

Treatment of AIS patients in
multidisciplinary teams in UK

Tratamiento del SIA en equipos
multidisciplinares en el Reino Unido

Gerard Conway

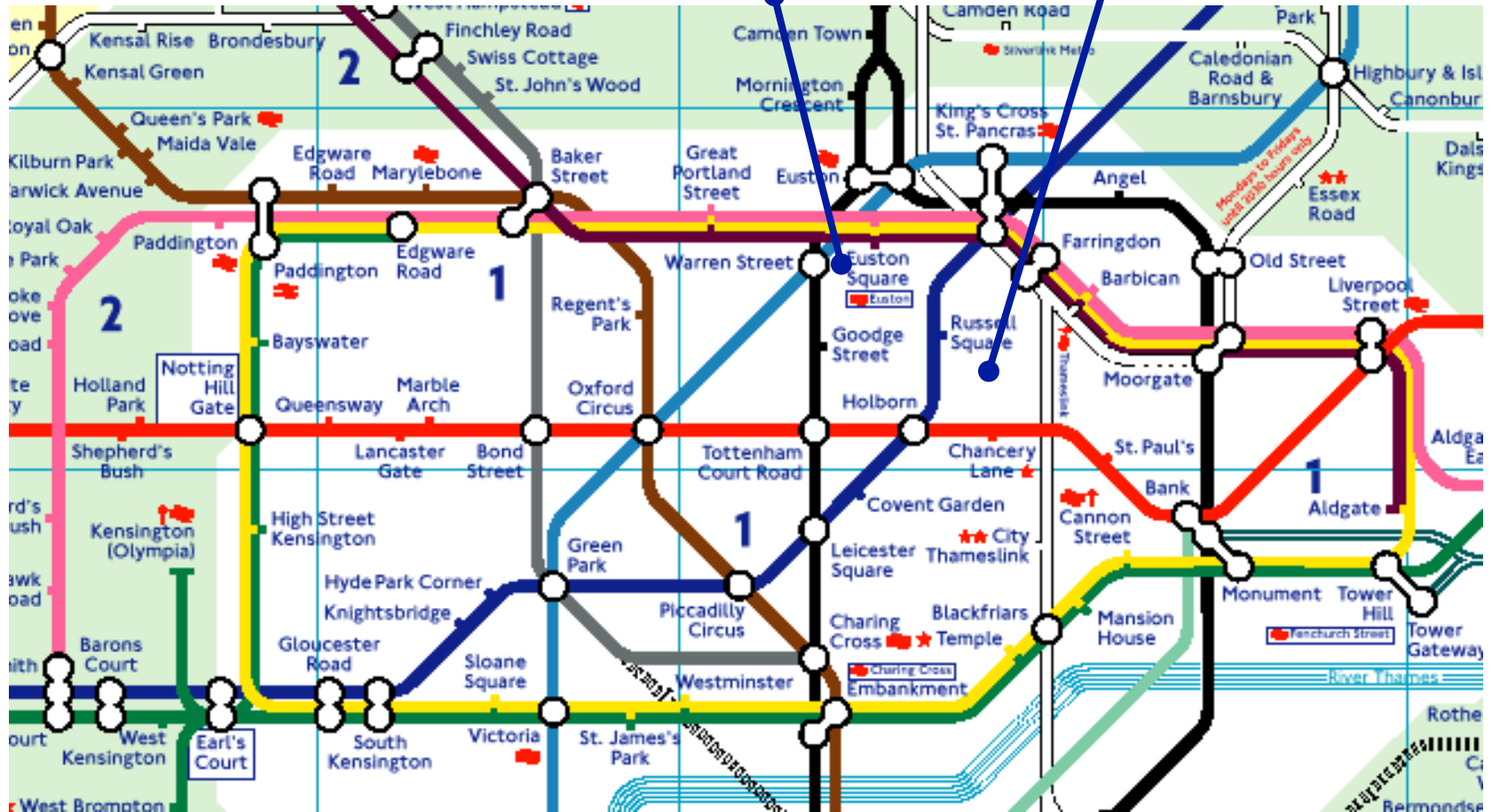
London, UK

