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Case Report

A Case Report On Prune Belly Syndrome

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Abstract: 'Eagle-Barrett syndrome' is the other name for prune belly syndrome. The classical triad of urinary tract anomalies, deficient abdominal musculature, and bilateral cryptorchidism. It is a congenital anomaly of unknown aetiology, most commonly seen in males. It is seen in one and 30,000 live births.96% of them are males. In this case report, we present a boy child born to A 38-year-old multigravida mother with a previous history of three vaginal deliveries. On examination of the baby, the following features are to be found-hypotonia, deficiency of abdominal muscle, undescended testis, a palpable bladder and kidney. Radiological imaging showed gross hydronephrosis on both sides and big ureter. The baby was diagnosed with Prune Belly syndrome. Ultrasound investigations done during the antenatal period, helps in directing anomalies of the kidney which is to be followed in the postnatal period. The aim of the writing this case report is to establish the importance of antenatal diagnosis of congenital malformations including Eagle-Barrett Syndrome among many others. Prenatal diagnosis of PBS should be considered whenever the following ultra- sound anomalies are clearly identified: oligohydramnios, urinary abnormalities (dilatation of the urinary tract, megacystis, bilateral hydroureteronephrosis), and the absence of abdominal musculature. Routine screening during the antenatal period with ultrasound helps in detecting anomalies of the kidney at an earlier stage . This help in initiating optimal treatment to avoid the fatal cause of Prune Belly Syndrome

Keywords: Hydronephrosis, Big Ureter, Undescended Testis, Prune Belly Syndrome

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1. INTRODUCTION

Prune belly syndrome (PBS), also called Eagle-Barrett syndrome, is a multisystem condition with rare occurrence. Disease severity is staged by the extent of renal dysplasia and pulmonary involvement. Most of the PBS patients also have various concomitantly associated with non-genitourinary systems involving cardiopulmonary, gastrointestinal and musculoskeletal anomalies.[1–4]. It is seen in one and 30,000 live births.[5] About 96% of them are males.[6] Characteristic features include defective development of the muscles present in the abdominal region which leads to wrinkling of the abdomen skin like a prune, Undescended testis, gross hydronephrosis, big ureter, Megacystitis. [5] The aetiology of prone Valley syndrome is unknown but some studies have shown possible genetic inheritance and association with trisomy.[6,7] The prognosis is poor in stillbirth children and early infant deaths have been reported. No best practice guidelines exist for management of PBS, owing to the scarcity and complex nature of the disease, along with diverse multisystem co-morbidities. Since the incidence of Prune belly syndrome is rare, the diagnosis is often missed during the antenatal period and adds to the burden do the disease. Hence this case report has been made with the aim to establish the importance of antenatal diagnosis of congenital malformations including Eagle-Barrett Syndrome among many others to prevent disease burden in any form.

2. CASE REPORT

A 38-year-old G4 P3 L3 A0 with a just gestational age of 38 weeks 4 days was admitted in view of labour pain. All three previous deliveries were normal vaginal delivery. She is an uncooked case; she belongs to a lower socio-economic status. There is no history of exposure to radiation or drug intake or fever presenting with rash during the pregnancy period. There is no history of gestational hypertension, diabetes or genetic and congenital anomalies in the family. On examination, it was found that she was in her second stage of labour, the presentation of the fetus or cephalic with the regular fetal heart rate. The weight of the baby at birth was 2.6 kg, Apgar score was Eight and nine at first and fifth minute. Hypotonia was present at birth. On examination of the abdomen - the skin was wrinkled and peristalsis was visible. The kidney and bladder was palpable. On examination of the scrotum, the rugae was little and testis was absent in the sac. Other systems are normal. USG abdomen was gross hydronephrosis on both kidneys, and big ureter. Hence the baby was referred to the paediatric surgery department for further management.

