

CASE REPORT

A Case of Prune Belly Syndrome with Uro-rectal Malformation Sequence

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ABSTRACT

Background: Prune belly syndrome (PBS) is a congenital and rare condition which presents a partial or complete triad of deficiencies or absence of muscles of abdomen, abnormalities in the genital organs like cryptorchidism and abnormalities of the urinary tract. This condition is usually not compatible with life.

Case presentation: A 26-year-old female, fourth gravida with 1 alive and well child and 2 abortions, a known case of hypothyroidism, delivered a stillborn baby at 19 weeks of gestation. On autopsy there was lax, "prune belly" like abdominal wall, low set ears with club foot, hypoplastic lungs, cryptorchidism, blind cloacal sac, urachal sac, hypoplastic left kidney, hydroureter, urethral and anal atresia. A case of prune belly syndrome with urorectal malformation sequence was diagnosed.

Conclusion: Anomaly scan should be advised wherever a routine obstetric scan shows abnormality of the lungs or urinary system to rule out prune belly syndrome.

KEYWORDS

- Absent abdominal muscle • Genito-urinary abnormality

BACKGROUND

Prune belly syndrome (PBS) is a congenital and rare condition which presents a partial or complete triad of deficiencies or absence of muscles of abdomen, abnormalities in the genital organs like cryptorchidism and

abnormalities of the urinary tract. This condition is usually not compatible with life.

In utero, these cases present with oligohydramnios, which causes Potter's facies, due to pressure effects of the fetus against the uterine wall. The features of Potter's facies

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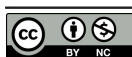
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include hypertelorism, flattened nasal bridge, micrognathia and epicanthal folds. Complete URSMS consists of ambiguous genitalia, urological, colorectal and lumbosacral abnormalities. There is absence of any anal/perineal opening. Partial URSMS is a milder form and has only single perineal opening which serves to drain the urine and faeces.¹

CASE PRESENTATION

This study was conducted at Chettinad Hospital and Research Institute, Kelambakkam, Chennai, India. A 26 year old female, fourth gravida with one alive and well child and two abortions, a known case of hypothyroidism, presented at 19 weeks of gestation at Chettinad Hospital and Research Institute, Kelambakkam, Chennai, India. A Second trimester anomaly obstetric scan showed a fetus gestational age by ultrasound-18 Weeks 5 days, with biparietal diameter of 3.80 cm, head circumference of 15.92 cm, fetal length

of 3.11 cm and expected fetal weight of 1188 ± 173 grams. There was a single live fetus in variable presentation. Placenta was posterior in location. Lower margin of placenta was seen covering the internal os. Liquor volume was absent. No liquor pockets were seen. Fetal cardiac activity and somatic movements were observed. Fetal heart rate was normal. Thoracic and abdominal organs appeared normal. Urinary bladder was not visualized. A large cystic structure measuring 9.6×5.0 centimeter was noted in midline with communication to bilateral kidneys, possibly urinary bladder. The right kidney was hydro nephrotic. The left kidney showed multiple non communicating cysts, possibly dysplastic (*Figure 1*). Ultrasound findings were suggestive of bladder outlet obstruction leading to a massively enlarged cloacal sac (*Figure 2*) with bilateral hydro ureters.

The patient expelled a dead-born male fetus, weighing 540 g and placenta weighing 270 g.



Figure 1: Ultrasound images showing dysplastic kidneys



Figure 2: Ultrasound images showing massively enlarged cloacal sac

Autopsy findings

On external examination, the abdomen was massively enlarged. The skin was lax in the lower abdomen and resembled a "Prune". The fetus had low set ears, the crest of the pinna was at a lower level than that of outer canthus on both sides. Club foot was present (*Figure 3*). Phallus could be made out. Imperforate anus and no urethral opening were seen. The fetal weight was 542.5 g (expected weight: 313±139 g). The crown-rump length, crown-heel length and head circumference were normal. The chest circumference was 15.5cm (expected chest circumference: 14.2 cm) was slightly increased for the corresponding gestational age. The abdominal circumference was 21.5 cm (expected abdominal circumference: 2.4 cm) and was markedly increased. The biparietal diameter was normal. Hand length, foot length, inner canthal distance were normal. The outer canthal distance was increased. The nipple was not developed on one side.



