

Picture of the Month

Prenatal diagnosis of fetal genital prolapse

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Neonatal genital prolapse, an uncommon condition, usually presents as a tumor-like mass protruding from the ventral part of the vulva. All cases that had been previously reported were diagnosed after birth. We present the first case report of fetal genital prolapse that presented prenatally by ultrasonography as a solid protruding lesion on the ventral part of the vulva. Postnatal magnetic resonance imaging (MRI) confirmed the diagnosis of an isolated neonatal vaginal prolapse. At follow-up examination at 6 months of age, the infant's external genitalia appeared normal, with no evidence of genital prolapse.

A 33-year-old woman, gravida 2 para 1, attended our prenatal care clinic at 12 weeks' gestation to request first-trimester ultrasound screening to evaluate the risk of fetal Down syndrome. Ultrasound examination revealed an increased nuchal translucency thickness of 3 mm. After genetic counseling, chorionic villus sampling was performed uneventfully; testing revealed a normal female karyotype (46,XX). The patient presented at 20 weeks' gestation to complain that an error had been made in the ultrasonographic fetal gender deduced at a local obstetric clinic. Consequently, we performed a detailed fetal sonographic examination, which found a phallus-like structure protruding over the ventral part of the fetal vulva (Figure 1). The covering of the protruding mass was continuous with the lining of the vaginal wall (Figure 2). Fetal growth was appropriate for gestational age, but the amniotic fluid index was 29.2 cm, indicating polyhydramnios. Fetal ambiguous genitalia or genital prolapse were suspected.

Sex-determining region Y gene analysis was negative after a follow-up amniocentesis was done to provide rapid fetal sex determination (ruling out an XX male or maternal cell contamination) and ensure that no error had occurred in the original fetal karyotyping. Androgen levels were measured in maternal blood and found to be normal. Both parents were negative for the carrier test for the CYP21 gene. A diagnosis of fetal genital prolapse was finally made. The parents were made aware of the implications of the diagnosis

and made a fully informed choice to continue the pregnancy.

At 33 weeks of gestation, the patient was admitted with preterm rupture of membranes and emergency Cesarean section ensued owing to breech presentation. At birth, the baby weighed 2080 g and had a mass protruding between the infant labia minora. Close inspection revealed rugae consistent with vaginal epithelium (Figure 3). A normal urethral orifice was confirmed and spontaneous voiding was observed along with a normal outer anal appearance with passage of meconium. The prolapsed lesion was reducible with a Q-tip. Two weeks later, transabdominal ultrasonography depicted a normal infant uterus measuring $2.2 \times 1.0 \times 1.0$ cm, and showed the presence of two normally positioned and sized kidneys with no evidence of hydronephrosis or hydroureter. MRI confirmed the diagnosis of an isolated neonatal vaginal prolapse (Figure 4). The mother was instructed on the management of the prolapsed vaginal vault and the application of topical estrogen cream. The infant was discharged in good health 3 weeks after delivery. At follow-up examination at 6 months of age, the infant's external genitalia appeared normal with no evidence of genital prolapse.

Genital prolapse occurs when pelvic organs slip from their normal anatomical positions and either protrude into the vaginal wall or press against it. Various degrees of neonatal genital prolapse have been reported, ranging from total genital prolapse to a lesser degree of prolapse that may include the uterine cervix, corpus, and vaginal vault, to isolated vaginal prolapse^{1,2}.

