10.5005/jp-journals-10009-1217

CASE REPORT

3D, Vocal and Tomographic Ultrasound Image in Prenatal Diagnosis of Hypospadias

Francisco Raga, Francisco Bonilla Jr, Juan Carlos Castillo, Fernando Bonilla-Musoles

Department of Obstetrics and Gynecology, Valencia School of Medicine, Hospital Clinico Universitario, Valencia, Spain

Correspondence: Fernando Bonilla-Musoles, Professor, Department of Obstetrics and Gynecology, Valencia School of Medicine Hospital Clinico Universitario, Blasco Ibañez 17, 46010 Valencia, Spain, e-mail: profesorbonillamusoles@hotmail.com

ABSTRACT

We report a case of anterior hypospadias, diagnosed at 26th week in a 37 years-old primigravida with normal 46XY kariotype through amniocentesis carried out at 16th week.

Sonographic examination with 2D showed a short and curved penis. The use of three orthogonal planes, Tomographic Ultrasound Image (TUI) and VOCAL allowed an exact prenatal diagnostic, showing the "tulip" sign and defining localization, situation and extension of the urethral orifice.

Keywords: Hypospadias, 3D ultrasound (3D US), Vocal and TUI modes.

INTRODUCTION

Apart from cardiac malformation, hypospadias is the most frequent congenital defect; this malformation is mainly sporadic and, except when it is part of a polymalformative syndrome (7-9%), its impact on the newborn is quite low (80%).

The anomaly occurs only in male fetuses as a result of failure of complete development of the anterior urethra, its diagnosis must alert the clinician to search if other defects are present.

The opening of the urinary tract is not localized on the tip of the penis but on its ventral region on glans penis. Hypospadias are classified according to the position of the meatus:

- Anterior (glandular, coronal, penile anterior), 50%
- Middle (middle penis axis), 30%
- Posterior (penile posterior, penoscrotal, scrotal, perineal), 20%. The most worrisome variant of hypospadias has received the beautiful name, worldwide known as the ecographic 'tulip sign'.[1]

The exact etiology remains unclear. It has been proposed that the defect of the urethral folds fusion might be a consequence of a deficit in testosterone receptors or to a partial insensibility of local conversion of testosterone to dehydrotestosterone.[2]

Recently, 3D US has been used for helping in improving the correct and most exact diagnosis.[1,3] Based on a recently observed case, we describe the new possibilities of 3D modes.

CASE REPORT

This is a 37 years old patient, who had an amniocentesis on week 16 showing a normal karyotype. All ultrasound parameters remained normal until week 22, when a 2D scan showed a short and shaft penis, also a tulip sign was evident, a suspect diagnosis of hypospadias was made. No other malformations were evident on scan. 3D ultrasound confirmed the diagnosis (Fig. 1).

Fig. 1: 3D orthogonal planes and 3D-rendering of the tulip sign (yellow arrow)

Since no other structural malformation or anomaly in growth, placenta or amniotic fluid were detected, patient was scheduled for a new scan at week 26. At this time, ultrasonography was then performed with a E-8 (GE Medical Systems, Kretztechnik, Zipf Austria). Multiplanar, surface-rendered images, Tomographic Ultrasound Image (TUI) and VOCAL of the gender were obtained in the midsagittal, axial and coronal planes to precisely delineate the ventral curvature and to observe the extension of the penile orifice (Fig. 2).

In order to carefully evaluate the exact localization and extension of the opening defect, TUI and VOCAL 3D modes were used focusing on the unique curvature of the sonographic picture of the tulip sign and the meatus.

As previously described,[3,4] TUI mode was specially useful, its tomographic cuts and the manipulation of the ecographic angles were perfect to clearly show the curve and size of the defect (Fig. 3).

