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Case Report

Prune-belly syndrome

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Prune-belly syndrome, also known as Eagle-Barret syndrome, is a congenital anomaly comprising three clinical findings: deficient abdominal musculature, urinary tract anomalies, and bilateral cryptorchidism. Other clinical findings involving respiratory, skeletal, digestion and cardiovascular system may also accompany the syndrome. The incidence is approximately 1 : 30,000 to 40,000 live births and 95% of cases occur in boys. Pulmonary hypoplasia and kidney failure are important prognostic factors that contribute to 60% of mortality rate. Treatment includes surgical correction of the abdominal wall and urinary tract, orchidopexy and other supportive managements.[1-4] We report 4 cases on typical Prune-belly syndrome, together with other clinical variants.

Case 1

A boy was born to a 26-year-old mother, P1A0, 39th week of pregnancy, assisted by obstetrician on indication of oligohydramnios. The baby's weight was 4000 grams, 49 cm, Apgar score was 6/8, and cried spontaneously on delivery. Five hours after delivery, he was referred to Sardjito Hospital due to abnormal abdominal wall, presence of meconium and had micturate. On examination, abdominal muscle hypoplasia (**Figure 1**) and bilateral cryptochirdism (**Figure 2**) were found, without any pulmonary abnormalities. Suspected to have Prune-belly syndrome, he underwent USG examination of urinary tract, and bilateral hydronephrosis stage 4 was found (**Figure 3**) without testis in the scrotum. On X-ray, scoliosis (deformity of the vertebral column) was also found (**Figure 4**). Diuresis on day 2 was only 0.3 ml/kg/day and urine was turbid. Urinalysis results were as follows: leukocyte 1-2, erythrocyte +3, leukocyte esterase 500 CFU/ml, protein 0, nitrite +, urine culture: *Candida albicans* (10^6), infection of urinary tract occurred due to stasis, and was treated with ketoconazole and catheterized via external urethral orifice. Patient was referred to pediatric surgeon and urology surgeon, and was planned to have abdominoplasty, surgical correction of urinary tract and orchidopexy. However due to financial problem, the patient was still on conservative treatment.

Case 2

A baby was born to a 31-year-old mother, G2P1A0, full term pregnancy, birth weight was 2200 gram, assisted by midwife with Apgar scores of 6/8, premature rupture of membrane was denied and she was referred to Sardjito Hospital three hours after delivery due to congenital anomalies.

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Figure 1. Peculiar wrinkled appearance of abdominal wall resembling "prune"

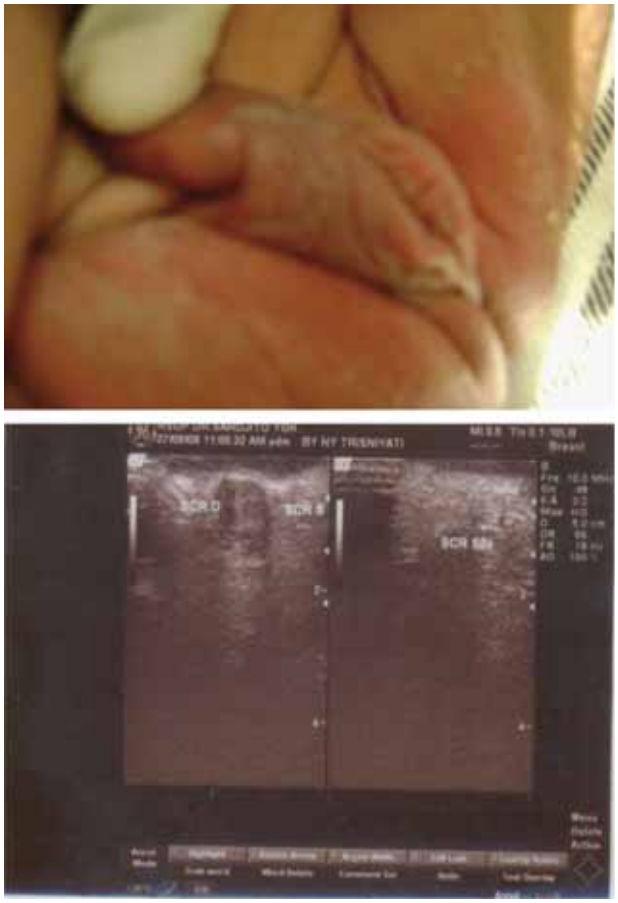

Figure 2. Neither testicles found in scrotum nor in USG examination (cryptochirdism)

