PIPADOR REVISION

MRCOG Part 1 Revision Notes

Congenital Urogenital Anomalies

EMBRYOLOGY • LECTURE 65

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 65: Congenital Anomalies of the Urogenital Tract

Embryology • Visual revision edition — integrating Lectures 55–64

COLOUR CODE USED THROUGHOUT THESE NOTES

INTERMEDIATE MESODERM	UROGENITAL SINUS	MÜLLERIAN	EXTERNAL GENITALIA	DSD / RED FLAGS

LEARNING OBJECTIVES — tick off as you go

- ☐ Place each urogenital anomaly in its embryological territory.
- ☐ Interpret the key antenatal findings (liquor, megacystis, hydronephrosis, kidneys, genitalia).
- ☐ Summarise Potter sequence, PUV, exstrophy and Müllerian anomalies.
- ☐ Classify DSD and explain CAH, CAIS, 5α-reductase deficiency and Swyer syndrome.
- ☐ Recognise the syndromic associations and plan counselling.

1 The Big Picture

IN ONE LINE: *Urogenital anomalies (CAKUT ~3–6 per 1000 births) share embryological territory — so they cluster. Find one, look for the other.*

The urogenital tract — three territories

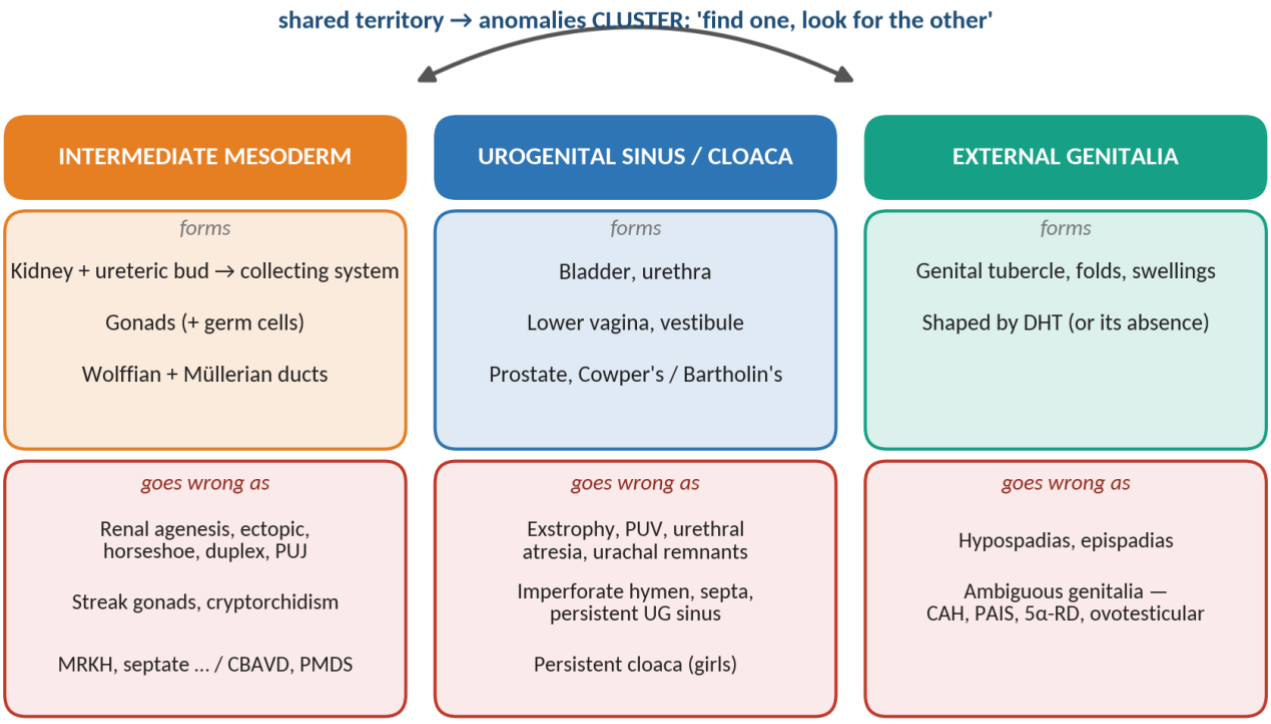


Figure 1. Three territories and their anomalies.

