International Journal of Research in Medical Sciences
Vani A et al. Int J Res Med Sci. 2013 May;1(2):148-152
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: 10.5455/2320-6012.ijrms20130523

Case Report

Prune belly syndrome (sequenze): a case report

A. Vani*, S. Saritha, K. Sangeetha, G. Supriya

Department of Anatomy, Medical College (KIMS), Hyderabad, Andhra Pradesh, India

Received: 27 March 2013
Accepted: 9 April 2013

***Correspondence:**
Dr. A. Vani,
E-mail: dr.achantavani@gmail.com

ABSTRACT

The Prune Belly syndrome (PBS) also known as Eagle Barret syndrome is a rare disorder. It is an abdominal muscles deficiency syndrome characterized by a Triad syndrome i.e. deficiency of abdominal wall muscles, failure of testicular descent and dilation of the urinary tract. This syndrome has derived its name from the wrinkled prune appearance of the abdominal wall. Prune Belly syndrome is a rare anomaly seen in one in 35,000-50,000 live births. It occurs in all races. Prune Belly syndrome almost exclusively occurs in males (M:F, 20:1). The diagnosis can be made in utero by ultrasonography at 21 weeks of gestation or in the Neonate with characteristic clinical findings. The present case was a dead male fetus of 20 weeks of gestation sent to Anatomy department after Medical termination of pregnancy, due to congenital anomalies identified in routine ultrasound examination during antenatal checkup.

Keywords: Ultrasound, Prune Belly syndrome (PBS), Genital system, Urinary tract, Cryptorchidism and Abdominal wall musculature

INTRODUCTION

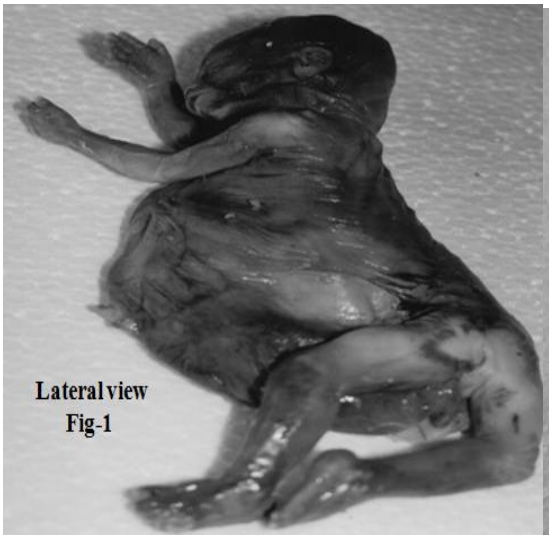
The Prune Belly syndrome (PBS) an uncommon congenital anomaly results from failure of lateral mesoderm to migrate or differentiate into the musculature of abdominal wall and urinary tract which normally occurs at about the tenth week of fetal period. Mainly it affects the males. Nearly one third of the babies is still born or dies within first few weeks of life. This PBS in its complete form is characterized by congenital absence of muscles of the abdomen wall and malformation of the urogenital tract. The lax excessive skin of the abdomen looks like the wrinkled skin of the Prune. Other features include persistent furrow like umbilicus, testes are intra-abdominal, cryptorchidism being attributed to the failure of development of the inguinal canal and gubernaculum, malrotation of gut, hip dislocation and lower limb abnormalities.

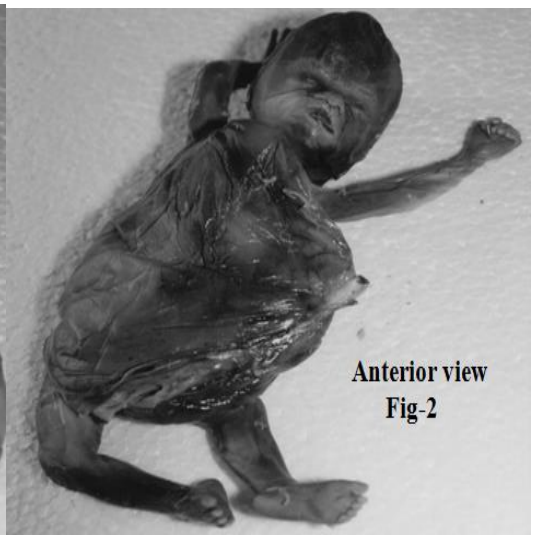
The present case was a dead male fetus of 20 weeks old, received from the department of Obstetrics and Gynecology. Postnatal it was diagnosed as PBS. The fetus was dissected after preservation. The gross appearance of our index case presented with complete features of various organs according to PBS. The embryological basis of the malformation and genetic concept were discussed in this article.

