DISORDERS OF SEXUAL DEVELOPMENT ● 1/2

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 exstrophy)
- Apparent female genitalia with enlarged clitoris, posterior labial fusion or inguinal/labial masses
- Apparent male genitalia with bilateral undescended testes, isolated perineal hypospadias, micropenis (normal penis ≥1.9 cm), or mild hypospadias with undescended testis
- Family history of DSD e.g. complete androgen insensitivity syndrome (CAIS)
- Discordance between genital appearance and antenatal karyotype
- Pseudo-ambiguity (atrophic vulva and clitoral oedema) in growth-restricted or preterm female babies

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
  - liaise with laboratory to enable evaluation without indicating gender in laboratory request forms
- Link with expert centre for appropriate evaluation
- Communicate openly with family
- Respect family concerns and culture
- DSD is not shameful
  - best course of action may not be clear initially
  - parents need time to understand sexual development

First line investigations

- Blood pressure
- Karyotype of QF-PCR (urgent)
- Imaging
  - abdominal and pelvic ultrasound by an experienced paediatric sonographer
  - assess presence and nature of internal genitalia, including gonads
- Blood tests
- cortisol short synacthen test
  - 17-OHP (delay until day 3 to allow maternal hormonal effects to decline)
  - testosterone and oestradiol
  - LH, FSH
  - U&E and glucose

Further investigations (following discussion with specialist endocrine advice)

- dHT (dihydrotestosterone)
- DHEA (dihydroepiandrosterone)
- Androstenedione
- ACTH
- LHRH and hCG stimulation
- ACTH stimulation test
- AMH (anti-mullerian hormone) imaging studies
- Molecular genetic studies [e.g. for complete androgen insensitivity syndrome (CAIS)]
- Urine: steroid profile
- Biopsy of gonad

DISORDERS OF SEXUAL DEVELOPMENT ● 2/2

TREATMENT

- Avoid unnecessary admission to NNU
- Check serum electrolytes and plasma glucose
  - in congenital adrenal hyperplasia electrolytes usually not abnormal until day 4
- Involves a multidisciplinary team with an identified person (usually consultant neonatologist) acting as primary contact with family
- Specific treatment dependent on many factors and diagnosis
  - discuss with specialists