

## Female pseudohermaphroditism

Published on 09.02.2011

**DOI:** 10.1594/EURORAD/CASE.8632

**ISSN:** 1563-4086

**Section:** Paediatric radiology

**Imaging Technique:** Ultrasound

Case Type: Clinical Cases

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**Patient:** 1 weeks, female

### Clinical History:

A one-week female newborn presented with genitalia hyperpigmentation, clitoromegaly and partial fusion of the posterior labioscrotal folds. Only two perineal orifices were detected, one of which was the anus and the other located posterior to the clitoris. A normal 46XX karyotype was previously obtained during the gestational period.

### Imaging Findings:

Ultrasonography revealed uterus and ovaries of normal size and echogenicity (Fig. 1a and 1b). The kidneys and the adrenal glands were also normal at ultrasonography (Fig. 1c).

Genitography depicted a persistent urogenital sinus and a male type urethra (Fig. 2a). The presence of the vagina and of the cervical imprint was established (Fig. 2b).

### Discussion:

The term ambiguous genitalia is used when the external genitalia do not have the typical anatomic appearance of normal male or female genitalia and correspond to the phenotypic appearance of some, but not all, types of disorders of sex development (DSD). DSDs have been defined as “congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical” and encloses five different entities.

Female pseudohermaphrodites (46,XX) have a female genotype and two ovaries for gonads, but their external genitalia show a variable degree of virilisation (as in our case); it correspond to 60%–70% of all cases of ambiguous genitalia during the neonatal period. The causes of female pseudohermaphroditism include congenital adrenal hyperplasia (CAH) and transplacental androgen exposure, being CAH the most common.

The term CAH encompasses a group of autosomal recessive disorders, each of which involves a deficiency of an enzyme involved in the synthesis of cortisol, aldosterone, or both. The most common form of CAH is due to mutations or deletions of CYP21A, the gene that codes for 21-hydroxylase, resulting in deficiency of this enzyme (>90%). CAH caused by 21-hydroxylase deficiency is found in all populations and has an overall prevalence of 1 case per 16,000 population. The clinical manifestation of each form of CAH depends on the nature and severity of the enzyme deficiency.

Females with severe forms of CAH due to deficiency of 21-hydroxylase have ambiguous genitalia at birth due to excess adrenal androgen production in utero, that range from complete fusion of the labioscrotal folds and a phallic urethra to clitoromegaly and partial fusion of the labioscrotal folds. Approximately 50% of patients with CAH due to CYP21A mutations or deletions have salt wasting due to inadequate aldosterone synthesis, which yields to

hyponatraemia and hyperkalaemia. The elevated serum levels of ACTH, due to cortisol deficiency, cause genitalia hyperpigmentation.

The diagnosis of CAH due to 21-hydroxylase deficiency is established with an elevated 17-hydroxy-progesterone serum concentration.

Imaging plays an important role in depicting the internal organs and urogenital anatomy.

Ultrasonography is the initial examination and is useful to confirm the presence and normality of the uterus and gonads. Morphological and dimensional evaluation of the adrenal glands should also be assessed, although the presence of normal-sized glands does not exclude the diagnosis.

Genitography is essential in the surgical work-up of children with female pseudohermaphroditism and remains the main investigation to display the urogenital anatomy, demonstrating the type of urethra and the presence of vagina, cervix, urogenital sinus and any fistulous communication with the vagina or rectum.

Urogenital sinus is the embryologic precursor of the bladder, urethra and distal third of the vagina in females. This structure may persist in patients with CAH, due to excessive androgens exposure in utero, and leads to the urethra and vagina opening through a single aperture. The urogenital confluence is measured in relation to the perineum and characterised in relation to the external sphincter. These anatomic characteristics assume a major impact for mapping surgical strategy.

**Differential Diagnosis List:** Female pseudohermaphrodites due to congenital adrenal hyperplasia.

**Final Diagnosis:** Female pseudohermaphrodites due to congenital adrenal hyperplasia.

## References:

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**Figure 1**

**a**



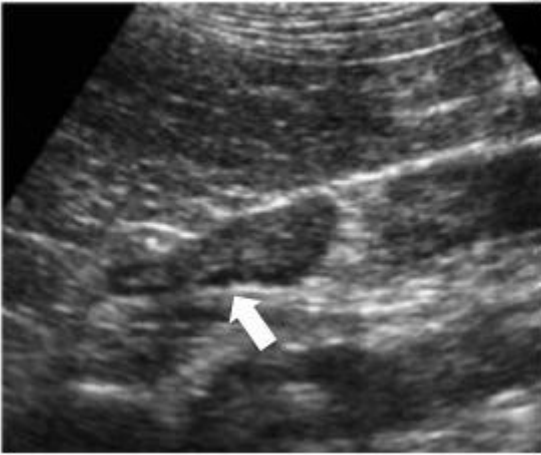
**Description:** Longitudinal and transverse US images show uterus (arrow) with normal size and morphology. **Origin:**