University College London Hospitals



University
College London
Hospital

Great
Ormond Street
Hospital



DSD team at UCLH

Gerard Conway – Endocrinology and Genetics



Lih Mei Liao – Consultant Psychologist



Sarah Creighton - Gynaecology



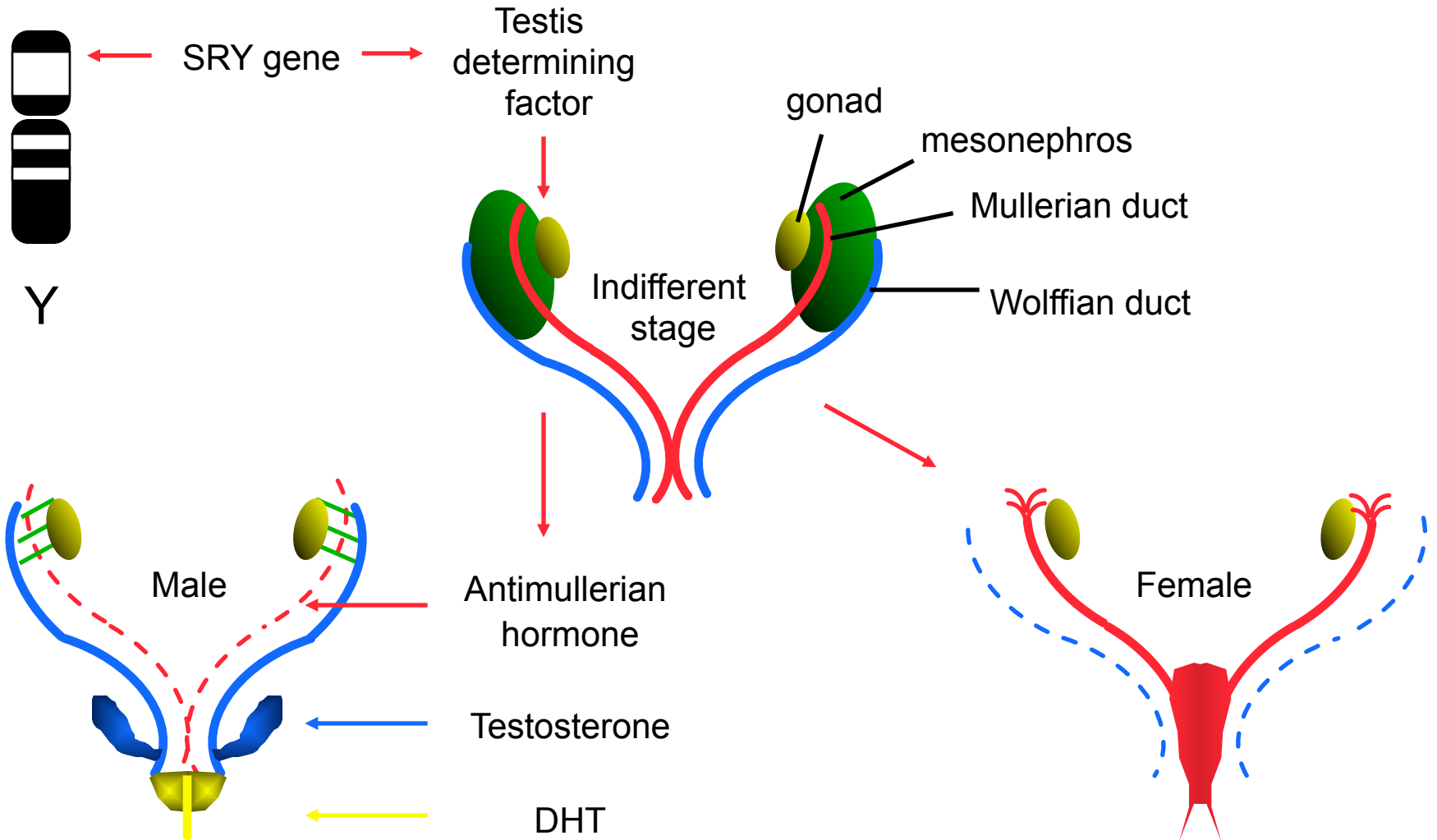
Determinants of sex and intersex

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

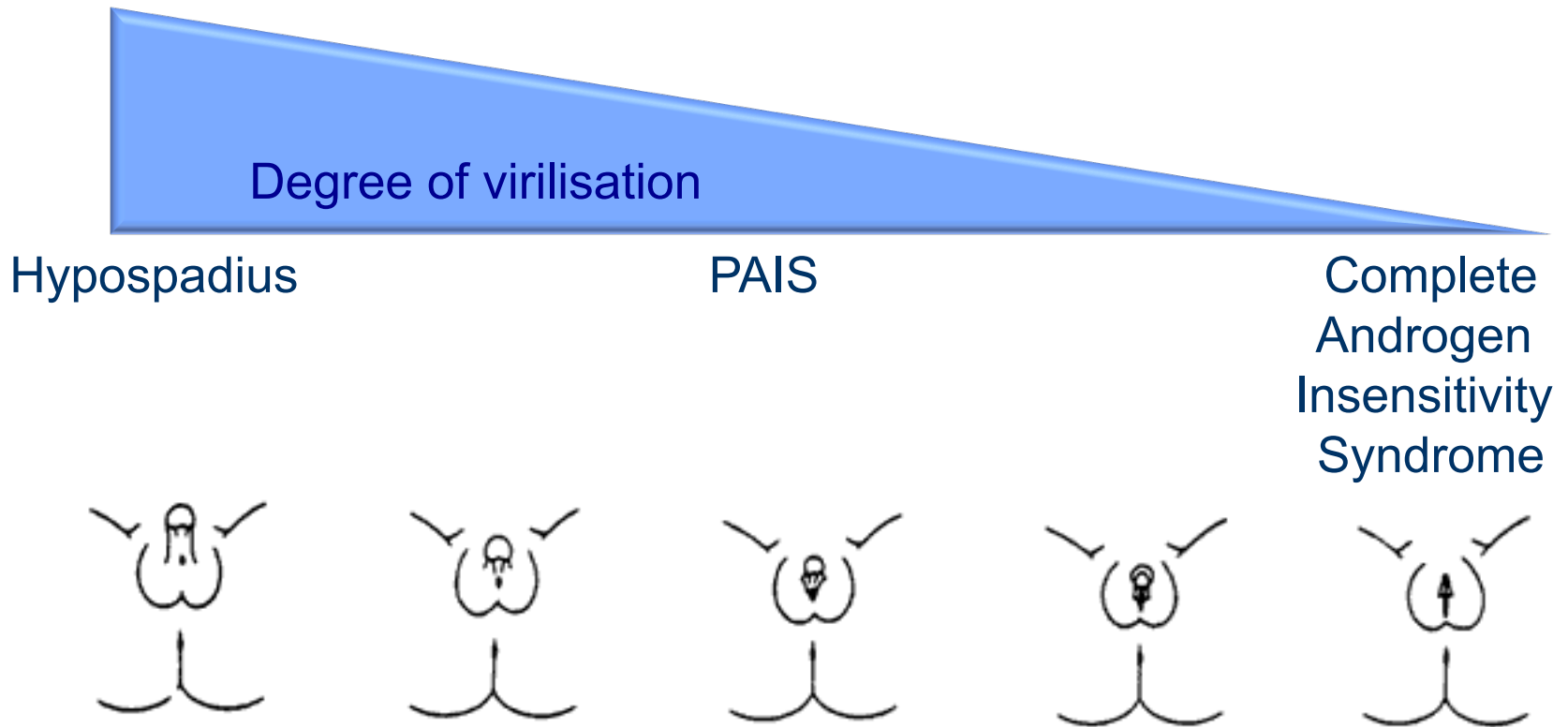
