

# The confirmation of the diagnosis of ambiguous genitalia with 4D ultrasonography

Aykan Yucel, Ayse Kirbas, Kudret Erkenekli, Ozgur Kara, Hulya Dede,  
Dilek Uygur  
Zekai Tahir Burak Training And Research Hospital  
Ankara, Turkey

## INTRODUCTION:

Ambiguous genitalia are external fetal genitals that cannot be differentiated as phenotypically male or female. Ambiguous genitalia are observed in 1/15.000 live births. Aberrant differentiation results from either genetic or hormonal influence. Ultrasound examination is efficient in the screening for ambiguous genitalia.

## CASE REPORT:

A 18 year-old primigravid woman referred to our Perinatology Clinics at 34 completed weeks with diagnosis of intrauterine growth retardation. The ultrasonographic examination revealed 4 weeks growth retardation with normal amniotic volume and Doppler findings. The 2 dimensional ultrasonography revealed no penis and the labia appeared to be swollen (Figure 1.). The 4 dimensional ultrasonography justified that there was no penis and the labia were swollen (Figure 2.). Cesarean section was performed two weeks later due to abnormal Doppler findings and oligohydramnios. An infant with 1180 g weight and with Apgar scores of 7 and 9, at 1 and 5 minutes, was born. In compliance with the sonographic findings, the external genitalia were ambiguous (Figure 3.).

## DISCUSSION:

Four dimensional ultrasonography can provide superior impression of genital ambiguity and depth perception in comparison to 2 dimensional ultrasonography, which may be helpful in differentiating a micropenis or enlarged clitoris from labia minora.

## References:

1. Pang, S, Clark, A. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: Newborn screening and its relationship to the diagnosis and treatment of the disorder. *Screening* 1993; 2:105.
2. Therrell BL. Newborn screening for congenital adrenal hyperplasia. *Endocrinol Metab Clin North Am* 2001; 30:15.



Figure 1. 2D ultrasonography revealed no penis and the labia appeared to be swollen



Figure 2. Swollen labia in 4D ultrasonography



Figure 3. Ambiguous external genitalia after birth