International Journal of Health Sciences and Research
www.ijhsr.org ISSN: 2249-9571

Case Report

Prune Belly Syndrome with Bilateral Hydronephrosis - A Case Report

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Received: 02/03//2014 *Revised: 29/03/2014* *Accepted: 02/04/2014*

ABSTRACT

Prune belly syndrome is a rare congenital anomaly of uncertain etiology almost exclusive to males. It is characterized by the triad of absent or incomplete abdominal musculature, undescended testes, and urinary tract abnormalities. A male baby with above characteristic triad was brought to our hospital. A diagnosis of prune-belly syndrome was made. Here we report a case of prune-belly syndrome, in order to highlight the occurrence of this rare syndrome in our environment and to review its pathogenesis, presentation and management.

Key words: Prune-belly syndrome, hydronephrosis.

INTRODUCTION

Prune belly syndrome was described for the first time in the 1800s.[1] It is a rare condition defined by the triad of abdominal muscle deficiency, severe urinary tract abnormality and cryptorchidism.[2-4] Other names for Prune Belly Syndrome are Eagle-Barret syndrome, Triad syndrome, Osler-Parker Syndrome. It is caused by urethral obstruction early in development resulting in massive bladder distension and urinary ascites, leading to degeneration of the abdominal wall musculature and failure of testicular descent. The urinary tract may have variable degrees of hydronephrosis, renal dysplasia, dilated tortuous ureters, enlarged bladder, and a dilated prostatic urethra.[5] The exact etiology is unknown. The case is reported for its rare congenital abnormality.

CASE REPORT

A 3.0kg male baby was brought to our hospital by an un-booked 23-year old Gravida 1, Para 1, mother with history of abdominal distension since birth, fever and lethargy since 2days and not taking feeds since one day. On examination vitals were stable. Abdominal examination revealed huge distension with thin abdominal skin, protruding most prominently on the right side (fig.1). Both the kidneys were palpable. Perineal examination showed bilateral cryptorchidism (fig.2). Other systems were within normal limits. Investigations showed Hb14.3gm%, Tc 17,000/mm, ESR 36mm/1hr, Serum creatinine 0.3mg, Blood

urea 20mg. Urine examination showed plenty of pus cells and klebsiella was grown on culture. Ultrasonography showed congenital posterior urethral valve with bilateral hydronephrosis with hydroureter and distended bowel loops. Diagnosis of prune belly syndrome with posterior urethral valve with bilateral hydronephrosis with urinary tract infection was made. Kyariotyping was normal. Baby was referred to higher center for further management.

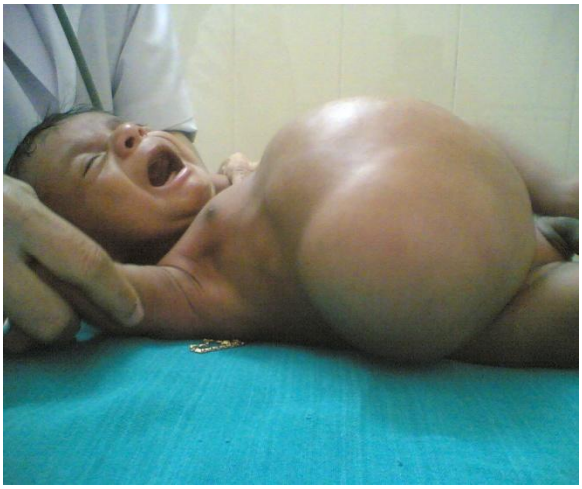

Fig-1. Baby with prune belly syndrome showing protruded abdomen with thin abdominal wall.

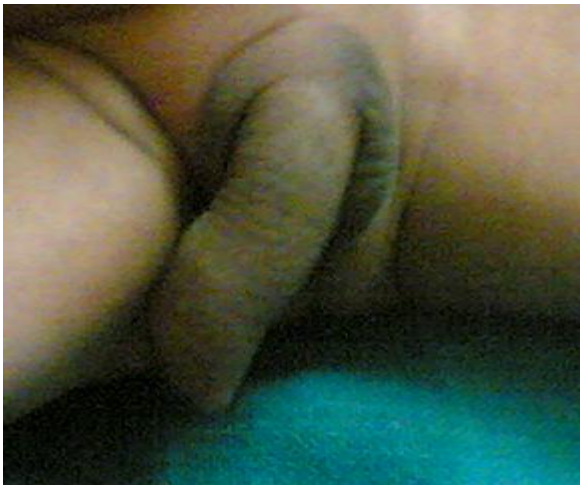

Fig 2. Baby with prune belly syndrome showing bilateral undescended testes.

