



Prenatal diagnosis of penoscrotal hypospadias and review of the literature

Penoskrotal hipospadiasın prenatal tanısı ve literatürün incelenmesi

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ABSTRACT

Urinary tract abnormalities are frequently detected during obstetrical ultrasonography (US). However, hypospadias is often missed on prenatal US, despite it being the most common congenital defect of the male external genitalia. The prenatal recognition of hypospadias is important because it will alert the physician to order karyotyping and to look for any possible associated dysmorphic syndromes. Here, we present a case of a penoscrotal hypospadias mimicking female genitalia and describe our experience of applying two-dimensional and three-dimensional US in the prenatal diagnosis of hypospadias. Prenatal ultrasound scans should include a study of the genitals and should not only be used for sex determination.

Key words: Obstetrics ultrasonography; penoscrotal hypospadias; prenatal diagnosis.

ÖZET

Gebelikte fetal üriner trakt anomalileri obstetrik ultrason taramasında kolayca saptanabilen gelişim bozukluklarıdır. Ancak prenatal hipospadias tanısı, erkek genital sisteminin en sık görülen konjenital anomalisi olmasına rağmen intrauterin dönemde sıklıkla konulamaz. Hipospadiasın prenatal dönemde tanınması, karyotipleme ve birlikte görülebilen dismorfik sendromların araştırılması açısından önemlidir. Bu çalışmada intrauterin dönemde dişi genital yapıyı taklit eden penoskrotal hipospadias olgusunun iki ve üç boyutlu ultrason ile antenatal tanısı sunulmaktadır. Gebelikte ultrasonografik tarama, fetal cinsiyet tayini dışında genital organların anomalileri açısından da ayrıntılı değerlendirmeyi kapsamalıdır.

Anahtar sözcükler: Obstetrik ultrason; penoskrotal hipospadias, prenatal tanı.

Introduction

Hypospadias is a birth defect found in boys. In hypospadias, the opening of the urinary tract is located on the ventral side of the penis, instead of the tip. It is the most common congenital defect of the male external genitalia, with a prevalence of approximately 0.2 to 4.1 per 1.000 live births.^[1-4] Despite hypospadias being the most common urogenital anomaly of male neonates, the diagnosis is often missed prior to birth.

Hypospadias are classified into 3 groups according to the location of the abnormal meatus along the expected course of the urethra: anterior (glandular or coronal), middle (penile shaft), and posterior (penoscrotal, scrotal, perineal). These groups account for 50%, 30%, and 20% of all hypospadias cases, respectively.^[5]

The postnatal treatment of hypospadias is always surgical and is usually carried out at the age of 4-18 months, depending on the type of hypospadias. The most frequent complications of untreated hypospadias are stenosis of the meatus, difficulties in sexual relationships due to an abnormal erection, and infertility due to ejaculation outside the vagina.^[6]

Although hypospadias may be suspected during the early second trimester or even the first half of pregnancy, a diagnosis at this time may be erroneous because the fusion of the labio-scrotal folds in a male fetus may be delayed. Because there is improved visualization of the external genitalia at the third trimester, the prenatal diagnosis of hypospadias is not usually made until then.^[2-7] Here, we report a successful case of the prenatal diagnosis of a penoscrotal hypospadias in the early second trimester of pregnancy.

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Case report

A 28-year-old woman (gravida 1, para 0) with no significant past medical history was referred to our tertiary center for US examination at 22 weeks of gestation. There was no history of congenital abnormalities or exposure to medication during this pregnancy. Based on the sagittal sign at 14 weeks of gestation, the fetus had been assigned as a female.

On the initial 2D sonogram, fetal growth was found to be appropriate for the gestational age, and no gross anomaly was found, except for an abnormally curved and shortened penis and a single umbilical artery (Figure 1). Due to the ambiguity of the genitalia and the presence of a single umbilical artery, a sample of the fetal chromosomes was obtained by chordocentesis and analyzed. This revealed a normal, male 46, XY karyotype. A repeat ultrasound scan at 23 weeks of gestation showed a shortened penis that was abnormally curved between the two scrotal folds in the coronal plane. An informed consent was taken from the patient. The possibility of hypospadias was raised, and to confirm the diagnosis of hypospadias, three-dimensional (3D) imaging in surface-rendered mode was used. Multiplanar and surface-rendered images were obtained in the midsagittal, axial, and coronal planes to precisely delineate the ventral curvature of the penis (Figure 2). At 30 weeks of gestation, both testes were visualized in the scrotum. At 38 weeks of gestation, a term newborn was delivered by cesarean section with a birth weight of 2950 g and Apgar scores of 9 and 10 at 1 and 5 minutes, respectively. The physical examination of the newborn revealed the presence of severe hypospadias, with the meatus in the penoscrotal position.



Figure 1. A two-dimensional ultrasound image showing the coronal view of the external genitalia at 23 weeks of gestation

Discussion

Hypospadias results from an incomplete fusion of the urethral folds that occurs between the seventh and fourteenth week of gestation. The exact etiology of the disorder remains largely unknown. The origin of hypospadias seems to be multifactorial, and previous research has identified several risk factors, such as genetic predisposition, placental insufficiency, and substances that interfere with natural hormones.^[8]

Although hypospadias usually occur as an isolated abnormality, up to 40% of the affected fetuses with hypospadias also have upper urinary tract anomalies. Cryptorchidism and inguinal hernias are the most common extragenital anomalies and are found in 7 to 10% of all hypospadias cases. Furthermore, triploidies, several syndromes (such as Fraser syndrome, and Smith-Lemi-Opitz syndrome), congenital heart defects, cleft lip and palate, neural tube defects, and anorectal malformations have also been described in combination with hypospadias. In addition, gene expression studies have identified CTGF, CYR61 and EGF as candidate genes that may contribute to the condition. The cause of hypospadias is most likely a combination of monogenic and multifactorial forms, involving both genetic and environmental factors.^[1-7]

In addition to the determination of fetal sex, the detection of external genitalia anomalies has become an integral component of second trimester anomaly scans. The conventional sonographic criteria for the diagnosis of hypospadias is a small, “blunt-ended” penis with two echogenic lines at the tip, which represent the prepuce lateral folds.^[8,9] Meizner reported the “tulip sign” for severe hypospadias, an US clue for the in utero diagnosis of severe



Figure 2. A surface-rendered three-dimensional image showing fetal hypospadias

hypospadias in which the tulip is formed by a ventrally orientated penis located between the scrotal folds.^[10] However, in certain cases, it is almost impossible to sonographically distinguish between the normal anatomy of a female and a male with severe penoscrotal hypospadias. In our case, there was a penoscrotal hypospadias, and the sonographic appearance of the genitalia was similar to the tulip sign reported by Meizner et al.^[10]

Although sonographic fetal sex determination is feasible in most pregnancies, in some cases, it may pose difficulties. Sonographic fetal sex determination in the late second trimester is based on the direct visualization of the external genitalia, whereas in the first and early second trimester, it is based mainly on the direction of the genital tubercle.^[11] Inaccurate fetal sex determination may occur when the external genitalia are malformed.

In this report, we have presented a case of the prenatal diagnosis of penoscrotal hypospadias using 2D and 3D US. Although 2D US is still the gold standard for diagnosing fetal genital abnormalities, accurate prenatal diagnosis is not possible in many of these cases. 3D US can improve the diagnosis by providing more detailed and realistic images.

Conflict of Interest / Çıkar Çatışması

No conflict of interest was declared by the authors.

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