PIPADOR REVISION

MRCOG Part 1 Revision Notes

Congenital Gut Anomalies

EMBRYOLOGY • LECTURE 69

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 69: Congenital Anomalies of the Gut

Embryology • Visual revision edition — integrating Lectures 66–68

COLOUR CODE USED THROUGHOUT THESE NOTES

FOREGUT	MIDGUT	HINDGUT	ABDOMINAL WALL	DIAPHRAGM

LEARNING OBJECTIVES — tick off as you go

- ☐ Place each gut anomaly in its embryological region.
- ☐ Recognise the antenatal ultrasound signs and liquor changes.
- ☐ Know the key features and management of each major anomaly.
- ☐ List the syndromic and chromosomal associations.
- ☐ Act on the newborn red flags and plan antenatal counselling.

1 The Map

IN ONE LINE: *Gut anomalies are common and often go with other anomalies — so a gut finding on scan means a full anomaly scan, karyotype discussion and fetal echo.*

Congenital gut anomalies mapped to their region

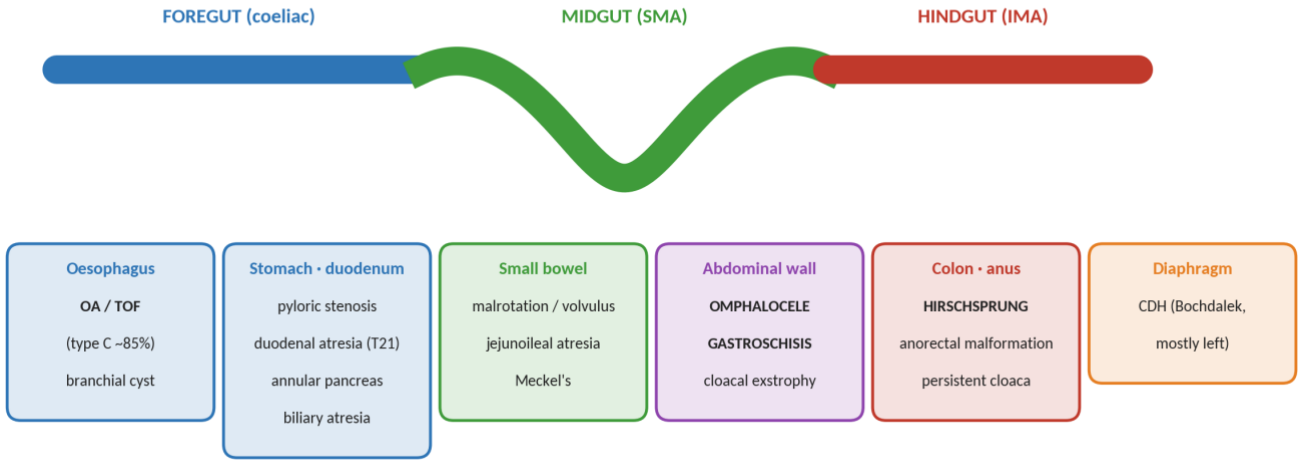


Figure 1. Anomalies by region.

