

## Potter's Syndrome associated with Pouch Colon Anomaly in Exomphalos

Supriya G\*, Saritha S, Suseelamma D and Praveen kumar M

Department of Anatomy, KIMS Narketpally, Nalgonda, Andhra Pradesh, India

### Abstract

Potter's Syndrome is a rare congenital malformation which is an atypical physical appearance of the foetus or neonate due to Oligohydramnios experienced in the womb with distinctive facial characteristics. Oligohydramnios is the cause of the various deformities observed in Potter's Sequence. It is characterised by bilateral renal agenesis, pulmonary hypoplasia, and skeletal defects of the neonate as a direct result of lack of amniotic fluid.

Congenital pouch colon (CPC) is an extremely rare variant of anorectal malformation (ARM), in which varying lengths of the colon is replaced by a dilated pouch accompanied by a fistula communicating with the genitourinary tract.

Complete Penoscrotal Transposition (CPST) is a rare and unusual malformation in which the scrotum is located cephalic to the penis. It is associated with major and often life threatening malformations.

The present case has multiple malformations which include, potter's syndrome, congenital pouch colon in omphalocele and complete penoscrotal transposition. The aim of this study was to determine the incidence, prevalence of these multiple malformations according to anatomical localization. This complex anomaly is very rare and interesting. Survival is extremely rare. In view of prognosis early diagnosis allows for earlier and less traumatic therapeutic abortion.

**Keywords:** Potter's sequence; Congenital pouch colon (CPC); Congenital penoscrotal transposition (CPST); Anorectal malformation (ARM)

### Introduction

Edith Potter, the pioneer in the field of human renal development, analysed 5000 autopsies on foetuses and new born infants over a period of 10 years. She was able to deduce the sequence of events that lead to Potter sequence. In 1946, she described the facial characteristics of infants with bilateral renal agenesis and pulmonary hypoplasia as a result of oligohydramnios due to compression in utero. Potter syndrome is fatal, incompatible with extra uterine life and 33% of foetuses die in utero.

Congenital pouch colon is commonly known as congenital short colon. This rare anomaly is counted with anorectal malformations (ARM). It is to be considered especially during the clinical evaluation of the children with cloacal malformations and anorectal anomalies. The pathogenesis and embryology of CPC are not well understood. It may be due to dietary, environmental factors and familial inheritance.

We report a rare variant of Potter sequence associated with congenital pouch colon in the omphalocele and penoscrotal transposition. These multiple malformations have not been reported in the literature. A number of abnormalities can add to morbidity and mortality in this case.

A still-born male foetus has been collected from the Department of OBG, KIMS, Narketpally during the month of October, 2011. After written consent was obtained from the parents, full autopsy examination was done.

### Case Report

A 21 years primigravida, presented to the Dept of Gynaecology, Kamineni Hospital, Narketpally with 34 weeks of gestation, diagnosed with severe oligohydramnios and intra-uterine death of the male foetus by antenatal Ultrasonography. Medical termination of pregnancy was done.

A detailed autopsy examination was undertaken and gross and microscopic examination was done in the Department of Anatomy.

### Observations: Study of the aborted foetus

**External features:** Weight - 1.6 kg

Crown to heel - 16 cm

Head circumference - 12 cm

Chest circumference - 10 cm

Crown to rump - 11 cm

Abdominal circumference - 10 cm

Typical Potters facies with skin fold extending from medial canthus across the cheek, redundant skin, flattened nose and low set large ears (Figures 1 and 2).

Limb anomalies - Absent right thumb and hyper extended left thumb (Figures 3 and 4).

Omphalocele present in the umbilical cord (Figure 5). External genitalia present penoscrotal transposition with empty scrotum (Figure 6). Anal orifice was absent (Figure 7).

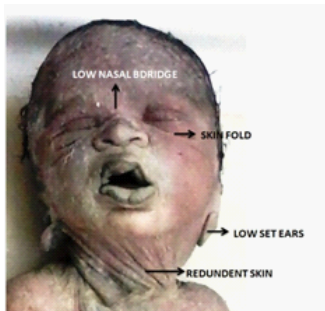
**Internal Features:** Brain - Normal

**\*Corresponding author:** Supriya G, Department of Anatomy, KIMS Narketpally, Nalgonda, Andhra Pradesh, India. E-mail: [dr\\_supriyaraj@yahoo.com](mailto:dr_supriyaraj@yahoo.com)

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**Figure 1:** Typical Potters facies with skin fold extending from medial canthus across the cheek, redundant skin and flattened nose.