The cause is uncertain. It has been suggested that genital prolapse is not just a local dysmorphic phenomenon but may be a local manifestation of systemic congenital anomalies that represent a defect in the musculature of the pelvic floor^{3,4}. Most cases are associated with other congenital anomalies, primarily neural tube defects^{1–3}. These spinal neuropathies are amenable to prenatal diagnosis for fetal genital prolapse using either ultrasonography or MRI. In addition, elevated intra-abdominal pressure, intrapartum trauma,

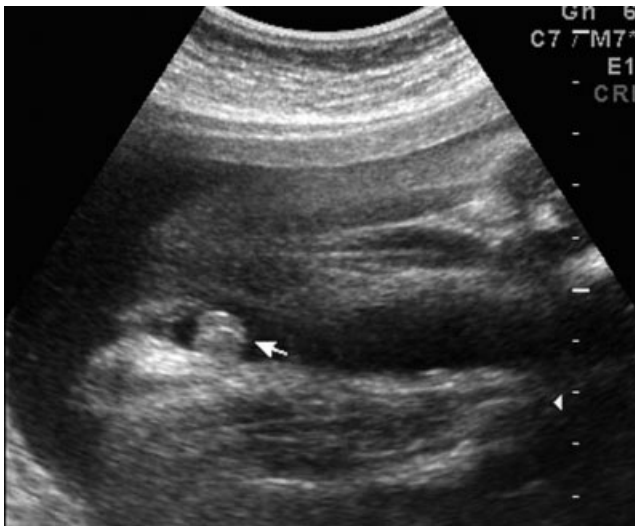


Figure 1 Prenatal sonographic image of female fetal genital prolapse. Axial section of the vulva shows a phallus-like structure (arrow) protruding over the ventral part.

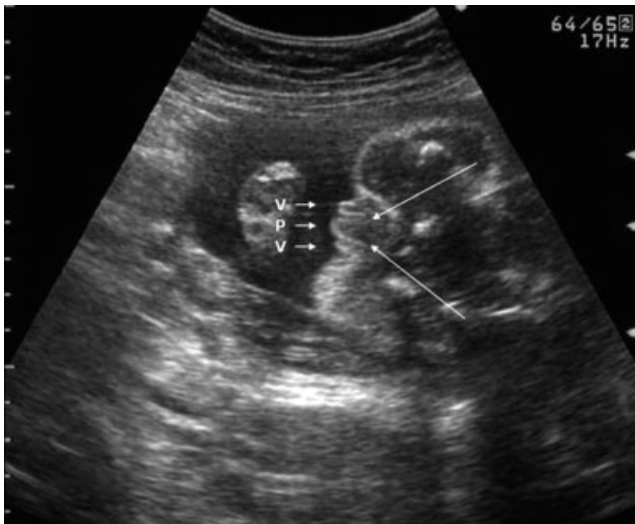


Figure 2 Coronal section of ultrasound image showing a mass bulging out of the vaginal canal. The covering of the protruding mass (P) is continuous with the lining of the vaginal wall. The long arrows mark the reflexion of the vaginal wall. V, vulva.

and fetal maldevelopment have been implicated as possible predisposing factors to neonatal genital prolapse in the absence of a congenital defect^{5,6}. These etiological factors, which lead to congenital muscular weakness and hypoplasia of the entire pelvic suspensory and supportive tissue, might explain the connection of neonatal genital prolapse with pregnancies of breech presentation, prolonged labor, preterm delivery, or intrauterine growth restriction⁵⁻⁷.

All previously reported cases have involved neonatal genital prolapse that was diagnosed shortly after birth⁷⁻⁹. To our knowledge, no reports prior to this case report have involved initial suspicion of diagnosis based on prenatal sonographic examination. Sonographically, this female fetus had grossly normal labia with a

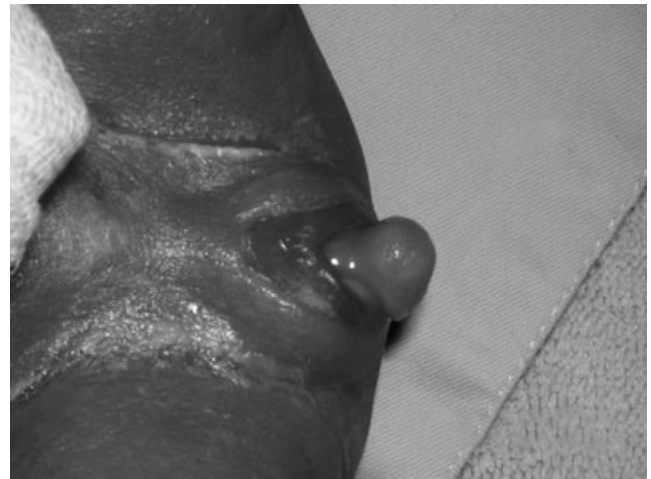


Figure 3 External genitalia at birth demonstrate vaginal prolapse.