Figure 3: External appearance of the fetus showing club foot and "Prune-Belly" abdomen

The placenta was intact with attached cord. Placental membranes were intact, with all cotyledons identified and unremarkable. Insertion of the cord to placenta was central.

Before dissection, the CSF was collected. Internal examination revealed brain with sulci and gyri not well formed and weighing 50 g. Before dissection of the thorax and abdomen, peritoneal fluid about 3 ml was aspirated. It was clear.

On dissection of the thorax, trachea and esophagus appeared unremarkable. No tracheo-oesophageal fistula was seen. Heart and lungs block were separated. Both lungs had not inspired. Lungs were hypoplastic; septations were not completely formed (*Figure 4*). Visceral pleural surfaces of both lungs were smooth and parenchyma appeared unremarkable. Serial cut sections through both lungs revealed solid lungs. The cardiovascular system was unremarkable. Diaphragm was complete and unremarkable.

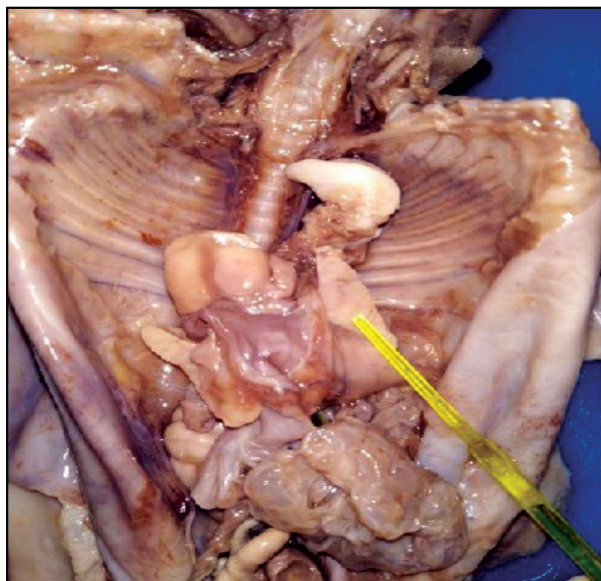


Figure 4: Dissected fetus showing Hypoplastic lungs

The gastrointestinal system showed meconium staining up to the rectum. The appendix measured 1 cm in length. The rectum was seen attached to a blind pouch-like sac (the cloacal sac), but no opening was visualized. The cloacal sac was blind and showed no external opening as the anus. The anus was imperforate.

The hepatobiliary system i.e., the liver and gall bladder showed no abnormality.

The genitourinary system and adrenal glands showed that the right kidney was lobulated and measured 2.6x1x0.6 cm. Cut surface was cystic, largest cyst measuring 1 cm across. The pelvicalyceal system was dilated. Normal kidney parenchyma was seen in the periphery and at the lower pole of kidney. The right kidney was hydro nephrotic. The right adrenal gland measured 0.8 g. Right ureter measured 5 cm in length and was dilated and tortuous. Maximum diameter of the dilated portion was 0.3 cm. Lower end of the right ureter was seen

opening into the cloacal sac. On microscopic examination, the right kidney showed a rim of normal kidney in the periphery and mesenchymal tissue in the medullary region.

Left kidney was hydro nephrotic and enlarged, and measured 4x3.5x1 cm. Cut surface of the kidney was gelatinous, normal kidney tissue was not identified grossly. Left adrenal gland weighed 1 g (*Figure 5*). Left ureter measured 7 cm in length and was markedly dilated and tortuous, maximum dilatation measured 1 cm and was seen opening in the cloacal sac (*Figure 6*). Microscopic section from the left kidney showed hydro nephrotic kidney with a few tubules.