2 Antenatal Detection

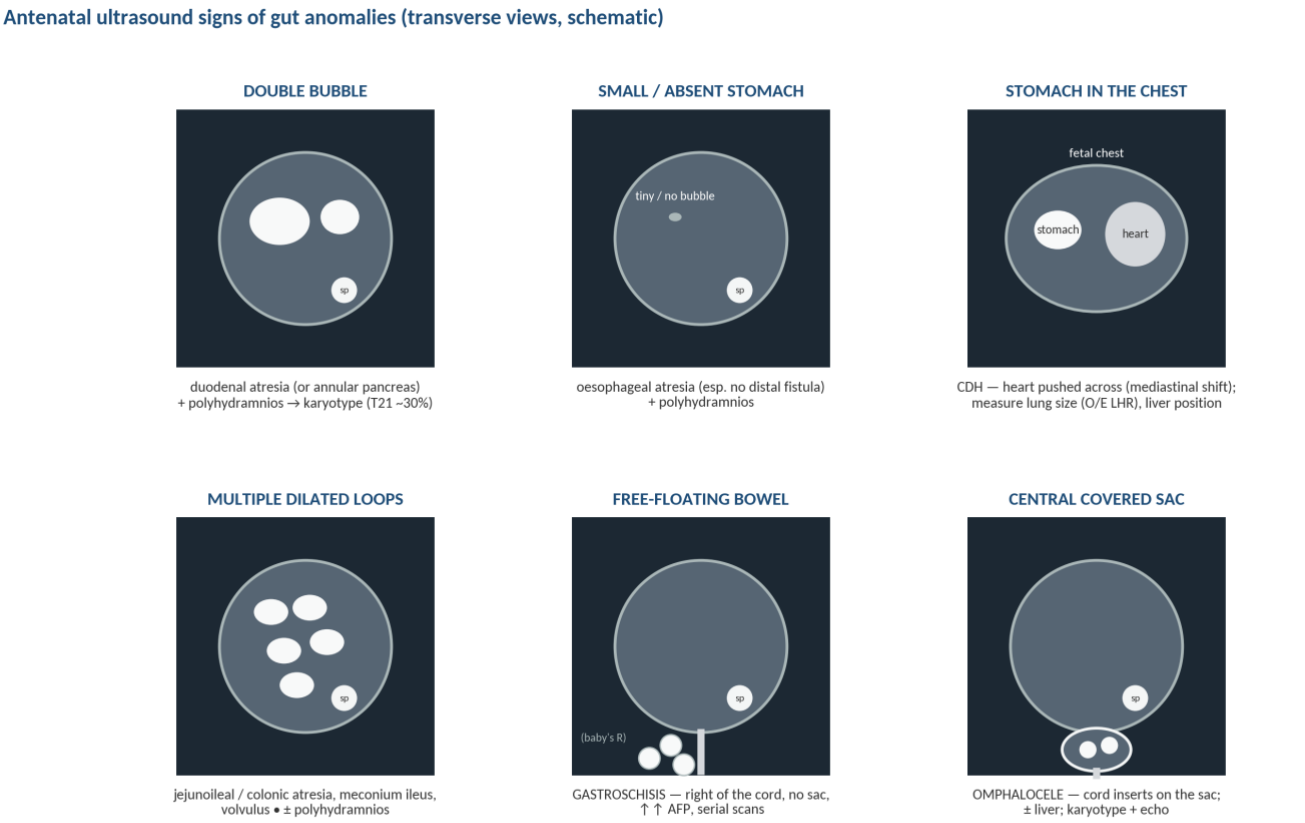


Figure 2. The key scan signs.

Clue	Meaning
Polyhydramnios	Upper-GI obstruction (OA/TOF, duodenal atresia) or poor swallowing (anencephaly, neuromuscular); also maternal diabetes
Oligohydramnios	Rarely gut — think renal anomaly, PPROM, placental FGR
Maternal AFP	Raised in open NTD and abdominal wall defects — markedly in gastroschisis
Echogenic bowel	CF, CMV (and other infections), T21, intra-amniotic bleeding, FGR

3 Foregut Anomalies

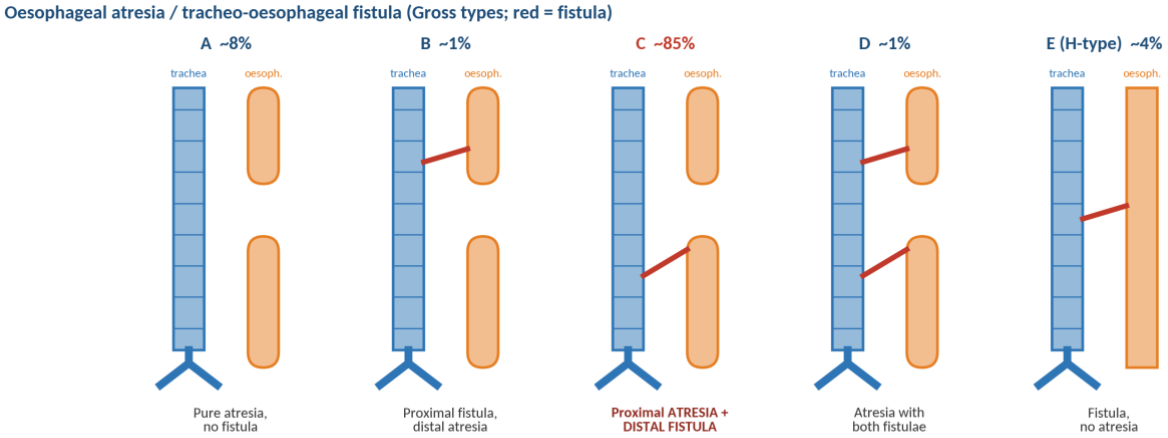


Figure 3. OA/TOF types (from Lecture 49).