Determinants of sex and intersex

- 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

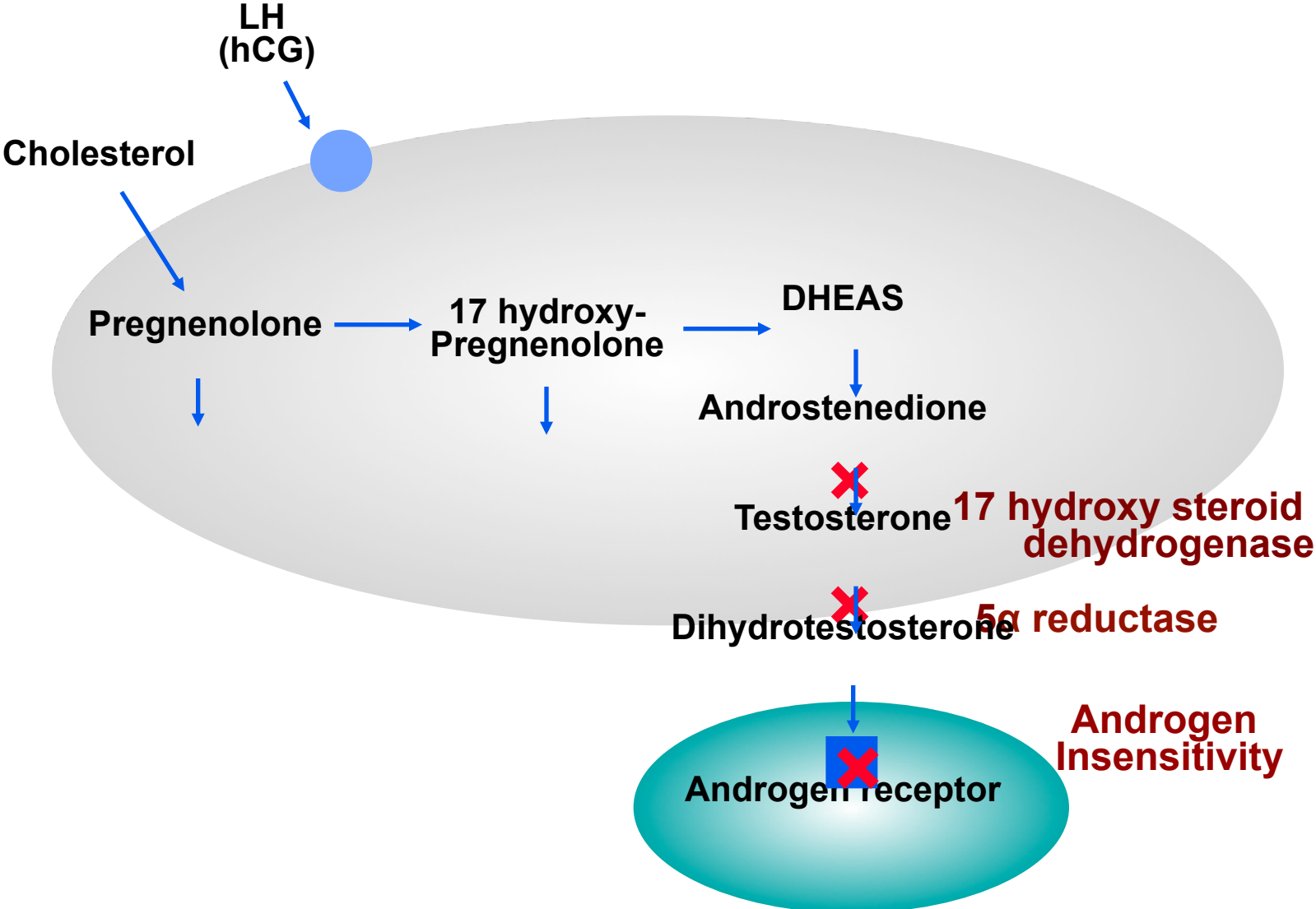
Embryonic sex differentiation



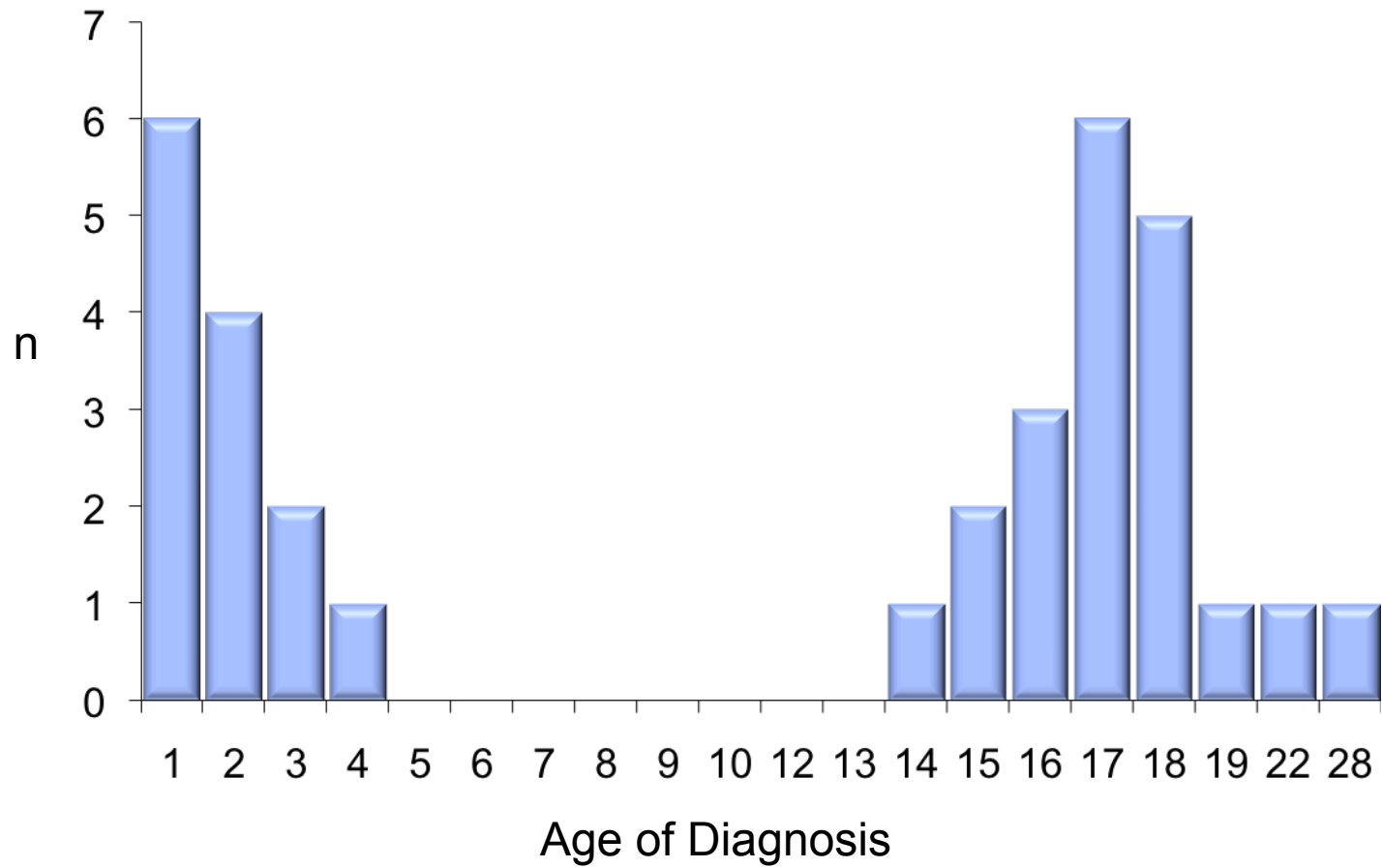
Spectrum of 46XY DSD



