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Clinical Image

The Prune Belly Syndrome with Overlapping Presentation of Partial Urorectal Septum Malformation Sequence in a Female Fetus

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Introduction

Prune Belly Syndrome (PBS) is a congenital syndrome which is also known as Eagle-Barrett syndrome, Obrinsky syndrome, Fröhlich syndrome or abdominal muscular deficiency syndrome. It is a rare congenital anomaly affecting 1:26.000 to 1:40.000 births. Approximately 97% of the affected cases occur in male gender. In males, it is characterized by a triad of abdominal muscle deficiency, bilateral cryptorchidism and urinary tract abnormalities. Nevertheless, a similar condition occurs in females, also called "pseudoprunes", but obviously cryptorchidism does not exist [1]. Actually, PBS is a multisystem disease with often coexists with a spectrum of extra–renal malformations, including cardiac, gastrointestinal, pulmonary and skeletal anomalies

The urorectal septum malformation sequence (URSMS) was first described as a separate entity by Escobar in 1987. It is a rare disorder with different degrees of complexity and results from a defect in the caudal mesoderm. Inadequacy to divide cloaca by urorectal septum into anterior primitive urogenital sinus and posterior rectum and persistence of cloacal membrane lead to URSMS. Also called female pseudohermaphroditism with caudal dysgenesis and cloacal dysgenesis sequence, this entity occurs in one in 50000 to 250000 neonates. The partial form is compatible with life, but complete form of this sequence is usually lethal [2].

URSMS is usually characterized by ambiguous genitalia, absence of urethral and vaginal openings, imperforate anus and urorec-

tal-vaginal fistulae. Mullerian duct defects, vertebral and renal defects are also described [2,3]. We report an extremely rare presentation of Prune Belly Syndrome with an unusual association with URSMS.

Case Presentation

A 43-year-old healthy pregnant woman, gravida 2, para 1, presented a fetal abdominal cyst with 22.79 mm in diameter at 12^{+1} weeks ultrasound (Figure 1). There was no history of consanguinity in the family or of an exposure to any known teratogen.

The chorionic villous sampling revealed a normal fetus (46, XX). At 15^{+4} weeks, the fetal abdominal cyst was increasing in size, reaching 57 mm in its longest diameter and occupying the whole abdominal cavity. Amniotic volume was normal and no other apparent anomalies were observed. We judged that the cyst was a bladder-enlarged due to a severe urethral stenosis or obstruction. Parents were counseled about the poor prognosis and termination of pregnancy was performed, after informed consent was obtained.

On autopsy, the abdominal wall was absent at the level of rectal muscles, with exteriorization of the liver, bowel, spleen and bladder. It also showed an ambiguous genitalia related to protruded fold skin with a median imperforated cleft, giving an impression of labial folds. Vaginal and urethral atresia and rudimentary uterus were documented. Ovaries and fallopian tubes were not well documented. Imperforate anus and short bowel along with recto-vesical fistula with dilated bladder were observed. Two short ureters and hydronephrosis were described. Other systemic examination revealed micrognathia, bilateral rocker-button foot and pulmonar hypoplasia, secondary to oligohydramnios and vertebral dysplasia (Figure 2 A-H). Tissues were subjected to histopathological examination, which confirmed the identity of the organs and documented ovary dysgenesis (Figure 3 A-C).

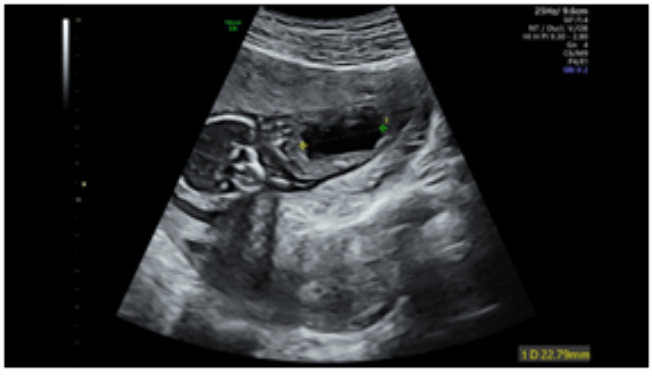

Figure 1: Fetal abdominal cyst (22,79 mm).

Placenta was normal and a single umbilical artery was observed. We concluded by a female fetus with overlapping features between PBS and URSMS partial type.

Discussion and Conclusion

Because of its rarity and complex multisystem variation, URSMS is difficult to diagnose prenatally. Obstetricians should be attentive when prominently dilated lower abdominal cyst is detected by ultrasonography. Etiology of PBS is uncertain, although some authors suggested early urethral obstruction as a main factor to the development of PBS leading to bilateral enlarged cystic kidneys and megacystis [3]. A possible role of genetic mutations in the development of URSMS is also suggested in the literature [4].

Moreover, the present case reports a gonadal female phenotype fetus with two out of the three features of the PBS (abdominal wall muscle deficiency and urinary tract abnormalities). It also had ambiguous genitalia with female internal genital organs and the third feature of the PBS i.e., cryptorchidism, was absent.The female internal genital organs consisted of vaginal and a rudimentary uterus. So, the present case was diagnosed as a female counterpart of the PBS, which accounts for 3-5% of all the recorded cases. Also, the urogenital tract and the lower intestinal tract, which were associated with a lack of the perineal opening and the presence of ambiguous genitalia were present in our case, like in the URSM sequence.