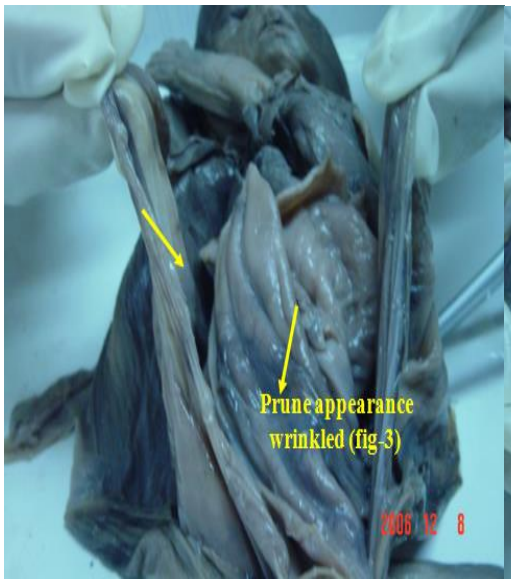
CASE REPORT

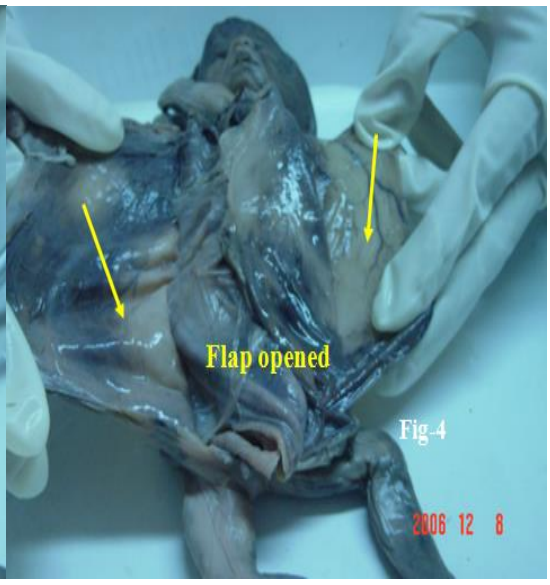
An 18 year old primi with 20 weeks of gestation presented to the hospital for routine antenatal checkup. Ultrasonography scanning was done (Figure 10). She was G1P0A0 at the time of scanning. She gave history of non-consanguineous marriage, did not take any drugs. The Ultrasound revealed live male fetus with 20 weeks of gestation. The scan also revealed that the fetus had large cystic swelling occupying the abdominal region and anterior abdominal wall was not formed and with rotated

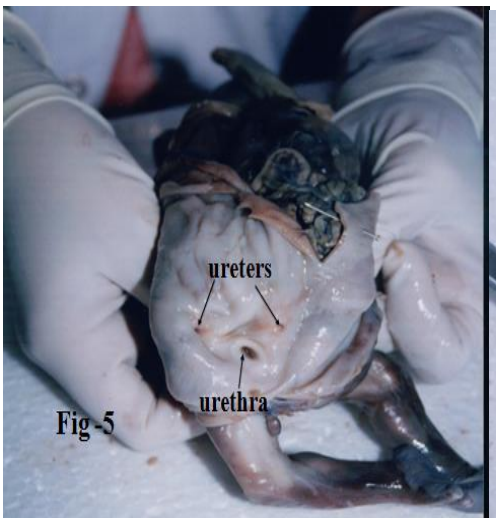
lower limbs. The mother was informed of poor prognosis and after counseling her, termination of pregnancy was done. The detailed autopsy was done in the Anatomy dissection hall and the diagnosis was made as Prune Belly syndrome.