Defects in Testosterone Synthesis



CAIS – Age of Presentation



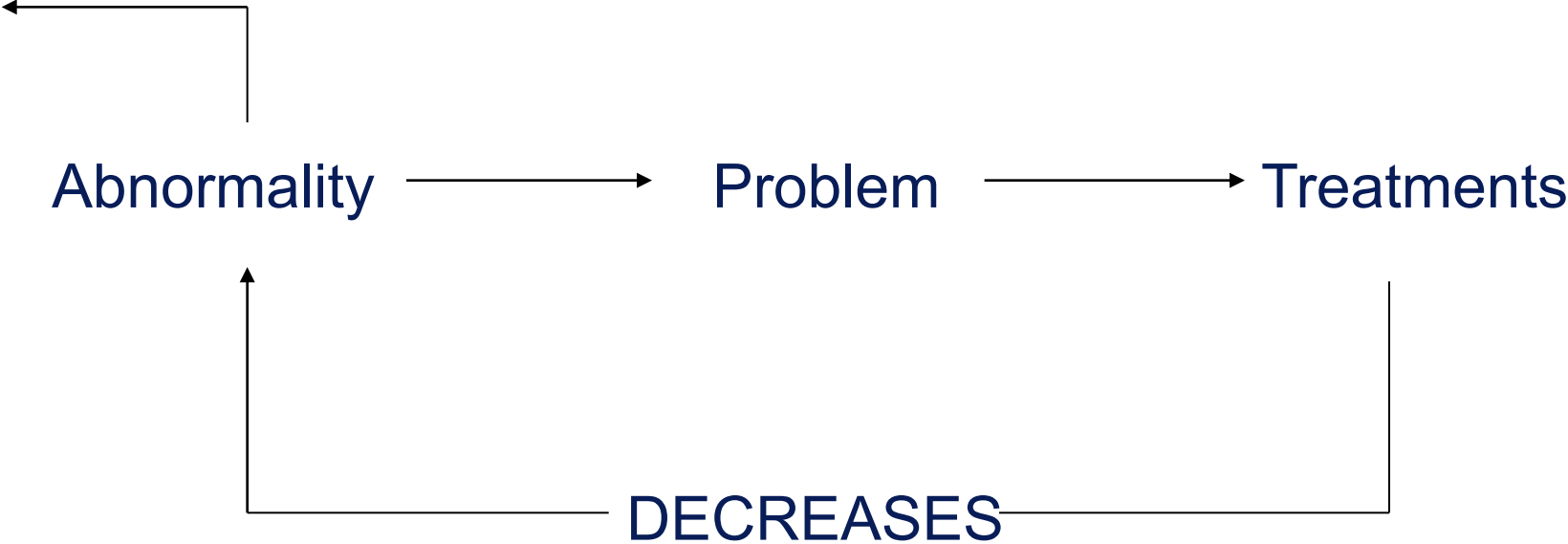
DSD Adults – Clinic problems

- 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