Fig 1 is an X ray picture showing loose abdominal wall

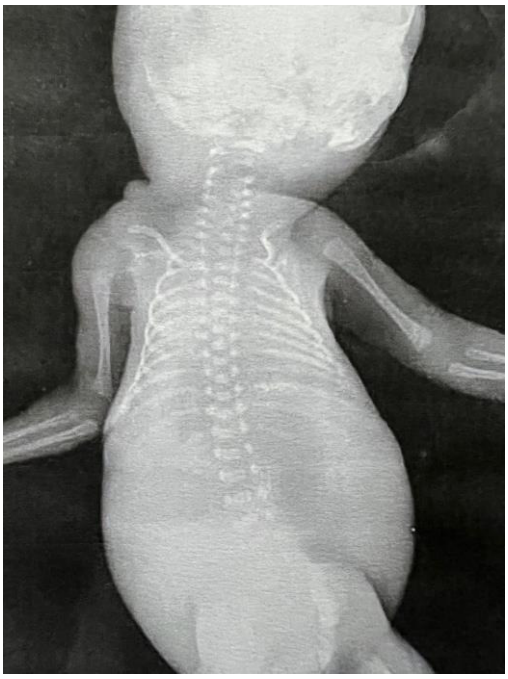
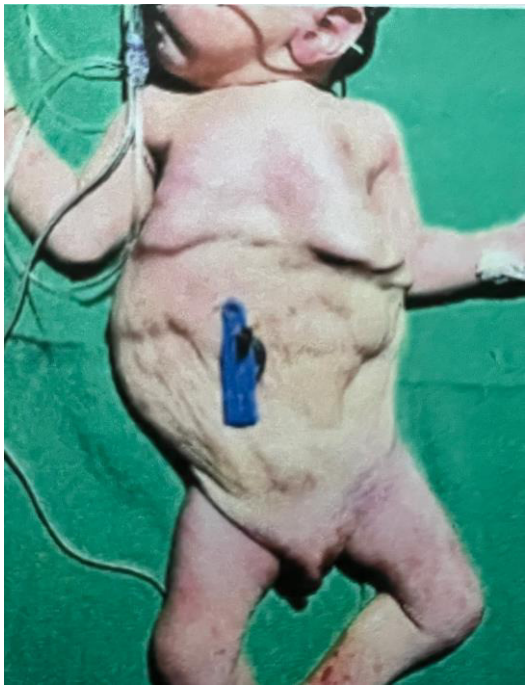


Fig 2 show the gross picture of loose abdominal wall



3. DISCUSSION

Eagle-Barrett syndrome is the other name for prune belly syndrome. The classical triad of urinary tract anomalies, deficient abdominal musculature, and bilateral cryptorchidism[8]. Cryptorchidism is one of the three key features. PBS most often occurs as a sporadic event, there is a high incidence rate in twins (12.2 per 100,000 live births), case reports of monozygotic male twins, familial case reports and males having higher incidence, all points towards a genetic contribution[9-12]. It is Associated Multi system involvement - musculoskeletal, heart, lungs and Genito urinary tract malformations. [6] The cause is not known, but familial inheritance seen in the past. [7] PBS is associated with a mutation of genomic HNF1β (hepatocyte nuclear factor) in 3 % of cases.[13-15] The association of PBS with de novo 1.3 megabase interstitial 17q12 microdeletion in which the hepatocyte nuclear factor- 1-beta gene at 17q12, and haploinsufficiency of hepatocyte nuclear factor-1-beta can lead to PBS phenotype by prostatic and ureteral hypoplasia which leads to severe obstructive uropathy with urinary tract and abdominal distension.[4] Elderly mother and history of consanguineous marriage indicates the hereditary cause. The abdominal wall muscle degeneration and the failure of testis descent is caused by huge distension of the urinary bladder and urinary ascites. Oligohydraminoa and Hypoplasia of the lungs is caused due to

improper elimination of urine. Hypothesis of Mesodermal defect and malformation due to urethral obstruction are the two main pathogenic hypotheses. Mesodermal defect hypothesis is aberrant development of the derivatives of the first lumbar myotome between 6 and 10 weeks of fetal life causes irregular patchy muscle deficiency or hypoplasia of the abdominal wall and sometimes include urinary tract abnormalities[16]. Urethral obstruction malformation hypothesis is where urethral obstruction leads to massive distension of the urinary bladder and ureters which in turn causes atrophy of the abdominal wall muscles due to the increase in pressure[17]. The exact mechanism remains elusive, as does the molecular basis for PBS. Bladder distension Interferes with the process of testis descent Thereby leading to bilateral Crytorchidism. Only about 30 cases have been in females. [18] A thorough clinical examination, ultrasound imaging of the abdomen, X-ray, IV pyelography and micturating cystourethrography can help in confirming the diagnosis.

