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CASE REPORTS

Prenatal Diagnosis of Prune Belly Syndrome: A Case Report

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Abstract Prune belly syndrome (PBS) is a rare congenital syndrome characterised by a triad of deficiency of abdominal wall muscles, undescended testis and dilated urinary tract. It may be related to lower urinary tract obstruction with renal dysfunction. Early and accurate diagnosis with proper treatment is important to prevent renal impairment. We report Ultrasound (US) and Magnetic Resonance Imaging (MRI) findings of a case of prune belly syndrome diagnosed at 20 weeks of gestation. US showed grossly dilated fetal urinary bladder, dilated ureters and kidneys with no identifiable renal parenchyma, mild ascites and severe oligohydramnios. Fetal MRI confirmed the ultrasound findings.

Case Report

A 30- years- old female (gravida 4 para 3) presented to our department for a fetal anomaly scan at 23 weeks of gestation (by last menstrual period). She had no complaints or significant past medical and family history. She was a non-alcoholic and non-smoker.

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Her previous ultrasound done elsewhere documented the presence of fetal gastroschisis and polyhydramnios.

Ultrasound revealed severe oligohydramnios with an amniotic fluid index (AFI) of 3 cm. A very large, smooth, thick-walled cystic lesion was noted communicating with dilated right ureter (Fig. 1). Fetal abdomen was severely distended with moderate free fluid in the peritoneal cavity in which small bowel loops were freely floating (Fig. 2). Both renal pelvicalyceal system were moderately dilated (left > right). Fetal anterior abdominal wall was severely thinned and visualized in the inferior quadrant only due to the presence of a small pocket of amniotic fluid. The right kidney showed a dilated pelvicalyceal system with normal cortico-medullary differentiation. The left kidney showed mild hydronephrosis with mild renal parenchymal thinning, a few tiny intra-renal cysts and loss of cortico-medullary differentiation. A provisional ultrasound diagnosis of prune belly syndrome with megacystis was made.

Fetal MRI performed on a 3 Tesla MR system, confirmed the ultrasound findings. The MRI demonstrated a severely distended abdomen with thinned out, irregular fetal abdominal wall and moderate ascites (Fig. 3 A &B). The large cystic abdomino-pelvic mass was a severely dilated urinary bladder. The communication of the urinary bladder with mild dilated bilateral ureters was clearly visualised. The posterior urethra was not dilated (Fig. 4). Fetal body parts were crowded due to severe oligohydramnios. Thoracic circumference was severely reduced indicating pulmonary hypoplasia.

The patient was counseled of options including termination of pregnancy by her treating physician. Postnatal fetal examination revealed a grossly distended abdomen and severely thinned abdominal wall (Fig. 5). The baby had associated club foot with cleft lip and palate.

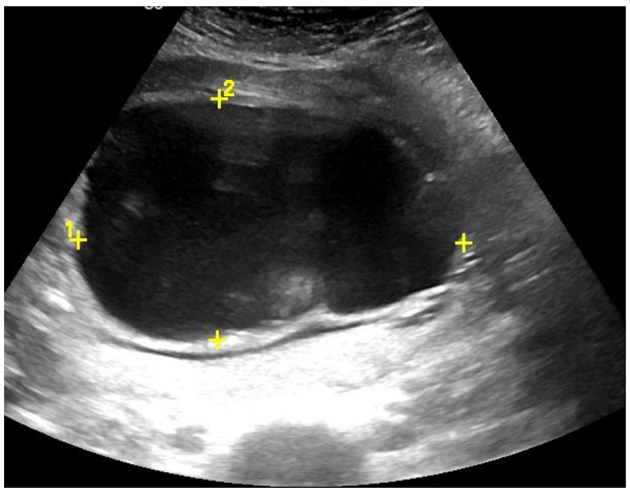

Fig. 1 A grossly dilated urinary bladder that occupies most of the ballooned-out abdomen with smooth and regular walls