**Figure 2:** Typical Potters facies with skin fold extending from medial canthus across the cheek, redundant skin, flattened nose and low set large ears.



**Figure 3:** Limb anomalies: Absent right thumb.

Thoracic region - The Heart was normal and the lungs were hypoplastic (Figure 8).

GIT: Stomach, liver, spleen and small intestine were normal

Large intestine: Large intestine beyond caecum was converted into a blind sac known as pouch colon, with caecum and appendix outside the pouch colon. The pouch colon is presented as omphalocele (Figures 9-11). Genitourinary system: Bilateral Renal agenesis with agenesis of ureters and urinary bladder (Figures 12 and 13). Bilateral undescended testis present in the lumbar region (Figures 12 and 14). Entire pelvic cavity was empty. Umbilical cord presents a dilated blind sac called pouch colon. It presents a single umbilical artery and a single umbilical vein (Figures 15 and 16). Placenta shows intervillous fibrinoid necrosis with syncytial knots (Figures 17 and 18).

## Discussion

Potter's sequence also known as Potter's syndrome or Oligohydramnios sequence is the atypical physical appearance of a foetus or a neonate due to oligohydramnios experienced in the womb.

Oligohydramnios causes disruptions in morphogenesis of the foetus [1].

It is characterised by bilateral renal agenesis, pulmonary hypoplasia, limb defects of the neonate as a direct result of lack of amniotic fluid. It is found in 0.2% to 0.4% of the autopsies performed on stillborn infants. There is no treatment available currently and they have a very poor prognosis with the respiratory distress which is the leading cause of death. Potter's facies is considered as typical wide set eyes, squashed nose, receding chin, prominent epicanthic folds and large low set ears.

In 1946, Dr. Edith Potter reported the necropsy findings of 20 infants dying in the perinatal period with bilateral renal agenesis [2].

Potter sequence has been defined into five distinct sub classifications

**Type I** is due to autosomal recessive polycystic kidney disease (ARPKD) which occurs at a frequency of approximately 1 in 16000 infants.

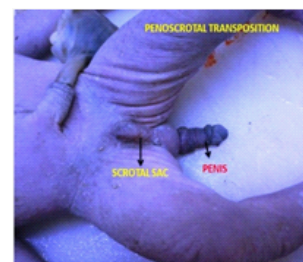
**Type II** is usually due to renal agenesis, which can also fall under the category known as hereditary urogenital adysplasia. This is characterized by the complete agenesis or absence of one kidney



**Figure 4:** Limb anomalies: hyper extended left thumb.



**Figure 5:** Omphalocele present in the umbilical cord.



**Figure 6:** External genitalia present penoscrotal transposition with empty scrotum.



Figure 7: Posteriorly displaced penis.

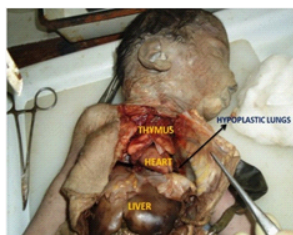


Figure 8: The Heart was normal and the lungs were hypoplastic.

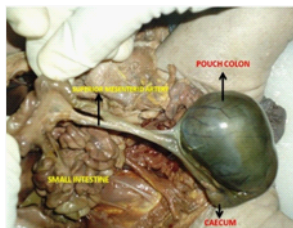


Figure 9: Large intestine beyond caecum was converted into a blind sac known as pouch colon, with caecum and appendix outside the pouch colon. The pouch colon is presented as omphalocele.

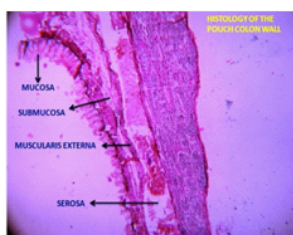


Figure 10: Histology of the pouch colon wall.

and the other kidney becomes small and malformed. Bilateral renal agenesis (BRA) is believed to be the most extreme phenotypic variation of Hereditary Renal Agenesis (HRA). However, BRA is often referred to as classic Potter's sequence.