**b**



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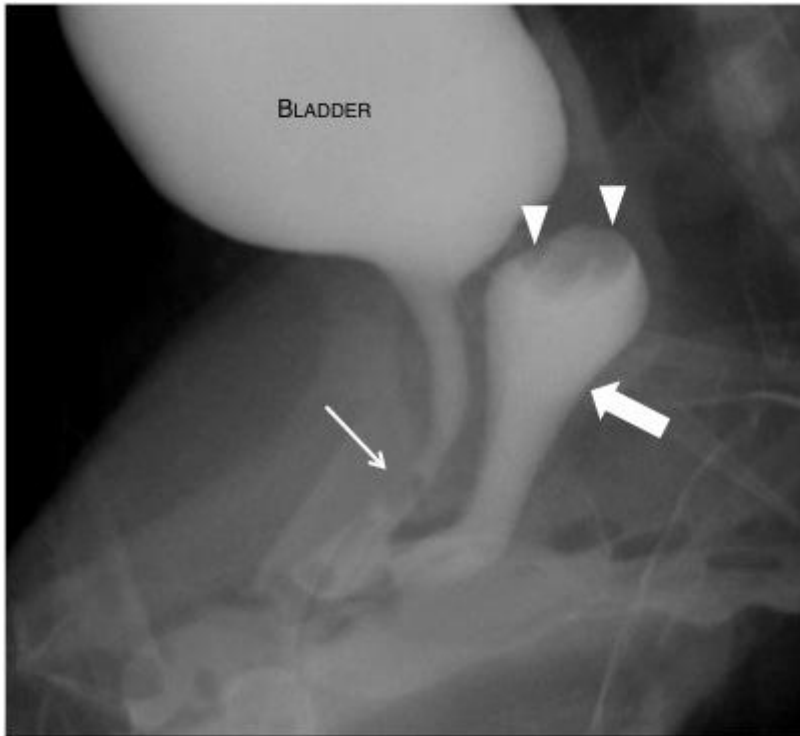
**c**



**Description:** Longitudinal image of the normal right adrenal gland (arrow). **Origin:**

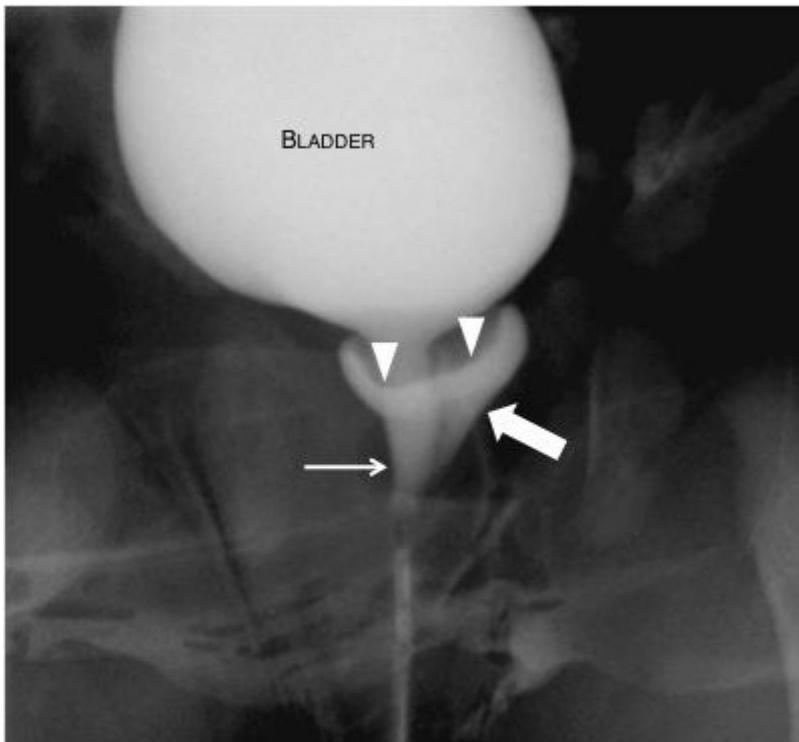
**Figure 2**

a



**Description:** Anteroposterior (2a) and lateral (2b) genitogram. Proper technique is mandatory. A perineal opaque marker may be used to allow for subsequent measurements and lateral projections images must be obtained. A complete retrograde cystography is performed after insertion of an 8-F Foley catheter in the anterior perineal orifice, followed by controlled-pressure retrograde injections along the urethro-vaginal route. Images show a persistent urogenital sinus and a normal vagina (arrow). The presence and the size of the vagina should be carefully assessed, along with the presence or absence of the cervical imprint on the vaginal dome (arrowheads), which are usually present in CAH because mullerian structures differentiation is not impaired. The configuration of the urethra (small arrow) indicates a male type urethra. **Origin:**

**b**



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