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Case Report

Fatal case of classic Potter's syndrome: a case report

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ABSTRACT

Potter's syndrome is a rare complication of oligohydramnios, incidence varies from 1 in every 2,000 to 5,000 fetuses with an average of 1 in 4,000. It is reported in 0.2-0.4% of autopsies in dead new-born who die immediately after birth. Potter's syndrome primarily affects male fetuses (2:1) and is characterized by pulmonary hypoplasia and renal failure. Here, we are presented a case of classic Potter's syndrome with fatal outcome. The baby was a live preterm male, born to a 28 years old multigravida, out of a non-consanguineous marriage via caesarean section. There was absolutely no liquor at the time of delivery. The baby did not cry immediately after birth and required resuscitation followed by mechanical ventilation. Multiple congenital anomalies suggestive of Potter's syndrome were noted including facial features such as low set ears with overturned helix, slanting eyes, redundant fold of skin beneath the cheeks, prominent epicanthal fold, receding chin, flattened broad ear, flat nasal bridge, parrot beak appearance of nose and triangular facies. Chest X-ray showed small volume lung fields suggestive of pulmonary hypoplasia, and both kidneys were absent on ultrasonography indicating bilateral renal agenesis. At 15 min of life, baby developed tachypnea and severe chest retractions and succumbed after 1 hour of birth due to respiratory insufficiency. This case report emphasizes upon the importance of regular antenatal check-ups in every patient as it picks up the suspicious cases, leading to further workup and diagnosis, resulting in a timely decision regarding management.

Keywords: Potters syndrome, Pulmonary hypoplasia, Oligohydramnios, Renal agenesis

INTRODUCTION

Potter's syndrome (or Potter's/oligohydramnios sequence) is a rare complication of oligohydramnios, incidence varies from 1 in every 2,000 to 5,000 fetuses with an average of 1 in 4,000 and a recurrence risk of 3-6% in subsequent pregnancies.[1] It is reported in 0.2-0.4% of autopsies in dead new-born who die immediately after birth.[2] Potter's syndrome primarily affects male fetuses (2:1) due to increased incidence of Prune Belly syndrome and obstructive uropathy secondary to posterior urethral valve in them and is characterized by pulmonary hypoplasia and renal failure.[3] After 16 weeks of gestation, the amount of amniotic fluid which is present, depends mainly on the foetal urine production. Normally during foetal development, foetus continuously swallows the amniotic fluid, which after getting reabsorbed by the gastro-intestinal tract, is again reintroduced into the amniotic cavity by foetal kidneys in the form of foetal urine. If the volume of amniotic fluid is below normal for the period of gestation, oligohydramnios develops. The possible causes could be decreased production of urine which is caused by bilateral renal agenesis or obstruction of urinary tract and prolonged rupture of the membranes.[4] Foetal urine production is crucial for adequate development of lungs, resulting in the expansion of airways and alveoli by exerting hydrodynamic pressure and also by supplying proline (a critical amino acid for lung development). At birth, if the alveoli and lungs are underdeveloped, the neonate will soon land-up in respiratory distress due to pulmonary hypoplasia, which is

the principal cause of mortality in Potter's syndrome. Foetal urine also cushions the foetus from being compressed by the mother's uterus as it grows. The resulting oligohydramnios is the cause of the typical facial appearance of the foetus, known as 'Potter's facies' which consists of a flattened nose, recessed chin, epicanthal-folds and low-set abnormal ears.[5]

The main cause of this condition is unknown; this syndrome has a genetic background in some cases, and is more common in neonates with a positive family history of kidney malformation.[6] It has a fatal outcome and is incompatible with life, but the Potter's sequence due to a non-renal cause has a higher survival rate. Though it is rare, it is believed to be more common because the infants are either stillborn or may die soon after the birth. There is no known method for preventing this deadly disease.[6]

Therefore, an ultrasound screening for oligohydramnios and the absence of the foetal kidneys is recommended for couples with a previous affected pregnancy between 16-18 weeks of gestation, so that the termination of the pregnancy may be offered before it becomes viable. If the baby survives, it has to be resuscitated at delivery and treated for any urinary outlet obstruction, but the outcome is poor.[6] In this report, we present a case of Potters syndrome with fatal outcome.