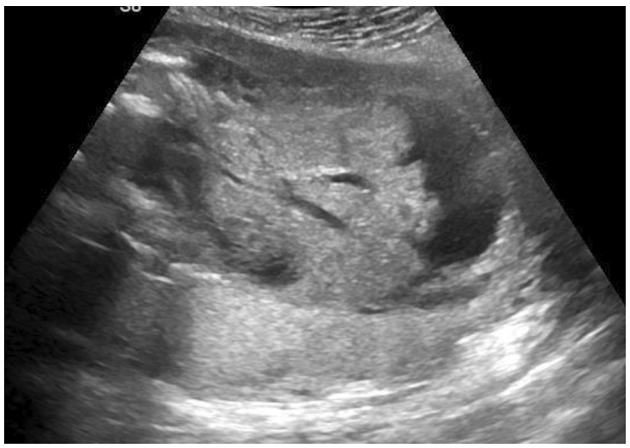

Fig. 2 US shows moderate free fluid in the peritoneal cavity. The anterior abdominal wall is not separately seen from the uterine wall due to severe oligohydramnios giving a false impression of a large fetal anterior abdominal wall defect with herniation of liver and small bowel

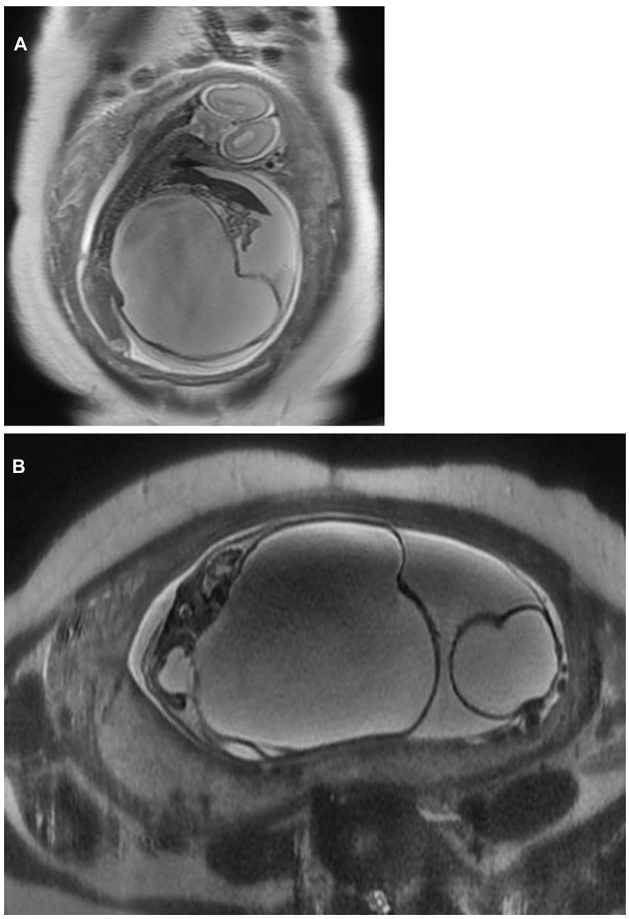

Fig. 3 A & B: Fetal MRI confirmed moderate free fluid in the peritoneal cavity with severe oligohydramnios. Fetal abdominal wall is severely thinned out. Large cystic structure involving abdomino-pelvic cavity representing grossly dilated urinary bladder. The right renal pelvis is moderately dilated. The dilated ureter is seen opening in the lateral wall of the urinary bladder. Left renal parenchyma shows altered signal intensity

Discussion

PBS is a rare disorder with an incidence of one in 35,000–50,000 live births [1]. PBS is a specific constellation of anomalies consisting of deficiency of abdominal wall muscles, failure of testicular descent and dilation of the urinary tract. PBS almost always involves boys, females are rarely affected. The exact pathogenesis of PBS is unclear but mostly due to a combination of a primary mesodermal developmental defect, severe bladder outflow obstruction and teratogenic effect [2].

The presence of hypoplastic or dysplastic prostate results in obstruction of the urethra and overdistension of the urinary bladder and the upper urinary tract. Poor renal function leads to oligohydramnios and subsequent lung hypoplasia. Concomitant ureteral anomalies like megaloureter and bulbar or penile urethral diverticulum may occur [3].

