

Short rib-polydactyly syndrome: a case report

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Short rib-polydactyly syndrome (SRPS) is a group of rare, lethal skeletal dysplasias characterized by short ribs and limbs, polydactyly, hypoplastic thorax and visceral anomalies.

Our case had coarsening of facial features, low-set ears, lobulated tongue, cleft palate, and hypoplastic epiglottis. Short proximal parts of upper limbs, bilateral postaxial polydactyly of hands, and bifid big toe with zygodactyly were additional findings. Chest was narrow. Ambiguous genitalia was noted but testicles were in scrotum. Choroid plexus cyst and coarctation of aorta were found in autopsy.

Radiographies of the skull revealed occipital horn accompanied by prominent external occipital protuberance. The thoracic cage was narrow and elongated with short and iliac wings, pubic and ischial rami were hypoplastic, and both acetabula were shallow and trident shaped. All tubular bones had wide and rounded metaphyses.

Because clinical and radiological features of the four established subtypes are very similar, there are difficulties in the classification. We report an infant whose radiological, clinical and postmortem features were consistent with type IV SRPS (Beemer-Langer).

Key words: skeletal dysplasia, short rib-polydactyly syndrome (SRPS), Beemer-Langer syndrome.

Short rib-polydactyly syndrome (SRPS) is a descriptive category or a group of lethal skeletal dysplasias characterized by hypoplastic thorax, short ribs, polydactyly, and visceral abnormalities. Classically, four different types have been described: SRPS I (Saldino-Noonan); SRPS II (Majewski); SRPS III (Verma-Naumoff); and SRPS IV (Beemer-Langer)¹⁻³. All forms of the SRPS described to date are thought to be inherited in an autosomal recessive manner.

There are difficulties in the classification of these disorders because of the frequent overlap. There is controversy, however, as to whether this is due to variable expression or to genetic heterogeneity^{2,4,5}.

We report an infant whose radiological, clinical and postmortem features were consistent with SRPS IV (Beemer-Langer).

Case Report

After a completed 35 weeks of gestation, complicated with polyhydramnios, an infant was delivered by cesarean section. The baby presented low Apgar (2/3) scores. The baby died of respiratory insufficiency due to pulmonary hypoplasia despite an immediate resuscitation and then ventilation treatment for eight hours.

The mother did not suffer from any known infection during pregnancy. She had three earlier pregnancies that resulted in a normal male and two spontaneous abortions of unknown cause. The baby was a product of consanguineous parents (second cousins), a 21-year-old healthy mother and 24-year-old healthy father.

On the physical examination: weight was 2750 g (50-90th percentile), length was 46 cm (10-50th percentile), and head circumference

was 34 cm (50-90th percentile). The baby presented with coarse face, low-set ears, and cleft palate and lobulated tongue with hypoplastic epiglottis (Fig. 1). There was a 1 cm long bony tuberculum on the occipital area. The thorax was narrow in spite of large abdomen. External genitalia were ambiguous but both testes were palpable. Proximal upper limbs were short. There was bilateral postaxial polydactyly of hands and bifid big toe with zygodactyly (syndactyly of toes 2-3) (Fig. 2).



Fig. 1. General appearance of the infant.



Fig. 2. Bifid big toe with zygodactyly.

In addition, cerebral choroids cyst, hypoplastic stomach and coarctation of aorta were found during autopsy. X-ray investigation revealed: low mineralization of the skull vault with prominent protuberantia occipitalis externa under an occipital horn (Fig. 3). Thorax was narrow and long with short horizontal ribs. Iliac bones were hypoplastic with trident pelvic appearance and low acetabula shallow (Fig. 4). All of the metaphyses of the tubular extremity bones were widened with a round appearance. Nutritional foramens were widened. On the lower limbs cortical thickening was obvious. There were incomplete postaxial polydactyly on both hands.



Fig. 3. Prominent protuberantia occipitalis externa under an occipital horn.



Fig. 4. Narrow and long thorax with short horizontal ribs and hypoplastic iliac bone.

Discussion

The presence of hypoplastic thorax, short horizontal ribs and polydactyly with facial dysmorphism places these cases in the category of SRPS. Table I compares the features of our patient to those seen in the various types of this syndrome.