2 Antenatal Findings

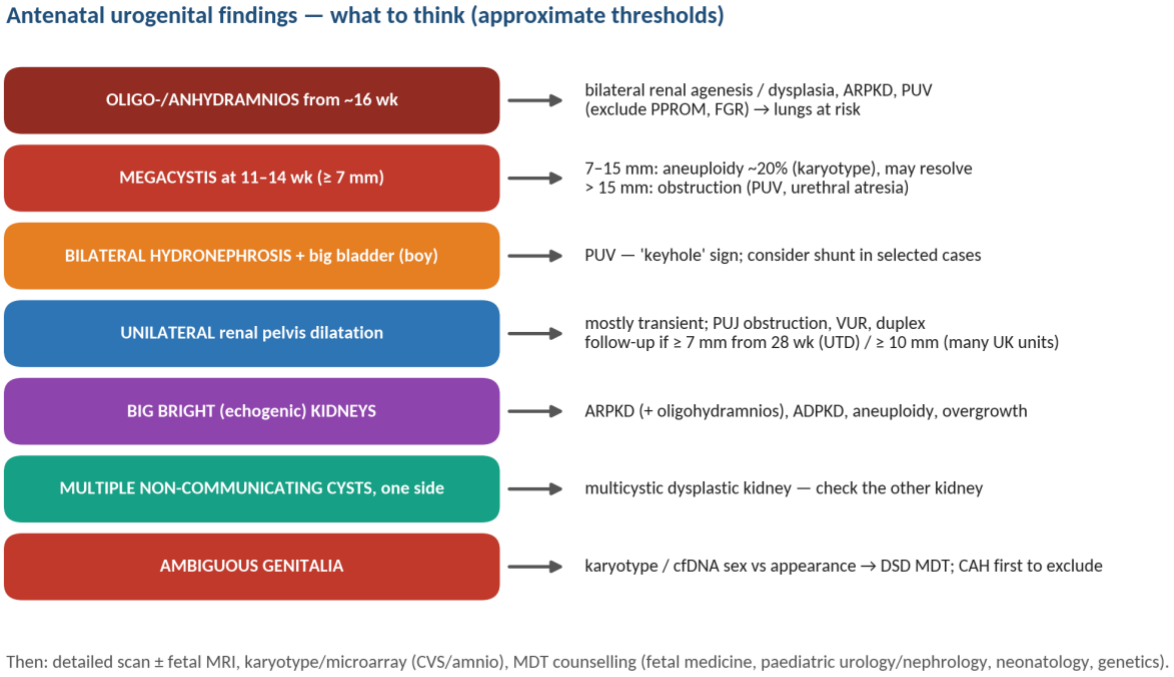


Figure 2. What each finding suggests.

Scan point	Timing
Bladder	Visible from ~12 weeks
Kidneys	From ~12–14 weeks; checked at the 18^{+0}–20^{+6} anomaly scan
Genitalia	From ~14–16 weeks; cell-free DNA sex from ~9–10 weeks
Liquor	Mainly fetal urine from ~16 weeks — so renal failure shows as oligohydramnios from then

3 Kidneys, Bladder and Outflow

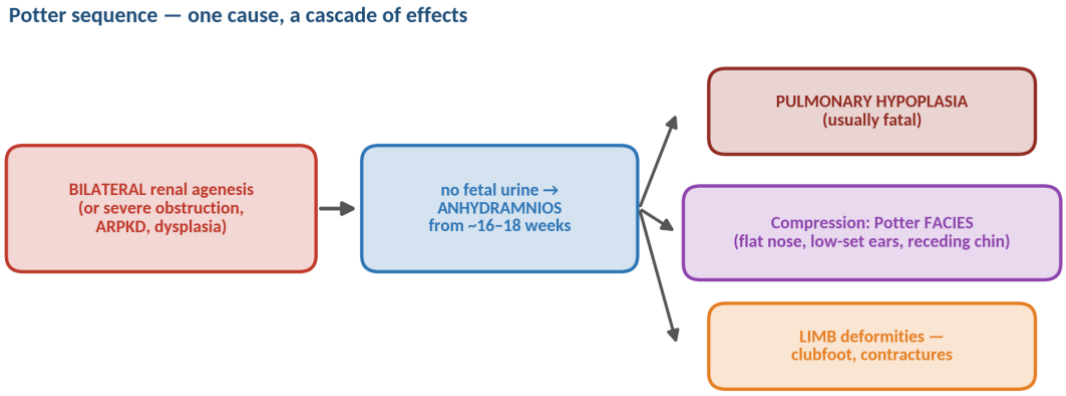


Figure 3. Potter sequence (from Lecture 55).

