International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2024: 7.101

A Case Report of Fatal Potter's Syndrome / Sequence

Garima Agrawal[1], Basudev Agarwal[2], Aman Gupta[3], Alok Sao[4]

[1]Consultant Radiologist, Shree Narayana hospital, Raipur, Chhattisgarh, India
Email: *agarwal21390[at]gmail.com*

[2]Assistant Professor, Radiology Department, Shri Balaji Institute of Medical Science, Raipur, Chhattisgarh, India
Email: *dr.basudevagrawal[at]gmail.com*

[3]Professor and Head, Dept of Radiodiagnosis, M. G. M. Medical College, Indore, Madhya Pradesh., India
Email: *dramangupta68[at]gmail.com*

[4]Consultant Radiologist, Shree Narayan Hospital, Raipur, Chhattisgarh, India
Email: *aloksao2014[at]gmail.com*

Abstract: ***Potter's syndrome is a rare congenital disorder characterized by characteristic atypical appearance of a newborn due to the oligohydramnios experienced during the intrauterine life. Potter's syndrome refers to the typical facial characteristics and associated pulmonary hypoplasia of a neonate as a direct result of oligohydramnios due to the renal pathology. Severe respiratory insufficiency leads to a fatal outcome in most of the infants. Though it is not inherited, sometimes the primary cause may have a genetic reason like autosomal dominant polycystic kidney, which may run in families. As there is no known method of prevention, the mortality rate is high. This reported case of Potter's syndrome was fatal. In this case, reported by us, a 19years old primigravida female came to antenatal ultrasound, which was of 29 weeks gestational age by last menstrual period. On ultrasound the gestational age was 25 to 26 weeks and typical findings of Potter's syndrome were detected in ultrasound. On follow up, after 1 week, a preterm male child was spontaneously delivered through vaginal route in severe respiratory distress and died immediately after birth. This case report emphasizes upon the importance of regular antenatal check-ups in every pregnant woman.***

Keywords: Potter's syndrome/sequence, Potter's facies, oligohydramnios sequence, pulmonary hypoplasia, polycystic kidney disease

1. Introduction

Potter's syndrome (or Potter's/oligohydramnios sequence) is a known but rare complication of oligohydramnios, incidence of which varies from 1 in every 2,000 to 5,000 foetuses 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 performed in dead new-born who died immediately after birth.[2] Potter's syndrome predominantly affects male foetuses more than female foetuses (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 can be caused by bilateral renal agenesis, cystic kidney disease, obstructive uropathy, renal hypoplasia and prolonged rupture of the membranes.[4] Hence the term potter's sequence or oligohydramnios sequence emerged. 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 includes flattened nose, recessed chin, epicanthal-folds and low-set abnormal ears.[5] The etiology 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 usually has a fatal outcome and is incompatible with life, but 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]

2. Case Report

In this case report of fatal potter's syndrome, a 19years old primigravida female came to Sri Aurobindo medical college and PG institute, Indore, M.P, India for routine antenatal ultrasound, which was of 29 weeks gestational age by last menstrual period. 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. On ultrasound, single alive intrauterine foetus was identified with ultrasound gestational age of 25 to 26 weeks and typical findings of Potter's syndrome were detected in ultrasound. Foetal heart rate and Doppler flows were normal. There was complete absence of liquor (anhydramnios) and growth discrepancy of 3 to 4

weeks. Foetal bilateral kidneys were grossly enlarged in size with diffusely increased parenchymal echogenicity. On HRUS, multiple tiny cysts were present in both renal parenchyma which suggested bilateral polycystic kidney disease. Urinary bladder was not visible. There was crowding of foetal parts and foetal face and limbs were sub-optimally visualised due to anhydramnios. Severe pulmonary hypoplasia was detected. Thoracic circumference was markedly reduced as compared to abdominal circumference. Distinctive facial features were observed including recessed chin, flattened depressed bridge of nose, hypertelorism, low set ears that lack cartilage (Potter's ears), prominent epicanthal folds, and a crease beneath the lower lips. This collection of facial features is referred to as Potter's facies. Also, cleft lip, bell shaped thorax and spastic deformities of limbs were detected.

During follow up, after 1 week, a preterm male child was spontaneously delivered through vaginal route in severe respiratory distress and died immediately after birth. Typical findings detected on ultrasound was found at birth as well.

