CASE REPORT - ANATOMY

Short Rib - Polydactyly Syndrome Associated With Oro-Facial-Digital Syndrome

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ABSTRACT

Introduction: Short Rib - Polydactyly Syndrome (SRPS)is a group of malformations inherited as autosomal recessive condition. It is well recognized but confusion exists over the characteristics and nomenclature as different phenotypic findings are recorded due to variable gene expression. It may be associated with oro-facial-digital syndrome as they are mild and severe expressions of the same disorder.

Case Report: A 28-week old fetus was diagnosed to have Short Rib - Polydactyly Syndrome following an antenatal USG screening of a multigravida by the OBG department, VMKVMCH, Salem. The pregnancy was terminated fearing other major congenital anomalies usually associated with the syndrome.

Aim: To confirm the diagnosis/ arrive at a more precise diagnosis by radiological & morphological(gross & histological) study.

Objectives: A. To identify skeletal abnormalities in the X-ray. B. To measure various morphological parameters of the fetus. C. To interpret histopathological slides.

Materials and Method: The following were taken for the study –28-week dead fetus, USG Images, X-Rays, measuring tape & Vernier's calipers, histopathological slides.

Result:USG images indicated polydactyly & syndactyly and short ribs.Gross inspection of the fetus showed a number of anomalies, notably short limbs, polysyndactyly, cleft lip and hairy nevus. X-ray showed tibia shorter than fibula.All the morphological parameters were lesser than normal values.Histopathology indicated mononuclear infiltration of cerebellum, alveolar hemorrhages in lungs& inflammatory changes in the liver.

Conclusion: Following the study the diagnosis arrived at is Short Rib – Polydactyly Syndrome associated with Oro-Facial-Digital Syndrome.

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INTRODUCTION:

Short Rib - Polydactyly Syndrome (SRPS)is a group of malformations inherited as autosomal recessive condition characterized by the triad of micromelia, polydactyly and short horizontal ribs.[1]It is well recognized but confusion exists over the characteristics and nomenclature as different phenotypic findings are recorded due to variable gene expression.[2]Oral-Facial-Digital Syndrome (OFDS) is an extremely variable congenital condition and its diversity has led to considerable investigations and discussions.[3]Cases have been reported where constitutional dwarfism with short limbs, short ribs and polydactyly presented with additional congenital anomalies including cleft palate, notching of upper lip and small tongue[4]. Phenotypic overlap between SRPS and OFDS has been reported in the literature, and it has been suggested that the two syndromes may be mild and severe expressions of the same autosomal recessive disorder.[4]

CASE REPORT:

The female patient aged 28 years, had a parity index of G-3, P-2, L-1 with a marital history of II degree consanguinity. Her obstetric history indicated in her first pregnancy which was unbooked, she came at 7 months antenatal period with pregnancy induced hypertension. The scan showed lethal anomalies. The pregnancy was terminated. The dead born female baby had very short limbs, polydactyly and cleft lip. Her second pregnancy was uneventful & she delivered a normal healthy female baby.

In the present pregnancy, she had an early USG done at 4 months which showed a single live intrauterine fetus of 15-17 weeks. She developed polyhydramnios at 7th month. Repeat scan was done. The biparietal diameter (Fig. 1), head circumference & abdominal circumference corresponded to 28 weeks. Femoral length, tibial length & humeral length corresponded to 20 weeks. This was an incidental finding of shortened extremity during a routine USG. Other findings were narrow thorax with short ribs, polydactyly (Fig. 2), syndactyly and ascites. A diagnosis of SRPS was made.The pregnancy was terminated fearing other major congenital anomalies usually associated with the syndrome.

Figure 1. Fetal ultrasound – Biparietal diameter.

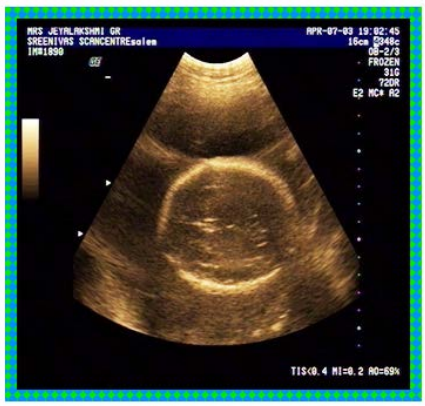
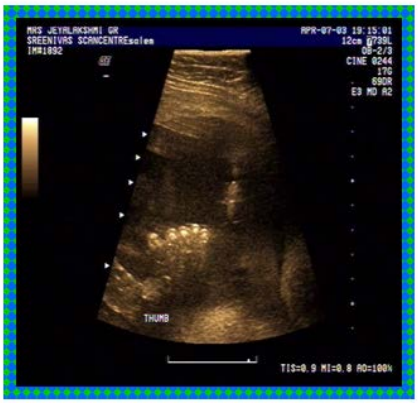


Figure 2. Fetal ultrasound showing polydactyly of left hand.