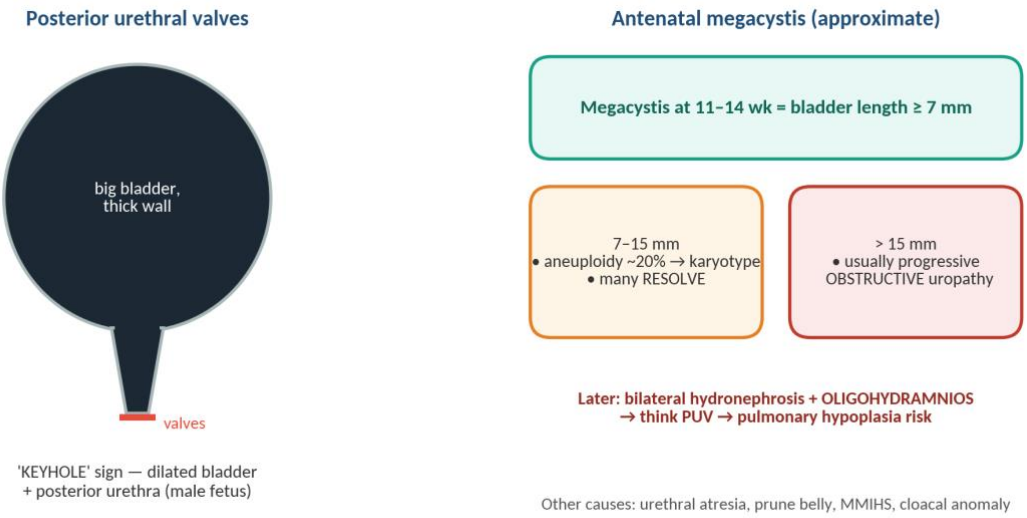


Figure 4. Megacystis and PUV (from Lecture 56).

Condition	Key points
Bilateral renal agenesis	Anhydramnios from ~16 wk → pulmonary hypoplasia (cause of death), Potter facies, limb contractures, breech — lethal
PUV	Commonest cause of obstructive megacystis + bilateral hydronephrosis in boys; keyhole sign; shunting in selected cases (PLUTO: possible survival gain, poor renal outcomes); lifelong CKD follow-up
PUJ obstruction	Commonest pathological antenatal hydronephrosis (most mild dilatation is transient); pyeloplasty if persistent/severe
VUR	Commonest congenital cause of recurrent childhood UTI; MCUG, DMSA for scars; selective prophylaxis, endoscopic dextranomer–hyaluronic acid injection or reimplantation
Bladder / cloacal exstrophy	Failed mesoderm ingrowth; epispadias, pubic diastasis • OEIS for cloacal exstrophy
Persistent cloaca	Girls only; one perineal opening for urinary, genital and bowel tracts

