Recurrent Short Rib-Polydactyly Syndrome (Majewski Syndrome)

TEKRARLAYAN KISA KOSTA POLİDAKTİLİ SENDROMU (MAJEWSKİ SENDROMU)

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Summary

Objective: Short rib-polydactyly syndrome (SRPS) is manifested by short-limb dwarfism, short ribs with thoracic hypoplasia, and polydactyly.

Case Report: In this report, a recurrent case of SRPS type II (Majewski) was diagnosed in a consanguineous couple. In two cases, sonography revealed a small thorax, markedly short ribs, micromelia, polydactyly, a cleft lip. The postnatal appearance and radiographs confirmed the prenatal diagnosis.

Conclusions: The termination of pregnancy is justified at any gestation when diagnosed. The mother of an affected fetus must be told of the risks for subsequent pregnancies.

Key Words: Skeletal dysplasia, Short rib, Polydactyly, Dwarfism, Majewski syndrome

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Özet

Amaç: SRPS (short rib polydactyly syndrome); ekstremitelerde kısalıkla karakterize cücelik, dar göğüs kafesi, kısa kostalar ve polidaktili ile kendini gösteren bir sendromdur.

Olgu Sunumu: Bizim vakamızda yakın akraba evliliği yapmış olan çiftin tekrarlayan SRPS, tip II Majewski sendromu sunulmuştur. İki vakada da ultrasonografide küçük toraks, belirgin kısa kostalar, mikromeli, polidaktili ve yarık damak saptanmıştır. Doğum sonrası görünümleri prenatal tanıyı doğrulamıştır.

Sonuçlar: Tanı konulduğu anda gebelik terminasyonu önerilmelidir. Böyle çocuk doğurmuş anneler bir sonraki gebeliklerinde tekrarlama riski açısından uyarılmalıdır.

Anahtar Kelimeler: İskelet displazisi, Kısa kosta, Polidaktili, Cücelik, Majewski sendromu

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Skeletal dysplasias represent 1% to 3.5% of the fetuses detected sonographically with congenital malformations (1). Short rib-polydactyly syndrome is a rare autosomal recessive and lethal skeletal dysplasia. This syndrome is classified into four subtypes and manifested by short-limb dwarfism, short ribs with thoracic hypoplasia, and polydactyly. Diagnosis can be based on the radiological features. In addition to short ribs and pre-and/or postaxial polydactyly of the hand and feet, there is shortening of the long bones, with smooth rounded metaphysis and a striking oval configuration of the tibiae (2). Various miscellaneous anomalies accompanying the syndrome include: cleft lip-palate, small cerebellar vermis, and renal anomalies. The skull, clavicles, pelvic and vertebral bones are usually normal (3). We present a case with recurrent type II-Majewski syndrome and a review of the recent data about this syndrome.

Case Report

A female infant, the third child of apparently healthy parents (father 35 years old, mother 32 years) was stillborn at 9 months of gestation. The infant had narrow thorax, polydactyly and short limbs. The sixth child was born after detection of the short rib polydactyly syndrome (SRPS)–Majewski Type II with ultrasonographically at 36 weeks of gestation. The parents are first degree relatives and the chromosomal analysis was normal. The infant delivered after induced labor for intrauterine death. Examination of the body showed pulmonary hypoplasia, narrow thorax, bilateral cervical rib, symmetrical shortness of the

upper and lower limbs, bilateral dislocation of the radius, preaxial polydactyly of the hands and postaxial polydactyly of the feet, unilateral incomplete cleft lip-palate, ventricular septal defect, unilateral segmental renal dysplasia, short intestine, two umbilical cord vessel, with these findings the diagnosis of SRPS type II was confirmed (Figure 1). Her seventh pregnancy was two years later. At the 18th week of the gestation detailed ultrasound revealed a single live fetus with polyhydramnios, short limbs (tibia), polydactyly of the feet and hand, cleft lip and palate, bilaterale echodense kidneys (Figure 2). In view of the past history and sonographic findings prostaglandin induction was done and the patient delivered a male fetus weighing 330 gr. The death infant showed extremely short upper and lower limbs, complete cleft lip and palate, postaxial polydactyly of the hands and preaxial polydactyly of the feet (Figure 3). There were 7 digits on the left hand (incomplete syndactyly between 1 and 2 digits, complete syndactyly between 6.-7. digits), 6 digits on the right hand (syndactyly between 5.-6. digits), and 7 digits on the left and right foot (syndactyly between the first three of them). In addition bilateral aplasia of the fibulae was detected. The autopsy confirmed the diagnosis as on previous occasions of SRPS type II-Majewski syndrome and no anomaly of the viscera was detected. Radiographic studies showed generalized shortening of all the long bones and ribs (Figure 4).