According to the international osteochondrodysplasia classification¹, there are at least four types of SRPS: Type I (Saldino-Noonan), Type II (Majewski), Type III (Verma-Naumoff) and Type IV (Beemer-Langer).

There are also publications that include asphyxiating thoracic dysplasia (ATD) and Ellis-van Creveld syndrome (EVC)². It is difficult to distinguish these distinct entities from one another and the findings frequently overlap.

Table I. Comparison of Features in Accepted Types of Short Rib-Polydactyly Syndrome (+ = presence of trait)^{4,5,7}

	Current Case	Type I	Type II	Type III	Type IV
Short horizontal ribs	+	+	+	+	+
Short limbs	+	+	+	+	+
Polydactyly pre/postaxial	+	+	+	+	±
Ambiguous genitalia	+	-	+	+	+
Hydrops	+	+	+	+	+
Polyhydramnios	+	+	+	+	+
Median cleft palate/lip	+	-	+	+	+
Situs	-	+	-	+	-
Coarctation of the aorta	+	-	+	-	-
Vertebrae abnormalities	-	+	-	+	±
Hypoplastic epiglottis	+	-	+	-	-
CNS abnormalities	+	-	+	-	+
Oval tibia	-	-	+	-	-
Smooth metaphyses	+	-	+	+	+
Gastrointestinal and genital abnormalities	+	+	+	+	±

Asphyxiating thoracic dysplasia is characterized by severely narrowed chest with acromelic shortening of the limbs. Polydactyly is observed in some cases. Radiologically the vertebrae are normal and the iliac bones are vertically shortened. In ATD, the ribs are usually placed horizontally, as in our case. EVC is characterized by ectodermal dysplasia. The extremities are acromelically short and hands exhibit polydactyly. The skeletal symptoms of our case differ from this syndrome and rather resemble those of SRPS.

Saldino and Noonan first reported Type I SRPS^{5,6}. This type is characterized by hypoplasia in vertebrae and iliac bones, in addition to the characteristic properties of SRPS. Various abnormalities of the heart have been defined, including complete transposition of the great vessels^{2,6}. In our case, the fact that the vertebrae were normal and the iliac bones relatively well developed led us away from such a diagnosis. In addition, anomalies of the larynx and epiglottis have not been reported for Type I. Hypoplasia of the larynx and epiglottis and lobulated tongue are observed in Majewski type (Type II). However, in this type, the iliac wings are normal and, radiologically, the tibia often appears oval. In our case, the tibia did not have an oval appearance^{3,7,8}.

Larynx anomalies, bifurcation of the toe, and postaxial polysyndactyly are common findings of the Majewski syndrome and oral-facial-digital (OFD) syndrome Type II (Mohr syndrome)⁹. The Majewski syndrome can be distinguished from the Mohr syndrome as it exhibits normal thorax and shortened extremities.

The Verma-Naumoff type was first published in 1983 as a subtype of Type I, since most of its characteristics are the same as those of Type I. It differs from Type I as the bones of the extremities and the iliac bones show better ossification, and visceral organ anomalies occur less often². Generally, hypoplasia of the larynx and epiglottis, cleft lip and cleft palate are not observed in this type. None of the features of this syndrome match with the case presented here.

The most characteristic properties of the Beemer-Langer type (Type IV) are hydrops, ascites, macrocephaly, cleft palate, narrow thorax, short extremities and polydactyly^{1,10,11}. Beemer et al.¹⁰ pointed out that the oral manifestations resemble those of OFD with median cleft lip. This type should also be differentiated from the Majewski type as it differs through the radiological appearance of the tibia.

In SRPS, polyhydramnios or oligohydramnios is frequently encountered in prenatal ultrasonography scans during pregnancy¹². Polyhydramnios was observed in the prenatal examination of this case.

In conclusion, we think that clinical, radiological and autopsy findings of our case match with SRPS, particularly Beemer-Langer type. In addition, the presence of a horn-like protrusion on the occipital bone of our case has not been reported before in SRPS and, if not coincidental, makes this case an interesting one. As the overlapping clinical features make SRPS cases difficult in differential diagnosis, revealing the gene loci responsible for them seems necessary¹³.

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