

Treatment of AIS patients in multidisciplinary teams in UK

Tratamiento del SIA en equipos
multidisciplinares en el Reino Unido

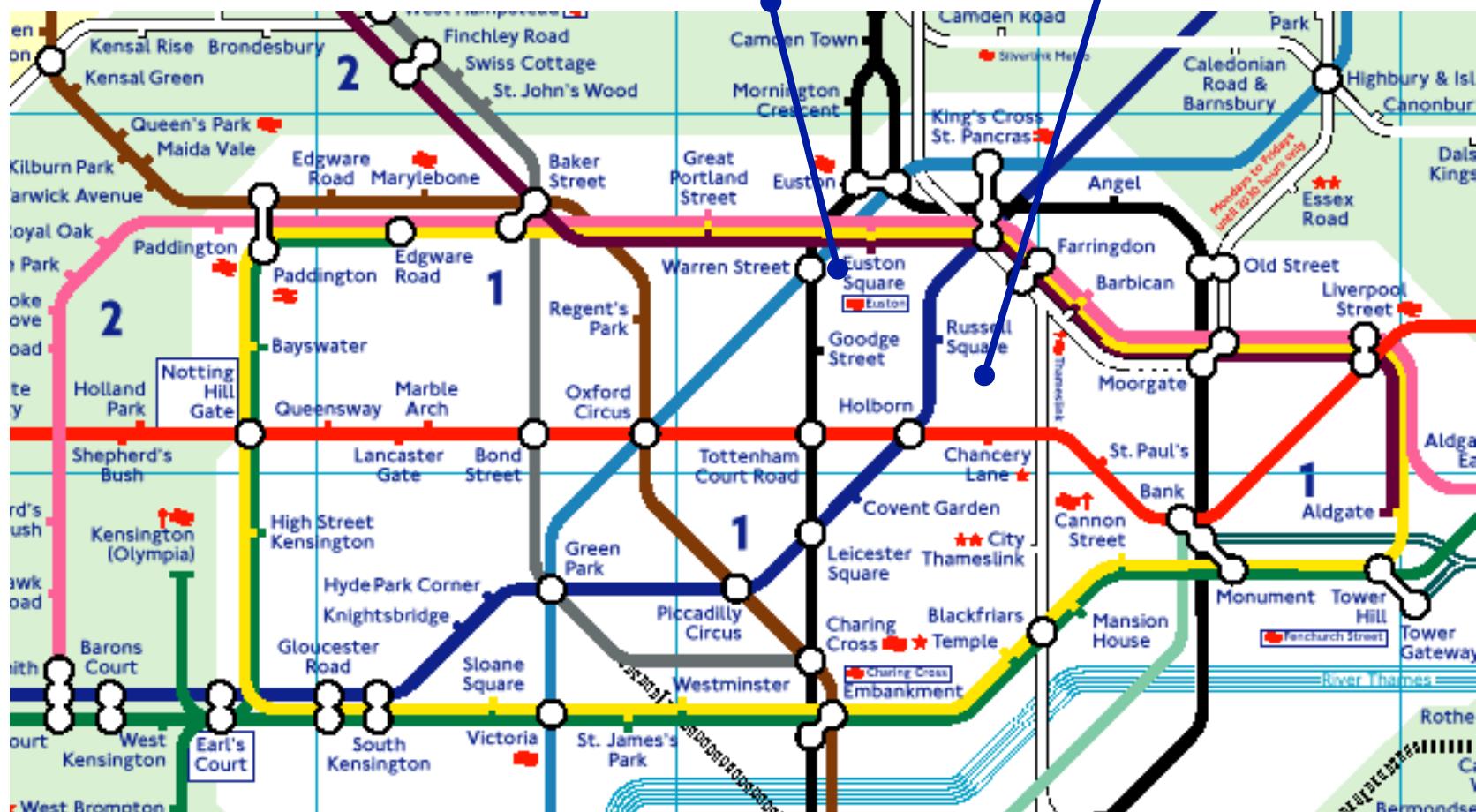
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University
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DSD team at UCLH