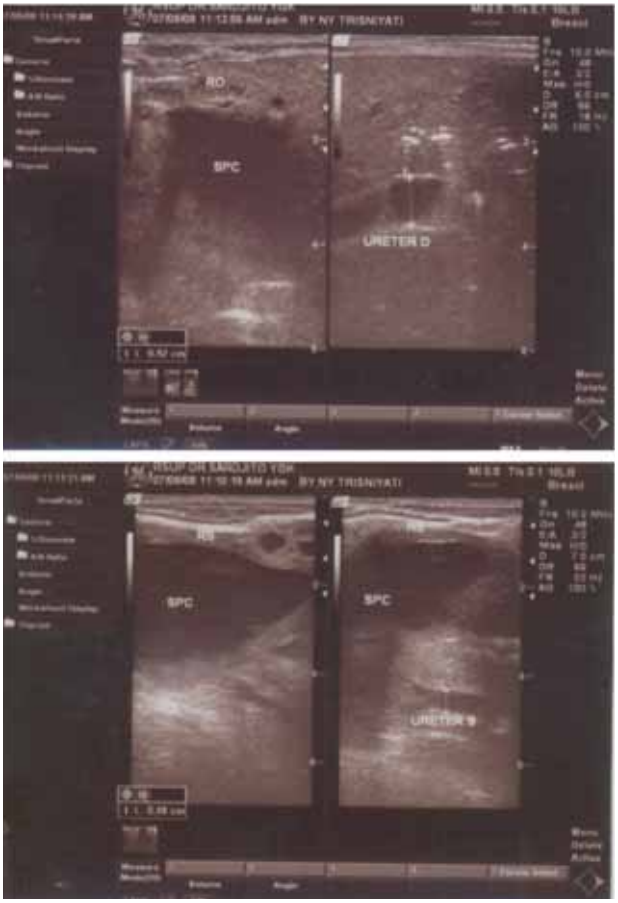

Figure 3. Right and left hydronephrosis on USG of urinary tract

On examination at Sardjito Hospital, disclosed thin, loose and wrinkled abdominal skin, omphalocele 3 x 3 x 4 cm, anal atresia, urethral atresia and vaginal atresia that led her being on plain photo of abdomen. She was diagnosed as having Prune-belly syndrome.

On the second day of life, the patient underwent colonostomy to repair anal atresia and cystotomy in order to insert suprapubic catheter. At the age of 15 months old, she underwent ureteroplasty, and anal reconstruction surgery by 18 months old. She suffered