Woodard classification of Prune Belly Syndrome Classification (1985) [19]:

3.1 Category 1

3.1.1 Renal dysplasia
3.1.2 Oligohydramnios
3.1.3 Pulmonary hypoplasia
3.1.4 Potter's facies
3.1.5 Urethral atresia

3.2 Category 2

3.2.1 Full triad features
3.2.2 Minimal or unilateral renal dysplasia
3.2.3 No pulmonary hypoplasia
3.2.4 Renal failure

3.3 Category 3

3.3.1 Incomplete or mild triad features
3.3.2 Mild to moderate uropathy
3.3.3 Absence of renal dysplasia
3.3.4 Stable renal function
3.3.5 Absence of pulmonary hypoplasia.

Our patient discussed here falls under category 2. Neonatology, Nephrology, Urology departments play a major role in treating babies with prune belly syndrome. Voiding cystourethrography with antibiotic usage in cases with suspected renal insufficiency or bladder outlet obstruction (BOO), suprapubic catheter in BOO helps in preventing urinary tract infections, orchidopexy and Chest X-ray PA view to exclude pneumothorax, pulmonary hypoplasia and pneumomediastinum. Monfort technique can be performed for those who fall under category 2. The procedure includes reduction cystoplasy, ureteric shortening tapering and reimplantation with or without abdominoplasty. Differential diagnosis of PBS, includes causes of LUTO including posterior urethral valves, ureterocele and urethral atresia, megacystis-microcolon-intestinal- hypoperistalsis syndrome (MMIHS). Prenatal diagnosis of PBS should be considered whenever the following ultra- sound anomalies are clearly identified: oligohydramnios, urinary abnormalities (dilatation of the urinary tract, mega- cystis, bilateral hydroureteronephrosis), and the absence of abdominal musculature. Early, accurate diagnosis allows not only for prompt multidisciplinary management of new- borns in a tertiary Center at birth, resulting in improved survival, but also allows for the option of voluntary termination if desired. Prognosis is poor because those babies who are still born die in the early neonatal period.The most common complication is chronic failure of kidneys. 6 Renal shutdown is the reason for death in prune belly syndrome babies. [20] There is good prognosis when the Serum Creatinine levels are less than 60 μmol/l. When diagnosed in the antenatal period by ultrasonography intrauterine therapeutic option including in utero placement of a vesicouterine shunt can be done to prevent the development of PBS. [21]

4. CONCLUSION

Prune belly syndrome is a congenital anomaly that is rare and there is no preventive measure. Also, routine screening during the antenatal period with ultrasound helps in detecting anomalies of the kidney at an earlier stage. This help in initiating optimal treatment to avoid the fatal cause of Prune Belly Syndrome. Like other congenital anomalies which are complex in nature, the key to management of PBS is a multidisciplinary team-based approach providing individualized care. Long-term surveillance of the urinary tract is crucial as bladder dynamics and renal function can change over time. As technological advances, continue to improve the overall survival and life expectancy of PBS patients, the challenge remains to develop best practice standards and provide comprehensive care while mitigating potential negative disease sequelae.

5. AUTHORS CONTRIBUTION STATEMENT

Dr.Clince conceptualized and gathered the data with regard to this work. Dr. Ravanagomagan, Dr.Shanthi R and Dr.Sundari S analyzed these data and necessary inputs were given towards the designing of the manuscript. All authors discussed the methodology and contributed to the final manuscript.

6. CONFLICT OF INTEREST

Conflict of interest declared none.

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