Gerard Conway – Endocrinology and Genetics



Lih Mei Liao – Consultant Psychologist



Sarah Creighton - Gynaecology



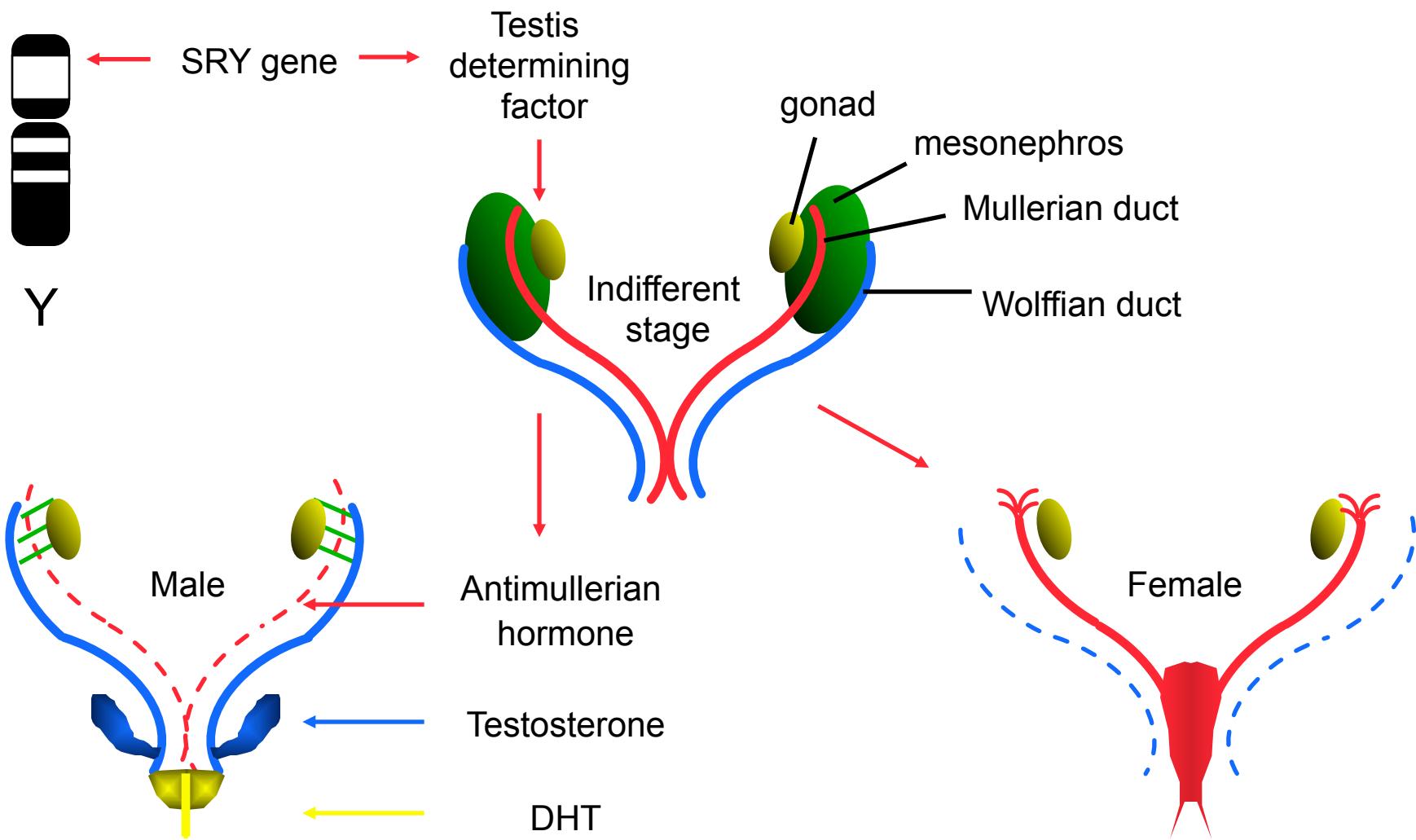
Determinants of sex and intersex

- Genetic sex
male
46XY female, 45X, 46XX
- Gonadal sex
Hermaphroditism / CAIS
- Genital sex
Micropenis / Hypospadias
Cliteromegaly - CAH