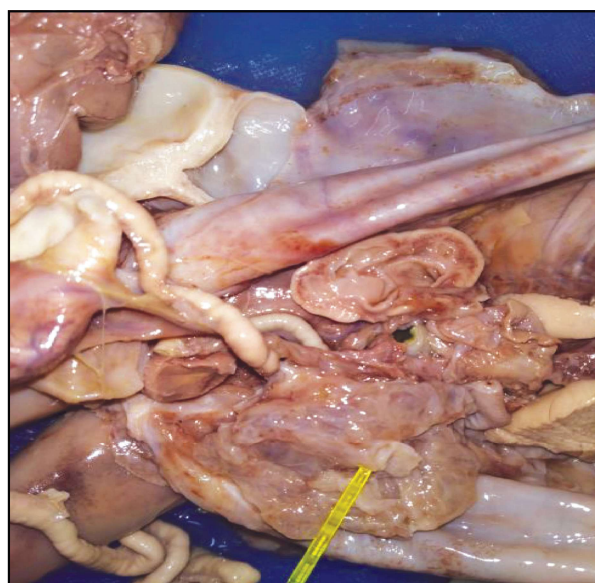


Figure 5: Dissected fetus showing dysplastic kidneys

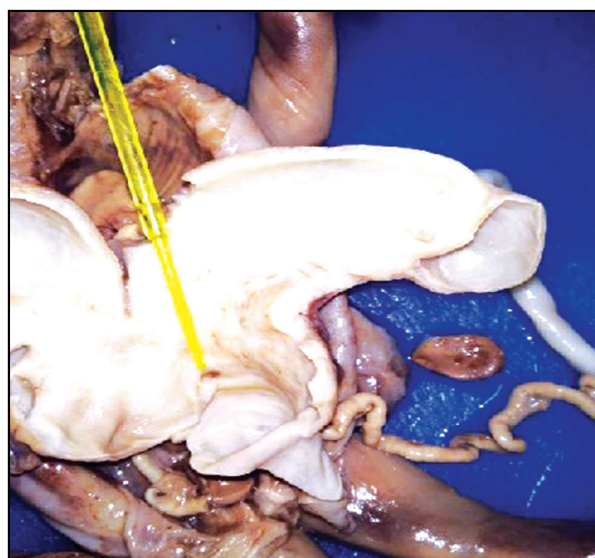


Figure 6: Dissected fetus showing cloacal sac with enlarged opening of the left ureter

Cloacal sac measured 10x5 cm. Wall thickness ranged from 0.3 to 0.5 cm. The rectum was seen on the right side of the cloacal sac and there was anal atresia. Both the ureters were seen opening into the cloacal sac. Right ureteric opening measured 0.2 cm and left ureteric opening measured 1 cm in diameter. No trigone of the bladder was seen. Attached urachal sac measured 2.5 cm across. It was seen attached to the umbilicus and was continuous with the cloacal sac (*Figure 7*). The inner wall of the cloacal sac and urachal sinus were smooth. No septations were seen. No urethra or anus were seen. There were urethral and anal atresia. The cloacal sac contained 0.5 ml of urine. Smears from the urinary sediment showed epithelial cells in a background of few RBCs and few inflammatory cells. Blood urea nitrogen and creatinine in urine were below detectable limits. Microscopic section from the cloacal sac showed lining by transitional epithelium, muscle hypertrophy and many nerve bundles.

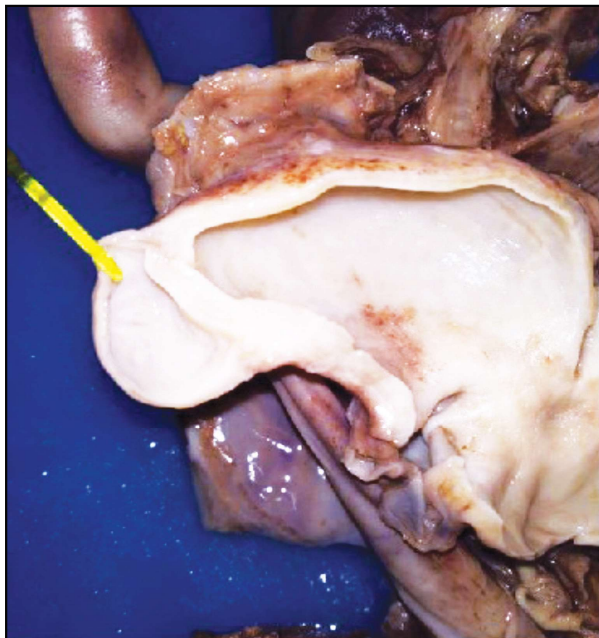


Figure 7: Dissected fetus showing cloacal sac with urachal sac

The endocrine system showed thyroid gland weighing 0.5 g. Right lobe measured 0.6x0.5 cm and left lobe measured 0.7x0.5 cm. The pancreas weighed 0.1 g.