CASE REPORT

28 years old G5P3L3A1 mother presented to our center in emergency unit with complaints of bleeding per vaginum for 12 days with occasional passage of clots and pain in abdomen. The mother did not have any antenatal checkup done since conception and no ultrasound scans were available. The marriage was non-consanguineous. Family history was insignificant for any medical or surgical illness including renal disease. Emergency Doppler sonography done at our center showed single live intrauterine fetus of 32 weeks 4 days gestation with severe oligohydramnios (amniotic fluid index-1 cm), single loop of cord around neck with placenta placenta previa and focal placenta accreta. Fetal heart rate and Doppler flows were normal. The mother was immediately taken up for caesarean section (CS) and a live male baby was delivered with a birth weight of 2000 g. There was absolutely no liquor at the time of delivery. The baby did not cry immediately after birth and was flaccid with flexed hip posture. After tactile stimulation, bag and mask ventilation was started, however, in view of poor respiratory effort baby was intubated and put on mechanical ventilation. Apgar score of the baby was 5/10 and 6/10 at 1 and 5 min, respectively.

Multiple congenital anomalies were noted. The facial features noted in this baby were low set ears with overturned helix, slanting eyes, redundant fold of skin beneath the cheeks, prominent epicanthal fold, receding chin, flattened broad ear, flat nasal bridge, parrot beak appearance of nose, triangular facies and other features of Potter's sequence including narrow chest, limb deformities (bowing of legs, bilateral club feet), laxity of skin, under developed genitalia (Figure 1). At 15 min of life, baby developed tachypnea and severe chest retractions. He was not maintaining saturation and the heart rate began falling, cardiopulmonary resuscitation was started and continued for 45 min, however, the baby succumbed after 1 hour of birth due to respiratory insufficiency.

Meanwhile, urgent chest X-ray was done which showed small volume lung fields, bell shaped thorax, and a chest wall that was disproportionately small with respect to the abdomen suggestive of pulmonary hypoplasia (Figure 2a). Ultrasonography of abdomen showed complete absence of renal tissue suggesting bilateral renal agenesis (Figure 2b and c). The diagnosis of Potter syndrome was based on visible physical findings of the baby, pulmonary hypoplasia on chest X-ray and presence of bilateral renal agenesis on ultrasonography (Figure 2). Post-mortem autopsy of this baby could not be done as parents refused for it.

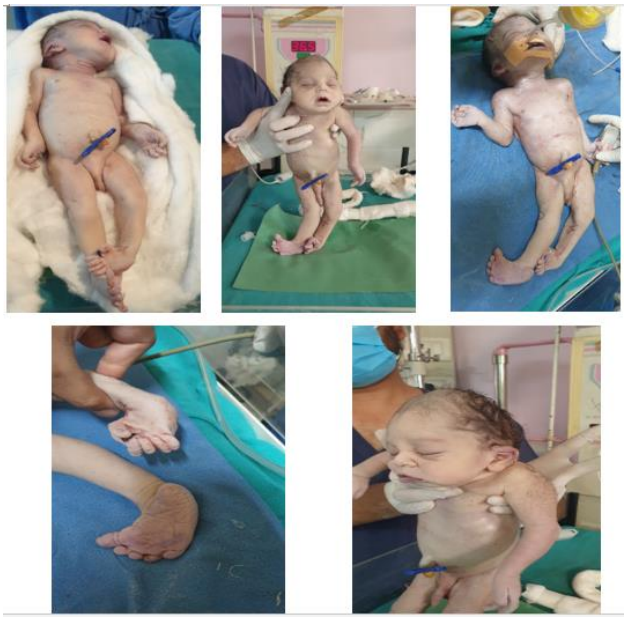

Figure 1: Showing facial and limb deformities.

