Prune Belly Syndrome: Extremely Rare Case in a Female Baby

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Abstract

Prune Belly syndrome is usually found exclusively in males. I report an extremely rare case of a female baby who died shortly after birth with cystic dysplasia of kidneys, gross ureteric dilation, distended hypertrophied urinary bladder with no internal or external openings, but communicated with a distended abnormal mullerian structure into which both ureters opened. Under development of the anterior abdominal wall cause 'prune belly' appearance with associated findings of coxa vara, talipes equinovarus right foot, malrotated gut, absent spleen, variant of tetralogy of Fallot with pulmonary atresia.

Keywords

RARE PRUNE BELLY IN FEMALE, MUSCULOSKELETAL, GASTROINTESTINAL, CARDIAC AND PULMONARY ANOMALY

Introduction

Prune belly syndrome (PBS) is a rare genetic disorder with incidence of 1:30,000-50,000 live births, occurs more frequently in twin pregnancies [1]. 97% in males being extremely rare in females with only about 30 reported cases [2, 3]. I report Prune belly in a female baby who died shortly after birth with PBS triad of distended lax abdominal wall, cystic dysplasia of kidneys, gross ureteric dilation, distended hypertrophied urinary bladder with no internal or external openings but communicated with a distended abnormal mullerian structure into which both ureters opened. Under development of the anterior abdominal wall cause 'prune belly' appearance with associated findings of coxa vara, talipes equinovarus right foot, malrotated gut, absent spleen, variant of tetralogy of Fallot with pulmonary atresia.

Case Report

A single live preterm girl baby born by assisted breech delivery was severely asphyxiated at birth and expired at one hour of age. External malformations included microcephaly with head circumference of 28.5 cm, Potters facies and single palmar crease on hands, bilateral coxa vara and calcaneovarus of right foot. The abdominal musculature was deficient and the abdomen grossly distended. Masses were palpable in both the lumbar and hypogastric regions. The external genitalia consisted of vulva, no identifiable clitoris with common cloaca.

Autopsy revealed deficient abdominal musculature, cystic dysplasia of both kidneys with markedly dilated pelvis and ureters. The urinary bladder was hypertrophied and distended with no internal or external openings, but communicated with a distended abnormal mullerian structure into which both ureters opened. The gut was malrotated with an imperforate anus, absent spleen, pulmonary atresia and cyanotic cardiac malformation tetralogy of Fallot (TOF) with hypertrophied right ventricle, defect of ventricular septum with overriding of aorta [4].

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Discussion

Parker in 1895 [5] first described triad of deficient abdominal musculature, resembling a wrinkled prune, renal and urinary tract dysplasia with cryptorchidism also known as Eagle Barret syndrome or triad syndrome [6]. Other synonyms are abdominal muscle deficiency syndrome, Obrinsky syndrome [7], Frolich syndrome [8]. Though exact aetiology is unknown some studies reveal genetic inheritance with possible association with trisomy 18 and 21 [1]. However two main pathogenic hypotheses by clinical studies indicate mesodermal defect occurring early in embryogenesis, sixth to tenth weeks involving first lumbar myotome causing patchy deficiency of abdominal wall musculature with abnormalities of kidneys, ureter and bladder [9], secondly the abdominal wall atrophy occurs due to chronic intrauterine abdominal distension as a result of severe obstructive uropathy caused by haploinsufficiency of 1-beta hepatocyte nuclear factor due to de novo 1.3 megabase interstitial microdeletion of 17q12 with ureteral hypoplasia [10].

Characteristic triad is deficient development of abdominal muscles that causes the skin of the abdomen to wrinkle like a prune with abnormalities of the urinary tract such as bilateral gross hydrenephrosis, megaureter and megacystitis, in males with bilateral cryptorchidism, associated with other anomaly such as respiratory with pulmonary hypoplasia, gastrointestinal such as intestinal malrotation, imperforate anus, musculoskeletal including polydactyl/syndactyly and talipes equinovarus, pectus excavatum, scoliosis, congenital joint dislocation including hip and cardiovascular anomalies such as variant of tetralogy of Fallot. Prognosis is usually poor with stillbirth or early neonatal deaths.

Other renal anomalies include hydronephrosis, cystic or hypoplastic dysplasia with elongated tortuous extremely dilated ureters, distended hypertrophied bladder due to obstruction at the vesical neck with hypoplasia to absence of urethra or a persistent patent urachus causing urinary ascites, the increased pressure causing lax abdominal wall, oligohydramnios with pulmonary hypoplasia and bilateral cryptorchidism by interfering with descent of testes [2, 11].

Prenatal diagnosis by ultrasonography usually confirms diagnosis, however a differential diagnosis is posterior urethral valve with hydroureter and hydronephrosis confirmed by key hole sign with no evidence of cryptorchidism or megacystitic microcolon, intestinal hypoperistalsis syndrome who tend to have polyhydraminos usually in males. Prenatal therapeutic option include in utero placement of vesicoureteric shunt to prevent development of PBS [12]. After birth clinical examination and investigations such as ultrasonography of abdomen, plain X-ray, intravenous pyelography and micturating cystourethrography can confirm diagnosis, chest X-ray to rule out pneumothorax, pulmonary hypoplasia and pneumomediastinum.

Management requires a multidisciplinary approach between neonatology, nephrology, urology and other departments. Treatment depends on severity of symptoms. Severe PBS in category I described by Woodard in 1985 [13] with renal dysplasia, urethral atresia causing oligohydraminos, pulmonary hypoplasia/oligohydramnios usually in males. Prenatal therapeutic option include in utero placement of vesicoureteric shunt to prevent development of PBS [12]. After birth clinical examination and investigations such as ultrasonography of abdomen, plain X-ray, intravenous pyelography and micturating cystourethrography can confirm diagnosis, chest X-ray to rule out pneumothorax, pulmonary hypoplasia and pneumomediastinum.

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In category II patients may survive infancy have mild triad or unilateral renal dysplasia that may progress to renal failure but without pulmonary hypoplasia. Management options includes voiding cystourethrography with antibiotic cover in suspected cases of renal insufficiency or suprapubic catheter in bladder outlet obstruction (BOO) to prevent urinary tract infections (UTI) and in boys, orchidopexy to move testes into scrotum. In addition reduction cystoplasty, ureteric shortening tapering and reimplantation with or without abdominoplasty (Monfort technique), which may prove ineffective as the bladder will again stretch due to lack of musculature. Vesicoureteral reflux is common in those with poor bladder contractility, dilated posterior urethra without urethral obstruction result in frequent urinary tract infections due to inability to expel urine often lead to progressive renal insufficiency.

However those in category III with mild features of PBS and stable renal function without renal dysplasia who may have mild to moderate uropathy with no pulmonary hypoplasia have better prognosis and even survived into adult life after urinary tract repair and abdominal reconstructive surgery, may in later life experience a common symptom of post-ejaculatory discomfort lasting for about two hours probably due to bladder spasm [15].
Follow-up with careful surveillance and intervention for repeated febrile UTI or renal deterioration is recommended.

**Conclusion**

Extremely rare case report of a female baby with Prune belly unamenable to surgery with pulmonary atresia, cystic dysplasia of kidneys markedly dilated pelvis and ureters, massive hypertrophied and distended bladder with no internal or external openings but communicated with a distended abnormal mullerian structure into which both ureters opened. Other associated anomalies included microcephaly with simian crease could indicate aneuploidy or trisomy 21; other associated anomalies were talipes equinovarus, dislocation of hips, gastrointestinal anomalies of malrotation of gut with imperforate anus and splenic agenesis with Tetralogy of Fallot.

**References**