Condition	Key points
OA / TOF	Type C ~85%, pure OA ~8% (no gas in the stomach), H-type ~4% (later — recurrent chest infections, choking). VACTERL, CHARGE. Primary anastomosis where possible
Pyloric stenosis	3–6 weeks, boys; projectile non-bilious vomiting; olive; hypochloraemic hypokalaemic alkalosis; US muscle > 3 mm, channel > 16 mm; correct fluids → Ramstedt pyloromyotomy
Duodenal atresia	Failed recanalisation; double bubble + polyhydramnios; ~30% T21; bilious vomiting day 1; duodenoduodenostomy
Annular pancreas	Bifid ventral bud encircles D2 → same picture as duodenal atresia
Biliary atresia	Conjugated jaundice, pale stools; Kasai before ~60 days; many later need a transplant
Choledochal cyst	Todani I (fusiform CBD) commonest; malignancy risk → excise

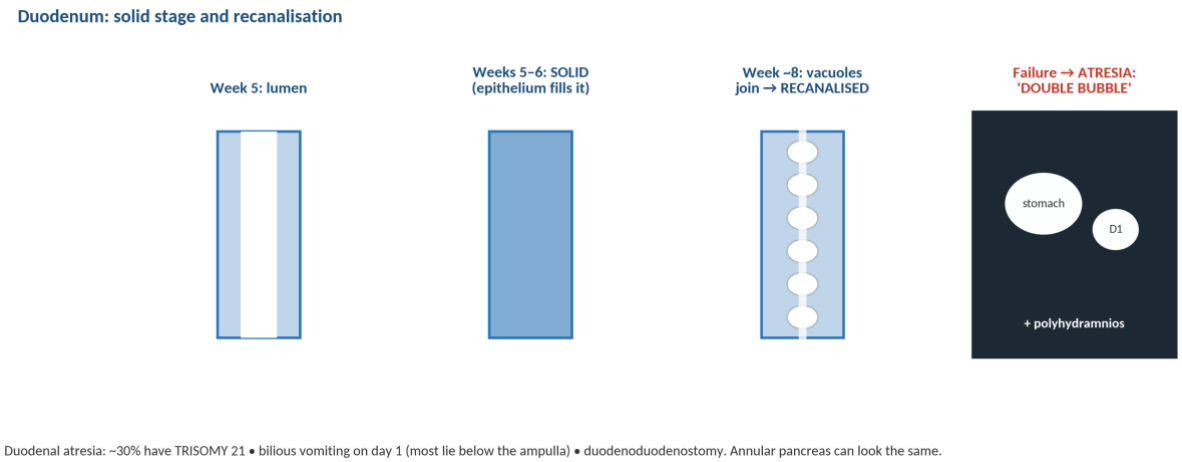


Figure 4. Duodenal recanalisation (from Lecture 66).