In fact, our autopsy findings carried a female fetus with overlapping phenotypes between PBS and a partial URSMS.

The occurrence of the PBS in females is rare.PBS was identified in 5 female newborns and 13 male newborns in over half a million consecutive life births in British Colombia from 1964 to 1978 [5-7]. Moreover, the overlapping with URSMS makes it an extremely rare entity. Goswami et al. were the first to report a case of PBS with URMS [8].

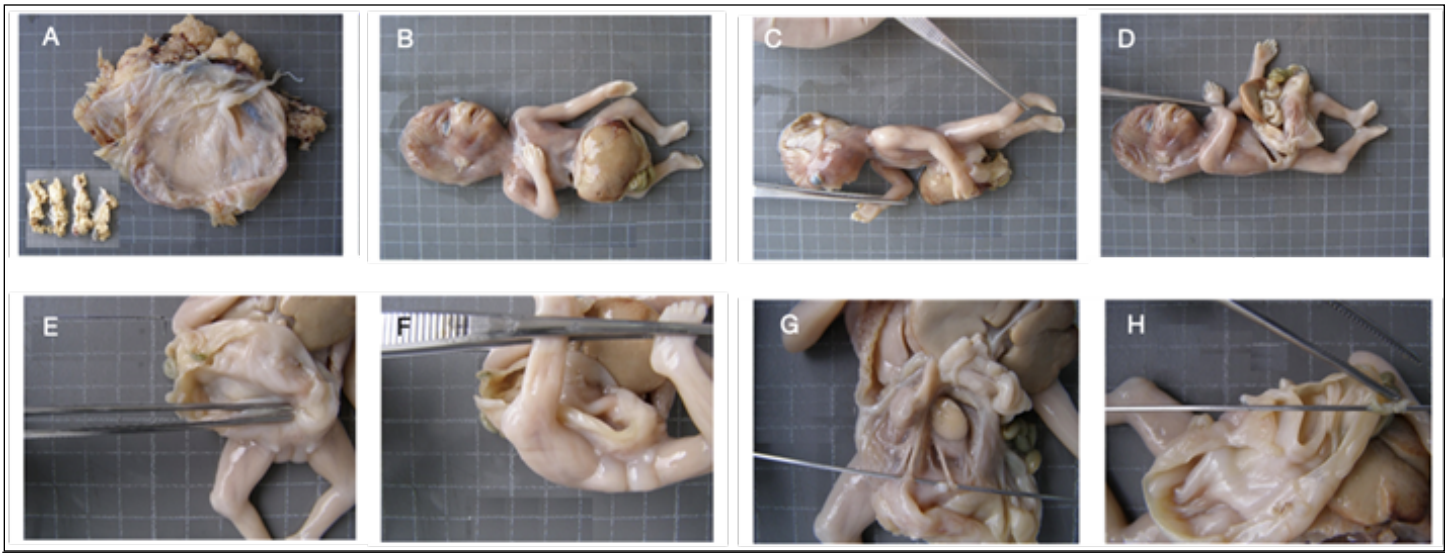

Figure 2: A: Short umblical cord; B,C,D,E,F: Abdominal wall absent at the level of rectal muscles with exteriorization of liver and bowel. It continues to bladder; F,G: Ambiguous genitalia with skin folds protruded on both sides of the mucus membrane gave an impression of labial folds with central imperforated cleft; G: Colo-Vesical fistula; H: Dilatated and multiseptated bladder.

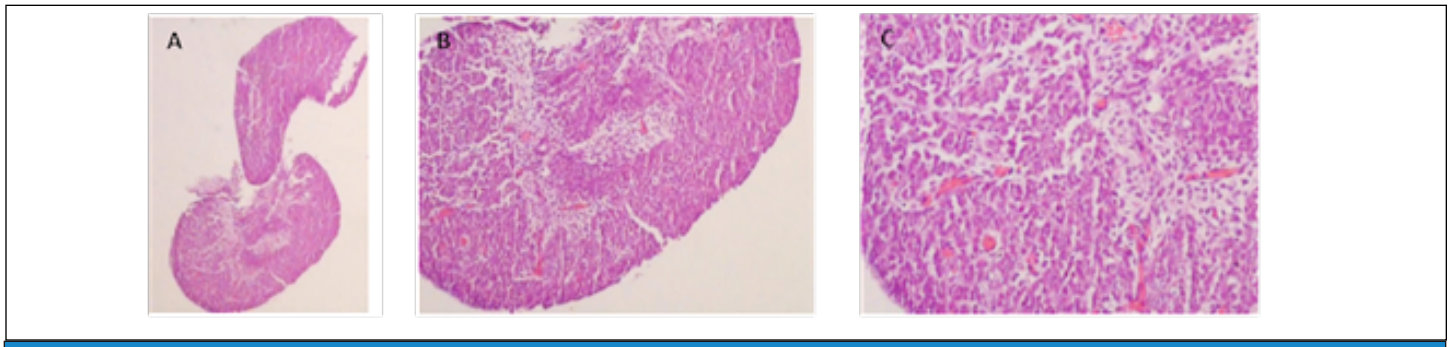

Figure 3: A,B,C Gonadal dysgenic ovaries. H&Ex40(A)x100(B)200(C).

To the best of our knowledge, this is the fourth case of an overlap between PBS and URSM in literature [7].

A chance for an accurate autopsy evaluation should never be missed.This case has been reported with the intention to contribute to the present knowledge as regards to PBS, along with URSMS and its poor prognosis.

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