**Type III** is due to Autosomal dominant polycystic kidney disease (ADPKD) linked to mutations in the genes PKD1 and PKD2.

**Type IV** occurs when a long standing obstruction in either the kidney or ureter leads to cystic kidneys or hydronephrosis.

It has been estimated to occur at a frequency of approximately 1:4000 to 1:8000 fetuses and neonates. No racial predilection is known.

This condition has been reported to occur twice as common in males as in females. In males the testes will begin to form at the same

time the kidneys do, whereas in females the ovaries begin to form after the kidneys.

Al-Haggag M et al. [3] reported one foetus of sirenomelia sequence with Potter's syndrome which showed oligohydramnios and symelia Apus. The infant showed absent urinary tract and external genitalia, the legs were fused by skin and had separate bones associated with Potter's syndrome. The mother had a history of gestational diabetes mellitus.

### Genetics

During nephrogenesis, multiple genes, transcription factors and growth factors control the essential interaction between the ureteric bud and the metanephric mesenchyme. LIM1 and PAX2 transcription factors are essential for the formation of the mesonephric duct, from which ureteric bud develops. LIM1-deficient individuals have complete renal agenesis [4].

The index case presents a male foetus of type II classification, a classic Potter sequence with pulmonary hypoplasia and absence of thumb and associated Congenital pouch colon (CPC) present in umbilical cord. It also presents congenital penoscrotal transposition (CPST) and single umbilical artery.

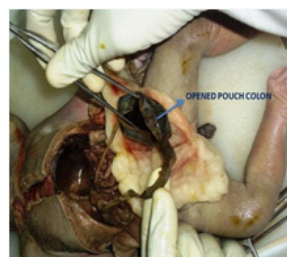


Figure 11: Opened pouch colon.

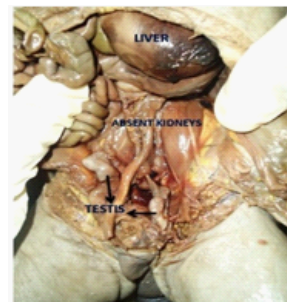


Figure 12: Bilateral Renal agenesis with agenesis of ureters.



Figure 13: Bilateral Renal agenesis with agenesis of urinary bladder.

Potter syndrome coexists with CPC and penoscrotal transposition has not been reported in the literature. Congenital pouch colon associated with anorectal malformations is an extremely rare variant in which part of the colon is replaced by a pouch like dilatation [5]. They are divided into four types [6]:

**Type I-** Normal colon is absent and ileum opens directly into the colonic pouch.

**Type II-** The ileum opens into a short segment of caecum, which then opens into the pouch.

**Type III-** Presence of normal colon of significant length between the ileum and the colonic pouch.

**Type IV-** Presence of near normal colon with only the terminal portion of colon (sigmoid and rectum) converted into a pouch.

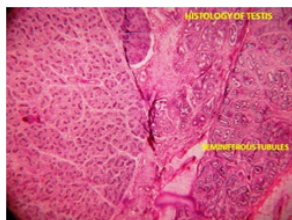


Figure 14: Bilateral undescended testis present in the lumbar region.



Figure 15: Umbilical cord presents a dilated blind sac called pouch colon.

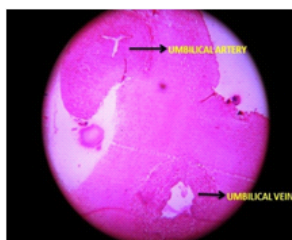


Figure 16: Umbilical cord presents a dilated blind sac called pouch colon. It presents a single umbilical artery and a single umbilical vein.



Figure 17: Placenta shows intervillous fibrinoid necrosis with syncytial knots.

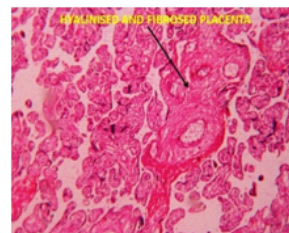


Figure 18: Hyalinised and Fibroid Placenta.

The present case is type II with entire colon beyond caecum opens into a blind pouch which is present in the umbilical cord. The exact embryogenesis of the pouch colon syndrome has not been worked out.

