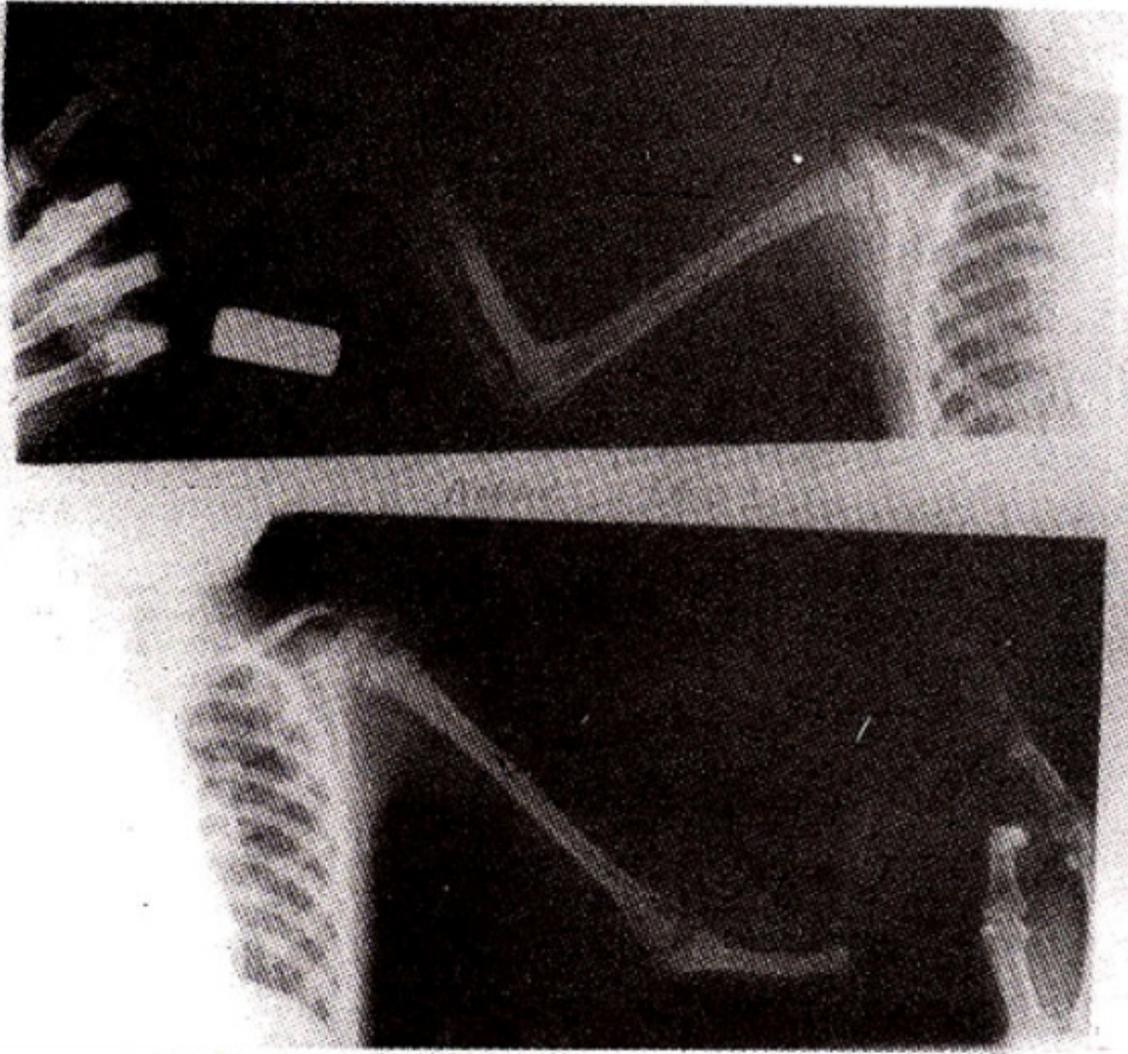


Figure 2. Radiological examination shows ankylosed right elbow joint with absent radius of left side.

