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## Ambiguous genitalia in neonates

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### Key messages

- Be aware of associated metabolic problems.
  - Palpable gonads imply the presence of testicular tissue.
  - Be very careful in your choice of words when talking to parents.
  - **The situation should be treated as a medical emergency**, with paediatric endocrine advice being sought immediately.
  - There is some ongoing controversy as to when is the appropriate time to make decisions as to sex of rearing.
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Ambiguous genitalia occur in approximately one in 4,500 births. **This situation is rarely anticipated and can be a source of great distress for parents, delivery room and nursery staff.**

### Do not assign gender

Often there can be pressure on medical staff to 'make it better' and assign a gender to the child arbitrarily in the first few hours after birth. This must be avoided at all costs.

### Choose your words carefully

Staff must be careful in their choice of words when discussing the baby with parents. It is unnatural not to discuss a baby without using the terms 'he' or 'she' and it is easy to accidentally refer to the baby in a gender-orientated way. Parents who are greatly distressed may assume that medical and nursing staff 'know' what the gender of the baby 'really is'.

Consequently any terminology used (deliberately or accidentally) will be given great emphasis by parents. This may lead to confusion and distress later if the suggested sex of rearing is at odds with initial 'off the cuff' remarks.

### Advice on naming

Parents may seek advice regarding the naming of their infant. The usual advice is to select non-ambiguous names (use gender-specific names) since it is thought that by encouraging the use of ambiguous (non-gender specific) names, staff are implying an ongoing sense of 'ambiguity'.

Parents might not name the baby until the gender is known.

## Management

Be very careful in your use of terms when discussing the baby with ambiguous genitalia.

Use the appropriate, non-gender orientated terms listed in Table 1.

**Table 1: Suggested phenomenology when dealing with babies with ambiguous genitalia**

Female	Ambiguous	Male
She Clitoris Labia Ovaries Vagina, urethra	Your baby Phallus Folds Gonads Urogenital sinus	He Penis Scrotum Testes Urethra

Never refer to the baby in question as 'it'.

The situation should be treated as a medical emergency, with paediatric endocrine advice being sought immediately.

## Clinical evaluation

Genital ambiguity can be quantified according to the Prader scale (see Figure 1).

Other relevant clinical details include:

- Are gonads palpable in the labioscrotal or inguinal regions?
- Is penile length and width normal? (length > 2.5 cm and < 4.5 cm in full-term infant)
- Where is the urethral opening?
- Is there pigmentation of the genitalia?
- Are there syndromic features?
- What is the metabolic condition of the baby (paying particular attention to glucose, sodium and potassium)?
- Examine the baby's mother for signs of hyperandrogenism.

Care should be taken in the interpretation of examination findings in growth retarded or premature female neonates. These children often have very little labial fat, with a prominent clitoris, giving an appearance of 'pseudo-ambiguity'. In addition biochemical parameters are different from preterm infants.

Breech delivery can result in clitoral and vulval oedema, again with the appearance of pseudo-ambiguity.

It is a moot point where the boundary lies between severe perineal hypospadias and genital ambiguity. Inability to palpate the gonads in this situation may be indicative of a diagnosis other than isolated hypospadias.

**Figure 1: Prader staging system for the degree of virilisation of the external genitalia**

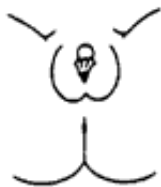
Prader 0: Normal female external genitalia



Prader 1: Female external genitalia with clitoromegaly



Prader 2: Clitoromegaly with partial labial fusion forming a funnel-shaped urogenital sinus



Prader 3: Increased phallic enlargement. Complete labioscrotal fusion forming a urogenital sinus with a single opening



Prader 4: Complete scrotal fusion with urogenital opening at the base or on the shaft of the phallus.



## Prader 5: Normal male external genitalia



Source: Prader Von, A. (1954). 'Der genitalbefund beim Pseudohermaproditismus femininus des kongenitalen adrenogenitalen Syndroms. Morphologie, Hausfigkeit, Entwicklung und Vererbung der verschiedenen Genitalformen. Helv. PEDIATR. ACTA. 9: 231-248.)

## Investigations

Blood should be sent for:

- chromosome analysis
- gonadotropins (LH, FSH)
- testosterone, 4 androstenedione (after day 3)
- serum electrolytes and glucose
- serum 17-hydroxyprogesterone (17OHP) levels (after day 3 of life).
- dihydrotestosterone (note: current assays may be inaccurate – seek specialist advice) (after day 3).

Pelvic/abdominal ultrasound, carried out by an experienced sonographer, should be undertaken as soon as possible to determine:

- pelvic structures
- the presence or absence of gonads.

Other investigations which may or may not be subsequently relevant include:

- sinugram
- human chorionic gonadotropin stimulation test (to assess testosterone and dihydrotestosterone synthesis capability). AntiMullerian hormone is now taking over from this investigation.

## Differential diagnoses

Gonads palpable, 46XY:

- gonadal dysgenesis
- partial androgen insensitivity
- biosynthetic defect in either testosterone or dihydrotestosterone production.

Gonads impalpable, 46XX:

- **congenital adrenal hyperplasia (CAH)**
- gonadal dysgenesis
- exogenous androgen exposure.

Often only one gonad palpable, Mosaic karyotype:

- gonadal dysgenesis.

## Ongoing management

Decision as to sex of rearing is made after opinions have been sought from the endocrine and surgical teams.

It should be undertaken with the baby's parents after all the relevant investigation results have been discussed.

Decisions as to sex of rearing may have no relationship to karyotypic, gonadal or genital status in isolation.

The decision will be influenced by an amalgam of:

- the baby's karyotype
- gonadal status
- internal and external genital duct status
- potential for fertility and adequate sexual function
- cultural influences.

## Birth certificate - wait until decision is made

Do not complete the baby's birth certificate until the sex of rearing has been decided. There is a 60-day period of grace between the birth of a child and when their birth certificate needs to be completed, so there is no rush.

If the 'wrong' sex is entered on the form it is extremely difficult to correct and requires judicial intervention.

## Long-term care

Issue to note:

- Families require long-term medical and psychological support.
- Corrective surgery is usually undertaken within the first year of life but timing can be controversial. Very early surgery at under six months of age is less commonly performed than in the past.
- Infants with **CAH** and congenital syndromes have additional requirements for ongoing medical therapy.
- Disclosure to the patient about their diagnosis is usually undertaken in mid to late adolescence when they have the ability to understand complex issues such as chromosomes, hormones etc, and possess some degree of emotional maturity.

## More information

### Clinical

- UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development. Ahmed SF, Achermann JC, Arlt W et al. Clin Endocrinol (Oxf). 2011 Jul;75(1):12-26
- Changes over time in sex assignment for disorders of sex development. Kolesinska Z, Ahmed SF, Niedziela M, et al Pediatrics. 2014 Sep;134(3):e710-5

### Consumer

- [Mayo clinic Ambiguous genitalia](#)
- [What is DSD?](#)

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<https://www2.health.vic.gov.au:443/hospitals-and-health-services/patient-care/perinatal-reproductive/neonatal-e handbook/congenital-abnormalities/ambiguous-genitalia>

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