Clinical Case Reports

The Roberts Syndrome

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Summary. Four siblings with the autosomal recessive Roberts syndrome are reported, and we discuss the phenotypic overlap of this syndrome with other similar radial aplasia syndromes.

Introduction

Roberts syndrome (RS), a very rare nosologic entity, was first described in 1919. Typically, the syndrome is characterized by prenatal and postnatal growth retardation, severe hypomelia, bilateral cleft lip and palate with protruding premaxilla, eye abnormalities, and large phallus. This combination of malformations was recognized as a syndrome in 1966 by Appelt and co-workers. Inheritance is autosomal recessive.

We report four affected siblings (three males, one female) from northeast Brazil, and comment on the clinical overlap of Roberts syndrome with the so-called pseudothalidomide (SC phocomelia) and radial aplasia-thrombocytopenia (TAR) syndromes.

Case Reports

The parents, a 31-year-old man and a 23-year-old woman, were first cousins. Both were healthy. Family pedigree, pictures, and X-rays of the patients are shown in Figs. 1, 2, 3, and 4.

Case 1 (III.14). Born in a breech position on August 30, 1979, after a full-term pregnancy, birth weight was 1300 g, length 31 cm, head circumference 27.8 cm, and chest circumference 25 cm. The physical examination soon after birth showed severe microbrachycephaly, midfacial capillary hemangioma, prominent eyes, mild ocular hypertelorism, cloudy corneas, hypoplastic ear lobules, bilateral cleft lip and palate with protruding premaxilla, small phallus, hypoplastic first metacarpals, partial fusion of the第三 and fifth metacarpals, abnor-malities of the thumbs, and clinodactyly of the fifth fingers, and dislocation of the hip. Routine hematological investigations demonstrated thrombocytopenia (32,800 platelets/mm³) and all other blood counts were within normal limits. In the first 48 h of life the general condition of the infant was normal. He presented jaundice on day 3 with serum bilirubin 13.14 mg/dl (direct bilirubin 0.5 and indirect, 12.64 mg/dl). After 4 days of phototherapy, he showed a clinical improvement. However, on day 13 the jaundice became worse and an apneic crisis occurred. Death occurred on day 14.

Case 2 (III.16). This child was born at term on April 15, 1982. Pregnancy and delivery were normal. Birth weight was 1060 g, length 32 cm, head circumference 25 cm, and chest circumference 25 cm. He had severe microbrachycephaly, prominent eyes, upward-slanting palpebral fissures, ocular hypertelorism, cloudy corneas, low nasal bridge, and bilateral cleft lip and palate with protruding premaxilla. The ears were malposed with hypoplastic lobules and prominent antihelix. The phallus was large. He also showed mesomelic hypomelia, hypoplasia of the first and fifth fingers, limitation of the movement in most joints with flexion contractures at the knees, and cutaneous syndactyly of the fourth and fifth right toes. X-ray examination showed absence of the fore-arm bones on both sides and of the phalanges of the thumbs, and absence of the fibula bilaterally. Routine hematological investigations were normal. He lived only 5 days and a specific cause of death was not determined.

Case 3 (III.12). This child was born in a breech position on March 2, 1977, after a full-term pregnancy. At 3 years and 6 months of age her weight was 5400 g, while height, head circumference, and chest circumference were 70, 37.5, and 43.5 cm, respectively. These values, especially the first three, were well below the third percentile. Physical examination revealed a very retarded infant...
Fig. 2. Front (a) and side (b) views of case 1; case 2 (c)

Case 4 (III.15). This boy was born at term on November 8, 1980. Pregnancy, delivery, and the neonatal period were normal. He was first seen at 10 days of age when weight was 1830 g and length, head circumference, and chest circumference were 41, 29, and 29 cm, respectively. His general condition was normal. He presented microbrachycephaly, midfacial capillary hemangioma, high arched palate, cloudy corneas, hypoplastic alae nasae, unilateral cryptorchidism, large phallus, mesomelic hypomelia, hypoplasia of the thumbs and 5th fingers, clinodactyly of the 5th fingers, unusual pattern of palmar creases, limitation of the movement of the wrists, elbows, hip, and knees, and clinodactyly of the 2nd toes. The radiological findings were radial aplasia and cubital hypoplasia on the left side, and absence of both radius and ulna on the right and dislocation of the hip. Routine laboratory tests showed no abnormalities.

Discussion

Only a few papers about RS have been published since 1919. The literature up to 1976 recorded just 26 cases, including questionable ones (Zergollern and Hitrec 1976). At least two autosomal recessive dysmorphogenetic syndromes present clinical overlap with RS, the so-called pseudothalidomide (SC phocomelia) syndrome (SCS) and TAR syndrome (TARS).

Our cases 3 and 4 presented features of both RS and SCS. According to the case report and comments by Herrmann et al. (1969) and Herrmann and Opitz (1977) about these syndromes, we assumed that these two patients presented more clinical similarities with SCS, since they did not have facial cleft and the craniofacial anomalies were capillary hemangioma, moderately sparse scalp hair (only case 4), cloudy corneas, and hypoplastic