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Case Report

A RARE CASE OF ULBRIGHT-HODES SYNDROME

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ABSTRACT

A primigravida presented with severe oligohydramnios and breech presentation with fetal distress. She underwent emergency cesarean section. The baby had multiple anomalies including Potter's facies, phocomelia, mesomelia, renal dysplasia and anomalies of external genitalia. Baby died immediately after birth. MRI and foetal dissection showed findings correlating with Ulbright-Hodes syndrome or Renal dysplasia – limb defects syndrome (RL Syndrome). This is a very rare autosomal recessive congenital disorder. Here we report a case of Ulbright-Hodes syndrome as only four cases have been reported so far.

INTRODUCTION

Ulbright-Hodes syndrome is characterized by renal dysplasia, growth retardation, phocomelia or mesomelia, anomalies of external genitalia and potter like facies (Ulbright *et al*., 1984). Ulbright Hodes syndrome is a very rare anomaly with a prevalence of <1/1000000 (http://rarediseases.info.nih.gov/GARD/Disease.aspx?PageID=4&disease). This syndrome was described in three infants, all of whom died shortly after birth. The mode of transmission appears to be autosomal recessive (Schrander-Stumpel, 1990). A 19 years primigravida was referred to the department of Obstetrics at 34 weeks pregnancy with severe oligohydramnios with breech presentation having foetal distress. An emergency LSCS was done. A male baby was born, died immediately after birth. Multiple congenital abnormalities were noted including potters facies, phocomelia of all four limbs and external genital abnormality.

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After obtaining a written consent from the parents, radiological imaging like x ray and MRI scan was done. Full autopsy examination of the foetus was done in the department of anatomy. Absence of ulnae and hypoplastic radius Fig.1 A Absent fibulae Long thin ribs, anterior rounding of lumbar vertebrae Fig. 1 B Micrognathia Scoliosis.

MRI findings-Fig.1 C & D

Edema of scalp, Brain: Calcific foci in right cerebral hemisphere Face: micrognathia craniofacial dysmorphisms, depressed nasal bridge. Thorax: slender ribs, pleural effusion Abdomen: liver displaced to midline, both kidneys not visualized, ascites present Spine: scoliosis convexity to right Limbs: bilateral distal limb aplasia.

Findings on autopsy

Weight-1800gms CR length-29cms Head circumference-32.2cms. Thoracic circumference-28cm Abdominal circumference-25cms External observations: Figure 2 Wide palpebral fissure, depressed nasal bridge, low hair line, short

neck, undescended testes and scoliosis on right side. Both hands show phocomelia, mesomelia. Both lower limb show mesomelia, Anal opening present External genitalia appears abnormal.

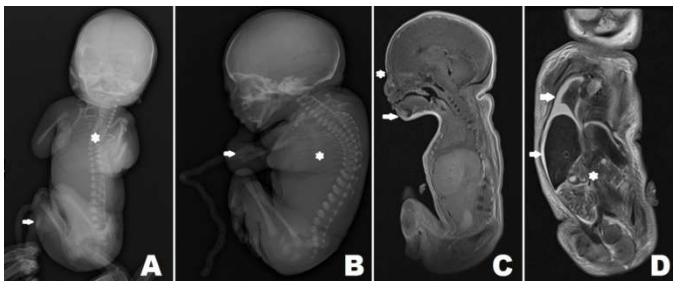

Figure 1. Fetus diagnosed as Ulbright - Hode Syndrome. Radiological Imaging was performed (plain radiographs and MRI on 1.5T scanner). A) Frontal radiograph showing scoliosis (*) and mesomelia in the lower limbs (arrow). B) Lateral radiograph showing long, slender ribs (*) and mesomelia in the upper limbs (arrow). C) T1 sagittal MR image showing craniofacial dysmorphism with micrognathia (arrow) and mildly depressed nasal bridge (*). D) T2 coronal MR image showing pleural effusion and ascites (arrows) and bilateral empty renal fossae (*).