Fig. 2: The tulip sign in 3D-rendering and the macroscopic finding

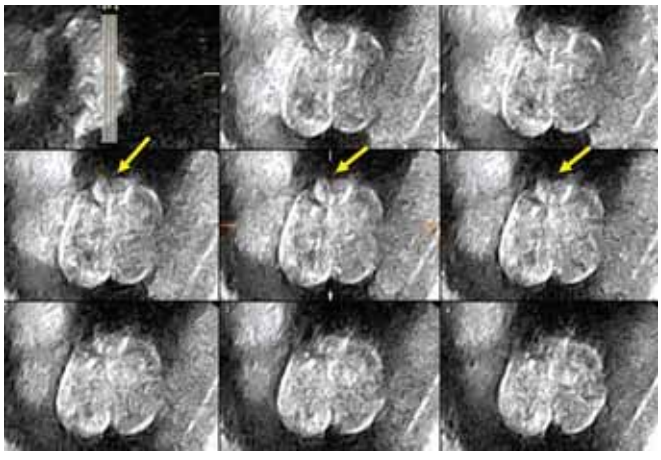

Fig. 3: TUI to see inferior and anterior zones of the penis (up and left). The distal bulbous tip of the penile shaft is showed clearly. In center images, the urethral opening and its extension (arrows). Notice the tulip sign (upper images)

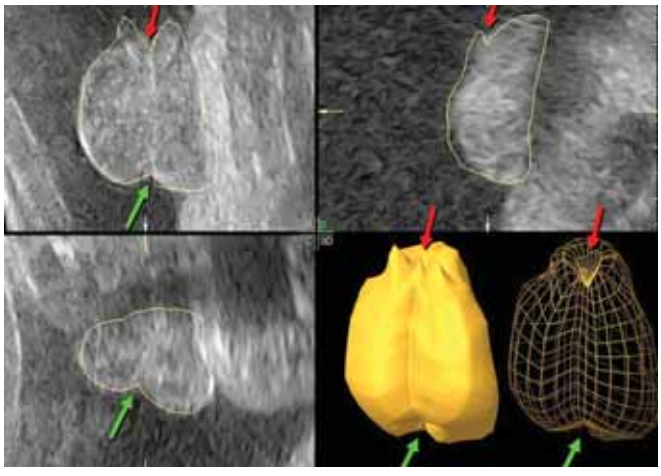

Fig. 4: VOCAL mode showing a small urethral opening (yellow arrows) and a very marked middle raphe (green arrows)

VOCAL mode was useful to study the localization and size of the opening defect, the scrotal middle part and the lateral folds (Fig. 4).

A 40th week normal male baby, weighing 3.400 gm was vaginally delivered.

DISCUSSION

Sonographic diagnostic of fetal sex is part of the so-called morphological ultrasound scan at 20th week. However, antenatal diagnosis of hypospadias is difficult (or unnoticed) before third trimester when genitals can be better observed.[5] In fact, 3D US has allowed an accurate differential diagnosis in complex cases in female fetuses.[3] Moreover, sometimes fusion of vulvar/scrotal folds can be delayed, leading to confusion or misdiagnosis.[4]

Sonographic markers for prenatal diagnosis include:[1]

- Blunter and bulbous tip of the distal part of the penile shaft rather than the normal pointed morphology
- Penile duct anomaly short or small
- Shaft morphology of the distal part of the penis with severe ventral curvature which results from a fibrous band, the chordee, occurring in the projected course of the urethra
- Observation of small lateral folds, epithelial remnants of the foreskin[3] and the presence of an abnormal urine stream[1,6]
- The tulip sign[1]
- Using TUI mode focusing on the ventral zone of the penis, the urethral opening and its extension can be seen.

2D US scan is the first-line method for diagnostic suspicion.[7] However, recent publications about 3D show that a more accurate ecographic diagnosis can be made, using surface, multiplanar, VOCAL and TUI modes.[8] Our experience indicates that 3D should be included for the diagnosis of this genital anomaly; this conclusion is also supported by others.[4,9]

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