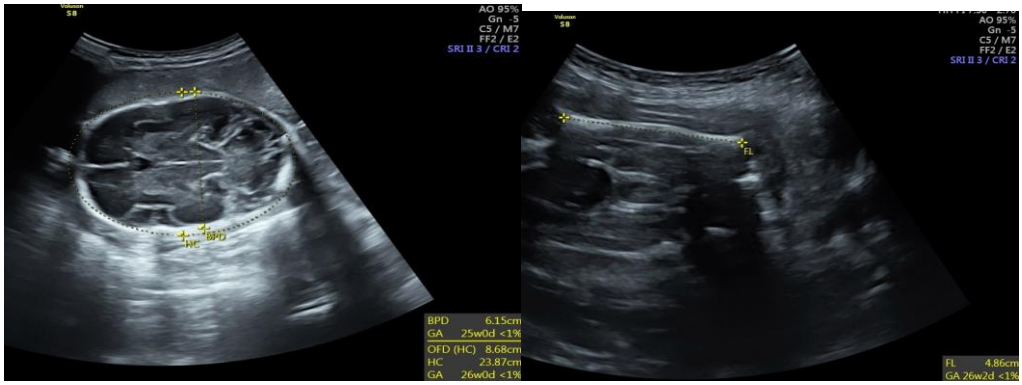

Figure 1: Growth discrepancy in BPD, HC and FL length measurements and anhydramnios

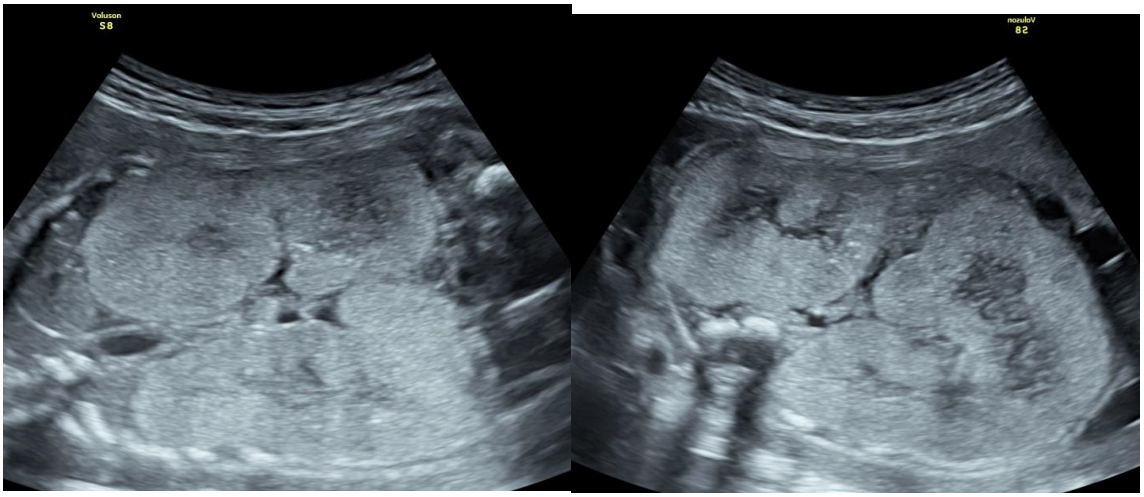

Figure 2: Foetal bilateral kidneys markedly enlarged and diffusely echogenic

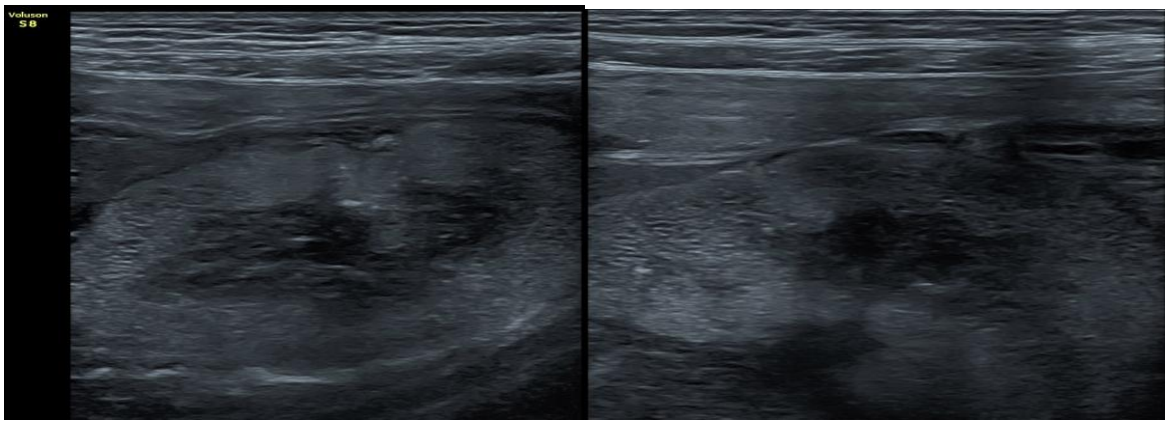

Figure 3: HRUS demonstrating bilateral multiply tiny renal cysts.