DISCUSSION

This anomaly was first described by Frolichs in 1839[6]. Parker in 1895 presented a case with deficient abdominal wall musculature and dilatation of the urinary tract.[7,8] The term Prune Belly was first coined by Oslar in 1901, as the skin of the patient had dried plum appearance with skin creases.[8] In 1950, Eagle Barret described a syndrome involving congenital deficiency of the abdominal muscles with associated genitourinary abnormalities called the "Triad`` or Eagle Barret Syndrome.[8] History says that infants born with a full blown syndrome had a poor prognosis for long term survival.[6] Incidence: 1: 35,000 to 1:50,000 live births. Twins, blacks and children born to younger mother appear to be at higher risk in epidemiological studies.[6,9] The defect is almost exclusively seen in males. A fully developed syndrome is seen only in this sex.[7] Clinically significant non-urological features are noted in 7.3% of cases, among which cardiac 10%, pulmonary 55%, gastrointestinal 31% , and orthopedics being 40-63% .[10,11] The genetic basis has been explored but no clear inheritance pattern has emerged. The observation of 100% discordance among all twins in whom monozygosity has been proven, goes against a genetic aetiology.[12] However, the reported association with Turner's syndrome, monosomy 16, Trisomy 13 and Trisomy 15 confound identification of a simple gene locus.[13]

The pathogenesis is not clearly known. The mesodermal defect theory suggests that a defect exists in the mesoderm of the anterior abdominal wall and urinary tract. Between 6 and 10 weeks of gestation, aberrant development of the derivatives of the first lumbar myotome leads to a patchy muscular deficiency or hypoplasia of the abdominal wall as well as to urinary tract abnormalities.[14,15] An alternate theory, proposes that pressure atrophy of the

abdominal wall muscles occurs when urethral obstruction leads to massive distension of the bladder and ureters. Bladder distension would also interfere with descent of the testes and thus be responsible for the bilateral cryptorchidism. This mechanism is responsible for the urinary tract dilatation and distension.[16]

The higher incidence of this syndrome in males has been explained on the basis of the more complex morphogenesis of the male urethra, possibly resulting in obstructive anomalies at several levels. Ultrasound, plain X-ray, and intravenous pyelogram are more useful investigations to diagnose the condition. Many neonates with this syndrome have difficulty with effective bladder emptying because the bladder musculature is poorly developed. When no obstruction is present, the goal of treatment is the prevention of urinary tract infection with antibiotic prophylaxis. When obstruction of the ureters or urethra is demonstrated, temporary drainage procedures, such as a vesicostomy, may help to preserve renal function until the child is old enough for surgery. Urinary tract infections occur often and should be treated promptly. Correction of the undescended testes by orchidopexy can be difficult in these children because the testes are located high in the abdomen. Reconstruction of the abdominal wall offers cosmetic and functional benefits.[5]

The prognosis ultimately depends on the degree of pulmonary hypoplasia and renal dysplasia. The most common complication is chronic renal failure that occurs in 25–30% of cases. Many infants are either stillborn or die within the first few weeks of life from severe lung or kidney problems, or a combination of congenital anomalies. There are cases of this syndrome who survived into adult life after abdominal reconstruction and urinary tract repair.[17]

CONCLUSION

Early diagnosis of this syndrome and determining its optimal treatment are very important in helping to avoid its fatal course. These patients need multidisciplinary management of a neonatologist, nephrologists, and pediatric urologist for an optimal outcome. Pregnant ladies should have regular antenatal check up with anomaly scan and health education regarding intake of nutritious food, folic acid, required vitamins and minerals. Genetic counseling of parents is important, especially those with a history of abortion, perinatal death, or with a family history of genetic diseases.

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How to cite this article: Kumar GV, Shivaleela C, Viswanathakumar HM et. al. Prune belly syndrome with bilateral hydronephrosis - a case report. Int J Health Sci Res. 2014;4(4):233-236.

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