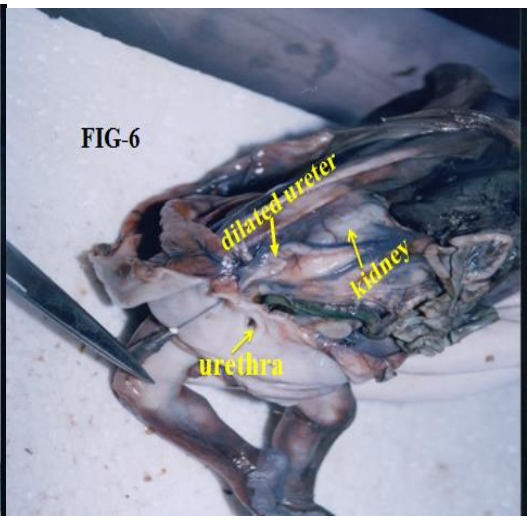


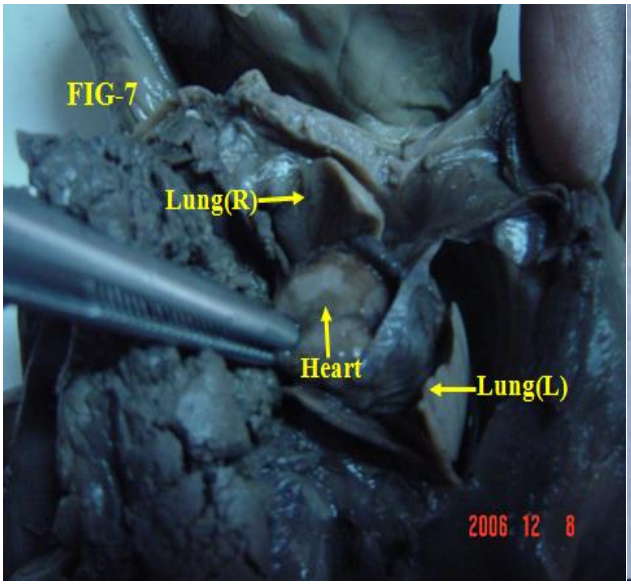


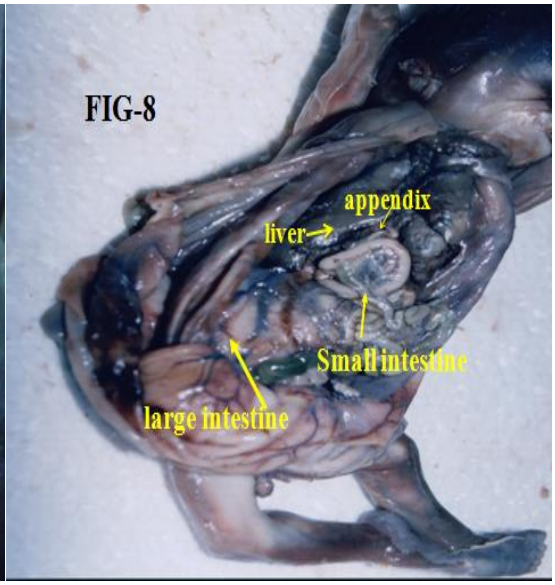


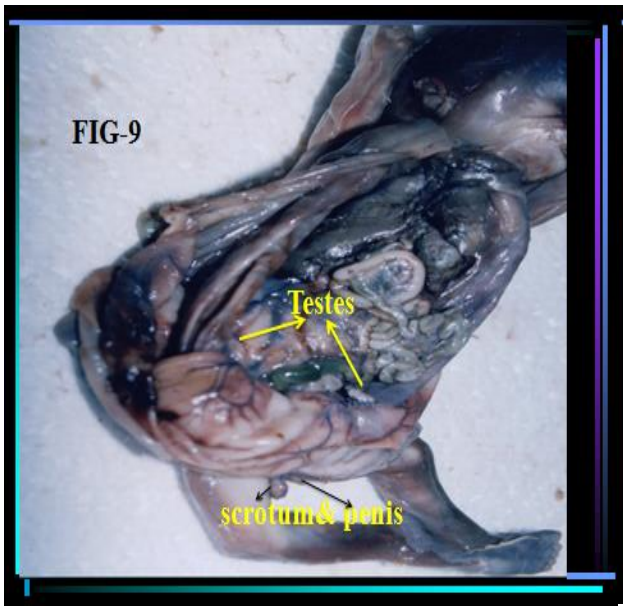


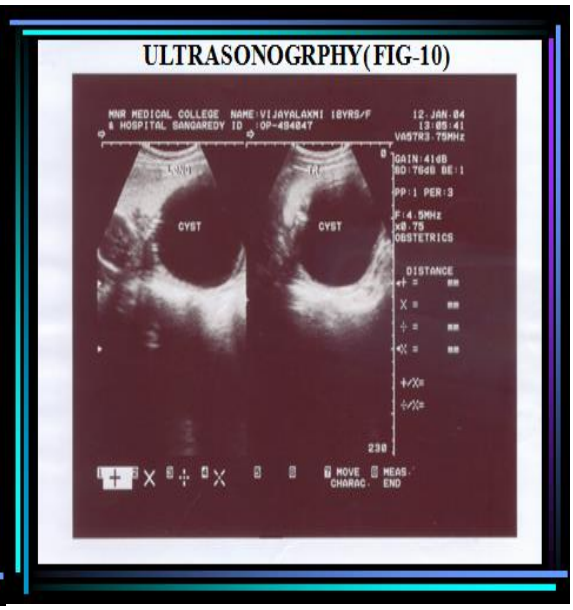












External features (Figure 1, 2 & 3)

The present case was dead male fetus aged 20 weeks. On examination there was a large cystic swelling occupying the entire abdominal region with bulging flanks and thin flabby wrinkled abdominal skin. The lower limbs were contracted and malrotated with bilateral club feet. The upper limbs also showed contractures. The abdominal wall showed complete absence of musculature and scanty subcutaneous tissue. There was extreme laxity of the abdomen and it looked wrinkled skin of a prune. It was therefore diagnosed as Prune Belly Syndrome. Later the diagnoses were confirmed by the dissection of the internal structures. External genitalia showed small penis with undeveloped scrotum and bilateral testes not palpable within the scrotum (Figure 9).

Internal features

The anterior wall of the cyst was opened and it was found to be large dilated urinary bladder (Figure 4). The urinary bladder showed three external openings i.e. two ureteric openings and single opening of urethra (Figure 5). The kidneys were both lobulated, dilated and ureters were also tortuous and dilated (Figure 6). Lungs were hypoplastic and Heart with no anomalies. The Diaphragm was present (Figure 7). Malrotation of the gut was seen. The Large intestine on the right side and Small intestine on the left side with appendix just below the Heart but separated by the diaphragm (Figure 8). Bilateral testes were seen intra-abdominal and at the sacroiliac level (Figure 9).

Ultrasonography observations (Figure 10)