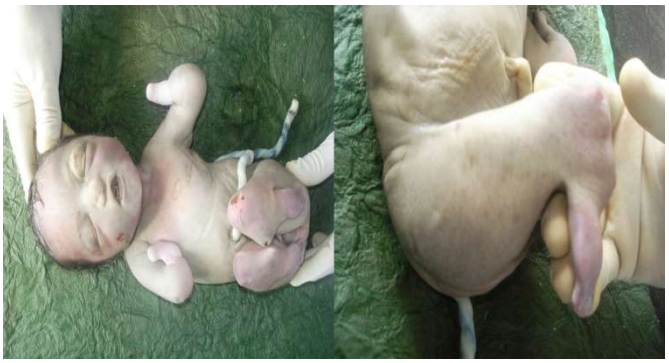

Figure 2. Wide palpebral fissure, depressed nasal bridge, low hair line, short neck, undescended testes and scoliosis on right side. Both hands show phocomelia, mesomelia. Both lower limb show mesomelia. External genitalia appears abnormal.

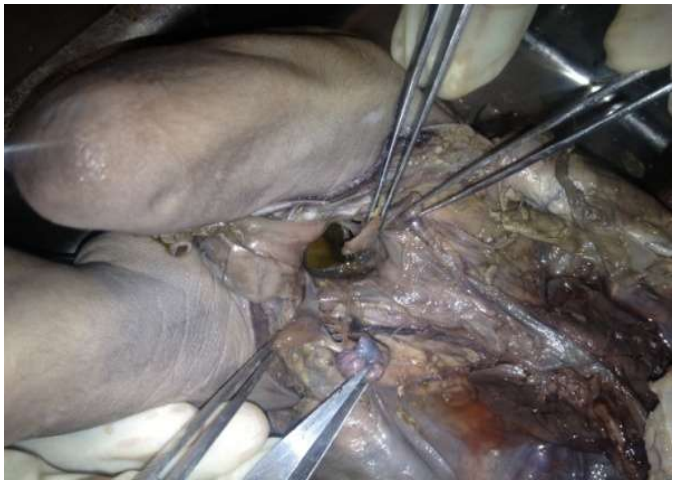

Figure 3. Small hypoplastic unascended kidneys seen in supra-pubic region in close conjunction with undescended testes bilaterally.

DISCUSSION

Ulbright *et al* described a new syndrome in 1984 in a male neonate with mesomelia, radiohumeral fusion, renal dysplasia, high palate, congenital shortness of forearms with fusion of humerus and radius, absence of ulnae and severe hypoplasia of radius, absent fibulae, bilateral talipes equinovarus, long thin ribs, anterior rounding of lumbar vertebrae, potters facies (Ulbright *et al*., 1984). In Schrander Stumpel *et al*. (1990) reported two siblings a male and a female, who died of respiratory failure shortly after birth. The siblings had, Phocomelia of upper limbs, rib anomalies, renal dysplasia, external genital abnormalities, potter like facies with growth restriction. Edith Potter studied 5000 autopsies on fetuses and newborns. She described typical facial look with bilateral renal agenesis, pulmonary hypoplasia associated with oligohydramnios (Dunn, 2007). In 2008 Maruotti *et al* described a successful prenatal diagnosis of the condition at 21 weeks of gestation with a few additional intra cranial anomalies apart from those described above (Dunn, 2007). Ulbright Hodes syndrome also known as Renal dysplasia-limb defects syndrome is very rare. In the Online Mendelian Inheritance in Man database coexistence of renal dysplasia and limb reduction defects was found in very few cases (McKusick, 2010).

But all the abnormalities described in our case were correlating with Ulbright –Hodes syndrome (OMIM-266910) (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB &Expert=3404). International code of diseases ICD-10 classified it as Q87.7. Orphanet-about rare diseases ORPHA3404) (http://www.wrongdiagnosis.com/medical/rl_ syndrome.htm). It is inherited in an autosomal recessive manner. The defective gene responsible for the disorder is located on an autosome, and two copies of the defective gene are required for the disorder to manifest (http://www.orpha. net/consor/ 2010). This syndrome is lethal because severe renal dysplasia resulting in oligohydramnios and pulmonary hypoplasia which is fatal (Schrander-Stumpel, 1990). The presence of most of the characteristic features including renal dysplasia, mesomelia and oligohydramnios with pulmonary hypoplasia suggests that our case was none other than Ulbright-Hodes syndrome.

Conclusion

We can say that rare anomalies may many times be missed in fetuses of parents with negative history of any congenital anomaly. A high degree of suspicion and careful sonography scan or a targeted scan can help us detect such anomalies at an early stage.

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