4 Müllerian Anomalies

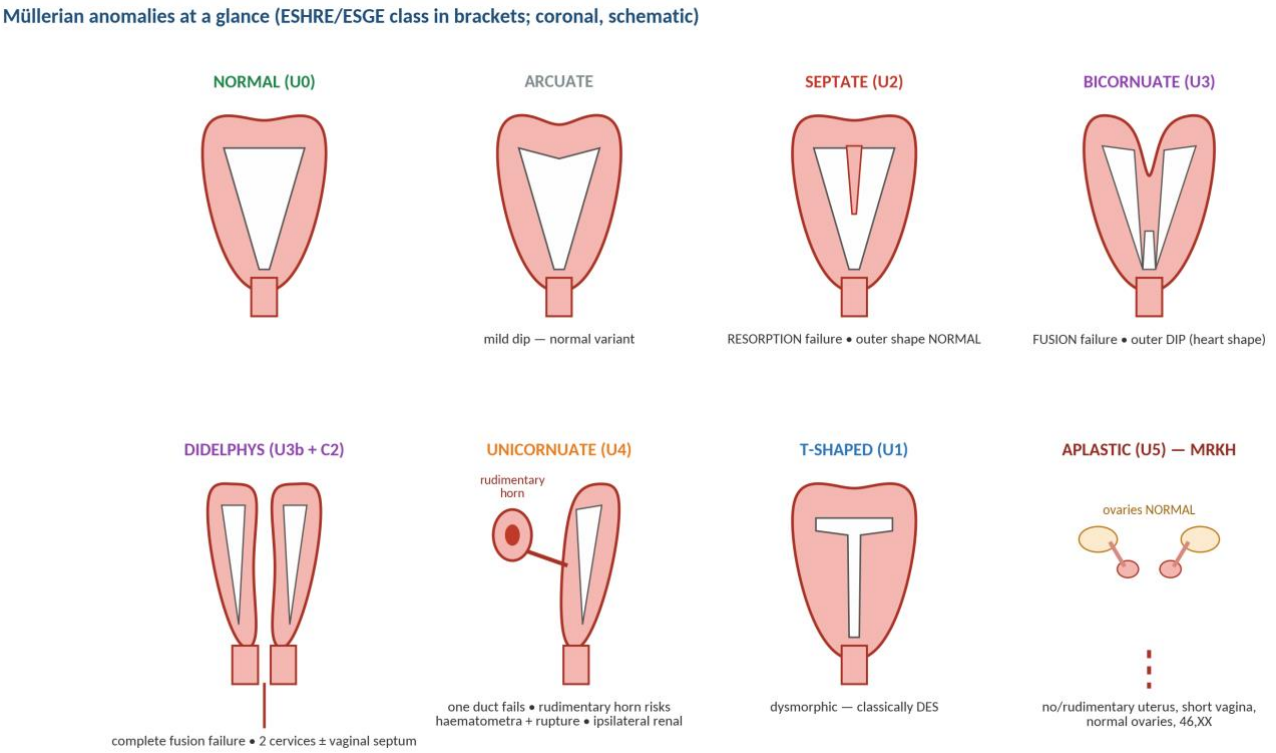


Figure 5. Müllerian anomalies (from Lecture 62).

Anomaly	ESHRE/ESGE	Error	Key point
MRKH	U5	Both ducts absent	46,XX, normal ovaries; renal anomalies ~30–40%
Unicornuate	U4	One duct absent	Ipsilateral renal agenesis; rudimentary horn risks
Bicornuate	U3a / U3b	Partial / complete fusion failure	Outer fundal dip; preterm, breech
Didelphys	U3b + C2	Complete fusion failure	Two cervices, vaginal septum; OHVIRA
Septate	U2	Resorption failure	Commonest; miscarriage link; resection benefit uncertain

5 Differences of Sex Development

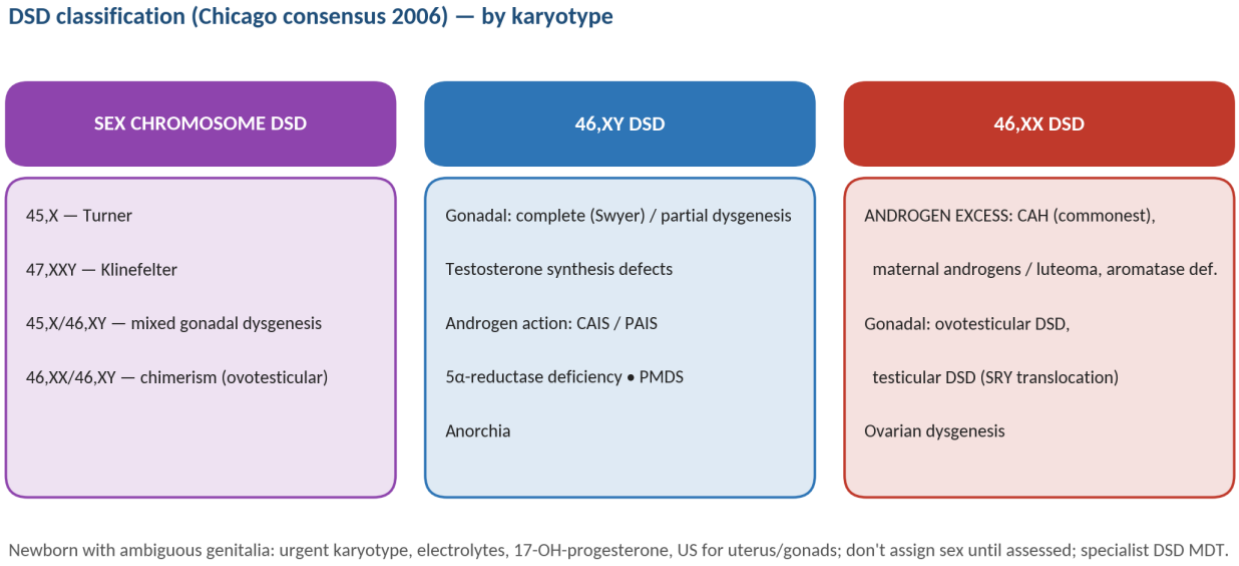


Figure 6. Chicago classification.