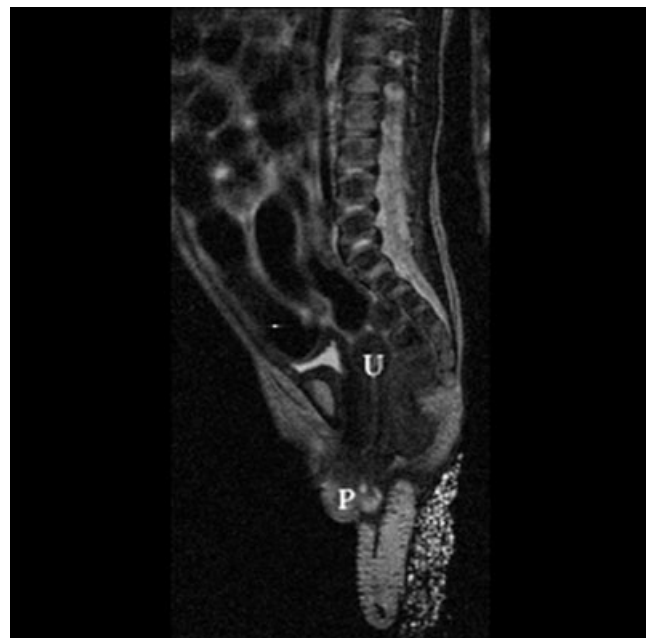


Figure 4 Sagittal section on postnatal magnetic resonance imaging reveals a neonatal vaginal prolapse (P). The covering of the protruding mass is continuous with the vaginal mucosal lining. The size and location of the neonatal uterus (U) are normal.

protruding solid lesion on the ventral part of the vulva. Postnatal examination confirmed the diagnosis of fetal vaginal vault prolapse and ruled out other fetal congenital anomalies. In addition, polyhydramnios, causing increased intra-abdominal pressure for the fetus and fetal malpresentation, was evident in our case^{10,11}.

Interlabial masses in the female fetus and newborn may be caused by urethral prolapse, ureterocele, vaginal introital cyst, imperforate hymen, urogenital sarcoma, and genital prolapse¹²⁻¹⁴. Abnormalities of the female genitalia on prenatal ultrasonography may be an isolated finding or part of a more complex syndrome. An accurate prenatal diagnosis based on

targeted ultrasound examination following a conspicuous finding may allow timely postnatal assessment. Regarding postnatal determination of the origin of an interlabial mass, an easy technique is to examine the area under appropriate lighting and evaluate the mass with Q-tips while the labia majora are separated⁹. If the mass is manually reducible by Q-tips, as in our case, prolapse is of vaginal rather than urethral origin. Treatment modalities of neonatal genital prolapse vary and should be tailored to the degree of prolapse encountered. Successful correction has been achieved with conservative care, digital reduction with a Q-tip and application of a small pessary, as well as primary surgical correction. The non-complicating vaginal vault prolapse in this case was overcome spontaneously in the neonatal period through conservative treatment with application of topical estrogen cream.

In conclusion, the current case is the first in which prenatal diagnosis of fetal genital prolapse was made (at 20 weeks' gestation). Furthermore, this child's case was unusual in that it was associated with prenatal polyhydramnios as a possible predisposing factor. Families can benefit from thorough prenatal work-up of such a pathological finding so that appropriate postnatal medical care can be planned.

Acknowledgments

This work was supported by research grants NSC 93-2314-B-182A-099 from the National Science Council, Taiwan, ROC.

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