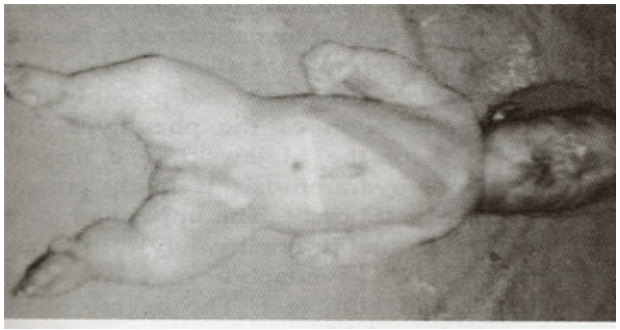

Figure 1. Full body frontal view. Narrow thorax, symmetrical shortness of the upper and lower limbs, preaxial polydactyly of the hands and postaxial polydactyly of the feet, cleft lip-palate (case 1, fetus at 36 week).

Discussion

Skeletal dysplasias (osteochondrodysplasias)

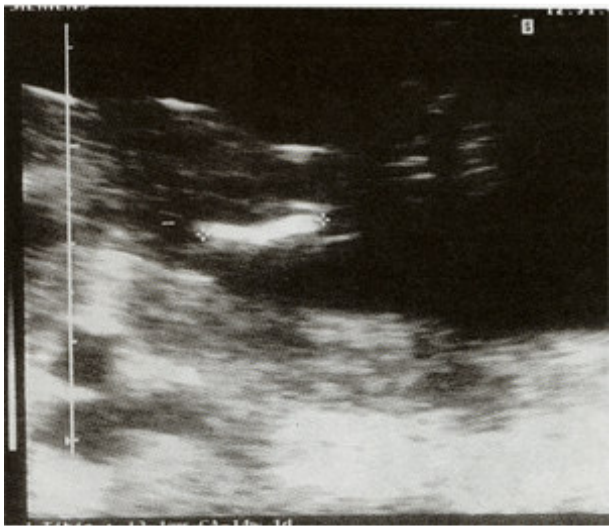

Figure 2. Longitidunal sonograms of a lower extremity show a severely short tibia (fetus at 18 week, case 2).

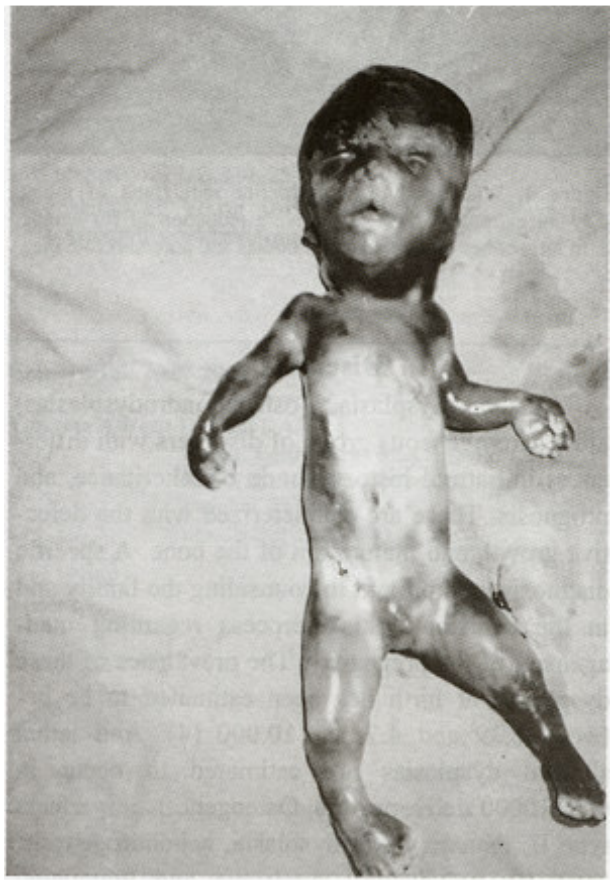

Figure 3. The fetus showed extremely short upper and lower limbs, complete cleft lip and palate, postaxial polydactyly of the hands and preaxial polydactyly of the feet. SRPS type II Majewski syndrome (case 2, fetus at 18 week).

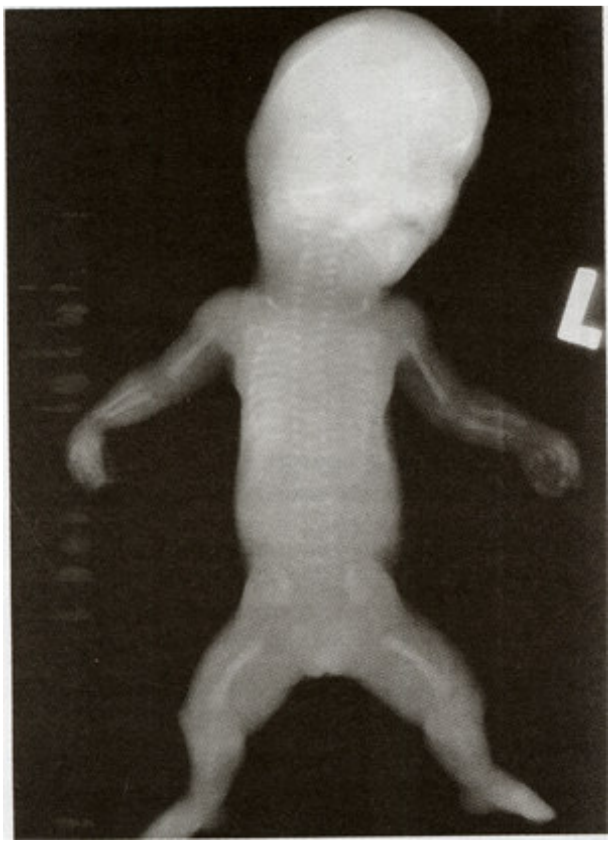

Figure 4. Whole-body anteroposterior radiograph of infant showing extremely short ribs, severe reduction of rib length, short long bones, square vertebral bodies and unremarkable ilia.

