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Case Report

Symphony of challenges-prune belly syndrome with congenital heart disease

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ABSTRACT

Rare congenital anomaly prune belly syndrome (PBS) affects male infants. Usually characterized by anterior abdominal wall muscle aplasia, cryptorchidism, and urinary tract defects. This is a similar case of a term male baby admitted to our NICU. Cryptorchidism, anterior abdominal wall weakness, and massive bladder dilation were found. The diagnosis was made clinically and then we confirmed the diagnosis by ultrasonography which revealed massive urinary bladder dilation with bilateral hydronephrosis, and thinning of the renal cortex. On clinical evaluation murmur could be heard for which 2D echo was planned and a large arterial septal defect (Left to right shunt) was present. Treatment mainly in volves orchidopexy, anterior abdominoplasty as well as urinary tract reconstruction.

Keywords: PBS, Atrial septal defect, Hydronephrosis, Congenital heart disease, Genitourinary tract defect, Abdominal wall defect

INTRODUCTION

The rare prune belly syndrome (PBS) or triad syndrome affects male neonates. There are bilateral undescended testes, complex urinary tract anomalies, and absent anterior abdominal wall muscles.[1]

Approximately 1 in 30,000 to 1 in 40 000 live births have the anomaly, with >95% in males and three to 5% in females.

PBS babies often die early or have stillbirths. The rare and term birth of a male baby with PBS requires reporting.

CASE REPORT

A newborn male was delivered in our setup of an elderly mother of 36 years, multipara, born out of non-consanguineous marriage with no significant genetic history. Delivery was done via vaginal route cephalic presentation. Apgar of 6 and 8 at 1 and 5 mins respectively. Meconium passed immediately after birth dribbling of urine present so the patient was catheterized.

Vital parameters were within normal limits. No facial dysmorphism was seen. A nasal catheter was passed to rule out choanal atresia and tracheoesophageal fistula but no coiling was present. A mild systolic murmur could be heard.

The abdomen was distended, and asymmetrical, with visible bowel loops a lump could be visualized in the suprapubic region extending to the umbilical region.

On further examination, anterior abdominal wall hypoplasia was appreciated. A humongous lump was palpable in the suprapubic region extended to the umbilical region with smooth surface and round margins all findings suggestive of the urinary bladder.

On the left flank, kidneys were palpable which was

ballotable. Intestines were felt beneath the abdominal wall.

Scrotum with good rugae present but scrotal sac was empty due to undescended testis.

Figure 1: Anterior abdominal wall hypoplasia.

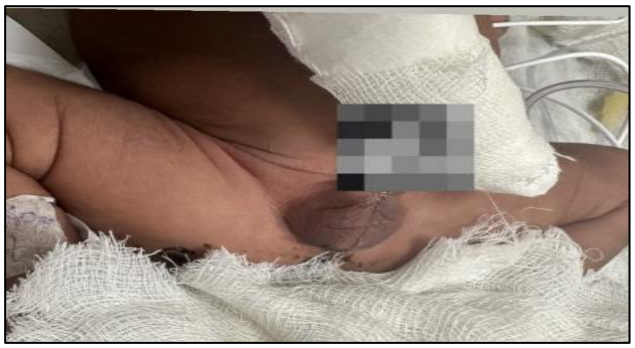

Figure 2: Bilateral undescended testis.

USG whole abdomen and KUB done which was suggestive of left-sided gross hydronephrosis with thinned-out renal parenchyma.

Right-sided moderate hydronephrosis. Over distended urinary bladder up to the epigastric region and bilateral flanks causing compression of bowel and adjacent structures.

Bilateral testis was abdominally placed.

2D ECHO was done which was suggestive of acyanotic congenital heart disease, parge atrial septal defect (ASD) 11 mm left to right shunt right atrium as well as right ventricle dilated.

Management consisted of maintaining electrolyte balanced Enteral feed was started via an orogastric tube. Orchidopexy, urinary tract reconstruction, and abdominoplasty were scheduled but the baby died on the eighth day.

Figure 3: 2D-ECHO suggestive of large ASD.