An ultrasound showed a male fetus of 20 weeks gestation with a disproportionately large ballooned out distended

abdomen probably cystic in nature with over all abdominal diameter several times that of the thoracic diameter.

DISCUSSION

The Prune belly syndrome was first described by Frolich in 1839 and summarized later in detail by W. Obrinsky in 1949. Prune Belly syndrome is a rare genetic birth defect affecting about 1 in 40,000 births. About 97% of those affected are males. 4% of all cases are seen more frequently in twin pregnancies with 4 time's higher incidence than singleton and 2 times higher if woman is less than 25 years old. There have been reports of siblings being born with prune belly syndrome, indicating that there is a potential genetic factor. While in the womb, the developing baby's abdomen swells with the fluid. The fluid disappears after birth, leading to a wrinkled abdomen that looks like a prune. Prune Belly syndrome is also called Eagle-Barrett syndrome. Other names for the syndrome include Abdominal Muscle Deficiency syndrome, Obrinsky syndrome, Frohlich syndrome or rarely Triad syndrome.[1-4] Prune belly syndrome is a congenital disorder with a group of birth defects that involve three main problems: 1. Poor development of the abdominal muscles, causing the skin of abdomen wrinkle like a prune, 2. Undescended testicles (cryptorchidism) and 3. Urinary tract problems. In addition to this classic triad, a broad spectrum of associated defects includes musculoskeletal, cardiovascular, pulmonary and genital malformations have also been documented.[1,2] The present case was male fetus and had typical triad features of PBS and also associated with hip malrotation with talipes of the feet and pulmonary hypoplasia with no cardiac defects.