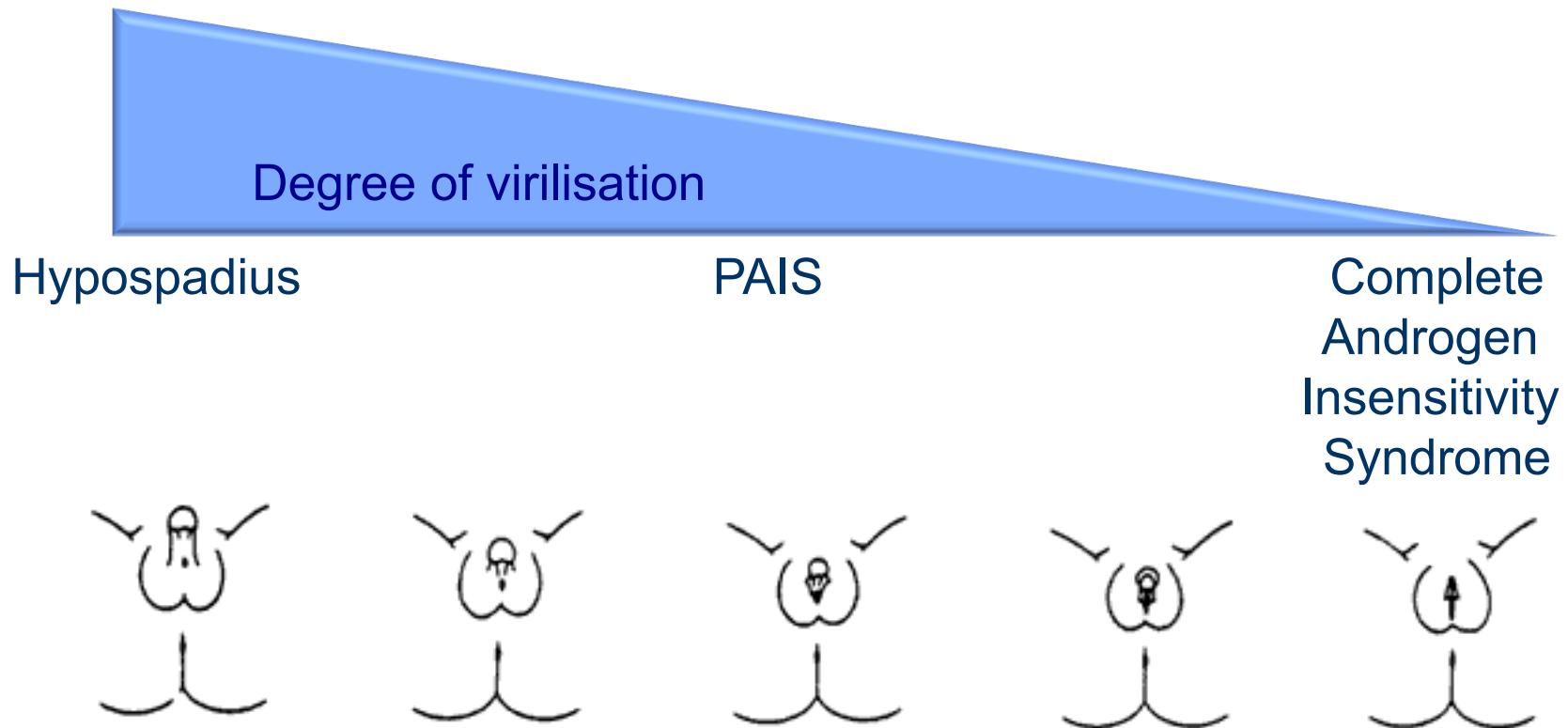
Determinants of sex and intersex

- Genetic sex
 - Turner Syndrome – 650
 - 46XY gonadal dysgenesis - 60
 - Klinefelter syndrome 50
- Gonadal sex
 - CAIS – 110
 - Partially virilised XY DSD - 94
- Genital sex
 - Adrenal Hyperplasia - 260