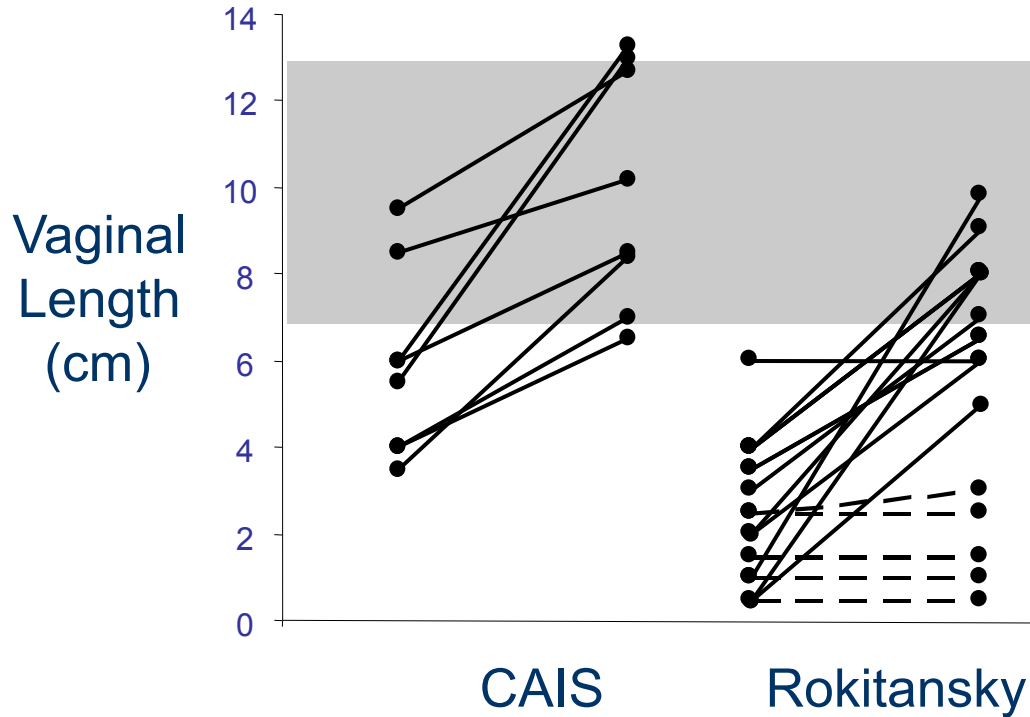
- Which hormone?
oestrogen
testosterone
progesterone
- Which route?
tablets
patch or gel
vagina
- Which dose?
up a little / down a little
- How long for?
until the of 50?