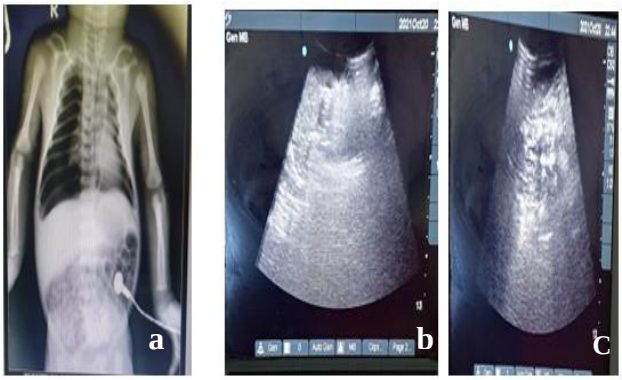

Figure 2: Showing B/L (a) pulmonary hypoplasia; and (b, c) renal agenesis.

DISCUSSION

The 'Potter's syndrome' was described by Edith Potter in new-borns with bilateral renal agenesis or other kidney abnormalities, including renal aplasia, dysplasia, hypoplasia, or multicystic disease.[7] Initially, the term was applied to the cases caused by bilateral renal aplasia (true Potter's sequence), however, nowadays, the term refers to atypical morphological appearance of the baby due to any underlying cause of oligohydramnios (foetal growth restriction, premature rupture of membranes, foetal chromosomal anomalies or post-maturity etc.).[7] Potter's syndrome may be classified into various types, causes being renal and non-renal (Table 1).

Table 1: Classification of potter's syndrome.

Classification	
Classification types	Bilateral renal agenesis (malformation of the ureteric bud)
Type 1	Autosomal recessive polycystic kidney disease (ARPKD)
Type 2	Complete agenesis/absence of one kidney and the remaining solitary kidney being small and malformed
Type 3	Autosomal dominant polycystic kidney disease (ADPKD)
Type 4	Longstanding obstruction in either the kidney/ureter resulting in cystic kidneys or hydronephrosis
Non classic	Results from rupture of the foetal membranes

Potter's sequence is thought to result from intrauterine compression of the growing foetus due to severe oligohydramnios leading to physical deformities, commonest being 'Potter's facies'. The latter is characterized by low set ears, receding chin, redundant fold of skin beneath the cheeks, flattened nasal bridge, parrot beak appearance of nose, prominent epicanthal fold. Other features of Potter's sequence include: limb deformities (which include clubbed feet, bowing of legs, limb hypoplasia etc.); ophthalmological malformations (cataract, prolapsed of lens, angiomatous malformation of optic disc area etc); pulmonary hypolpasia; cardio-vascular abnormalities (patent ductus arteriosus, ventricular septal defect etc.); VACTERL (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula, Renal defects, Limb defects), caudal dysgenesis, caudal dysplasia syndrome, and isolated anomalies of skeletal, and central nervous systems.[8-14] Skeletal anomalies frequently associated with this condition are hemi-vertebra and sacral agenesis. These abnormalities can add to the increased morbidity and mortality in such infants.

Classical Potter's sequence is said to occur in foetuses with bilateral renal agenesis leading to oligohydramnios. Ultrasonography of this baby showed bilateral renal agenesis and chest X-ray showed bilateral pulmonary hypoplasia as the cause of Potter's sequence. Hence, our case seems to be a case of classical Potter's sequence which is a very rare occurrence. Relatives did not give consent for foetal autopsy and karyotype, due to which we could not ascertain the details of the associated genetic or chromosomal abnormalities of the baby.

Medical management of neonates with Potter's sequence depends on their renal function, respiratory status and associated congenital anomalies. In cases of classical Potter's sequence with bilateral renal agenesis, further treatment may not be helpful and prognosis is grave. However non-classical Potter's sequence due to rupture of membranes during pregnancy have higher chances of survival rate, and demands proper assessment, resuscitation and management for better neonatal outcome.

This case report emphasized upon the importance of regular antenatal check-ups and examination in each and every patient as it picks up the suspicious cases which can lead to further workup and definite diagnosis of the condition and taking timely decision regarding management.

CONCLUSION

Potter's sequence is a rare but known complication of chronic oligohydramnios and is associated with grave foetal prognosis. Prevention and early diagnosis of underlying cause of oligohydramnios leads to improvement of neonatal outcome. Whereas, Classical Potter's sequence requires termination of early pregnancy, owing to underlying defects like bilateral renal agenesis, cases of non-classical Potter's sequence can usually be prevented and treated to help in providing better neonatal outcome .

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