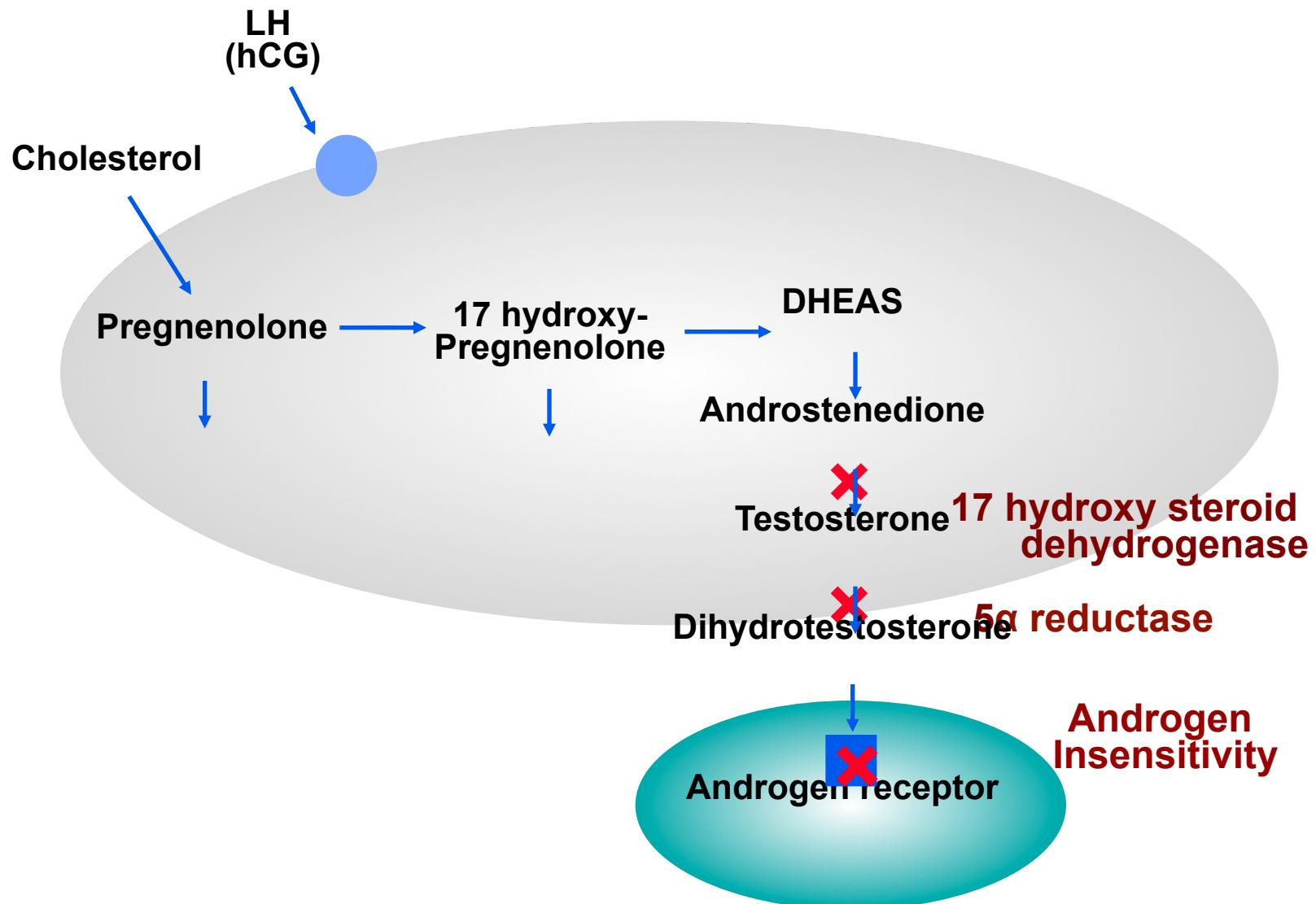
Embryonic sex differentiation



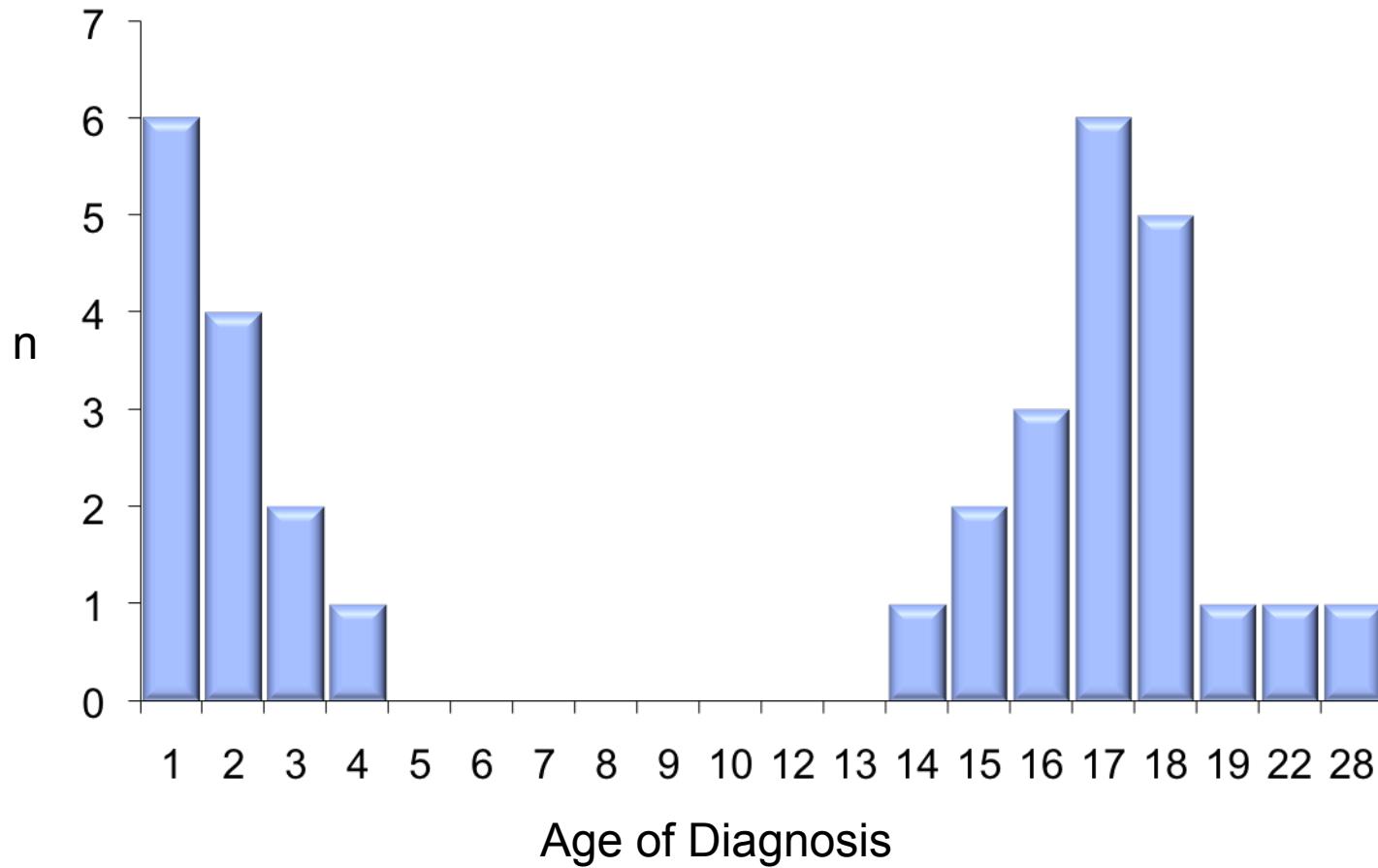
Spectrum of 46XY DSD



Defects in Testosterone Synthesis



CAIS – Age of Presentation



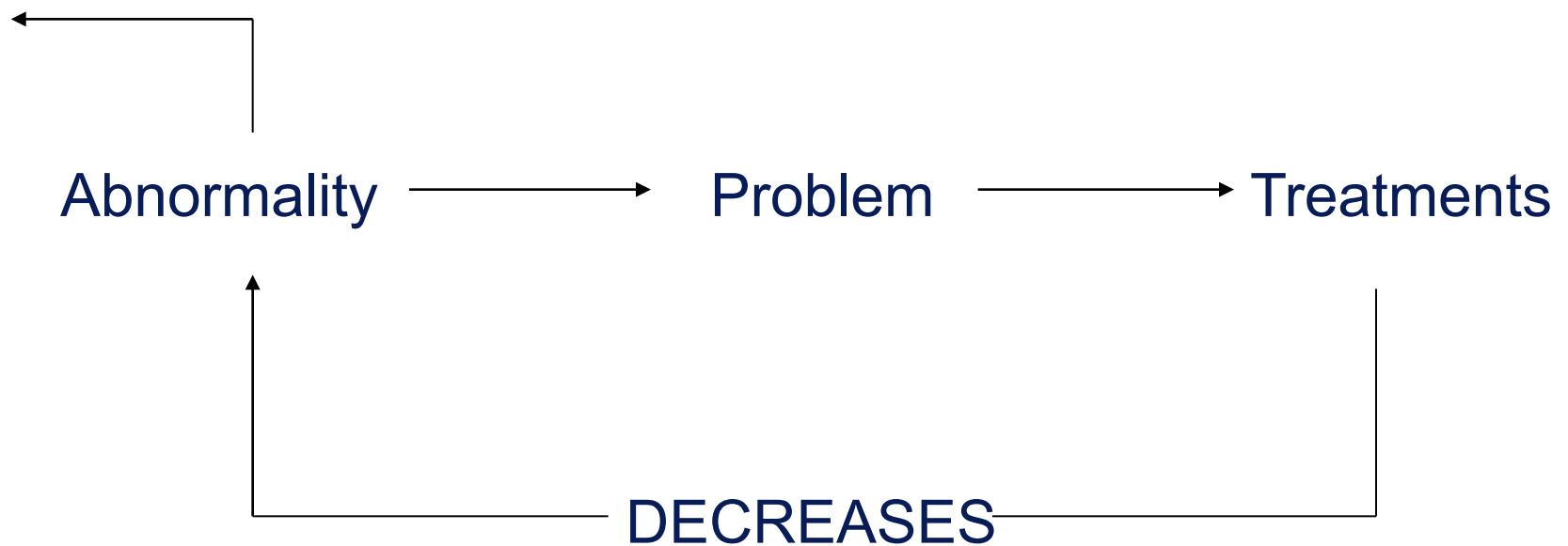
DSD Adults – Clinic problems

- 50% of DSD presents as adult
- Variable early experiences
 - age < 25 very different from age > 30
- Dispersed care
- Little psychology provision

DSD Adults

- Empowerment through knowledge and choice
- Choice of surgical / fertility / HRT options
- Pathogenesis and Genetics revisited
- Tracking long term consequences
- Group forum – ‘open days’

Managing anatomy: the old medical approach



Initial assessment

- Readiness to discuss
- Establishing a shared language
- Exploring relationship to diagnosis:
 - Knowledge of diagnosis
 - Meanings
 - Emotional responses to diagnosis