4 Midgut and Abdominal Wall

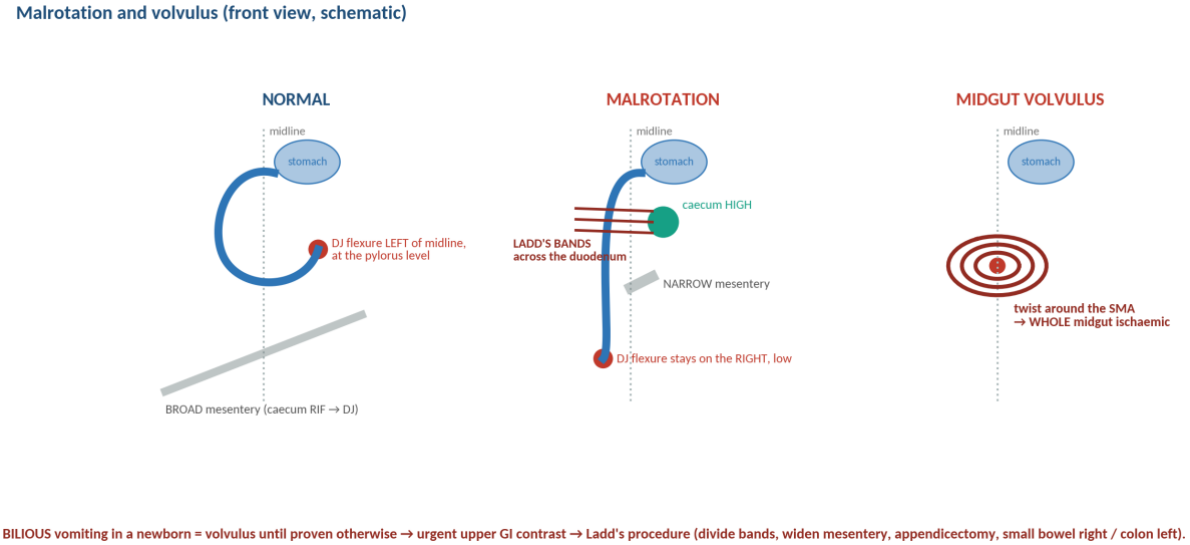


Figure 5. Malrotation and volvulus (from Lecture 67).

Abdominal wall defects compared

	OMPHALOCELE	GASTROSCHISIS
Sac	YES — amnion + peritoneum	NO — bowel floats free
Site	CENTRAL, through the umbilicus	to the RIGHT of an intact cord
Contents	bowel ± LIVER	bowel (thickened, matted)
Other anomalies	COMMON — cardiac, NTD, BWS; chromosomal ~30% (T13, T18, T21)	UNCOMMON — bowel atresia ~10–15%
Maternal AFP	raised	MARKEDLY raised
Who	older mothers	YOUNG mothers, smoking, deprivation
Plan	karyotype/microarray, echo, MDT	serial scans (bowel, growth), deliver ~37 wk in a surgical centre

Don't diagnose omphalocele before ~12 weeks' gestation — the midgut is normally in the cord until then. Vaginal birth is fine for both unless other reasons.

Figure 6. Omphalocele vs gastroschisis (from Lecture 67).

- **Jejunoileal atresia**: vascular accident; **apple-peel (type IIIb)** severe. **Meckel's**: rule of 2s, painless bleeding, Tc-99m scan.

5 Hindgut and Diaphragm

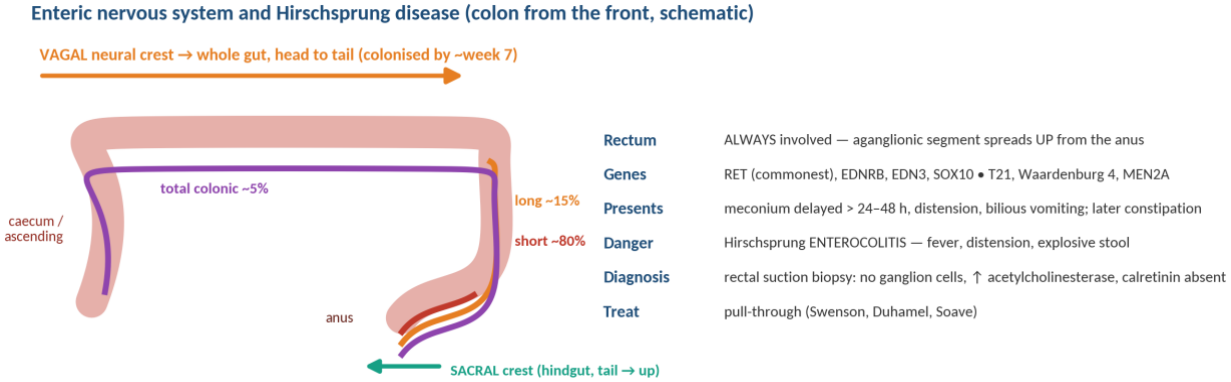


Figure 7. Hirschsprung (from Lecture 68).