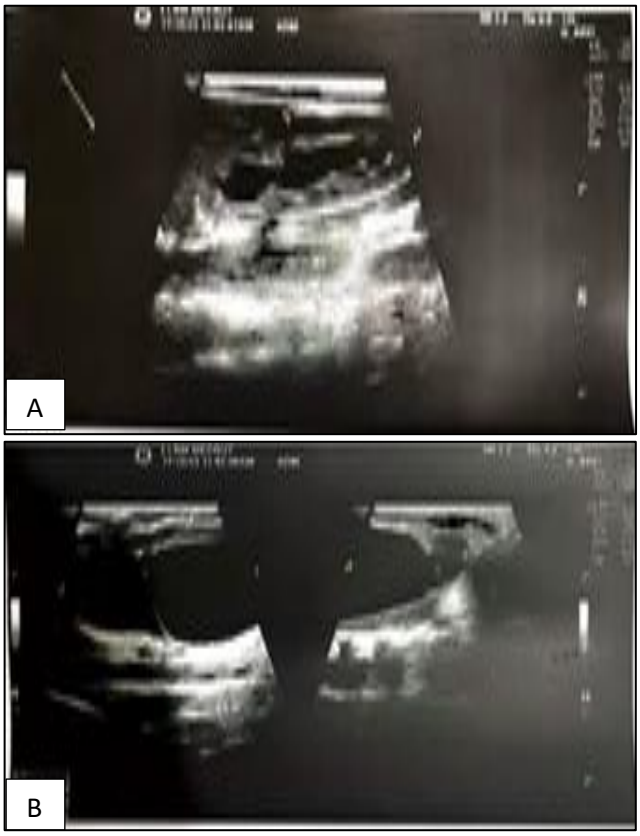

Figure 4 (A and B): Ultrasonography suggestive of gross hydronephrosis, renal parenchyma thinning, and undescended testis.

DISCUSSION

Prune means a dried fruit (plum-like). PBS is a rare congenital malformation that involves deficient abdominal wall muscle, undescended testicles, and urinary tract malformations.[2]

The global prevalence is anticipated at 1 case per 40,000 live births.[3] PBS was previously associated with other malformations, but VACTERL was rarely associated.[4]

In our case patient has all features of PBS described as cryptorchidism, abdominal muscle hypoplasia, and urinary tract abnormalities with additional features of a large 11 mm ASD. As reported by Routh et al there is a significant incidence of twenty-five percent for cardiovascular defects, twenty-four percent for gastrointestinal birth defects, twenty-three percent for musculoskeletal defects, fifty-eight percent for respiratory defects, and fifteen percent for genital malformations. These are only a few of the many other malformations that may be linked to PBS.[5]

The most prevalent associated respiratory malformation, pulmonary hypoplasia, may be a significant contributor to precocious neonatal mortality in PBS and may cause varying degrees of respiratory failure. It is not uncommon to have digestive malformations like Hirschsprung disease, gastroschisis, atresia, stenosis, volvulus, anal imperforation, and splenic torsion. Additionally, there may be birth defects related to the bones, such as scoliosis, hip dysplasia, clubfoot, and vertebral malformations. Potential cardiovascular malformations include ductus arteriosus and Fallot tetralogy.

There is still debate regarding the pathogenesis of PBS, with the following theories dominating the field: According to the first theory, there is a urinary tract blockage during pregnancy, which results in fetal abdominal distension, urinary tract dilatation, and subsequent muscle wall hypoplasia as well as cryptorchism.[6] A theory based on embryology suggests that between the sixth and tenth week of gestational age (GA), primary mesodermal differentiation fails, resulting in abnormal muscularization of the urinary tract as well as the abdominal wall.[7] In fact, the yolk sac theory suggests that PBS is caused by a dysgenesis of the yolk sac and allantois.[8]

Prenatal ultrasound can detect a PBS as early as twelve weeks of gestational age. Usually, abnormal bladder dilatation, decreased amniotic fluid, bilateral hydronephrosis, and absent abdominal muscles result in a lower abdominal cystic echo.[9]

An antenatal scan performed in the 22nd week of pregnancy in our case also suggested gross hydronephrosis with a distended bladder.

Postnatally, the sonomorphologic classification listed below is utilized: Grade I°-Kidneys that are dysplastic and lack distinct renal parenchyma around them. Grade II°-Significant ureteric dilatation and little to no renal pelvic and calyces dilatation. Grade III°-milder involvement, ranging from grade II sonographic results to normal-appearing urinary tract findings.[10]

Based on the sonomorphological classification described above, our case is classified as grade 2.

Intrauterine shunt therapy has been developed by a study; however, vesico-amniotic shunt therapy has to meet certain requirements in order to have a good chance of success. These include renal function being normal, as determined by serial analyses of fetal urine, a normal karyotype, and careful sonographic examination ruling out other malformations. It is generally best to insert the shunt as soon as feasible.[11]

CONCLUSION

Other than early, routine prenatal screening for fetal abnormalities, there is no known preventive measure for PBS, a rare congenital condition. Early screening and treatment can prevent the fatal course of PBS. Patients who survive the neonatal period now have a better survival rate thanks to modern care; 25-30% of these patients experience end-stage renal failure and chronic renal failure.

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