

Prune Belly Syndrome - A Rare Case with Complex Malformations

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Abstract: *Prune Belly Syndrome is a congenital triad of abdominal muscle deficiency, malformations of urinary tract and cryptorchidism forms a rare congenital malformation. The disorder affects males and exact etiology is unknown. The prognosis of babies with Prune Belly Syndrome is poor. We report a case of Prune Belly Syndrome in a 19 weeks male abort.*

Keywords: Prune Belly Syndrome, abdominal wall musculature deficiency, bilateral multicystic renal dysplasia, cryptorchidism

1. Introduction

Prune Belly Syndrome (PBS) is a rare congenital anomaly affecting 1 in 30,000 births¹. It is characterized by triad of abdominal muscle deficiency, malformations of urinary tract and cryptorchidism. The syndrome is seen almost exclusively in males². The exact etiology of PBS is not known. However some studies reveal possibility of genetic inheritance³. Many infants are either still birth or die within first week of life. We present a rare case of Prune Belly Syndrome in a 19 weeks male abort.

2. Case Report

A 21 years old lady, G3 P1 A1, presented with 19 weeks amenorrhea for antenatal check up. She had history of intrauterine foetal death during 1st pregnancy and 12 weeks spontaneous abortion in 2nd pregnancy. There was no history of consanguineous marriage. The ultrasonography revealed complex malformations in a foetus with oligohydramnios. The pregnancy was terminated and the foetus was sent for autopsy.

3. Autopsy Report

External examinations revealed a male foetus weighing 1200 gms with signs of prematurity, Potter's facies, and imperforated anus. The abdomen was distended showing parchment like thin and wrinkled skin (Fig-1). Umbilical cord revealed two blood vessels. Systemic examination revealed hypoplastic lungs. Both kidneys revealed multiple cysts on gross examination. Microscopy of both kidneys showed features of multicystic renal dysplasia i.e. cystically dilated tubules with immature disorganized mesenchymal tissue and nephrons (Fig-2). Both the ureters were dilated. Urinary bladder was distended, thick walled filled with considerable amount of residual urine (Fig-3). Both the testis were rudimentary and lying in pelvic region (bilateral cryptorchidism)

4. Discussion

Prune Belly Syndrome also known as Eagle Barret Syndrome is a rare congenital malformation consisting of a

triad of deficient development of abdominal muscle, abnormalities of urinary tract and bilateral cryptorchidism.

The syndrome is associated with other malformations including pulmonary hypoplasia as well as gastrointestinal, musculoskeletal and cardiovascular defects². PBS was initially reported by Frolich in 1839 although the term Prune Belly Syndrome was coined by Osler⁴. Eagle and Barret reported 9 cases in 1950 and described the condition as Eagle Barret Syndrome⁵. Other names used in the literature include triad syndrome and abdominal musculature deficiency syndrome⁶.

5. Pathogenesis

The syndrome is exclusively seen in males and the exact etiology is not known. Different pathogenic theories have been proposed for the malformation in PBS. The theory of urethral obstruction states that bladder distention and urinary tract dilatation is because of urethral obstruction. The Bladder distention prevents development of normal abdominal musculature and descent of testis⁷. One hypothesis is the presence of prostatic hypoplasia which may result in transient urethral obstruction⁸. The other main theory is failure of development of mesoderm resulting into complex congenital malformation⁹. Our case presented in a male foetus with a classical triad of PBS along with bilateral hypoplasia of lungs, multicystic renal dysplasia, single umbilical artery and an imperforate anus. Bad obstetric history was revealed in mother which suggests the possibility of genetic inheritance.

6. Diagnosis

Antenatal diagnosis of PBS can be done in second trimester of pregnancy. Oligohydramnios associated with PBS is secondary to reduced urine output, poor renal function and subsequent lung hypoplasia.

Differential Diagnosis – Include posterior urethral valve and Megacystic Microcolon Intestinal Hypoperistalsis Syndrome (MMIHS)

Management – Neonatal management includes serial evaluation of serum electrolytes, blood urea nitrogen and creatinine levels during the first week of birth. Primary aim

is to preserve renal function. Other necessary interventions are abdominal muscular wall reconstruction and orchiopexy for prevention of testicular malignancy¹⁰.

Prognosis - Prognosis of PBS is poor. Many Infants are either still born or die within first few weeks of birth due to renal failure and pulmonary hypoplasia². Perinatal mortality rate for PBS is between 10- 25%. The coexisting morbidity is due to cardiovascular and pulmonary problems and prematurity¹¹⁻¹².

7. Conclusion

Prune Belly Syndrome is a rare congenital anomaly seen in male foetuses. Routine antenatal care with ultrasonography helps in detecting complex malformations seen in Prune Belly Syndrome. Optimal treatment thereafter can minimize fatal course of Prune Belly Syndrome.

References

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Figure 1: Gross findings-Distended thinned out anterior abdominal wall.

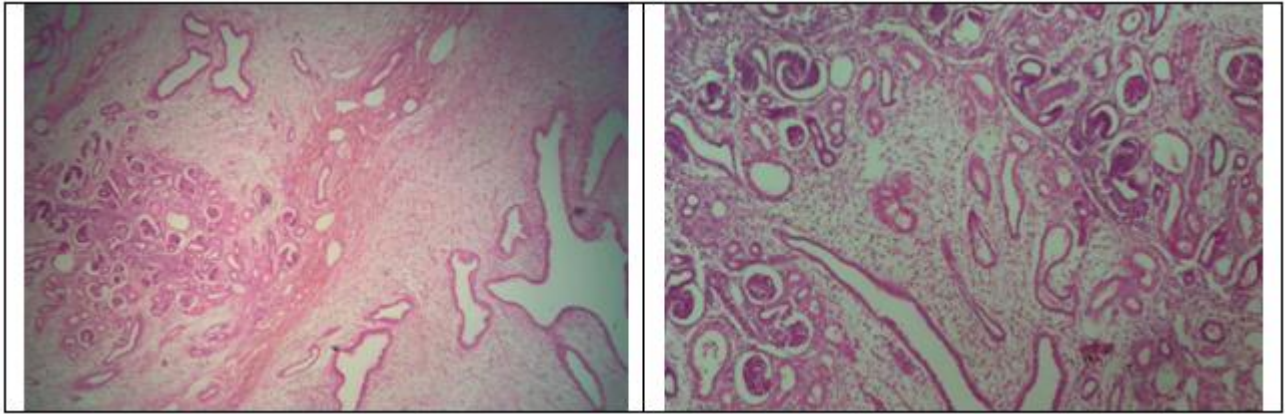


Figure 2: Microscopic findings: Cystic renal dysplasia showing cystically dilated tubules, disorganized nephrons and immature mesenchyme. (40x, 100x H&E.)



Figure 3: Thinned dilated bladder and Potter's facies.