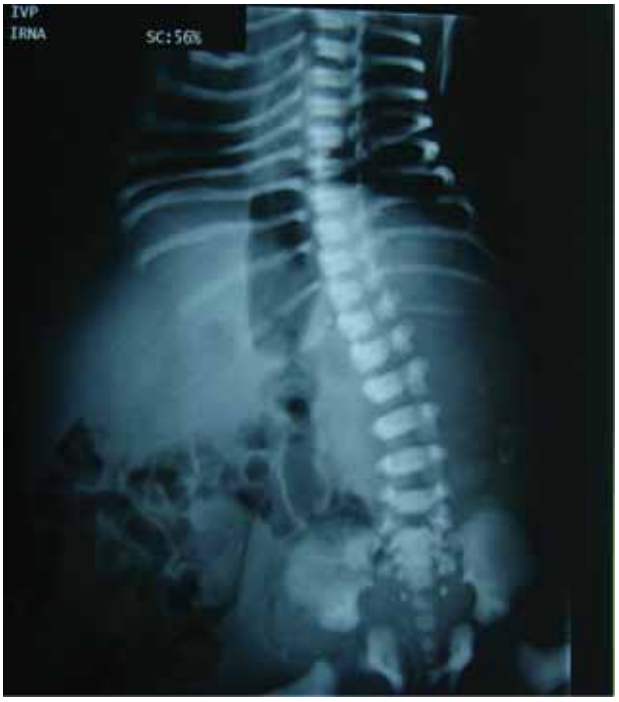

Figure 4. Scoliosis appearance

from urinary tract infection and recurrent respiratory tract infection. Recently at the age of three years old, she was treated for urinary tract infection caused by *Escherichia coli*, and suffered from malnutrition with body weight of 7.5 kg. She was treated for malnutrition and was given antibiotic cotrimoxazol and nalidixate acid. Body weight on discharge was 8 kg and she was recovered from urinary tract infection. The patient was asked to do follow-up treatment for colonostomy and ureteroplasty at pediatric surgery and urologic surgery clinic for further management, but the patient did not come.

Case 3

A boy was born to a 24-year-old mother, P1A0, 37th week of pregnancy via spontaneous vaginal delivery with severe asphyxia (Apgar scores 2/4), premature rupture of membrane was denied, birth weight was 2600 grams. Physical examination revealed wrinkled abdominal skin, absence of testicles in the scrotum, asymmetrical extremities (hip dislocation), and bilateral equinovarus. Fifteen minutes after delivery, his condition worsen, cyanosis occurred, subcostal and suprasternal retracted, vesicular sound of lung decreased, inadequate breathing, oxygen saturation decreased from 86% to 71% to 60%. In addition, blood gas analysis revealed respiratory acidosis and severe hypoxia. Resuscitation was done, but the patient was confirmed dead four hours after delivery due to respiratory failure.

Case 4

Patient was a referred patient from midwife, presented with asphyxia and congenital anomalies. He was a baby boy born to a 25-year-old mother, P1A0 via spontaneous delivery. He was delivered at term with birth weight of 3500 grams. His Apgar scores were 2/4, premature rupture of membrane was denied. He was sent to emergency ward of Sardjito Hospital 30 minutes after delivery, with bad general condition. In addition, he suffered from shortness of breath and cyanosis or mottled of his skin. Oxygen saturation was 60% with inadequate breathing. Blood gas analysis showed respiratory acidosis and severe hypoxia. Physical examination revealed wrinkled abdominal skin and absence of testicles in the scrotum. Skeletal anomaly was not found. Urinary tract anomalies were not examined since the patient had not yet micturated until he died at the age of three hours.

Discussion

The most essential characteristic of Prune-belly syndrome is loose and wrinkled abdominal skin, resembling prune. Thin abdominal muscle will result in visualization of intestinal peristalsis and easy palpation of internal organ,[5] as reported in these four mentioned cases. Abdominal wall musculature hypoplasia not only affects cosmetically but also causes chronic constipation due to ineffective Valsalva ability.[2]

Varying degrees of urinary tract abnormalities can be found associated with Prune-belly syndrome including hydronephrosis, ureteral atresia, megaureter, hypoplastic kidney, megacystic, and bladder atrophy. There are also cases of babies surviving during their neonatal period then suffering from recurrent urinary tract infection and stasis, in which 30% of the case will result in chronic or terminal renal failure

requiring dialysis or renal transplant in childhood or adolescence. Presence of renal dysplasia will worsen its function and lead to renal failure at younger age. This in fact is an important prognostic factor in patient diagnosed with Prune-belly syndrome.[6]

Prune-belly syndrome almost solely affects boys, presented with bilateral cryptochirdism and infertility, but normal testosterone level, normal libido and sexual function.[7] Three of four cases above are boys with absence of testicles in the scrotum. In girls, apart from abdominal musculature hypoplasia and urinary tract abnormalities, genital defects are also found, including vesicovaginal fistula, bicornus uterus and vaginal atresia,[7] as in Case 2.