The child with PBS is typically male with a thin or lax abdominal wall of variable severity and a long dilated prostatic urethra with prostatic hypoplasia. A large vertically oriented thick walled urinary bladder; an urachal remnant from the dome of the bladder with tortuous and dilated ureters. There is a large amount of hydronephrosis and varying amount of renal hypoplasia. All had cryptorchidism. The thinned abdominal wall has attributed to hydronephrosis distended urinary system which interferes with normal descent of the testes.[5] The index case was male fetus with all above features described by the author. The higher incidence of this syndrome in males has been explained on the basis of more complex morphogenesis of the male urethra and possible obstructive anomalies at several levels. Prune Belly syndrome is rare in females with fewer than 30 cases reported in literature.[6] Diagnosis is made in utero using ultrasound or neonate with characteristic clinical findings. Sonographic observations in PBS associated with oligohydramnios, primarily reflect the pathological changes and are similar to severe urethral obstruction. The fetus shows a disproportionately large ballooned out distended abdomen and grossly dilated urinary bladder with a dilated upper urinary tract. The abdominal wall appearing thin and membranous, the ureters are dilated and tortuous and involving dysplasia of the renal parenchyma. Oligohydramnios when present leads to a spectrum of anomalies secondary to pressure, which include flattened cranium, faces and nose, low set ears and chin and the pulmonary hypoplasia as well the testes are intra-abdominal. Malrotation of gut and anorectal malformations have been reported as associated anomalies. Spina bifida, Talipes, congenital dislocation of hips and other lower limb abnormalities are commonly found and are secondary to associated oligohydramnios.[8] The present index case was a male fetus of 20 weeks old with all three features of PBS i.e. wrinkled abdomen, intra-abdominal testes and dilated urinary bladder with lobulated kidneys and highly dilated, tortuous ureters. The associated anomalies includes malrotated intestines, hypoplastic lungs, scrotum empty with a small penis and lower limbs were both rotated with talipes feet.[7,8] Prune belly syndrome in the adult is scanty. A case of prune belly syndrome was diagnoses in a 24 year old Nigerian who had 3 years history of recurrent right loin pain. On examination showed wrinkled abdominal skin, bilateral undescended testes and a hypoplastic rectus abdominis, below the umbilicus. Further evaluation revealed enlarged bladder, bilateral mega ureters and right intra-abdominal testis. A diagnosis of Prune Belly Syndrome was made.[9]

Pathogenesis is mainly Urinary Tract Obstruction, Primary Mesodermal Developmental Defect and Teratogenic effect.

1. The urinary tract obstruction is mainly due to hypo plastic or dysplastic prostate leading to obstruction of the urethra and finally over distension of the bladder and the upper urinary tract. This further stretches the abdominal wall damaging the abdominal musculature and causes wrinkling of the abdomen and also interferes with the descent of the testicles (Wheatley et al. 1996).[10]

2. Primary Mesodermal Developmental Defect: Any insult between 6 and 10 weeks gestation which disrupts the development of the lateral plate mesoderm from which both anterior abdominal wall and genitourinary tract arises.

3. Teratogenic effect: It may be due to viral infection or any drug abuse during the early weeks of pregnancy.

The present case may be due to primary mesodermal developmental defects. There are a variety of non-urologic problems present in individuals with PBS. Musculoskeletal malformations seen in 20-60% (talipes deformities, congenital hip dislocation, club feet, flared iliac bones and polydactyl). Gastrointestinal anomalies like intestinal malrotation, imperforate anus and anal atresia seen in 30-40% of individuals. Pulmonary anomalies are hypoplasia of the lungs, Cardiac vascular anomalies are seen in 10% of the patients.[11] The present case had associated anomalies like pulmonary

hypoplasia, hip rotation with talipes feet and malrotation of the intestines.