The lymphoreticular system showed thymus weighing 1 g and measuring 2x1.5x0.3 cm. The spleen measured 2x1x0.5 cm and weighed 0.7 g.

The microscopic appearance of the abdominal wall showed no muscle layer and

was replaced by fibro-myxomatous tissue, which was confirmed by Masson's trichrome stain.

The final diagnosis was made as prune belly syndrome with uro-rectal septal malformation sequence.

Lax, "prune belly" like abdominal wall, low set ears with club foot, hypoplastic lungs, absence of internal sex organs, blind cloacal sac, absence of perineal openings, urachal sac, dysplastic kidneys, hydroureter, urethral and anal atresia.

DISCUSSION

Prune Belly Syndrome (PBS) is the Eagle-Barrett syndrome. PBS is associated with high morbidity and mortality. Cases of stillbirths like our case or early mortality especially within the first weeks of life are often seen. The etiology and pathogenesis of PBS is still not fully elucidated. Three theories regarding aetiology of PBS have been postulated. The first theory describes the obstruction of the urinary tract which leads to dilatation of the ureters, which in turn leads to hydronephrosis and distension of the abdomen. This distension of the abdomen leads to abdominal muscle weakness and cryptorchidism. The second theory states that there is failure of the mesoderm to differentiate (of the first lumbar myotome) which leads to defective muscle in both abdominal and urinary tract muscles. The third theory states that the cause could be yolk sac and allantois dysgenesis.^{1,2}

Renal dysplasia and the consequent hydronephrosis show renal dysplasia in varying degrees in > 50% of cases as was seen in our case. Renal dysplasia includes clubbing of the calyces, narrowing of the pelvi-calyceal system, and cyst formation. Often, the ureters show gross dilatation, increased length, and tortuosity all of which was seen in our case. This is because fibrous tissue replaces the smooth muscle cells as was seen in our case. The distal part of the ureter is more dilated and tortuous than the proximal part of the ureter as was seen in our case. Further, the bladder is dilated and non-trabeculated as the smooth muscle is replaced by fibrous tissue. In our case, it opened into a cloacal sac which was continuous with the urachal sac. The cloacal sac may show increased length and superiorly maybe continuous with the patent urachus or urachal diverticulum as was seen in our case.

Radiologic findings in PBS may show marked dilatation and tortuosity of the lower thirds of the ureters and urethral abnormalities in the anterior region than those seen in posterior urethral valve (PUV) cases. Rarely, arthroscopy may be of help to demarcate these cases.³

During fetal development, the larger abdominal cavity presses on the thoracic cavity leading to pulmonary hypoplasia, which if severe can lead to early death as was seen in our case. In the era following 1970s, the prognosis of PBS has markedly improved due to early diagnosis and treatment.

One of the older theories states that following obstruction of the urethra at the level of prostate leads to back pressure and over distension of the ureters which may cause dysplasia of kidneys. The dilated upper urinary tract leads to distension of the abdomen with weakened abdominal and ureteric musculature and also interferes with descent of testes leading to cryptorchidism. Newer hypotheses state that the syndrome is caused due to an insult in utero period causing a disruption in the mesodermal development. The muscles of the abdominal wall and of the genitourinary tract are thus destroyed and replaced by fibrous tissue. A genetic cause was postulated since males are affected by PBS probably an X-linked defect. However, then female cases of PBS were seen, casting doubt on the genetic basis of PBS.²

Oligohydramnios, which leads to pressing of fetus against the uterine wall causes Potter's facies which includes hypertelorism, epicanthal folds, micrognathia and flattened nasal bridge as was seen in our case. Also, the musculoskeletal abnormalities are postulated to be due to oligohydramnios. Our case had oligohydramnios, micrognathia, flattened nasal bridge, Potter's facies and bilateral clubfeet. The morbidity and mortality of PBS depends on the severity of lung and kidney hypoplasia and dysplasia respectively. Our patient had severe pulmonary hypoplasia and renal dysplasia and thus the high mortality.^{3,4}