The most credible theory for the development of prune belly syndrome is a primary mesodermal developmental defect caused by any insult between 6 and 10 weeks gestation. This results from failure of the lateral mesoderm to migrate or differentiate into the musculature of the abdominal wall and urinary tract.

Associated musculoskeletal malformations are seen in 20–60% of cases (congenital hip dislocation, club feet, talipes deformities, vertebral anomalies and polydactyly). Gastrointestinal anomalies like gut malrotation, anal imperforation and atresia are seen in 30–40% of individuals. Cardiac vascular anomalies are also seen in few cases [1].

The genetic basis of prune belly syndrome has not been well established. Recently the role of chromosome 17q12

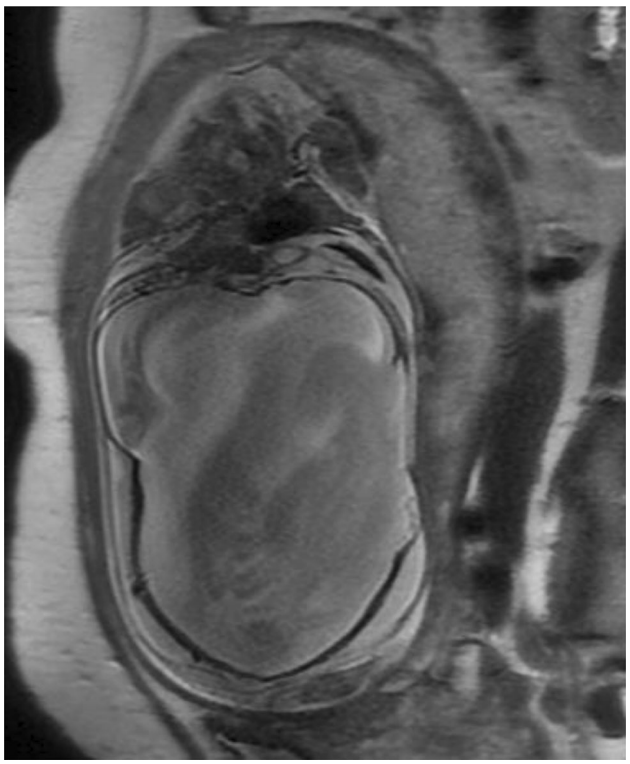

Fig. 4 Coronal HASTE MRI sections reveal typically smooth, nonuniform wall thickness of urinary bladder. No evidence of a dilated posterior urethra is noted. Thinned out abdominal wall is seen

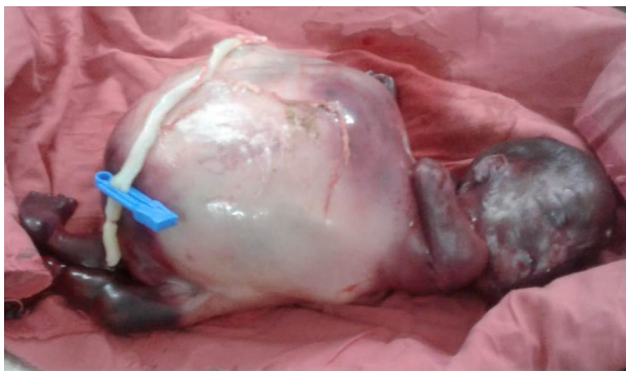

Fig. 5 Postpartum image of the neonate shows grossly distended abdomen with a membranous abdominal wall. Fetus shows Potter's facies: low set ears, dysmorphic chin and club feet

deletions encompassing the HNF1β gene as a candidate gene for prune belly syndrome has been suggested [2].

A classification of Prune belly syndrome into 3 categories was proposed by Woodard on the basis of antenatal and anatomical features [4]. Category 1 is characterised by the presence of severe renal dysfunction and pulmonary hypoplasia and carries the worst prognosis. Category 2 patients have a wide variation in the degree of renal dysplasia along with the classic features of PBS. Category 3 patients have a normal renal function and mild phenotypic features of PBS [5]. Our case was classified as category 1 because of the presence of severe oligohydramnios and severely reduced thoracic diameter.