AIM:

In order to confirm the diagnosis and arrive at a more precise diagnosis,a radiological & morphological(gross & histological) study was conducted on the dead female fetus.

OBJECTIVES:

A. To identify skeletal abnormalities in the X-ray.

B. To measure various morphological parameters of the fetus.
C. To interpret histopathological slides.

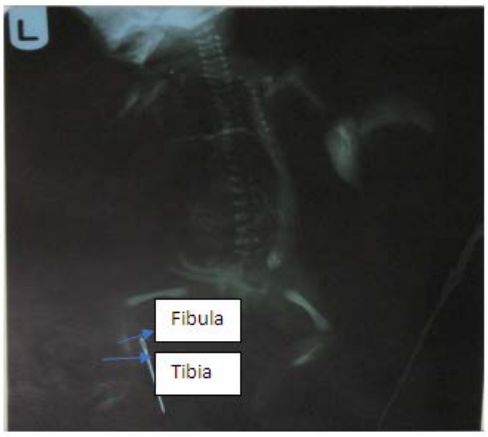
MATERIALS AND METHODS:

The following were taken for the study - the28-week dead female fetus, USG images, X-rays, measuring tape & Vernier's calipers, histopathological slides.

RESULTS:

It was difficult to ascertain short ribs on the X-ray. The tibia appeared shorter than fibula in both lower limbs (Fig. 3)

Figure 3. X-Rayshowing Fetus with short tibia & longer fibula.



On gross examination (Fig. 4), the fetus was found to have the following prominent facial features: low set ears, a median cleft upper lip and micrognathia. The other features were, flat bridge of the nose, hypertelorism, alveolar clefts and unilateral incomplete cleft palate. A hairy nevus was seen in the lower lumbar region (Fig. 5). The features in the limbs (Fig. 6 A, B, C, D) were: Short limbs, talipesequinovarus deformity of both feet and preaxial& postaxial polysyndactyly of both hands and feet.

Figure 4. Fetus with facial anomalies.

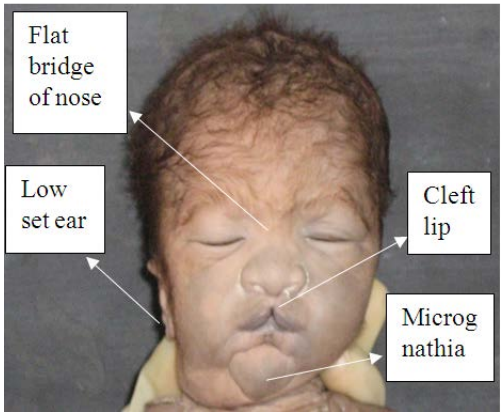
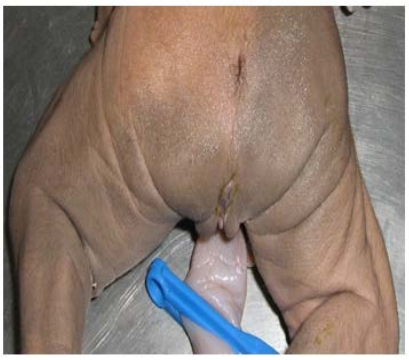


Figure 5. Hairy nevus in the lower lumbar region of the fetus.



Morphological study was extended to the measurement of various parameters (Table 1, 2. 3).Major parameters were found to be marginally lower in the fetus than normal values (Table 1).

Figure 6. Polydactyly of both hands (A & B), polysyndactyly in the feet (C), short lower limbs & talipes equinovarus deformity of both feet (D).

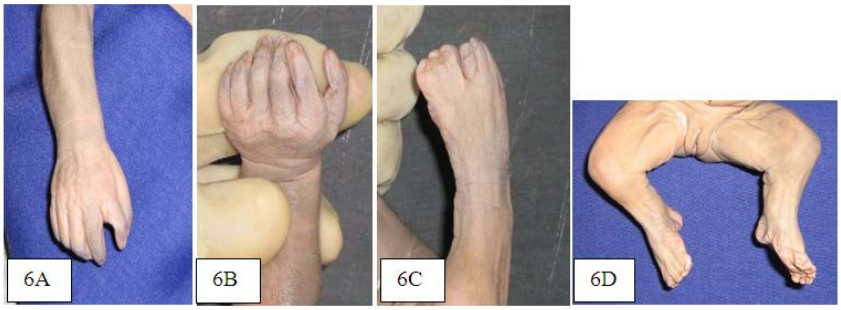


Table 1. Comparison of various measurements of the fetus with normal values.

