

## SHORT RIB POLYDACTYLY SYNDROME: A CASE REPORT

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**Abstract-** Short rib polydactyly syndrome (SRPS) is a very rare congenital anomaly that is classified into four subtypes. It is an autosomal recessive inherited disease. We report a case of this syndrome without a previous family history of congenital defects.

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*Acta Medica Iranica*, 45(6): 513-514; 2007

**Key words:** Rib polydactyly syndrome

### INTRODUCTION

Short rib polydactyly syndrome is a rare skeletal dysplasia that is manifested with short limb, short ribs with thoracic hypoplasia kpotyelactyly. Its birth prevalence is 2.5 to 3.3 per 10/000 births. We report a case of this syndrome without a previous family history of congenital defects.

### CASE REPORT

A 22-year-old gravid 2 para 1 woman was admitted in our hospital at 36 weeks of gestation because of rupture of membrane and a decreased amniotic fluid report in the sonographic studies. Her first pregnancy had been resulted in a live normal infant; without any anomaly or similar disorder; two years ago. She had negative history of infection or drug intake or radiation exposure during pregnancy. The first sonography performed in early pregnancy was normal, but the second one showed a cephalic fetus with oligohydramnios (Amniotic Fluid Index <3 cm). Fetal spine and skull were normal biparietal diameter corresponded to gestational age. There was no edema over the organs. Placenta was normal with a triple vessled structure cord.

Cesarean section performed because of prolonged rupture of membrane resulted in an infant weighing 2500 gm with a good Apgar score. The infant had short limbs, narrowed thorax and polydactyly. Radius, ulna, tibia, and iliac bone were hypoplastic. The other organs such as stomach, heart, lung, spleen, kidney and bladder were normal. The other organs such as stomach, heart, lung, spleen, kidney and bladder were normal. Karyotyping was not performed. Negative VDRL and TORCH profile and normal blood sugar was revealed in subsequent laboratory studies. Infant died due to lung hypoplasia 6 hour after birth (Fig. 1).



**Fig. 1.** Short rib infant with hypoplastic limbs

Received: 23 Jun. 2006, Revised: 22 Dec. 2006, Accepted: 17 Feb. 2007

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## DISCUSSION

Skeletal dysplasia represents 1 to 3.5% of the fetuses detected sonographically with congenital malformations. Its birth prevalence is 2.5 to 3.3 per 10'000 births. Many of the fetuses with this anomaly do not survive till term (1).

Short rib-polydactyly (SRPS) is a rare skeletal dysplasia that is manifested with short limb short stature, short ribs with thoracic hypoplasia, and polydactyly. Many of cases of this syndrome show manifestations of heart, intestines, genitalia, and cystic lesions of kidney, liver and pancreas. Polydactyly is greatly frequent but not a sure event (2).

Saldino-Noonan (SRPS type 1) is a rare and differs in shape of the long bones with a torpedo-shape appearance. It is manifested by metaphysial irregularity with periosteal spur formation. Cardiovascular disorders like ventricular septal defect and endocardial cushion defects, renal and pancreas cysts, genital anomalies and imperforated anus are the visceral anomalies that may accompany this type (2, 1). Verma-Naumoff (SRPS type 2) is much more common and has a banana-peel shape. Short oval tibia is the characteristic of Majewski syndrome (SRPS type 3). Beemer (SRPS type 4) resembles type 3 but the tibiae are not as short and polydactyly is rarely presented (2).

Due to impaired ventilation because of the chest tightness, type 1 and 2 are severely lethal in perinatal period. Type 3 is a milder variant with fewer visceral abnormalities (1). SRPS is an autosomal recessive inherited syndrome with a recurrent rate in 25% of cases. The different types of SRPS may be the result of several mutations of different genes or may be the consequence of heterogeneity, different mutant alleles, and secondary intrauterine modification of the phenotypes or all types may be a single genetic disorder with a widely variable clinical expression (2). Mutation in fibroblast growth factor receptor 3 (FGFR 3) gene; located in p16 region; is found to be responsible for the phenotype of achondroplastic hypochondroplasia and thanatophoric dysplasia 1 and 2. This gene is expressed mainly in skeleton and central nervous system (3).

Overlap between the 4 types of SRPS and orofacial syndromes is frequently seen. This may be as a result of deletions of different sizes in the same

chromosome region or because of compound heterozygosity for these syndromes (2).

Frequent complex consanguinity in the United Arab Emirates and similar areas attract more attention to consider the possibility of 2 recessive syndromes in the child when features overlap (3).

Potentially surviving conditions, like Jeune syndrome (asphyxiating thoracic dystrophy) and Ellis-van Creveld syndrome (chondroectodermal dysplasia); both recessively autosomal inherited; should be differentiated from 4 types of SRPS described above (2). Majewski syndrome is a lethal skeletal dysplasia. In cases of the 'Mohr-Majewski compound' the prognosis is mainly determined by the presence of lung hypoplasia, and cerebral malformations (3). Other report a complex consanguineous family in which short rib-polydactyly syndrome type III and congenital infection. By the second day the head circumference had increased to 36 cm and the fontanelle became more tense and bulging. CT scan showed a large hemorrhage in the middle of the brain which had destroyed the basal ganglia and showed extension into the ventricles. The baby was scheduled for ventricular drainage but deteriorated and died on the ninth day of life (4). There is a description of one case of a female child which suffered from a short rib-polydactyly-syndrome, a localized form of an osteo-chondrodysplasia, and died at the age of nine months as a result of the typical complications of this disease (5).

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