As the degree of renal dysplasia is the single most important factor that determines the prognosis, prenatal intervention to preserve renal function can prevent renal dysplasia, pulmonary hypoplasia and may improve postnatal quality of life.

Radiological Findings

Classical prenatal ultrasound findings of PBS primarily reflect sequelae of severe urethral obstruction. Ultrasound shows a large ballooned-out fetal abdomen which is disproportionate for thoracic diameter. A severely dilated urinary bladder (megacystis) with or without a dilated upper urinary tract is seen. The abdominal wall appears thin and membranous with irregular abdominal wall circumference. Diagnosis of PBS is possible in early pregnancy if megacystis is present. However, classic ultrasound features may not be identified until later in pregnancy. The appearance of kidneys varies widely ranging from variable degrees of hydronephrosis to severe renal dysplasia.

Fetal MRI is not commonly performed for PBS but confirms sonographic findings as in our case. In our case, it was very difficult to separate the anterior abdominal wall from chorio-amniotic layer on ultrasound due to severe oligohydramnios. On ultrasound, the absence of floating umbilical cord loops within free fluid suggested fetal ascites instead of abdominal wall defects with polyhydramnios.

Prenatal diagnosis of PBS should be considered when there is oligohydramnios, urinary abnormalities (dilatation of the urinary tract, megacystis, bilateral hydroureteronephrosis) and the absence of abdominal musculature. Differential diagnosis of PBS includes other causes of lower urinary tract obstruction including posterior urethral valves, ureterocele and urethral atresia and other syndromes with megacystis such as megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS). The absence of dilated posterior urethra (keyhole sign) is an important clue to rule out posterior urethral valves [5].

Pulmonary complications are prevalent in PBS neonates while chronic kidney disease is commonly seen during late childhood.

Antenatal intervention in PBS can be performed at 17–20 weeks of gestation in a fetus showing signs of lower urinary tract obstruction. Decompression of the urinary system in utero can be achieved via insertion of a vesico-amniotic shunt. The outcome is variable. Amniotic fluid

level is usually increased but renal function is variably restored.

PBS is characterised by the presence of low pressure dilation of the urinary tract with enlarged, hypotonic urinary bladder and low pressure vesicoureteral reflux (VUR) in approximately 75% of cases. [6].

There is no consensus regarding treatment or timing of surgery in PBS and treatment protocols are tailored according to each case. However, early simultaneous correction of PBS anomalies with urinary tract intervention, bilateral orchidopexy and abdominal wall reconstruction has been described in the literature. [7].

Author Contribution Shivya Parashar diagnosed the case, collected all the images, searched the literature and prepared the manuscript. Rajesh Malik confirmed the diagnosis, searched the literature and reviewed the manuscript. Radha S Gupta searched the literature and reviewed the manuscript. Kamaljeet S Randhawa collected patient's clinical information, followed up with the patient and collected postnatal information.

References

1. Aaronson IA, Cremin BJ (eds). Prune belly syndrome: Clinical paediatric uroradiology. London: Churchill Livingstone, 1984. pp. 255–65.
2. Vani A, et al. Prune belly syndrome (sequenze): a case report. Int J Res Med Sci. 2017;1(2):148–52.
3. Nunn IN, Stephens FD. The triad syndrome: a composite anomaly of the abdominal wall, urinary system and testes. J Urol. 1961;86:782–94.
4. KA Woodward PJ, Soahey R, Byrne JLB, Oh KY, Puchalski MD. Diagnostic imaging obstetrics. Salt Lake City: Amirsys Inc; 2005.
5. Hassett S, Smith GH, Holland AJ. Prune belly syndrome. Pediatr Surg Int. 2012;28:219–28.
6. Bellah RD, States LJ, Duckett JW. Pseudoprune-belly syndrome: imaging findings and clinical outcome. AJR Am J Roentgenol. 1996;167:1389–93.
7. Denes FT, Arap MA, AM S, FA A. Comprehensive surgical treatment of prune belly syndrome: 17 years' experience with 32 patients. Urology. 2004; 64:789–793.