AGE	mm							
28 weeks	Crown–heel length		Foot length		Biparietal diameter		Abdominal circumference	
	Normal	Specimen	Normal	Specimen	Normal	Specimen	Normal	Specimen
	395	300	55	Rt-50 Lft-56	75	73	250	320

*Normal values have been adapted from O'Rahilly. [5]

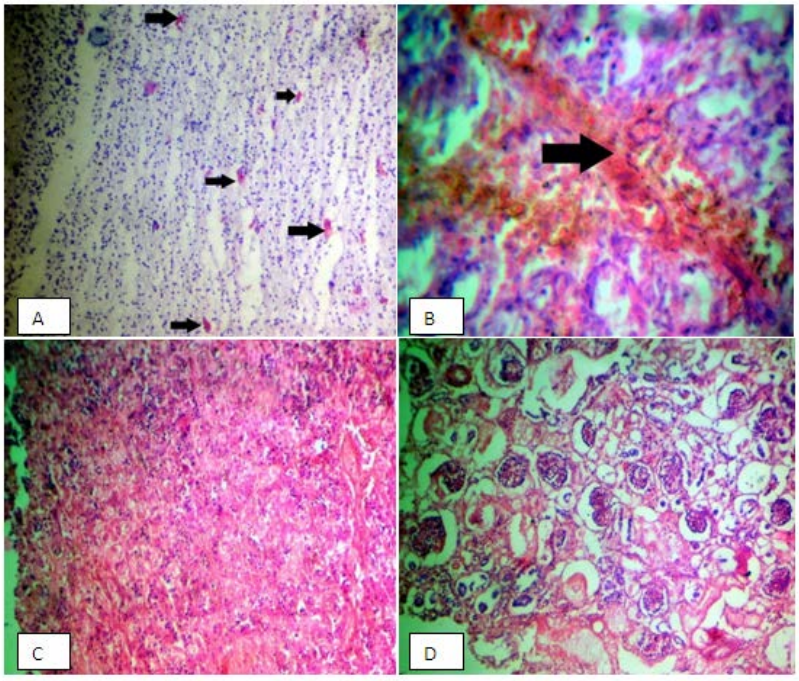
Table 2.Various parameters measured in the 28-week female fetus.

Sr. No.	Parameters	Measurement cms
1	Crown-rump	27.5
2	Lft iliac crest-lateral condyle of femur	8
3	Lftlat condyle femur-heel	6.5
4	Rt iliac crest-lateral condyle of femur	7
5	Rtlat condyle femur-heel	6
6	Rt acromion-olecranon	5
7	Rt olecranon-tip of middle finger	7.5
8	Lft acromion-olecranon	5
9	Lft olecranon-tip of middle finger	8
10	Nasion-chin	5
11	Vertex-nasion	8.3
12	Inion-nasion	16.3
13	Biauricular distance over vertex	17.1
14	Biauricular distance over orbit	13

Table 3.Various parameters measured in the 28-week female fetus.

Sr. No.	Parameters	Measurement cms
15	Nasion-tip of nose	2
16	Rt Mandibular condyle-chin	4.5
17	Lft Mandibular condyle-chin	5.5
18	Distance b/w two medial canthi	2.2
19	Distance b/w two lateral canthi	6.5
20	Dist b/w angles of mouth	1.6
21	Rt auricle Height Breadth	3 1.7
22	Lft auricle Height Breadth	2.9 2
23	Rt angle of mouth-base of mandible	2
24	Lft angle of mouth-base of mandible	1.7
25	Bialar distance	1.8
26	Rt root of auricle-base of mandible	4
27	Lft root of auricle-base of mandible	2.2

Figure 7. Mononuclear infiltration & capillary (arrows) proliferation in cerebellum (A). Alveolar hemorrhages (arrow) in lung (B). Inflammatory cells in liver (C). Normal kidney (D).



Histopathology indicated mononuclear infiltration of cerebellum, alveolar hemorrhages in lungs & inflammatory changes in the liver. The kidneys were normal (Fig. 7 A, B, C, D).

DISCUSSION:

SRPS are classified into various types. The fetus described here supports the hypothesis that clinical variability of this syndrome is indeed very wide. The fetus was noted to have short rib, polydactyly as well as cleft lip & cleft palate, micrognathia, hypertelorism and low set ears. Hence it was diagnosed to have SRPS associated with OFDS.

Collectively, SRPS & OFDS share a common underlying defect in primary cilium function and form a subset of the ciliopathy disease spectrum. Central nervous system, respiratory system & kidneys are affected in the SRPS & OFDS spectrum of conditions.

Mutations in the *OFD1* gene have a deleterious effect on primary cilia and alter several signalling pathways during development, which accounts for the wide variation in phenotypes and association with related ciliopathies. Hence, careful physical and genetic workups are necessary. The *OFD1* gene appears to play a critical role in the early development of many parts of the body, including the brain, face, limbs, and kidneys.[6] As the parents in this case are consanguineous, transmission of the syndrome is an autosomal recessive trait. The previous history of an affected offspring with an almost similar condition indicates phenotypic variability of this condition.

CONCLUSION:

Following the study the diagnosis arrived at is Short Rib – Polydactyly Syndrome associated with Oro-Facial-Digital Syndrome. The family needs to be provided with genetic counseling, after drawing out a pedigree chart which would help them plan a subsequent successful childbirth.

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