

DISORDERS OF SEXUAL DEVELOPMENT

RECOGNITION AND ASSESSMENT

Definition

- New nomenclature: disorders of sexual development (DSD) [known formerly as ambiguous genitalia](#)
- Congenital conditions in which development of chromosomal, gonadal or anatomical sex is atypical, most commonly:
 - congenital adrenal hyperplasia
 - gonadal dysgenesis
 - partial androgen insensitivity
- For DSD classification, see **Supporting information**

Factors suggesting DSD

- Overt genital ambiguity (e.g. cloacal extrophy)
- Apparent female genitalia with enlarged clitoris, posterior labial fusion or inguinal/labial masses
- Apparent male genitalia with bilateral undescended testes, isolated perineal hypospadias, mild hypospadias with undescended testis
- Family history of DSD [e.g. complete androgen insensitivity syndrome (CAIS)]
- Discordance between genital appearance and pre-postnatal karyotype
- Pseudo-ambiguity (atrophic vulva and clitoral oedema) in growth-restricted or preterm female infants

PRINCIPLES OF MANAGEMENT

This is a medical emergency: involve consultant immediately

- Avoid gender assignment before expert evaluation
- Consultant to discuss with parents
 - always use the term 'baby' and avoid using 'he', 'she' or, most importantly, 'it'
 - advise parents about delaying registration and informing wider family and friends until gender assignment complete
- Link with expert centre for appropriate evaluation
- Communicate openly with family
- Respect family concerns and culture
- DSD is not shameful
 - potential for well-adjusted individual and a functioning member of society
 - best course of action may not be clear initially
 - parents need time to understand sexual development

First line investigations

- [Blood pressure](#)
- Karyotype (urgent)
- Imaging
 - abdominal and pelvic ultrasound by an experienced paediatric sonographer
- [Steroid chromatogram/profile, especially 17-OHP](#) (delay until day 4-5 to allow maternal hormonal effects to decline)
- Testosterone and oestradiol
- LH, FSH
- [U&E and glucose](#)
- Cortisol

Further investigations (locally and/or in conjunction with specialist advice)

- dHT (dihydrotestosterone)
- DHEA (dihydroepiandrosterone)
- Androstenedione
- Urine steroid analysis

- ACTH
- LHRH hCG stimulation
- ACTH stimulation test
- AMH (anti-mullerian hormone) imaging studies
- Biopsy of gonad
- Molecular genetic studies (e.g. for CAIS)

TREATMENT

- Check [serum](#) electrolytes and [plasma](#) glucose
- [Involves a multidisciplinary team with an identified person \(usually consultant neonatologist\) acting as primary contact with family](#)
- Specific treatment dependent on many factors and the diagnosis
- discuss with specialists