are a heterogeneous group of disorders with differences in natural history, mode of inheritance, and prognosis. These are characterized with the defective growth and maturation of the bone. A specific diagnosis is important in counseling the family and in the decision-making process regarding management of the pregnancy. The prevalence of these dysplasias at birth has been estimated to be between 2.29 and 4.7 per 10.000 (4). And lethal skeletal dysplasias are estimated to occur in 0.95/10000 deliveries (5). Osteogenesis imperfecta type II, thanatophoric dysplasia, achondrogenesis, short rib polydactyly syndrome and hypophosphatasia are all lethal, shortly after delivery.

The association of polydactyly, a narrow thorax and short –limb dwarfism in the newborn leads to the following diagnostic considerations: SRP syndromes; chondroectodermal dysplasia (Ellis van Creveld syndrome) and asphyxiating thoracic dystrophy. Pelvic bone anomalies are a part of the last two syndromes, but not the Majewski type. The cases with chondroectodermal dysplasia may have nail dysplasia and the rate of the heart anomaly is 60%. Cleft lip and plate are usually not determined by the last two syndromes. In 1971 Majewski et al. described a stillborn girl with the definitive radiological features (3). In addition there was a cleft lip and palate, malrotation of the gut, hypoplasia of the lungs, a persistent left superior vena cava, patent ductus arteriosus and ascites. SRPS has an autosomal recessive mode of inheritance and 25% chance of recurrence in subsequent pregnancies (6). In addition to the dominant features of short – rib thoracic hypoplasia and polydactyly, fetuses with SRPS may have variable syndactyly, genital abnormalities, median cleft lip and visceral anomalies. The most of these abnormalities can be detected by transvaginal sonography beginning in the early second trimester. Four types of SRPS have been described: Saldingo-Noonan (7), type I; Majewski, type II; Verma-Naumoff (8), type III and Beemer-Anger type IV. Each type of the syndrome is characterized with certain anomalies: narrow metaphysis by type I, cleft lip and palate and shortened tibia by type II, wide metaphysis by type III, and medially cleft palate, very short rib, thoracic hypoplasia, umbilical hernia and ambiguous genitalia by some male infants with type IV. The overlap of anomalies among the different SRPS types has led investigators to theorize that SRPS results from mutant alleles and a single locus with variable expressivity and possibly from intrauterine modification of the phenotype. The fetus with SRPS of the Majewski type may be hydropic and polyhydramniosis is likely present. The abdomen is protuberant. The upper and lower limbs short (mezomelic brachymely). Pre- and postaxial polydactyly -syndactyly may be present on both hands and both feet. The degree of the polysyndactyly is variable, some cases have 9 digits. The necropsy findings of the cases are: hypoplasia of the epiglottis, larynx and lungs. About %50 exhibit imperforate anus and intestinal malrotation and/or short bowel and fibrocystic pancreas,

cardiac anomalies include persistent superior vena cava, ventriculer septal defect, coarctation of the aorta. Genitourinary anomalies observed are small or absent anogenital opening, hypospadias, micropenis, cryptorchidism, septate uterus, and septate or rudimentary vagina. The kidneys are hypoplastic with multiple glomerular cyst and focal cystic dilatation of the tubules. Often there is cystic and/or hypoplastic uterus. Radiographic changes include very short horizontal ribs, pre-and postaxial polydactyly and short rounded tibiae. The iliac and vertebral bodies appear essentially normal. There is premature ossification of the proximal epiphyses of the humeral and femora. The middle and distal phalanges are poorly ossified. Our cases were the third and sixth female and seventh male child of the parent with 4 normal girls and a normal boy. The affected infants are stillborn and after detailed examination of the last two bodies a diagnosis of SRPS type II (Majewski) was confirmed. Cleft lip and palate are the most important characteristics in distinguishing the syndrome of Majewski type from the others. Because of the rarity of this syndrome and its recurrence, we wanted to announce our tree cases from the same family. The chromosome analysis of the fifth fetus was performed via cordocentesis at 36. Week of gestation revealed 46XX. Because of the autosomal recessive inheritance we decided not to do the chromosomal analysis. The mother of an affected fetus must be told of the risks for subsequent pregnancies. Accurate measurements of limb length can be made and it should be possible to detect skeletal dysplasia earlier than before. Perhaps femoral length should be measured routinely as is the biparietal diameter.

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