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 synch 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

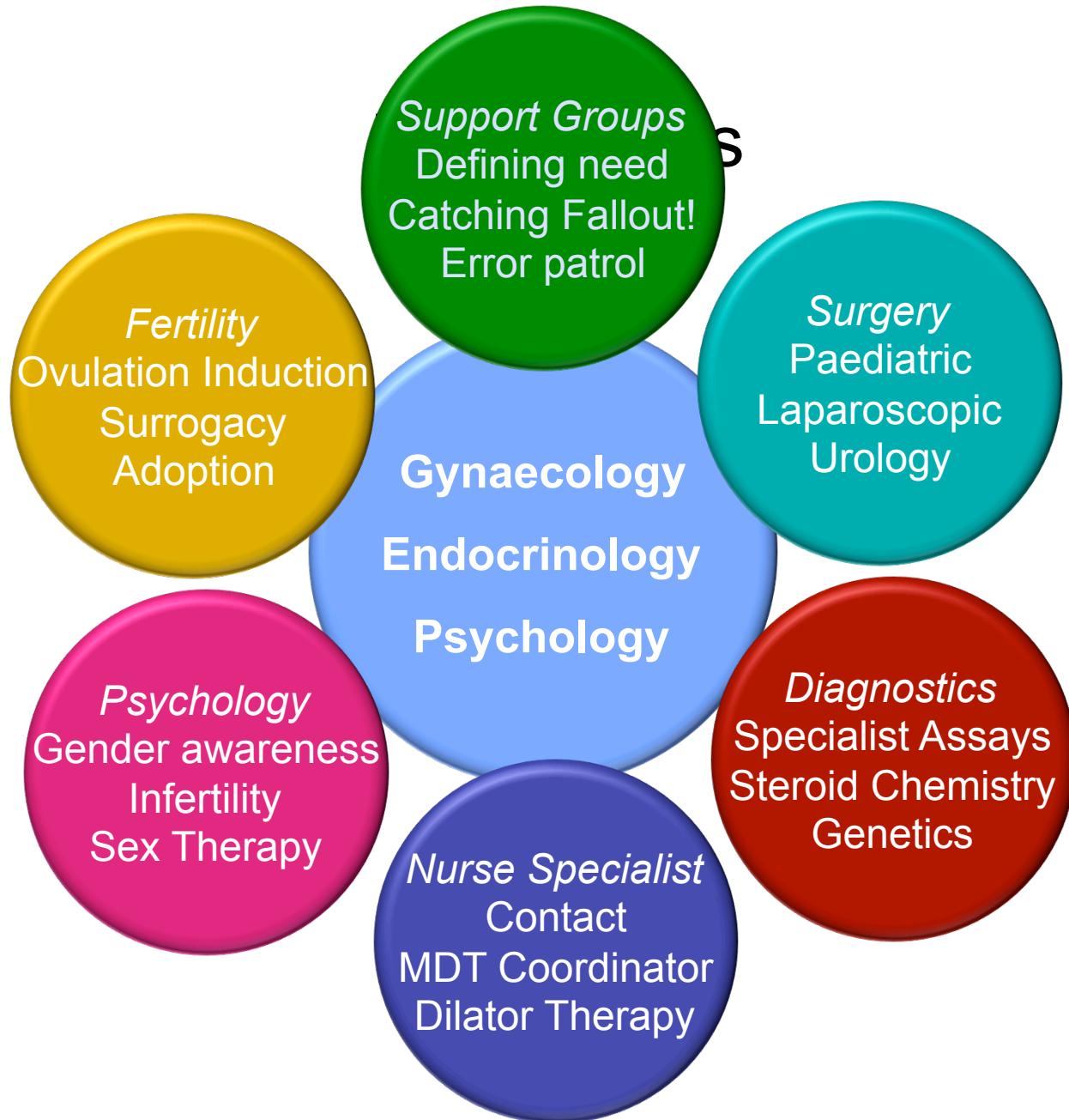
- gender identity
- gender(s) of preferred sexual partners
- body perceptions
- sexual experiences
- sexual and relationship aspirations
- knowledge of a range of sexual activities

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