A cloaca which is persistent or a partial urorectal septal malformation sequence (URSMS) shows a single opening in the perineum which drains a common urinary and gastrointestinal system. The incidence of URSMS is 1 in 50,000 to 250,000 births. A complete URSMS anomaly consists of ambiguous genitalia, urologic, colorectal abnormalities, absence of any anal/perineal opening and lumbosacral

abnormalities. Our patient also lacked perineal opening and had urologic abnormalities. This defect is more common in females and has a male to female ratio of 0.87. Complete URSMS has a mortality rate of 95%. The high rate of mortality is due to the oligohydramnios leading to pulmonary hypoplasia and Potter's facies.⁵ Incomplete URSMS is generally associated with a perineal opening for draining of urine and cycling in utero leading to normal lung development. There is no oligohydramnios. In such cases, urogenital and gastrointestinal surgeries offers a good prognosis. Since our patient had absence of anal opening and a Potter's facies we classified our case as PBS with complete URSMS. Complete URSMS is associated with 95% mortality. The concurrence of both URSMS and Prune Belly syndrome has been rarely described in literature with only 4 cases being reported previously.^{3,4}

PBS with URSMS is characterized by renal agenesis and renal dysplasia of one kidney, absence of internal genitalia and absence of urethra, vagina and opening of the anus. Bilateral clubfeet, as was seen in our case are an additional feature. Lumbar scoliosis is commonly seen but was absent in our case.^{5,6,7} These defects are commonly seen in females but our patient was a male and had Potter's facies with club foot, dysplastic kidneys, cloacal sac with communication to urachal sac with absence of urethral, anal openings. A female fetus is described with female internal genital organs. The distal gastro-intestinal tract, ureters and fallopian tubes opened into a common cloacal sac. The ovaries showed Leydig cells microscopically. Farooqui *et al* reported a 34-week old twin, female fetuses with absence of abdominal wall musculature, dilatation of the vagina, distended urinary bladder, moderate hydroureter and hydronephrosis and streaks of linear calcification in the hypoplastic left kidney. This baby was lost in the 3rd postpartum month despite many corrective surgeries. This case represents the third case of overlap between PBS with URSMS.⁵⁻⁷

Like PBS the etiopathogenesis of urorectal septum malformation sequence has been postulated to impaired mesodermal development.

The treatment of PBS includes kidney transplantation and surgery for the malformed urinary tract abnormalities due to dysplastic

kidneys, correction of the abdominal wall musculature and surgery for undescended testes. Despite the corrective surgeries the prognosis of PBS is poor which is because of the hypoplasia of the lungs which is associated with oligohydramnios. The prognosis of partial URSMS is also poor because of the numerous corrective surgeries required in the new-born period. Occurrence of PBS together with URSMS has a poor prognosis.⁸

PBS has been described in seven female patients, who had abnormal abdominal wall musculature, uro-genital abnormalities. In addition, there was atresia of the urethra, duplication of the uterus, and anorectal abnormalities. The perinatal mortality was high as three patients expired; of the remaining four patients who survived, transplantation of the kidneys was required in two patients as they developed renal failure. Further, analysis of these cases postulates that urethral obstruction is also an important factor in addition to pulmonary hypoplasia contributes to the development of the Prune Belly syndrome in females.⁹⁻¹³

In this case report, a female fetus with Woodhouse category I PBS with URSMS has been described. Females with Prune belly syndrome (PBS) do not fulfill the characteristic features of PBS like anomalies of the genito-urinary tract, abdominal wall musculature deficiency and bilateral cryptorchidism. URSMS has a bad prognosis and is associated with ambiguous genitalia, absent openings for the urogenital and anal tract in the perineum and lumbo-sacral abnormalities.¹⁴

CONCLUSION

Cases of Prune-Belly Syndrome with URSMS have a bad prognosis and hence anomaly scan should be advised wherever a routine obstetric scan shows abnormality of the lungs or urinary system.