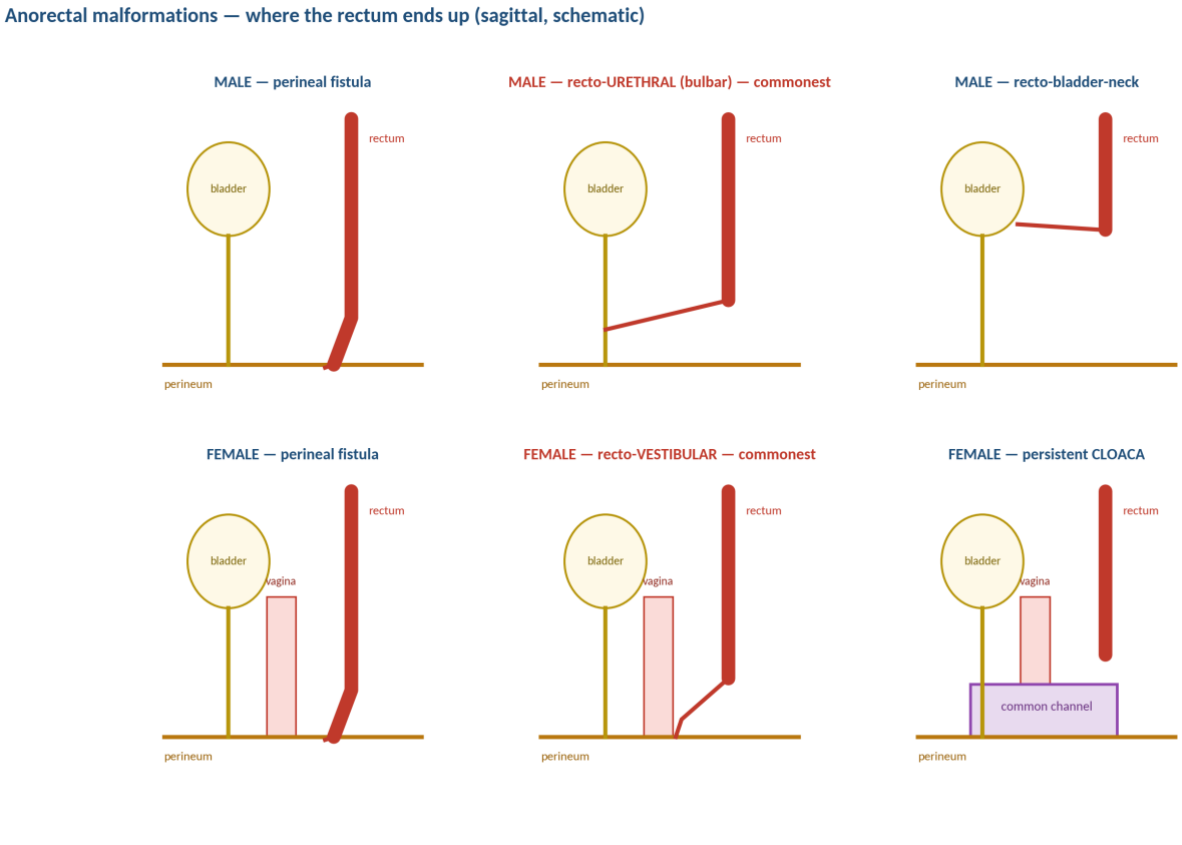


Figure 8. Anorectal malformations (from Lecture 68).

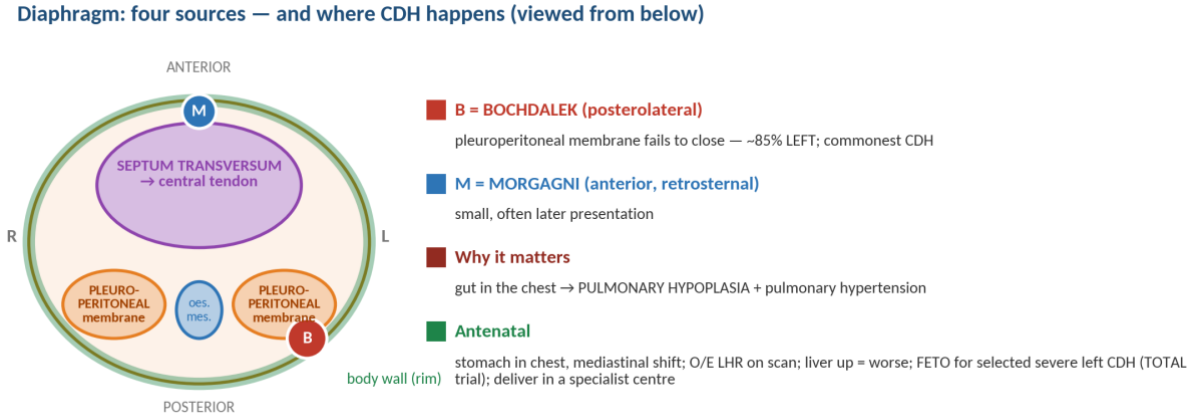


Figure 9. CDH (from Lecture 66).

