Journal of Clinical and Biomedical Sciences

Case Report

Case Report Of Short Rib Polydactyly Syndrome

Anshul Singh[1], Anil Kumar Sakalecha[2*], Rajeswari[3], Yashas Ullas L[4]

1. Postgraduate, Department of Radio-diagnosis,Sri Devaraj Urs Medical College, Sri Devaraj Urs Academy of Higher Education and Research, Tamaka, Kolar.
2. Professor, Department of Radio-diagnosis,Sri Devaraj Urs Medical College, Sri Devaraj Urs Academy of Higher Education and Research, Tamaka, Kolar.
3. Assistant professor, Department of Radio-diagnosis,Sri Devaraj Urs Medical College, Sri Devaraj Urs Academy of Higher Education and Research, Tamaka, Kolar.
4. Postgraduate, Department of Radio-diagnosis,Sri Devaraj Urs Medical College, Sri Devaraj Urs Academy of Higher Education and Research, Tamaka, Kolar.

Abstract

Short rib polydactyly syndrome (SRPS) is a group of skeletal dysplasias manifested by short-limb dwarfism, short ribs with thoracic dysplasia and polydactyly. SRPS is an inherited autosomal-recessive disorder with different prenatal sonographic and postnatal clinical, histological and radiologic findings. In this report, we present a case of SRPS type 3 (Verma-Naumoff) with thoracic hypoplasia, short limbs and postaxial polydactyly in a 30-week fetus.

Keywords: Short rib polydactyly syndrome, Verma-Naumoff, Ultrasonography, Narrow thoracic cage

Introduction

There are 7 types of short-rib polydactyly syndrome (SRPS) classified in the literature according to clinical, pathological and radiological differences. Type 1 (Saldino-Noonan), type 2 (Majewski), type 3 (Verma-Naumoff) and type 4 (Beemer-Langer) are accepted to be the 4 main SRPSs. However, Spranger and Maroteaux reported 2 other types in 1990. [1–6] The frequency is 1 in 200,000 live births or less. In the current literature, different variants of SRPS have been identified with overlapping ultrasonographic, pathological and radiological features. [7–10] In this case report, we aim to discuss a case of SRPS type 3 (Verma-Naumoff)

Case Report

A 25 year-old woman, gravida 2, para 0, abortion 1 with 30 weeks of gestation as per her last menstrual period, was referred to our institution with a prenatal sonographic suspicion of fetal skeletal dysplasia. Her previous pregnancy was terminated at 23 weeks in view of cardiac defects, details of which were not available. Personal history: 1st degree consanguinous marriage.

Sonographic evaluation revealed B Parietal Diabetes, Head Circumference (fig. 1) and AC measurements consistent with 30 weeks. Intraorbital distance was normal. Spine, facial profile, lips and chin revealed no abnormality. Fetal thorax was narrow with short ribs (bread loaf appearance) and cardia showed single atrium. Two vessel umbilical cord was noted (fig. 2). Femur Length and humerus measurements corresponded to 20 weeks (fig. 3 & 4). Tibia, fibula and radius were also short and postaxial polydactyly was noted in both hands. Foot length was normal.

Chest abdominal circumference < 5th percentile, heart area–chest area ratio < 5th percentile, chest –trunk length ratio < 0.32, femur length–abdominal circumference ratio = <0.16 Femur length–foot length ratio <1.

Termination of pregnancy was induced with 50 g misoprostol (every 4 h q.i.d.) and oxytocin infusion. After termination of the pregnancy, fetus was consistent with 30 weeks according to weight and body measurements and had normal head and face. The thorax was narrow and bell-shaped because of short ribs and sternum; all extremities were short. Polydactyly (7 fingers) was present in both hands (fig.

***Corresponding Author**
Dr. Anil Kumar Sakalecha
Professor, Department of Radio diagnosis,
Sri Devaraj Urs Medical College,
Sri Devaraj Urs Academy of Higher Education and Research, Tamaka, Kolar-563101
Mobile No : 9844092448
E-mail: dranilsakalecha@gmail.com
Conflict of Interest: None
Financial Aid: Nil

5); feet were normal. External genitalia were of normal male type.