Helping with decision making

- Medical treatment
- Surgery
- Fertility
- Genetic testing

Hormone replacement

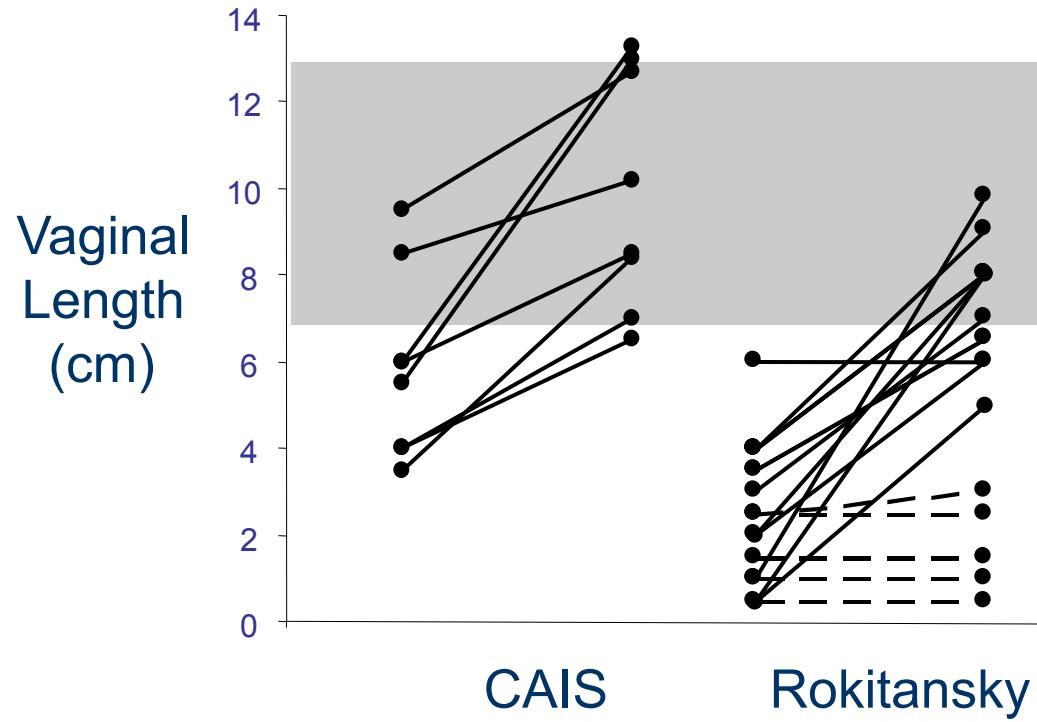
- Which hormone? oestrogen
testosterone
progesterone
- Which route? tablets
patch or gel
vagina
- Which dose? up a little / down a little
- How long for? until the of 50?

Central psychological concerns

- Self disclosure (diagnosis, potential sexual difficulties, continence, infertility...)
- Body image (breast development, genital appearance, hirsutism, short stature...)
- Relationships (being in it, not being in it)
- Parenthood
- Improve treatment compliance *



Avoiding surgery with vaginal dilatation



Ismail et al Human Reproduction 2006

DSD- Early experiences of health care

- Secrecy
- Over exposure (e.g. photography, repeat intimate examinations, psychological testing)
- (De-)sexualisation
- Childhood hospitalisation (separation from family, loss of control, invasion of privacy, out of sync with peer groups...)

DSD – family experiences

- Parental handling of information relating to diagnosis and treatment
- Emotional distress in siblings
- Financial hardship as a result of some DSDs
- Difficulties in separation/transition

Assessing sexual difficulties

- gender identity
- gender(s) of preferred sexual partners
- body perceptions
- sexual experiences
- sexual and relationship aspirations
- knowledge of a range of sexual activities

Sex therapy

- Improving understanding of past and present influences on identified problem(s)
- Increasing knowledge of wide variations in male and female sexualities; de-centralizing binary ideas of sex.
- Increasing control over social and sexual situations
- Self permission to explore a range of sexual activities – alone or partnered .
- Self permission not to pursue sexual activities where there is no barrier other than an absence of desire or even a wish to desire

Aims For DSD centres

- Building a mature team:
 - Clear vision that everyone shares
 - Genuine respect for each other's contributions
 - Psychology not as stand-alone treatment
 - Interdisciplinary working
 - Working with support groups to develop goals



DSD Team at UCL

| | |
|-----------------------|---------------------------------|
| Endocrinology | Gerard Conway |
| Gynaecology | Sarah Creighton |
| Psychology | Lih-Mei Liao |
| Fertility | Melanie Davies |
| Paediatrics | Carline Brain |
| Urology | Dan Wood, Christopher Woodhouse |
| Laparoscopic Surgeons | Tom Kurzawinski, Alfred Cutner |
| Molecular Genetics | Gill Rumsby, John Achermann |
| Steroid Biochemistry | John Honour |