IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 18, Issue 10 Ser.8 (October. 2019), PP 41-43
www.iosrjournals.org

Short Rib Polydactyly Syndrome Type 2 A Case Report

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Abstract: *short rib polydactyly syndrome is a group of rare lethal skeletal dysplasias, which occur in about 2.5 to 3.3 per 10,000 births.A 24 year old primigravida was admitted in MDM hospital at 24 weeks of gestation with preterm premature rupture of membrane.On USG - breech foetus with anhydramnios with severe micromelia with narrow chest ,hypoplastic lungs ,dolicocephlic head ,dysplastic kidneys ,gross ascites ,thickened nuchal fold suggestive of skeletal dysplasia.After delivery of alive baby with very low birth weight and poor APGAR score baby died soon and after consent x-ray and autopsy done which showed short limbs , narrow thorax and polydactyly . radius ,ulna ,tibia and iliac bones were hypoplastic .karyotyping was done.Rare skeletal dysplasis like short rib polydactyly syndrome should be reported early and to avert maternal jeopardy and for judicious management .*

Date of Submission: 04-10-2019 Date of Acceptance: 21-10-2019

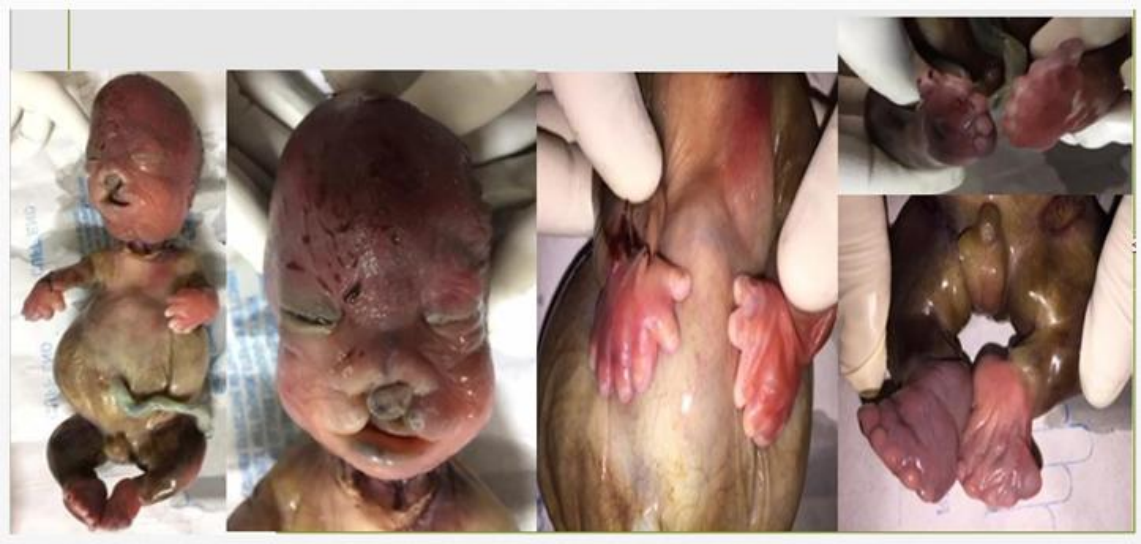
I. Introduction

- Short rib-polydactyly syndrome (SRPS) is 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).
- Its birth prevalence is 2.5 to 3.3 per 10,000 births.(1)
- We reported a case of that syndrome without a previous family history of congenital defects.

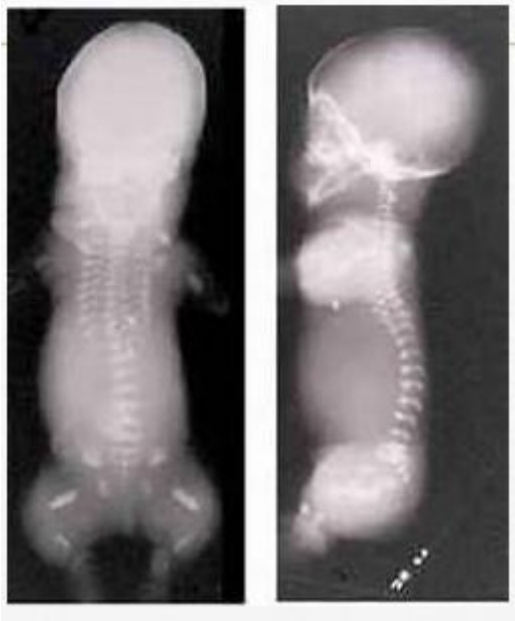
II. Case Report

- A 24-year-old primigravida woman was admitted in our hospital at 24 weeks of gestation because of rupture of membrane and absent amniotic fluid report in the ultrasonography. 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 anomaly scan at 19 weeks of gestation showed a live breech foetus with anhydraminos and severe micromelia with narrow chest, Hypoplastic lungs, Dolichocephalic head, Dysplastic kidneys, Gross ascites, Thickened NF s/o Lethal skeletal dysplasia. Placenta was normal with a triple vessel structure cord.
- Preterm vaginal delivary because of prolonged rupture of membrane resulted in an infant weighing 900 gm with a poor Apgar score. The infant had short limbs, narrowed thorax and polydactyly. Radius, ulna, tibia, and iliac bone were hypoplastic. infant died after 30 mins. On histopathological examination placenta and cord were normal, Karyotyping was normal. Negative VDRL and TORCH profile and normal blood sugar was revealed in subsequent laboratory studies. On fetal autopsy the diagnosis of SRPS type 2 (Majewskisyndrome) with cleft lip and cleft palate was made.

External features of newborn showing gross ascites with cleft lip and short limbs



Babygram showing short horizontal ribs with all four short limbs.



III. Discussion

- Skeletal dysplasia represents 1 to 3.5% of the foetuses detected sonographically with congenital malformations. Its birth prevalence is 2.5 to 3.3 per 10,000 births. Many of the foetuses with this anomaly do not survive till term. (1)
- Short rib-polydactyly (SRPS) is a rare skeletal dysplasia that is manifested with short limbs, 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. (2)
- SRPS is an autosomal recessive inherited syndrome with a recurrent rate in 25% of cases.
- 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 metaphysialirregulation 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).
- Type II SRPS (Majewski syndrome) was first described in 1971 (Majewski et al., 1971). These patients have very short ribs, severe pulmonary hypoplasia, micromelia, and polydactyly. A distinguishing feature of the syndrome is the presence of a median or midline cleft lip with or without cleft palate. These patients

also have a very high frequency of central nervous system abnormalities. The most common central nervous system abnormalities seen in Majewski syndrome include pachygyria, small cerebellar vermis, and absent olfactory bulbs. Other central nervous system changes that have been demonstrated in type II SRPS include arachnoid cysts, agenesis of the corpus callosum, and arrhinencephaly . In addition to the midline cleft lip and cleft palate, patients with Majewski syndrome can have a cleft tongue, oral frenulae, natal teeth, and abnormalities of the epiglottis.

- 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)
- All affected infants with short-rib polydactyly syndrome have severe pulmonary hypoplasia that prevents extrauterine survival.

IV. Conclusion

Awareness about potential occurrence and subsequent early and prompt judicious management approach may avert maternal jeopardy to some extent and help us to better understand about this lethal malformation.

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Dr Mangal Chand Yadav. "Short Rib Polydactyly Syndrome Type 2 A Case Report." IOSR Journal of Dental and Medical Sciences (IOSR-JDMS), vol. 18, no. 10, 2019, pp 41-43.