

# CASE REPORT

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## POTTER FACIES WITH POLYCYSTIC KIDNEY DISEASE IN ASSOCIATION WITH OTHER RARE CONGENITAL ANOMALIES: TWO CASE REPORTS

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**ABSTRACT:** Potter's sequence is more appropriate terminology than potter facies, since not every individual with this syndrome has exactly the same set of symptoms and signs but they share a common chain of events triggered by different causes, leading to the same endpoint of reduced or absent amniotic fluid. It has a characteristic facial appearance associated with other abnormalities as Ophthalmic(Cataract), Cardiovascular (Ventricular septal defect. Fallot's tetralogy, Patent ductus arteriosus), and musculoskeletal (Clubbed feet, Sacral agenesis) . Here we are presenting two cases of potter sequence due to polycystic kidney disease ( type-i ) in association with other congenital anomalies ( absence of left diaphragm ,pericardial effusion, pulmonary hypoplasia ) which is rare and incompatible to life .

**KEY WORD:** Potter Syndrome , Autosomal Recessive Polycystic Kidney Disease (ARPKD).

**INTRODUCTION:** 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 .Classic Potter Syndrome occurs when the developing fetus has bilateral renal agenesis, where as type-i due to autosomal recessive polycystic kidney disease(ARPKD).. In all five distinct types potter syndrome , a lack or reduced volume of fetal urine leads to oligohydramnios which causes physical deformities. 1 Potter syndromes have a characteristic facial appearance in association with other congenital abnormality. So serial ultrasounds should be done , to detect fetuses with other abnormalities, in suspected Potter's syndromes.

**CASE REPORT-1:** A newborn female baby born out of consanguineous marriage at term was admitted with a history of abnormal facies and respiratory distress .She did not required resuscitation at birth. Family history was essentially negative. On examination baby was grunting, cyanosed and respiratory distress score 4 with H.R-160/min,R.R- 74/min,Spo2-85% . Head to toe examination revealed depressed anterior fontanelle, short and snubbed nose, recessed chin(micrognathia), prominent epicanthal folds, low-set, cartilage-deficient flattened ears with deep eye creases, narrow thorax and distended abdomen ( fig-1(a). Respiratory systems revealed decrease breath sound bilaterally. Cardiovascular system being normal. Investigation revealed normal total and differential count . Urinalysis showed a specific gravity of 1.015, pH 7, and negative protein and cells. Serum urea nitrogen was 15 mg%, creatinine 0.9

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mg% and normal range electrolyte. Imaging typically showed large, diffusely increased parenchymal echogenic, multicystic, reniform kidneys, with pulmonary hypoplasia (Fig-1 (b)).

**CASE REPORT-2:** A 22 yrs old female, gravida 2 para1 was admitted, for elective termination of pregnancy with history of multiple congenital anomalies, and polyhydramnios, expelled a female baby spontaneously vaginally at 26 wks. She had no bad obstetric history before. She had done all routine examinations in first trimester. Whereas in second trimester, ultrasonography showed severe oligohydramnios and in fetus both diffusely increased parenchymal echogenic, right (56×40mm) and left (61×40mm) reniform kidneys with multiple non communicating cystic lesion, and pericardial effusion (fig-2(b)).

The newborn female baby was a product of consanguineous marriage, having abnormal facies. Family history was essentially negative. On examination baby was an aborted product with short and snubbed nose (parrot beaked nose), small chin, low-set, cartilage-deficient flattened ears, narrow thorax and distended abdomen (fig-2(a)). Ultrasonography of abdomen and thorax showed absence of left side diaphragm, minimal pericardial effusion with both enlarged, multicystic, reniform kidneys. Unfortunately autopsy could not be done due to non availability of parents consent.

**DISCUSSION:** Potter Syndrome Type I is due to Autosomal Recessive Polycystic Kidney Disease (ARPKD), which occurs at a frequency of approximately one in 40,000 infants and is linked to a mutation in the gene PKHD1.2 The male to female ratio was 2:1, suggesting that certain genes of the Y-chromosome could act as modifiers. The gene for ARPKD encodes a protein that is called fibrocystin which is localised to cilia on the apical domain of renal collecting cells. The primary defect in ARPKD may be ciliary dysfunction related to the abnormality of this protein. Potter's syndrome can also be seen in infants with normal kidneys due to the prolonged leakage of amniotic fluid during the middle gestational weeks. Potter's facies have a characteristic appearance of flattened 'parrot-beaked' nose, recessed chin, prominent epicanthal folds, low-set, cartilage-deficient ears (known as 'Potter's ears')<sup>3</sup> with other associated abnormalities as Ophthalmic (Cataract),<sup>4</sup> Cardiovascular (Ventricular septal defect, Fallot's tetralogy, Patent ductus arteriosus),<sup>5</sup> musculoskeletal (Clubbed feet, Sacral agenesis) and abnormalities in the development of the brain<sup>6</sup>. Kidneys develop 5 and 7wks of fetal gestation with ongoing urine production by week 14 which is the main contributor to production of Amniotic fluid from the second trimester. Any disease that impairs urine production causes oligohydramnios and its consequences. Though Ultrasound scanning may demonstrate oligohydramnios and/or a congenital urological malformation ( polycystic disease),<sup>7</sup> it does not reliably prove the aetiology of hyperechoic fetal kidneys without familial data: kidney size, and amniotic fluid volume which remain the best predictors of survival.<sup>8</sup> Potter's syndrome with bilateral renal agenesis is incompatible with life but babies with Potter's sequence due to other causes have a better chance of survival.<sup>4</sup> Doppler ultrasonography of pulmonary artery blood velocity waveforms can be used to monitor the development of pulmonary hypoplasia. Estimates of relative lung volume in fetuses suspected of pulmonary hypoplasia can be obtained using 3D ultrasound or MRI scanning. At birth, respiratory and renal support with a period of intensive care is required. In the longer-term, treatment of chronic renal disease is required including: nutrition and growth monitoring, electrolyte and renal monitoring, calcium and vitamin D supplementation, treatment of anaemia and antihypertensive medication. If a child progresses to end-stage renal failure, dialysis or transplantation will be needed. A study of antenatal

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ultrasound screening in 12 European countries has shown that a great many abnormalities of the renal tract may be detected in the second trimester, allowing termination of pregnancy to be considered.<sup>9</sup>

**CONCLUSION:** Potter syndrome due to different causes present with same characteristic facies associated with other congenital anomalies. So serial ultrasound should be done during pregnancy to detect these in early and their consequences.

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Fig-1 (a) Photograph showing Potter facies (short and snubbed nose, recessed chin, prominent epicanthi fold, low-set, cartilage-deficient flattened ears with deep eye creases, narrow thorax and distended abdomen)



Fig-1(b) Ultrasonography of abdomen showed large, diffusely increased parenchymal echogenic, multicystic, reniform kidneys.



Fig-2 (a) Photograph showing short and snubbed nose (parrot beaked nose), small chin, low-set, cartilage-deficient flattened ears, narrow thorax and distended abdomen



Fig-2 (b) ultrasonography showed both diffusely increased parenchymal echogenic, right (56.940mm) and left (61.940mm), reniform kidneys with multiple non-communicating cystic lesion.