Abbreviations

PBS - Prune belly syndrome - Fetus with absent musculature in anterior abdominal wall

URSMS - Uro-rectal Septal Malformation Sequence - Ambiguous genitalia, absence of any anal/perineal opening, urologic, colorectal and lumbosacral anomalies.

Declarations

Ethics Approval and consent to participate

This case report was written after getting consent from the institutional human ethics committee.

Consent for participation and publication

An informed written consent was obtained from the fetus' parents for being the subject in this case report and for publishing this article.

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REFERENCES

1. Valappil B., Lalu K., Ranjith S., Shetty A. Exploration of the fetus with gross anomaly: a case of pseudo prune belly syndrome. *Anat Cell Biol.* 2018; 51(3): 205–8. doi: 10.5115/acb.2018.51.3.205.
2. Metwalley K.A., Farghalley H.S., Abd-Elsayed A.A. Prune belly syndrome in an Egyptian infant with Down syndrome: a case report. *J Med Case Rep.* 2008; 2: 322. doi: 10.1186/1752-1947-2-322.
3. Wen C., Marquez D.J., Cohen A.J. Eagle-Barrett Syndrome (also known as prune-belly syndrome [PBS]) in an adult. *Appl Radiol.* 2004 Apr 13.
4. Akdag A., Arici S., Orhaner B., Dogan A. Prune belly syndrome associated with bilateral multicystic dysplastic kidneys and urethral obstruction: A case report. *Ped Urol Case Rep.* 2015; 2(4): 13–6.
5. Mashavave N.Z., Harrison D.S., Grieve A.D. Partial urorectal septum malformation sequence with prune belly syndrome and oesophageal atresia with a tracheoesophageal fistula. *J Pediatr Surg Case Rep.* 2019; 43: 83–6. doi: 10.1016/j.epsc.2019.02.016.
6. Sharma D.S.R., Shastri S. A case report of aphallia with urorectal septum malformation sequence in a newborn: a very rarely seen condition. *Int Med Case Rep J.* 2015; 8: 317–20. doi: 10.2147/IMCRJ.S92122.
7. Mirza B.M.S.M. Complete urorectal septum malformation sequence associated with prune belly syndrome and omphalocele. *J Paediatr Surg Specialties.* 2015 Apr; 9(2): 28–30.

8. Kunt Işgüder Ç., Kanat Pektaş M., Köseoğlu D., Takçi Ş. An Autopsy Case Report: Prune Belly Syndrome with Overlapping Presentation of Partial Urorectal Septum Malformation Sequence. *Turk J Pathol.* 2018; 1-4. doi: 10.5146/tjpath.2018.01440.
9. Reinberg Y., Shapiro E., Manivel J.C., Manley C.B., Pettinato G., Gonzalez R. Prune belly syndrome in females: a triad of abdominal musculature deficiency and anomalies of the urinary and genital systems. *J Pediatr.* 1991; 118(3): 395-8. doi: 10.1016/s0022-3476(05)82153-5.
10. Rabinowitz R., Schillinger J.F. Prune belly syndrome in the female subject. *J Urol.* 1977; 118(3): 454-6.
11. Straub E., Spranger J. Etiology and pathogenesis of the prune belly syndrome. *Kidney Int.* 1981; 20(6): 695-9. doi: 10.1038/ki.1981.198.
12. Pinto T., Baithun S.I., Giwan Y.A., Berry C.L. The prune belly syndrome--a possible pathogenesis. *Diagn Histopathol.* 1982; 5(3): 197-203.
13. Manivel J.C., Pettinato G., Reinberg Y., Gonzalez R., Burke B., Dehner L.P. Prune belly syndrome: clinicopathologic study of 29 cases. *Pediatr Pathol.* 1989; 9(6): 691-711. doi: 10.3109/15513818909022376.
14. Farooqui A., Alaqeel A., Habib Z. Prune belly syndrome with overlapping presentation of partial urorectal septum malformation sequence in a female newborn with absent perineal openings. *Case Rep Surg.* 2014; 2014: 746323. doi: 10.1155/2014/746323.