Impairment of intestinal organogenesis, primary dysplasia, congenital damage to myenteric plexus, intrauterine vascular catastrophe have all been considered as the reasons for abnormal dilatation of the bowel. Arrest of cloacal separation due to non-descent of urorectal septum along with faulty organogenesis of distal intestine close to descending urorectal fold would lead to Anorectal Malformation with abnormal dilatation of the bowel [7]. 90% of CPC are reported from the Indian subcontinent and predominantly from the Northern provinces. Earlier, type I CPC with high ARM was found to be common, but over years type IV CPC has been increasing [7].

The incidence of associated cardiac, vertebral and genitourinary anomalies is very high in CPC, so evaluation of other anomalies is important. The reported case was a male foetus with CPC in omphalocele associated with genitourinary anomalies (Potter's syndrome) and penoscrotal transposition, absence of left thumb and single umbilical artery.

Single umbilical artery is the most common developmental anomaly of the umbilical cord and is frequently associated with an increased incidence of atresia of the hollow visceral organs, gastrointestinal and urogenital abnormalities, musculoskeletal, cardiovascular and central nervous system malformations and limb reduction defects [8].

Complete Penoscrotal Transposition (CPST) is a rare and unusual malformation in which the scrotum is located cephalad to the penis, frequently associated with major and often life threatening malformations involving the urogenital, cardiovascular system, or skeletal systems. Parida et al. [9] have noted 90% complete agenesis of the urinary system. The most common non-urogenital abnormalities associated with CPST were mental retardation (60%), ano rectal malformations (33%), Central nervous system anomalies (29%), preaxial limb defects and congenital heart defects (19%).

It therefore appears likely that CPST may represent only one of the major localized or generalized embryological insult to the foetus during 4-6 weeks of development, when major organ system are passing through a crucial phase of development and differentiation [10]. The present case agrees with the above authors, that Potter sequence with Congenital pouch colon and ARM, CPST and absence of left thumb (i.e. preaxial limb defects) all due to generalized embryological insult during 4-6 weeks of gestation.

## Conclusion

The index case is very rare with multiple congenital malformations. The foetus presented with Potter's sequence, congenital pouch colon with ARM, absence of left thumb, Penoscrotal transposition and single

umbilical artery. These multiple malformations are not grouped in any of known classifications or syndrome.

The highlight of this study is these multiple malformations in a single foetus have not been reported in the literature so far. Anticipation of the frequency and multiplicity of this case is of paramount importance to the anatomists and particularly to the clinicians. Early antenatal sonographic diagnosis is important in view of dismal prognosis and allows for early, less traumatic termination of pregnancy.

#### References

1. Oligohydramnios sequence at Dorlands Medical Dictionary.
2. Dunn PM (2007) Dr Edith Potter (1901 1993) of Chicago: pioneer in perinatal pathology. *Arch Dis Child Fetal Neonatal* Ed 92: F419-F420.
3. Al-Haggar M, Yahia S, Abdel-Hadi D, Grill F, Al Kaissi A (2010) Sirenomelia (symelia apus) with Potter's syndrome in connection with gestational diabetes mellitus: a case report and literature review. *Afr Health Sci* 10: 395-399.
4. Gupta S, Araya CE (2010) Potter syndrome. *eMedicine*.
5. Pavai A, Pillai SD, Shanthakumari S, Sam CJ, Shylaja M, et al. (2009) Congenital pouch colon: Increasing association with low anorectal anomalies. *J Indian Assoc Pediatr Surg* 14: 218-220.
6. Herman TE, Coplen D, Skinner M (2000) Congenital short colon with imperforate anus (pouch colon). Report of a case. *Pediatr Radiol* 30: 243-246.
7. Narasimharao KL, Yadav K, Mitra SK (1984) Congenital short colon with imperforate (pouch colon syndrome). *Ann Pediatr Surg* 1: 159-167.
8. Sur M, Nayler SJ, Muc RS (2004) Association of Single Umbilical Artery with Common and Rare Congenital Malformations. *Internet J Pediatr Neonatol* 4: 44.
9. Parida SK, Hall BD, Barton L, Fujimoto A (1995) Penoscrotal transposition and associated anomalies: report of five new cases and review of the literature. *Am J Med Genet* 59: 68-75.
10. Shashidhar A, Rughani B, Keshavamurthy SR (2011) Complete penoscrotal transposition- a rare congenital anomaly. *Pediatric Oncall Child Health Care*