CASE REPORT

Urorectal Septum Malformation Sequence in a Newborn with VACTERL Association

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ABSTRACT

Urorectal septum malformation sequence (URSMS) is an extremely rare anomaly, consists of multiple system anomalies including ambiguous genitalia, absence of a perineal opening, an imperforate anus, and urological, colonic and lumbosacral defects. We describe a newborn with characteristic URSMS who also had features of congenital varus deformity of leg, polydactyly, tracheo-oesophageal fistula, cardiac defect, anal atresia and hydronephrosis in antenatal ultrasound characteristic of VACTERL association.

Key words: *Urorectal septum malformation sequence. VACTERL association. Neonate.*

INTRODUCTION

The urorectal septum malformation sequence (URSMS) is characterized by the absence of urethral, vaginal and perineal openings, ambiguous genitalia, internal genitourinary anomalies, and imperforate anus.[1] Although URSMS was first described in 1542, the pathogenesis still remains unclear.[1] The VACTERL association consists of vertebral defects, anal atresia, cardiac defects, tracheo-oesophageal fistula, renal and limb defects.[2] Though features of URSMS and VACTERL association overlap, but we are reporting this case of a newborn having features of both of these syndromes.

CASE REPORT

The case was born per vaginally at 34th week of gestation and was the first born of non-consanguineous healthy young parents. There were no known malformations, spontaneous abortion in the family. There was no history of any medication, alcohol intake, smoking, fever, or viral disease during pregnancy. The mother gained no weight and her abdomen was small for the gestational age during pregnancy. Fetal movements were first noted in the 18th week of gestation. The first ultrasonographic examination performed in the 20th week of gestation revealed a single fetus and repeat sonographic examination at 32 weeks showed oligohydramnios and mild bilateral hydronephrosis. There were no other gross congenital anomalies. The baby was born preterm due to premature rupture of membranes (PPROM) and beginning of spontaneous labour. Birth weight was 1800 grams. The neonate was of undetermined gender with normal placenta, membranes and umbilical cord.

Physical examination revealed head circumference of 25 cms and large fontanelles. There was no facial dysmorphism and neck was normal. The chest circumference was 18 cms. There were polydactyly of left upper limb (Figure 1) along with congenital varus deformity of lower limbs. The anogenital area showed complete absence of genitalia and anus (Figure 1). There were features of tracheo-oesophageal fistula as shown by excessive frothing from mouth and inability to pass nasogastric feeding tube with absence of air bubbling sound in stomach. The cardiovascular examination revealed a pansystolic murmur in the left lower parasternal area suggestive of ventricular septal defect. Though antenatal ultrasound detected bilateral hydronephrosis but kidneys were not palpable.

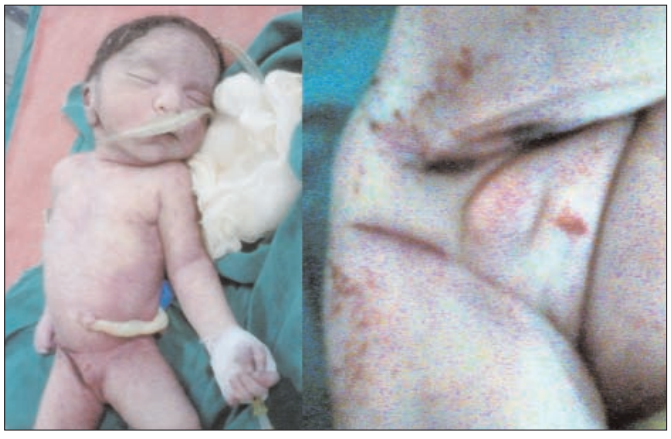

Figure 1: Photograph showing polydactyly of left upper limb along with complete absence of genitalia and anus.

On the basis of above anomalies, a diagnosis of URSMS with VACTERL association was made. The

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child had severe respiratory distress since birth and was oxygen dependent. The baby died after 3 hours of delivery. As permission for postmortem radiograph, autopsy and chromosomal analysis was refused, we could not investigate the baby further.

DISCUSSION

This patient had congenital varus deformity of lower limbs (may be due to vertebral defect), anal atresia, clinically cardiac defect and tracheo-oesophageal fistula, bilateral hydronephrosis (in antenatal ultrasound) and polydactyly (limb defect). All of these are classical features of spectrum of VACTERL association. This patient also had absent perineal and anal openings with ambiguous genitalia which are included under the 'full URSMS sequence'.[3] Those who have a single perineal or anal opening draining a common cloaca with an imperforate anus are called 'partial URSM sequence'.[3] The primary defect in this condition appears to be a deficiency in caudal mesoderm leading to the malformation of the URS and other structures in the pelvic region.[4] This combination of anomalies represents a recognizable and specific sequence that is due to failure of migration to and/or fusion of the urorectal septum with the cloacal membrane. This, in turn, leads to persistence of the cloaca and cloacal membrane and failure of normal differentiation of the external genitalia. Persistence of the cloacal membrane results in absence of the urethral and vaginal openings and an imperforate anus.[4]

The URSMS sequence is a lethal condition with long-term survival reported in only 3 of a total of 62 cases.[5] Although defects of the urorectal septum malformation sequence and the vertebral defects, anal atresia, tracheo-oesophageal fistula, renal defects and radial dysplasia association overlap, some authors believe that they are separate entities.[6] Urogenital anomalies like unilateral or bilateral cryptorchidism, hypospadiasis and micropenis in males and ambiguous genitalia and bladder exstrophy in females have been frequently described in VACTERL association.[7] VACTERL associations are derivatives of causally non-specific disruptive events acting on the developmental field which could be the entire embryo during first 4 weeks of life.[8] This pattern of malformation generally has a sporadic occurrence in an otherwise normal family. A disruption in differentiating mesoderm in first 4-5 weeks has been suggested to be the basis for such a non-random association.[9]

Differentiating the urorectal septum malformation sequence from vertebral defects, anal atresia, tracheo-oesophageal fistula, renal defects and radial dysplasia association is helpful to develop appropriate clinical investigations and search for the aetiology and pathogenesis of these diseases.

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