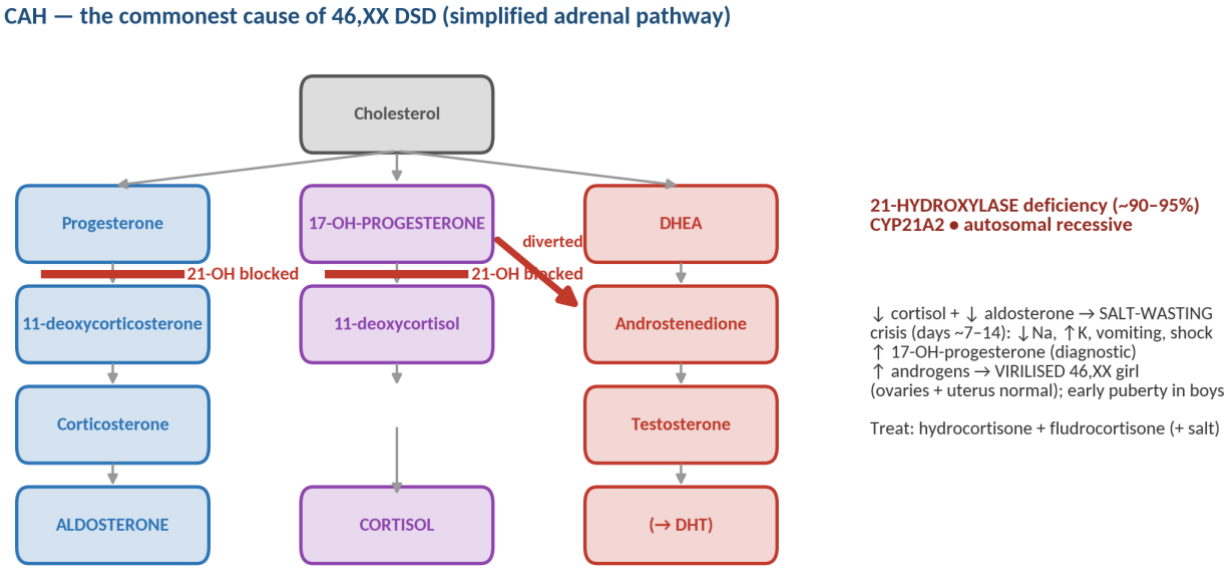


Figure 7. CAH — 21-hydroxylase deficiency.

46,XY differences of sex development — side by side

	Karyotype	Gonads	Müllerian (uterus/tubes)	Wolffian (vas, epididymis)	External genitalia	Key point
Complete AIS	46,XY	testes	ABSENT	absent	FEMALE	AR (X-linked); breasts, scant hair; gonadectomy often after puberty
5α-reductase deficiency	46,XY	testes	absent	PRESENT	AMBIGUOUS	SRD5A2; virilises at puberty; variable gender identity
PMDS	46,XY	testes (often undescended)	PRESENT	present	MALE	AMH / AMHR2; uterus found at hernia repair or orchidopexy
Swyer (46,XY CGD)	46,XY	STREAKS	PRESENT	absent	FEMALE	no AMH, no testosterone; gonadoblastoma → gonadectomy

Logic: Müllerian ducts vanish only with AMH (Sertoli); Wolffian ducts survive only with testosterone; external male genitalia need DHT.

Figure 8. 46,XY DSDs compared (from Lecture 60).

CAH (46,XX)

- **Commonest cause of 46,XX DSD**; 21-hydroxylase (CYP21A2), recessive
- Virilised girl, normal uterus/ovaries; **salt-wasting crisis** days ~7–14
- Hydrocortisone + fludrocortisone; carrier testing, antenatal diagnosis

46,XY CONDITIONS

- **CAIS**: female, no uterus, testes, primary amenorrhoea with breasts
- **5α-RD**: ambiguous at birth, virilises at puberty
- **Swyer**: streak gonads, **no breast development** without HRT, normal/tall; gonadectomy (gonadoblastoma); SRY changes in a minority only

- Management has moved towards **specialist MDT care, psychological support, shared decisions and deferring irreversible surgery** where possible.