- **CDH at birth**: intubate — **no bag-and-mask** (inflates the gut in the chest), NG decompression, gentle ventilation, delayed repair ± ECMO. **FETO** improved survival in severe left CDH (TOTAL trial); in moderate cases the gain was smaller and preterm birth rose.
- **Cloacal exstrophy (OEIS)**: most severe wall defect; survival now much better with specialist care.

6 Associations and Counselling

Syndrome	Gut features
Trisomy 21	Duodenal atresia, annular pancreas, Hirschsprung, ARM without fistula
Trisomy 18 / 13	Omphalocele, OA, malrotation
VACTERL	OA/TOF, anal atresia (+ vertebral, cardiac, renal, limb)

Syndrome	Gut features
Beckwith–Wiedemann	Omphalocele, macroglossia, organomegaly, neonatal hypoglycaemia
CHARGE (CHD7)	Choanal atresia, OA/TOF
Cystic fibrosis	Meconium ileus, echogenic bowel
Currarino (MNX1)	ARM + sacral defect + presacral mass

ANTENATAL COUNSELLING

- MDT: fetal medicine, paediatric surgery, neonatology, genetics
- Karyotype/microarray for omphalocele, duodenal atresia, multiple anomalies
- **Fetal echo** (heart defects are common)
- Deliver where there is **paediatric surgery + NICU**

POLYHYDRAMNIOS IN PRACTICE

- Check glucose (GTT), detailed scan for GI/CNS/neuromuscular causes
- Risks: preterm labour, malpresentation, cord prolapse, PPH
- Tell the neonatal team — early NG tube at birth if OA suspected

7 Newborn Red Flags



Figure 10. Think and do.

- **Prolonged jaundice** (NICE): jaundice beyond **14 days (term)** or **21 days (preterm)** → check **conjugated bilirubin**; pale stools or dark urine → urgent referral.

> ★ **PEARL**
> **Bilious vomiting = volvulus until proven otherwise. Double bubble = duodenal atresia (T21). Pale stools + prolonged jaundice = biliary atresia (Kasai by ~60 days).**

8 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

~85%	**~8% / ~4%**	**~30%**	**~30%**
OA/TOF type C	Pure OA / H-type	Duodenal atresia + T21	Omphalocele chromosomal

3–6 wk	> 3 / > 16 mm	~60 days	14 / 21 d
Pyloric stenosis	Pylorus muscle / channel	Kasai	Prolonged jaundice

HIGH-YIELD CHECKLIST

- ☐ Polyhydramnios → upper GI / swallowing; oligohydramnios → kidneys.
- ☐ Double bubble → duodenal atresia → karyotype.
- ☐ Small/absent stomach → OA; stomach in chest → CDH.
- ☐ Gastroschisis: right, uncovered, isolated, ↑↑ AFP. Omphalocele: covered, central, syndromic.
- ☐ Bilious vomiting → urgent upper GI contrast.
- ☐ No meconium > 24–48 h → Hirschsprung, ARM, meconium ileus (CF).
- ☐ Pale stools + prolonged jaundice → conjugated bilirubin → Kasai.
- ☐ Pyloric stenosis: alkalosis; fluids first.
- ☐ CDH: no bag-mask; FETO for severe left.
- ☐ Syndromes: T21, T18/13, VACTERL, BWS, CHARGE, CF, Currarino.

Self-assessment — cover the right-hand column

Question	Answer
Scan sign of duodenal atresia?	Double bubble (+ polyhydramnios)
Chromosomal association of duodenal atresia?	Trisomy 21 (~30%)
Which wall defect has markedly raised AFP?	Gastroschisis
Commonest OA/TOF type?	Proximal atresia + distal fistula (~85%)
H-type TOF presents how?	Later, with recurrent chest infections / choking
Acid–base in pyloric stenosis?	Hypochloraemic hypokalaemic metabolic alkalosis
First test for neonatal bilious vomiting?	Urgent upper GI contrast study
Why avoid bag-mask ventilation in CDH?	It inflates the bowel in the chest
When to check conjugated bilirubin?	Jaundice beyond 14 days (term) / 21 days (preterm)
Syndrome with omphalocele + macroglossia?	Beckwith–Wiedemann

9 References

- Sadler TW. Langman's Medical Embryology. 14th ed. 2019.
- NICE CG98. Jaundice in newborn babies under 28 days. 2010 (updated 2023).
- Deprest JA et al. TOTAL trials — severe and moderate left CDH. N Engl J Med 2021.
- NHS Fetal Anomaly Screening Programme Handbook (current edition).