Ultrasound examination of the abdomen was normal. No uterine abnormality could be detected as uterus was yet very small. Both the kidneys were normal. ECG and echocardiography did not reveal any cardiac anomaly.

Case 2

Second child was born five years after the above mentioned girl. During pregnancy mother had been using antihypertensive medicines but no record of any specific medicine was available. Child was born after a full term home delivery. It was a difficult vaginal delivery after a prolonged labour. Child presented at two months to us when he had bronchopneumonia after aspiration. At that time his weight was 2.2 kgs, length and head circumference both were 33.0 cm, all being less than third percentile for his age. He had bilaterally absent forearms, only had four digits in both hands with absent thumbs and single distal palmar crease. In lower limbs thighs were short, legs were deformed and hypoplastic, knees were fixed and ankylosed and feet had bilateral talipes equinovarus (Figure 3).



Figure 3. Bilateral cleft lip and palate with phocomelia in both upper limbs and deformed lower limbs.

There was webbing between fourth and fifth toes (syndactyly). On the face there was a glabellar hemangioma, very narrow anterior fontanelle, low set ears, deviated and deformed nasal septum, bilateral cleft lip with cleft palate, prominent premaxilla and bilateral corneal haziness with irregular outline. On auscultation a systolic murmur at left lower sternal border was audible. Complete blood picture showed TLC of 21,100 cmm, RBC count 3.8 million/cmm, Hb was 12.1 g/dl and platelet were 518,000/cmm. Serum electrolytes and urea were normal. Radiological examination showed absent radii and ulnae in both upper limbs (Figure 4).