6 Associations and Counselling

Syndromes and sequences to recognise

VACTERL
Vertebral, Anal, Cardiac, TracheoEsophageal, Renal, Limb

MURCS
Müllerian aplasia, Renal aplasia, Cervicothoracic Somite anomalies

OHVIRA
Obstructed HemiVagina + Ipsilateral Renal Agenesis (+ didelphys)

OEIS
Omphalocele, Exstrophy (cloacal), Imperforate anus, Spinal defects

WAGR (11p13)
Wilms, Aniridia, Genitourinary anomalies, Range of developmental delay

DENYS–DRASH (WT1)
Wilms + DSD (46,XY) + early nephropathy (diffuse mesangial sclerosis)

FRASIER (WT1)
FSGS + 46,XY complete gonadal dysgenesis (gonadoblastoma risk)

POTTER sequence
bilateral renal failure → anhydramnios → lung hypoplasia, facies, limbs

Figure 9. Syndromes and sequences.

ANTENATAL COUNSELLING

- MDT: fetal medicine, paediatric urology/nephrology, neonatology, genetics, psychology
- Oligohydramnios = lung prognosis
- Offer karyotype/microarray, fetal MRI where helpful; plan birth site

LATER LIFE

- CKD follow-up after PUV, severe reflux, dysplasia
- Fertility: MRKH (surrogacy, uterine transplant), Klinefelter (micro-TESE + ICSI), CBAVD (ICSI + partner CFTR test)
- Genetic counselling — CAH, PKD, CF, WT1

★ **PEARL**

Oligohydramnios from 16 weeks = think bilateral renal problem (after excluding membrane rupture and growth restriction).

Müllerian anomaly → scan the kidneys; unilateral renal agenesis in a girl → check the uterus and vagina.

7 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

3–6 / 1000 CAKUT	**~16 wk** Urine = main liquor	**≥ 7 mm** Megacystis 11–14 wk	**~20%** Aneuploidy, 7–15 mm
90–95% CAH = 21-OH	**Days 7–14** Salt-wasting crisis	**30–40%** MRKH + renal	**2006** Chicago DSD consensus

HIGH-YIELD CHECKLIST

- ☐ Three territories: intermediate mesoderm, urogenital sinus, external genitalia.
- ☐ Oligohydramnios from 16 wk → bilateral renal; Potter sequence.
- ☐ Megacystis ≥ 7 mm at 11–14 wk: karyotype; > 15 mm obstruction.
- ☐ PUV: boy, keyhole, bilateral hydronephrosis; PLUTO.
- ☐ PUJ = commonest pathological hydronephrosis; VUR → UTIs.
- ☐ Exstrophy; OEIS; persistent cloaca (girls).
- ☐ Müllerian: U2 septate, U3 bicorporeal (didelphys U3b + C2), U4 unicornuate, U5 MRKH.
- ☐ Chicago DSD: sex-chromosome / 46,XY / 46,XX.
- ☐ CAH: 21-OH, salt wasting, ↑17-OHP, virilised 46,XX.
- ☐ VACTERL, MURCS, OHVIRA, OEIS, WAGR, Denys–Drash, Frasier.

Self-assessment — cover the right-hand column

Question	Answer
Cause of death in Potter sequence?	Pulmonary hypoplasia
Antenatal features of PUV?	Megacystis, keyhole sign, bilateral hydronephrosis, oligohydramnios
Commonest pathological cause of antenatal hydronephrosis?	PUJ obstruction
What is OEIS?	Omphalocele, cloacal Exstrophy, Imperforate anus, Spinal defects

Question	Answer
ESHRE/ESGE class of didelphys?	U3b with C2 (double cervix)
Commonest cause of 46,XX DSD?	CAH (21-hydroxylase deficiency)
Diagnostic test for 21-OH deficiency?	Raised 17-OH-progesterone
CAIS vs Swyer at puberty?	CAIS develops breasts; Swyer does not (no ovarian oestrogen)
Components of MURCS?	Müllerian aplasia, renal aplasia, cervicothoracic somite anomalies
Gene in Denys–Drash and Frasier?	WT1

8 References

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- Grimbizis GF et al. ESHRE/ESGE classification of female genital tract congenital anomalies. 2013.
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