CASE REPORT

Prenatal Diagnosis of Short Rib-Polydactyly Type II by Ultrasound in Three Consecutive Pregnancies

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Introduction

Short rib–polydactyly syndrome (SRPS) type II is a rare lethal form of skeletal dysplasia, characterized by polydactyly, short limbs, short and horizontal ribs, a small ovoid tibia and major organ anomalies [1]. Prenatal diagnosis by genetic testing is not available; ultrasound plays an important role. With no prior history usually these cases are detected late in pregnancy; postnatal examination helps in definitive diagnosis and genetic counseling so that the patient can come early in subsequent pregnancy for prenatal diagnosis. We report a case which was followed in three consecutive pregnancies.

Case report

A 22-year-old, non-consanguineous, primigravida came to fetal medicine clinic at 34 week gestation with short limbs on ultrasound. The targeted scan showed polyhydramnios

Fig. 1 Baby with polydactyly, cleft lip and narrow thorax, infantogram showing oval tibia, polycystic kidneys, histopathology showing cortico-medullary cysts

with severe micromelia in all long bones (limb bones were 4 standard deviations below mean for gestation), there were no bending, no fractures, normal mineralization of bones, long and narrow thorax, thoraco-abdominal ratio was 0.4, and femur to abdominal circumference ratio was less than 0.16, all suggesting lethal skeletal dysplasia. There was polydactyly present in bilateral upper and lower limbs. On examination of other systems, bilateral kidneys were found to be echogenic, their size between 50th and 95th centile for gestational age, and bladder was seen. The examination of skull and spine was normal. The provisional diagnosis of SPRS was made. She delivered vaginally at 37 weeks, baby was male, stillborn weighing 1.5 kg, and external examination showed broad nose, depressed nasal bridge, low-set ears, small median cleft lip, no cleft palate, no natal teeth, normal tongue. The thorax was less than 5th centile for gestation, and abdomen was protuberant. There was postaxial polydactyly in all four limbs, and the nails were normal. On internal examination, the kidneys were bilaterally polycystic with small one millimeter cysts. The histopathological examination of the kidney showed cortico-medullary cysts. The infantogram showed short limb bones with flat epiphysis; the tibia was exceptionally small and oval in shape. The iliac bones appeared normal, were not hypoplastic and had no bony spurs (Fig. 1). The diagnosis of short rib–polydactyly type II or Majewski syndrome was made. As it has autosomal recessive inheritance, the chances of recurrence are up to 25 %.

During her second conception, patient came at 16 weeks, on ultrasound the digits appeared normal in number, the long bones including tibia were normal in length, and bilateral kidneys were normal. The baby was normal at birth. In the third pregnancy, the patient again came at 16 weeks for level II scan which showed

polydactyly, short long bones and absent tibial bone, with bilateral echogenic kidneys. The possibility of recurrence was explained, and termination followed by autopsy and infantogram confirmed the findings (Fig. 2).

Discussion

Short rib–polydactyly syndrome is a lethal skeletal dysplasia with marked limb reduction, narrow constricted thorax, short horizontal ribs, pre- and postaxial polydactyly and frequent cardiac defects and cystic renal dysplasia. The short rib–polydactyly syndromes must be distinguished from asphyxiating thoracic dysplasia and chondro-ectodermal dysplasia (Ellis–van Crevald syndrome). They are all inherited in an autosomal recessive fashion, but they may have different prognoses: In the latter two, infants may survive the neonatal period and the respiratory symptoms decrease with age. Particularly in cases with no family history, it is important to be able to make a precise diagnosis in order to provide effective prenatal testing.

The positive findings on antenatal USG in our index case were short limbs, narrow thorax, polydactyly and polycystic kidneys. There were micromelia and the absence of encephalocele making the diagnosis of Meckel–Gruber syndrome unlikely. Postnatal autopsy showed there were no features of chondro-ectodermal dysplasia making Ellis–van Crevald unlikely; there were no cleft palate and very short ribs ruling out Mohr Majewski syndrome. The infantogram showed the epiphysis of long bones was flat and not cone shaped, as seen in asphyxiating thoracic dysplasia. The distinctive feature suggesting short rib–polydactyly type II was very small tibia, almost oval in shape in the index case.

Fig. 2 USG showing narrow thorax, echogenic kidneys, short femur, polydactyly. Baby with polydactyly, cleft lip, absent tibia on infantogram

Type 2 SRPS (Majewski syndrome) was first described in 1971 [2]. Considering the limitations of therapy, prenatal diagnosis with selective termination of pregnancy is an important option for couples at risk. Although Chen et al. [3] provided evidence for a correlation of NIMA (never in mitosis gene a)-related kinase 1 (NEK 1) mutation with type II SRPS in their case report, at present the gene responsible for this disorder has not been identified, and therefore, the only method of prenatal diagnosis is the detection of its phenotypic manifestations. In some cases with familial recurrence, the transvaginal sonographic diagnosis at 13–14 weeks of gestation has been reported [4]. Fetoscopy and three-dimensional USG have also been tried in few reports [5, 6]. However, uncertainty about the phenotypic expression of such pathologies means that caution must be exercised when making a diagnosis at this early stage of gestation, and a targeted examination by an experienced sonographer is warranted.

Complete workup and autopsy of index case lead to the diagnosis, and the couple was counseled to come for follow-up. Prenatal diagnosis of this genetic disease was possible by ultrasound in subsequent pregnancies leading to normal outcome in second pregnancy and early diagnosis of anomaly in third pregnancy.

Compliance with Ethical Standards

Conflict of Interest There is no conflict of interest among authors. There is no funding from any source. There is no affiliation of any

authors with any organization with a financial interest, direct or indirect.

Human and Animal Rights There is no research involving human participants and/or animals.

Informed Consent Informed consent of the case was taken.

References

1. Spranger J, Grimm B, Weller M, et al. Short rib-polydactyly (SRP) syndromes, types Majewski and Saldino-Noonan. *Z Kinderheilkd.* 1974;116:73–94.
2. Majewski F, Pfeiffer RA, Lenz W, et al. Polysyndakty lie, verkürzte Gliedmassen und Genital fehlbildungen: Kennzeicheneinselfständigen Syndroms? *Z Kinderheilkd.* 1971;111:118–38.
3. Chen CP, Chang TY, Chen CY, et al. Short rib-polydactyly syndrome type II (Majewski): prenatal diagnosis, perinatal imaging findings and molecular analysis of the NEK1 gene. *Taiwan J Obstet Gynecol.* 2012;51(1):100–5.
4. Gabrielli S, Falco P, Pilu G, et al. Can transvaginal fetal biometry be considered a useful tool for early detection of skeletal dysplasias in high-risk patients? *Ultrasound Obstet Gynecol.* 1998;13:107–11.
5. Lee K, Lee JW, Chay DB, et al. Transabdominal embryo fetoscopy for the detection of short rib-polydactyly syndrome, type II (Majewski), in the first trimester. *J Korean Med Sci.* 2006;21:165–8.
6. Viora E, Sciarone A, Bastonero S, et al. Three-dimensional ultrasound evaluation of short rib polydactyly syndrome type II in the second trimester: a case report. *Ultrasound Obstet Gynecol.* 2002;19(1):88–91.