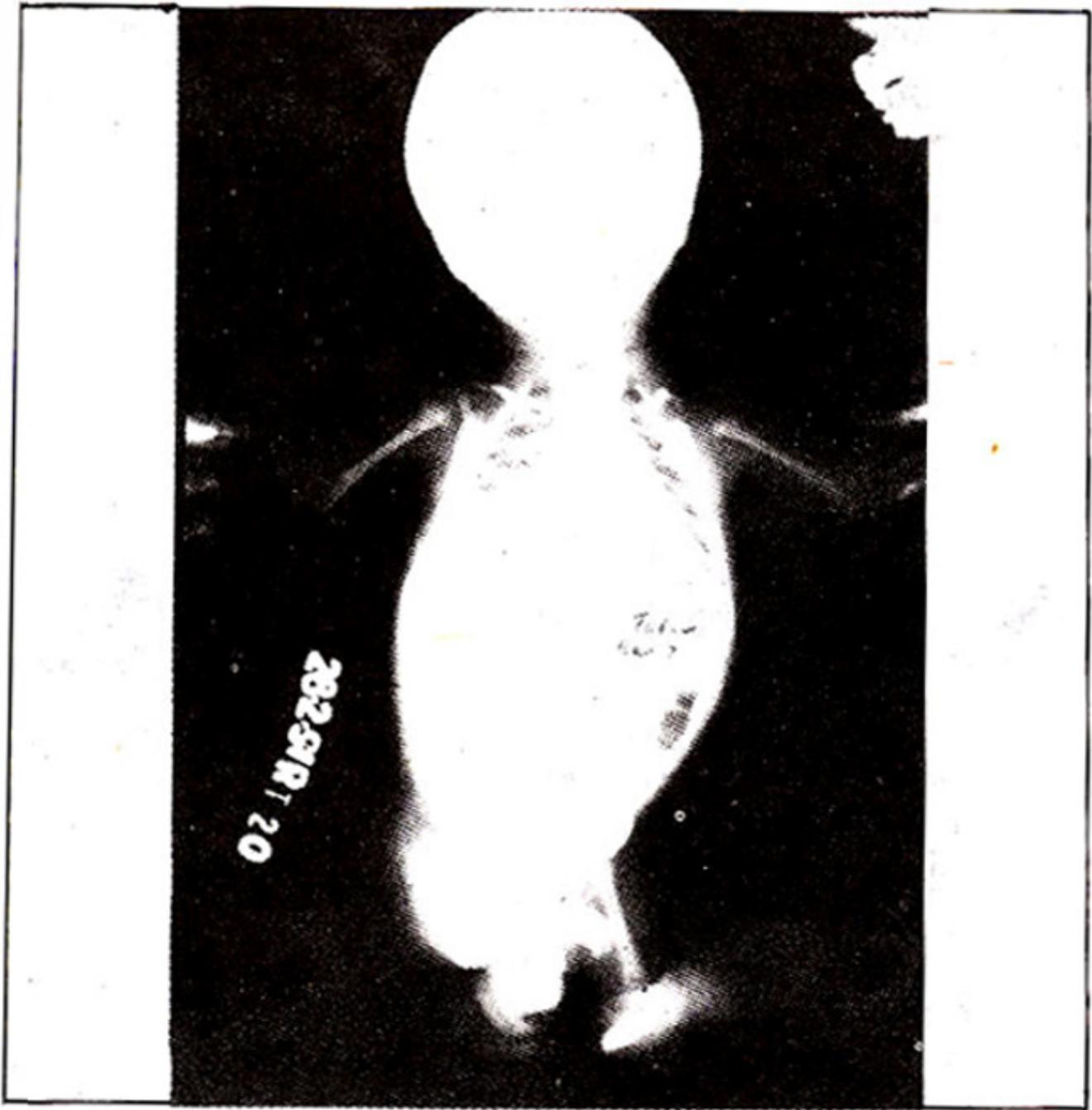


Figure 4. Bilaterally missing radius and ulna with deformed lower limb skeleton.

In both lower limbs femur were very short, in legs only tibia were seen which was deformed and bow shaped. Cardiac shadow was enlarged with bilateral soft shadows of pneumonia in chest x-ray. ECG showed right ventricular predominance and echocardiography depicted high VSD with left to right shunt. Ultrasound of abdomen did not pick up any abnormality. Both children did not return for follow-up, although they were advised to do so.

Discussion

Of the two sibs that we have presented the younger child has typical features of Roberts syndrome, while the older sister had less severe deformities and was less crippled. There is some overlap between

features of Roberts syndrome, pseudothalidomide syndrome (PTS) which has phocomelia, joint contracture, cleft lip and palate alongwith multiple minor anomalies and thrombocytopenia absent radius (TAR) syndrome, which is a combination of congenital heart failure, kidney malformation, thrombocytopenia and absence of radius. There has been some discussion whether all these syndromes can be grouped in a single entity with wide variety of symptoms or not^{4,5}. Grosse et al have voiced their reservations about discriminating atleast Roberts syndrome and Pseudothalidomide syndrome⁴, whereas Waldenmaier have hypothesized that the three syndromes cannot be considered as separate entities and probably represent only different degrees of the Roberts syndrome⁵. This argument is strengthened by the fact that, in the twenty-two cases reviewed by Freeman et al, the two sibs whose parents were double first cousins did not have cleft lip or palate and they classified the index case as Roberts syndrome and the sib as questionable Roberts syndrome². The same two sibs impressed Hermann et al, as possible cases of Pseudothalidomide syndrome. Previously, in some cases of Roberts syndrome thrombocytopenia has been described², which is also a feature of TAR syndrome⁵. Roberts syndrome has been described as autosomal recessive^{1,2,4}, but in this family seven sibs were born normal (two of them died due to neonatal sepsis). This does not favour autosomal recessive behaviour in our cases because only the eighth and ninth sib were affected. May be there is some other factor playing a role in its etiology. The role of drugs is doubtful, because in the present case the mother did not use any drugs during first affected pregnancy while she was regularly on medications for her hypertension during second affected pregnancy. According to the literature this may be the fifth family which has been reported with features of Roberts syndrome.

References

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