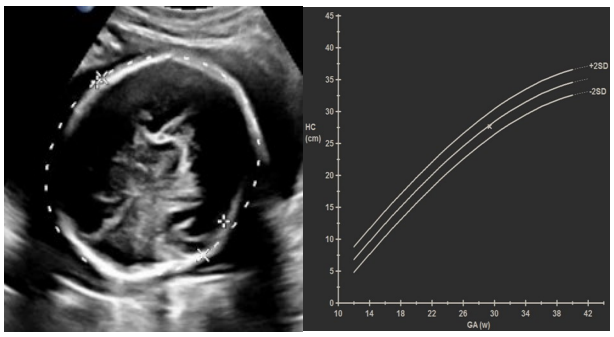

Fig. 1: HC and BPD corresponding to 30 weeks

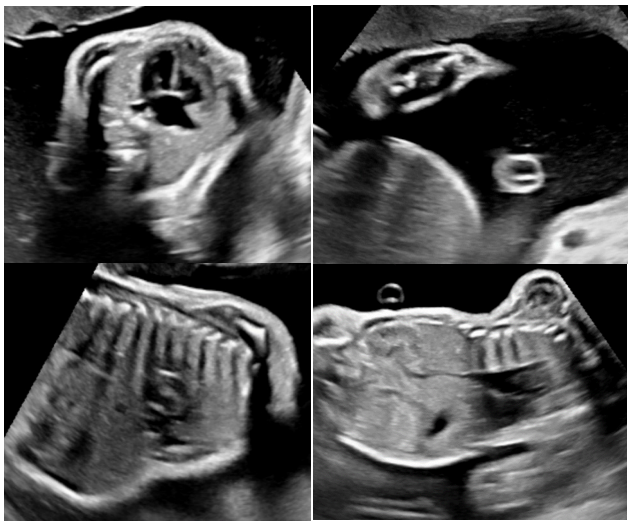

Fig 2: From top clockwise: 3 chamber cardia, 2 vessel cord, coronal and sagittal views of narrow thorax showing bell shape thorax and bread loaf appearance respectively.

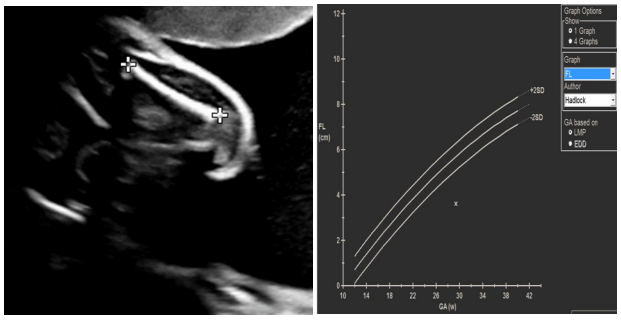

Fig 3: Femur length below 6 SD

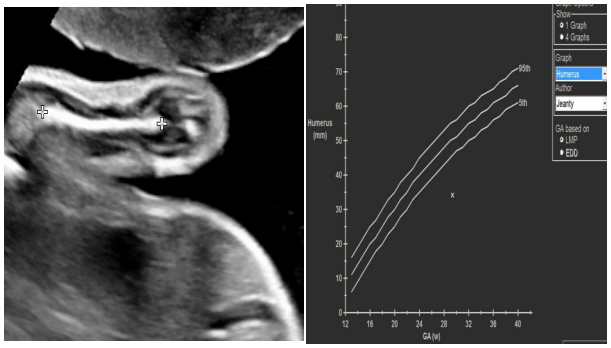

Fig 4: Humerus length below 6 SD

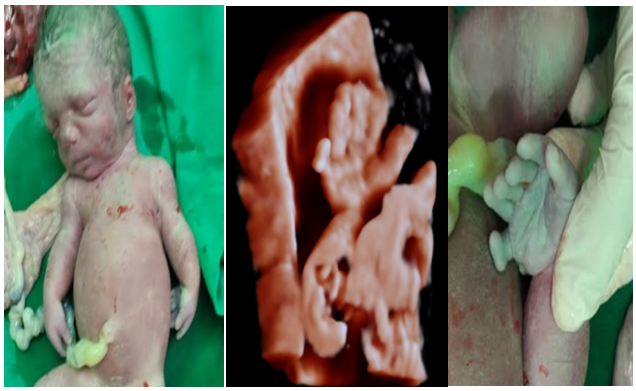

Fig 5: Terminated fetus with normal head and face, postaxial polydactyly in 3D USG image and corresponding fetus hand.

Discussion

Seven subtypes of SRPS, which are rarely encountered and difficult to diagnose, are described in current literature. The differences between types have not yet been determined at the molecular level. There are no specific biochemical and histopathological markers to differentiate between different subtypes of SRPS; however, studies support the value of ultrasonography in differential diagnosis. [11]

It is believed that different subtypes of SRPS are not single entities, but rather part of a continuous spectrum with variable expressivity. [12] The SRPS types which overlap most are types 1 and 3 with similar clinical phenotypes.

Type 3 is distinguished from type 1 by 50% male predominance, often no polydactyly, coarctation of the aorta, hypoplastic left and right side chambers of the heart, wide metaphyseal ends of the tubular bones, fan-shaped iliac bones and less severe radiologic manifestations. [4, 5, 7, 11, 13, 14]

Type 1 is distinguished from type 3 by more genitourinary, cardiovascular and gastrointestinal anomalies, female predominance, pointed femurs on both ends, flattened acetabular roofs, hypoplastic iliac bones, weak corticomedullary demarcation border and absent fibulae. [2, 8, 13, 14]

Ellis-Van Creveld syndrome and asphyxiating thoracic dystrophy (ATD) are types in which thoracic dysplasia is combined with short extremities and are recognized as autosomal-dominant syndromes. [9]

In prenatal ultrasonographic evaluation, our case showed short ribs, narrow chest, short extremities, postaxial polydactyly with short tubular bones. Life expectancy- Perinatal mortality because of respiratory distress due to pulmonary hypoplasia- which is lethal. Narrow thoracic cage is the reason for hypoplastic lungs. Postmortem examination was not done in our case due to lack of willingness from family.

Diagnosis of our case was type 3 SRPS.
The usual mode of inheritance is autosomal recessive and genes involved can be

a) Missense Mutation c.8077G > T (p.Asp2693Tyr) of paternal origin in DYNC2H1.
b) Frameshift Mutation c.11741_11742delTT (p.Phe3914X) of maternal origin in DYNC2H1.

Advances in molecular characterization in the future will facilitate differentiation between the SRPS types with certainty.

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