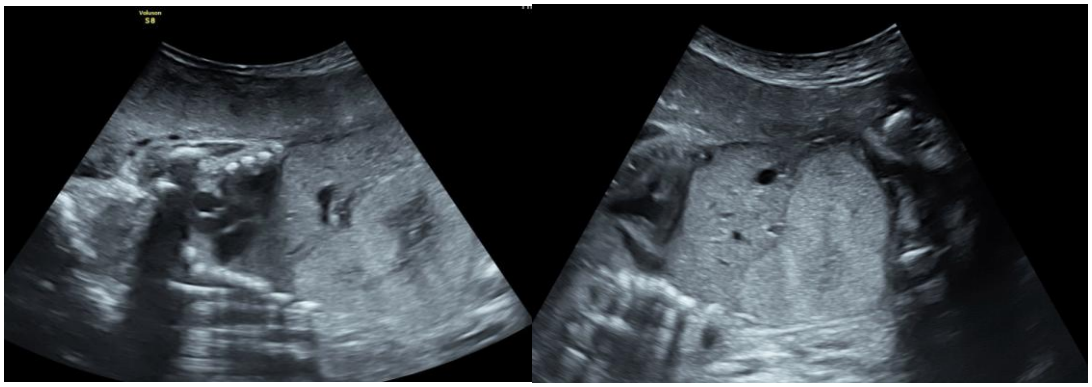

Figure 4: Marked foetal pulmonary hypoplasia.

