

Case Report

Prune Belly Syndrome Associated With Anorectal Malformation and Acyanotic Congenital Heart Defect: A Rare Finding

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Abstract: Prune belly syndrome is a rare congenital anomaly. It is characterized by triad of defective abdominal musculature, anomalous urinary tract and bilateral cryptorchidism. Association of other malformations is known. We are describing a case of Prune Belly Syndrome associated with High Anorectal Malformation and Acyanotic Congenital Heart Defect which is extremely rare.

Keywords: Prune Belly Syndrome, High Anorectal Malformation, ASD, PDA.

INTRODUCTION

Prune belly syndrome is a rare congenital anomaly with incidence of 1 in 29000-50000[1]. It is characterized by deficient or absent abdominal musculature, anomalous urinary tract and bilateral cryptorchidism. The name Prune belly is given due to resemblance of abdominal skin to a fruit called prune. It is also known as Eagle Barret Syndrome, Frohlich syndrome or Obrinsky syndrome. Cardiac, pulmonary, gastrointestinal and skeletal malformations often coexist. We are describing an association of High Anorectal Malformation and Acyanotic Congenital Heart Defect in a case of PBS.

CASE REPORT

A 23-Year-old mother came to obstetrics department of Lok Nayak Hospital, New Delhi with full term pregnancy and labour pains. Her first pregnancy was an abortion; second pregnancy was term gestation normal delivery. There was no history of bleeding or discharge per vaginum, no history of diabetes, hypertension, tuberculosis, asthma, and epilepsy in current or previous pregnancies. No history of consanguinity. Per abdominal examination showed 38-39 wks gestation with normal presentation of fetus. Fetal heart sounds present. Ultrasonography revealed a single viable fetus with bilateral renal hydronephrosis.

Baby delivered via vaginal route, cried immediately after birth, weight was 2710 gm. On examination, baby's face was normal, abdomen was distended with overlying skin wrinkled, there was swelling above umbilicus which protrudes on crying,

penis size was 5.5 cm with coronal hypospadias, bifid scrotum was present with testis not palpable in scrotum, neither palpable in inguinal canal and ring (figure 1). Externally there was no anal opening seen. Cardiovascular examination had short systolic murmur.

Investigations- Post natal USG shown bilateral hydronephrosis- Right kidney measuring 6cm x2.4 cm with parenchymal thickness of 3mm, Left kidney measuring 6.3cmx2.7cm with parenchymal thickness of 3.6 mm, bilateral PCS had internal echoes, urinary bladder reaching umbilicus, increased bladder wall thickness present, bilateral ureters were obscured. Post natal ECHO done shown- Tiny ASD with PDA. USG Cranium shown normal intracranial structures. Blood Investigations- Haemoglobin- 13mg%, Platelets- 1.9 lacs, Blood urea- 25, Serum creatinine-1.3, Sodium- 137meq, Pottasium- 4.2meq. X ray Abdomen (figure 3) showed no gas reaching in lower abdomen and rectum suggestive of intestinal obstruction.

Child was operated next day, Intra operative findings were- descending colon and sigmoid colon stenosis with distal end of stenosed colon communicating with bladder, and rectum was absent not visualized. Procedure done was exploratory laprotomy with divided right transverse colostomy with vesicostomy (figure 2). Treatment given was IV Fluids and Antibiotics. However, child died of renal failure due to associated congenital renal malformation.



Fig. 1: Bilateral Cryptorchidism



Fig. 2: Prune Belly, Umbilical Hernia (colostomy with vesicostomy)



Fig. 3: X ray Abdomen Showing no gas in lower abdomen and rectum

DISCUSSION

Prune Belly Syndrome is characterized by the triad of abdominal wall defect, urinary tract anomaly and Bilateral cryptorchidism. In our case findings were – large umbilical hernia (Abdominal wall defect), bilateral hydronephrosis with bladder involvement (Urinary tract anomaly), bilateral undescended testis (Bilateral Cryptorchidism), new findings to this case are High Anorectal malformation associated with Acyanotic Heart Defect- Tiny PDA with ASD which is extremely rare.

There are various hypothetical theories explaining the occurrence of this congenital defect, few theories described in earlier studies are mesodermal

arrest theory, early urethrourethral obstruction and yolk sac defect during embryogenesis [2]. Genetic basis for prune belly syndrome has also been described, it has been found that hepatocyte nuclear factor 1 β , and a transcription factor required for visceral endoderm are involved, their deletion is associated with PBS[3]. Embryogenesis of male urethra is complex so higher incidence is seen in males, it is extremely rare in females' with less than 30 cases reported [4]. Lower intestinal tract anomaly was first described by Escobar et al [4] in PBS. Association of PBS with Anorectal Malformation and Acyanotic Congenital Heart Defect is an extremely rare finding.

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