Renal dysplasia that has occurred since fetal period can lead to oligohydramnion related to skeletal deformity in 45% of Prune-belly syndrome cases. Oligohydramnion will cause fetal compression that results in variant skeletal deformations including clubfoot, hip dislocation, syndactily, torticollis and scoliosis,[8,9] as in Case 3.

Oligohydramnion can also result in severe complication as pulmonary hypoplasia that causes respiratory distress after delivery with higher mortality chance if associated with Prune-belly syndrome. In the last two cases, pulmonary hypoplasia was suspected to cause the breathing distress that led to perinatal death. Oligohydramnion will reduce the pressure in amniotic cavity that in turn will cause excessive leakage of lung fluid into the interstitial cavity of the baby's lungs during delivery. Death can occur due to respiratory distress within two hours after birth or few days if ventilator is used.[10]

Abnormalities of the digestive tract are also reported in 30% of Prune-belly syndrome cases. Among the abnormalities are malrotation, anal atresia, duodenal stenosis, volvulus, and chronic constipation.[5] In addition, congenital cardiac anomalies, facial dysmorphism, and single umbilical artery have also been reported in Prune-belly syndrome cases.[4]

The etiology of Prune-belly syndrome remains unknown, but there are two theories which predominate: (1) Intrauterine mesoderm growth defect theory, in which the defect of mesoderm plate occurs during 8-10 weeks of gestation, from which the abdominal wall and urinary tract arise,[9] (2) The urethral obstruction theory which proposes that distal urethral obstruction in early gestation causes dilation of the bladder and ureters, forming a physical barrier to abdominal wall musculature development and testicular descent.[7]

The major prognostic factor in Prune-belly syndrome is the degree of renal impairment. Patients with severe renal dysplasia and resultant pulmonary hypoplasia often die in the perinatal period from respiratory failure. Among those patients surviving the neonatal period, 30% will develop chronic renal insufficiency or end stage renal disease requiring dialysis or transplantation in childhood or adolescence. Patients with the mildest urinary tract involvement, without obstruction or impaired function, have a normal life expectancy. The overall mortality rate in Prune-belly syndrome is 60%.[11]

The diagnosis of Prune-belly syndrome is commonly made by prenatal ultrasound and also visualization of the prune-like abdominal wall at birth. Initial assessment should be aimed at identifying pulmonary or cardiac distress.[4] Further assessment should be done to detect infection or anatomical defect of urinary tract and renal function. Though the patients reported normal for the assessments, routine monitoring should always be performed.[12]

Treatment of Prune-belly syndrome includes abdominoplasty, orchidopexy, urinary tract reconstruction, and supportive management as dictated by associated pulmonary, gastrointestinal and skeletal manifestations.[3] Apart from cosmetic reason, abdominoplasty may also improve respiratory and bladder function and reduce scoliosis. Time of surgery is still debatable, some suggest surgery at younger age will reduce the diameter of ureter dilation which results in better appearance.[6] Others suggest conservative treatment, while surgery is only indicated if there is pyelonephritis and creatinine level of more than 0.7 mg/dl.[11] Several surgery techniques have been developed, including Monfort & Erlich techniques, which are reported with better outcome and lower post-operative mortality compared to standard techniques.[13] Abdominoplasty assisted by laparoscopy in combination with orchidopexy, cystoplasty, and peritoneal catheterization using Tenckhoff method was successfully done to under one year old patients[14].

Peritoneal dialysis is last necessary for Prune-belly syndrome patients with renal failure, because neonate's vessels are hard to find.[10] Due to loose

and wrinkled abdominal wall muscle, catheterization using automated peritoneal dialysis (APD) is more preferred that continuous ambulatory peritoneal dialysis (CAPD), using small single cuff and normal saline with flow rate of 100-150 ml/min, volume of 300-600 ml/m^2 and pressure of <10 cmH_2O.[15,16]

As a conclusion, early detection during neonatal with visualization of typical Prune-belly syndrome together with detection of abnormalities of other systems will result in better and more comprehensive management, reducing chances of complication, and lowering mortality rate of neonates with Prune-belly syndrome in absence of pulmonary hypoplasia.

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