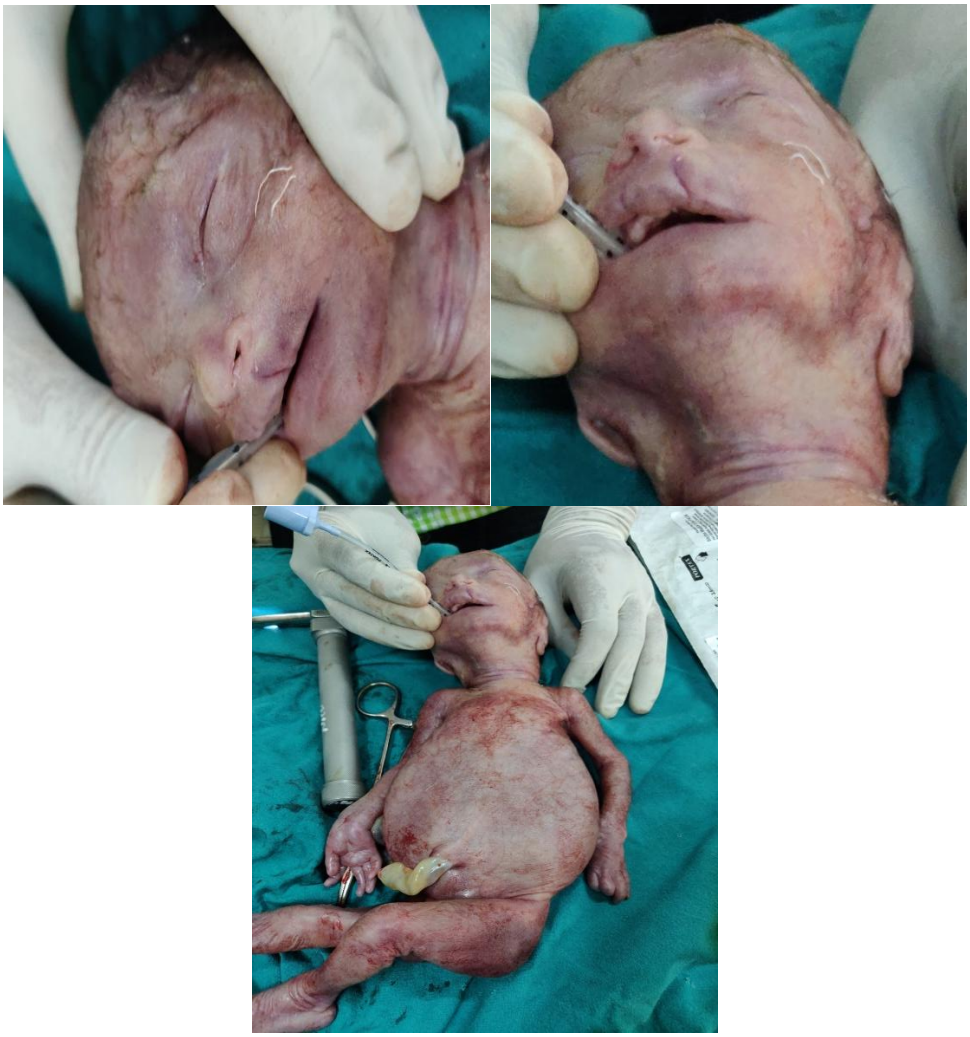

Figure 5: Potter's facies and bell shaped thorax

3. Discussion

The 'Potter's syndrome' was first 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) only, however, nowadays, the term refers to atypical morphological appearance of the baby due to any underlying cause of oligohydramnios.[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

Classical	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 (clubbed feet, bowing of legs, limb hypoplasia etc.); ophthalmological malformations (cataract, prolapsed lens, angiomatous malformation of optic disc area etc); pulmonary hypoplasia; 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] Hemi-vertebra and sacral agenesis are the skeletal anomalies frequently associated with this condition. These abnormalities can add to the increased morbidity and mortality in such cases.

4. Conclusion

The term Potter's sequence is most frequently associated with oligohydramnios sequence regardless of the root cause of absence or reduced volume of amniotic fluid. Not all cases of oligohydramnios will lead to Potter's sequence. This case report emphasizes upon the importance of regular antenatal check-ups and examination in every pregnancy as it picks up the suspicious cases which can lead to further workup and definite diagnosis of the condition and timely decision regarding management.

References

[1] Bhalla S, Ganjoo S, Kapoor P, Kaul V, Sethi A. Non classical Potter's sequence: a rare complication of chronic oligohydramnios.The New Indian Journal of OBGYN. 2019; 5:146-9.
[2] Gautam U, Kafley R, Chikanbanjar V, Shakya A, Basnet R, Manandhar SR. Rare manifestations of Potter Sequence: A Case Report. JNMA J Nepal Med Assoc. 2020;58(223):178-80.
[3] Amirshahi M, Badakhsh M, Hashemi ZS. Report of a deadly case of potter syndrome: A case report. Biomedical Res. 2019; 30:332-5.
[4] Rarediseases. National organisation for rare diseases, 2022. Available at: https://rarediseases.org/rare-diseases/potter-syndrome.
[5] Buchta RM, Viseskul C, Gilbert EF, Sarto GE, Opitz JM. Familial bilateral renal agenesis and hereditary renal adysplasia. Z Kinderheilkd. 1973;115(2):111-29.
[6] Khatami F. Potter's syndrome: A study of 15 patients. Arch Irania Med. 2004; 7:186-9.
[7] Potter EL. Facial characteristics of infants with bilateral renal agenesis. Am J Obstet Gynecol. 1946; 51:885-8.
[8] Fantel AG, Shepard TH. Potter syndrome. Nonrenal features induced by oligoamnios. Am J Dis Child. 1975;129(11):1346-7.
[9] Ginsberg J, Buchino JJ, Menefee M, Ballard E, Husain I. Multiple congenital ocular anomalies with bilateral agenesis of the urinary tract. Ann Ophthalmol. 1979;11(7):1021-9.
[10] Greenwood RD, Rosenthal A, Nadas AS. Cardiovascular malformations associated with congenital anomalies of the urinary system. Observations in a series of 453 infants and children with urinary system malformations. Clin Pediatr (Phila). 1976;15(12):1101-4.
[11] Prouty LA, Myers TL. Oligohydramnios sequence (Potter's syndrome): case clustering in northeastern Tennessee. South Med J. 1987;80(5):585-92.
[12] Kadhim HJ, Lammens M, Gosseye S, Gadisseux JF, Evrard P. Brain defects in infants with Potter syndrome (oligohydramnios sequence). Pediatr Pathol. 1993;13(4):519-36.
[13] Preus M, Kaplan P, Kirkham TH. Renal anomalies and oligohydramnios in the cerebro-oculofacio-skeletal syndrome. Am J Dis Child. 1977;131(1):62-4.
[14] Tonni G, Azzoni D, Ventura A, Ambrosetti F, Felice C. "Multicystic dysplastic kidney (Potter type II syndrome) and agenesis of corpus callosum (ACC) in two consecutive pregnancies: a possible teratogenic effect of electromagnetic exposure in utero". Fetal Pediatr Pathol. 2008;27(6):264-73.