The genetic basis has not been established. It may occur sporadically as an X-linked transmission associated with chromosomal abnormalities. A multifactorial or polygenic inheritance has also been proposed. Additionally, areas of Nigeria have a high prevalence of PBS.[12] Though the cause of prune belly syndrome is unknown**,** familial evidence suggests a genetic component. Recently 2 non familial cases of PBS with chromosome 17q12 deletions encompassing the HNF1β gene have made the candidate gene for prune belly syndrome. The HNF1β mutation was detected in 3% of patients with prune belly syndrome, but found to be functionally normal. The functionally significant HNF1β mutations are uncommon in prune belly syndrome.[13] No genetic background was identified in the family of the present case.

Prune belly syndrome can be diagnosed via ultrasound while a child is still in-utero. An abnormally large abdominal mass is the key indicator, as the abdomen swells with the pressure of accumulated urine. While the fetus grows, fluid develops in its abdomen, which stretches larger and larger. The fluid is reabsorbed before birth, so when born the infant has a sagging or wrinkled abdomen thus known as "prune belly".[14] For the present case an ultrasound was done during pregnancy, the baby had a large cystic swelling occupying the entire abdominal region, later after the delivery it was found to be a dilated urinary bladder with absence of the abdominal wall.

CONCLUSION

An early surgery is recommended to fix weak abdominal muscles, urinary tract problems, and undescended testicles. The baby may be given antibiotics to prevent urinary tract infections .The type of treatment, depends on the severity of the symptoms. A surgical "remodeling" of the abdominal wall and urinary tract is done. Boys may have an orchiopexy, which moves the testicles to their proper place in the scrotum. Even with treatment, many patients experience renal failure. Prune belly syndrome is a serious and often life-threatening problem. There is no known way to prevent this condition.

REFERENCES

1. Baird PA, MacDonald EC. An epidemiologic study of congenital malformations of the anterior abdominal wall in more than half a million consecutive live births. Am J Hum Genet 1981;33:470-8.
2. Eagle JF, Barrett. Congenital deficiency of abdominal musculature with associated genitourinary abnormalities: A syndrome. Report of 9 cases. Pediatrics 1950;6:721-36.
3. Obrinsky. Agenesis of abdominal muscles with associated malformation of the genitourinary tract; a clinical syndrome. Am J Dis Child 1949;77:362-73.
4. Frolich F. Der Mangel der Muskeln, insbesondere der Seitenbauchmuskeln. Dissertation: Wurzburg 1839.
5. Beverly P Wood et al “Pune Belly syndrome” http//www.emedicine.com/radio/topic; 1-10; 2001.
6. Reinberg Y, et al. Prune-Belly syndrome in females: a triad of abdominal musculature deficiency and anomalies of the urinary and genital systems. J Pediatr 1991;118:395-8.
7. Aaronson IA, Cremin BJ (eds). Prune Belly Syndrome: Clinical paediatric uroradiology. London: Churchill Livingstone, 1984:255-65.
8. Ganesan S, Indrajit I K. Images : Prune belly sydrome : Antenatal ultrasound. Indian J Radiol Imaging 2001;11:25-8.
9. Salako AA, Takure AO, Olajide AO, Aarowolo OA, Egberongbe AA. Prune belly syndrome in an adult Nigerian: A case report. Afr J Med Med Sci 2009;38:357-60.
10. Wheatley JM, Stephens FD, Huston. Prune-Belly syndrome: ongoing controversies, pathogenesis and management. Semin Pediatr Surg 1996;5:95-106.
11. Manivel JC, et al. Prune-Belly syndrome: clinicopathologic study of 29 cases. Pediatr Pathol 1989;9:691-711.
12. Nakayama DK, Harrison MR, Chinn DH, et al. The pathogenesis of Prune-Belly. Am J Dis Child 1984;138:834-6.
13. Granberg CF, Harrison SM, Dajusta D, Zhang S, Hajarnis S, Igarashi P, Baker LA. Genetic basis of prune belly syndrome: screening for HNF1β gene. J Urol 2012;187:272-8.
14. Williams DI, Burkholder GV. The Prune Belly Syndrome. J Urol 1967;98:244-51.

DOI: 10.5455/2320-6012.ijrms20130523
Cite this article as: Vani A, Saritha S, Sangeetha K, Supriya G. Prune belly syndrome (sequenze): a case report. Int J Res Med Sci 2013;1:148-52.