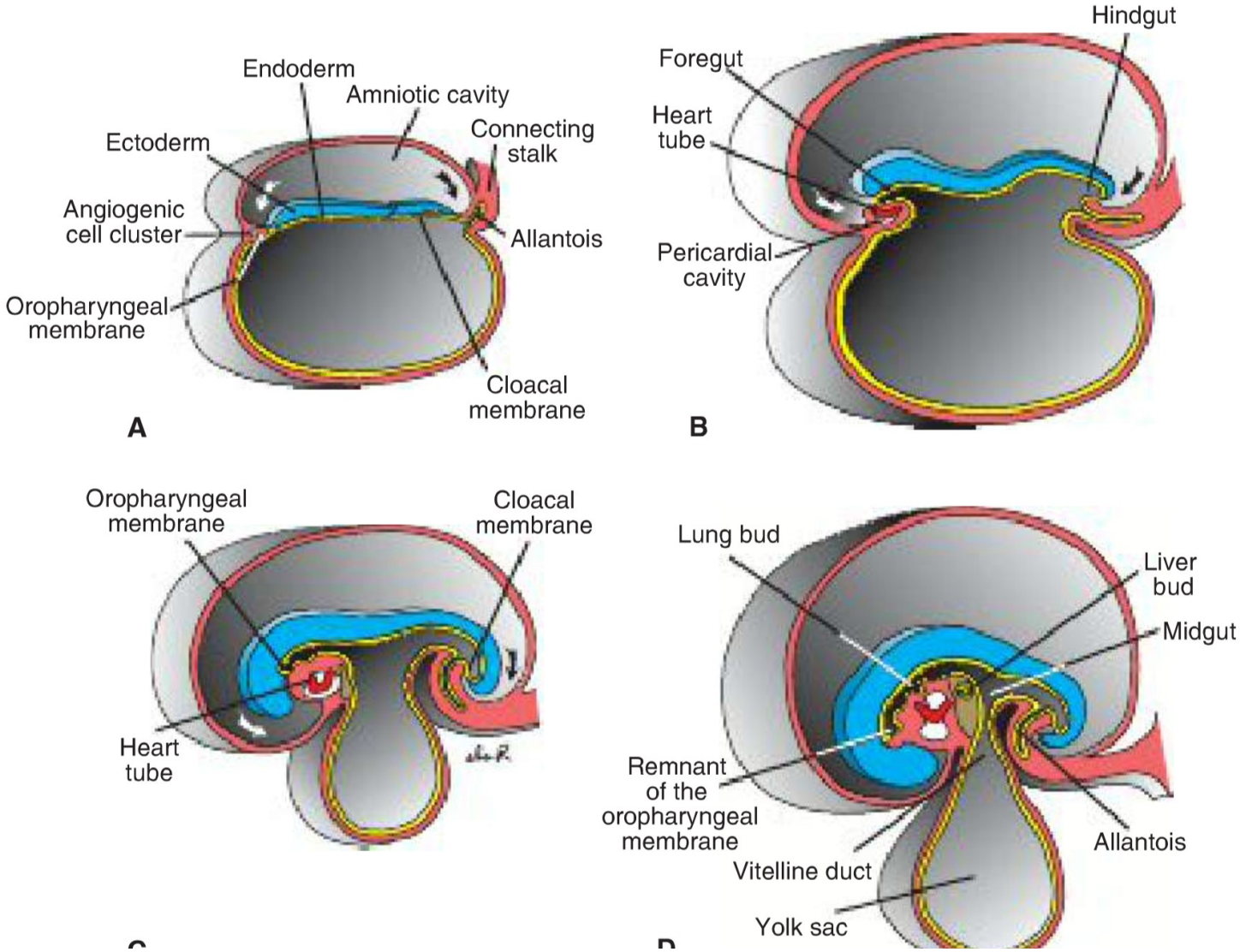
Development and teratology of digestive system.

Development of facial and cervical region, face clefts.

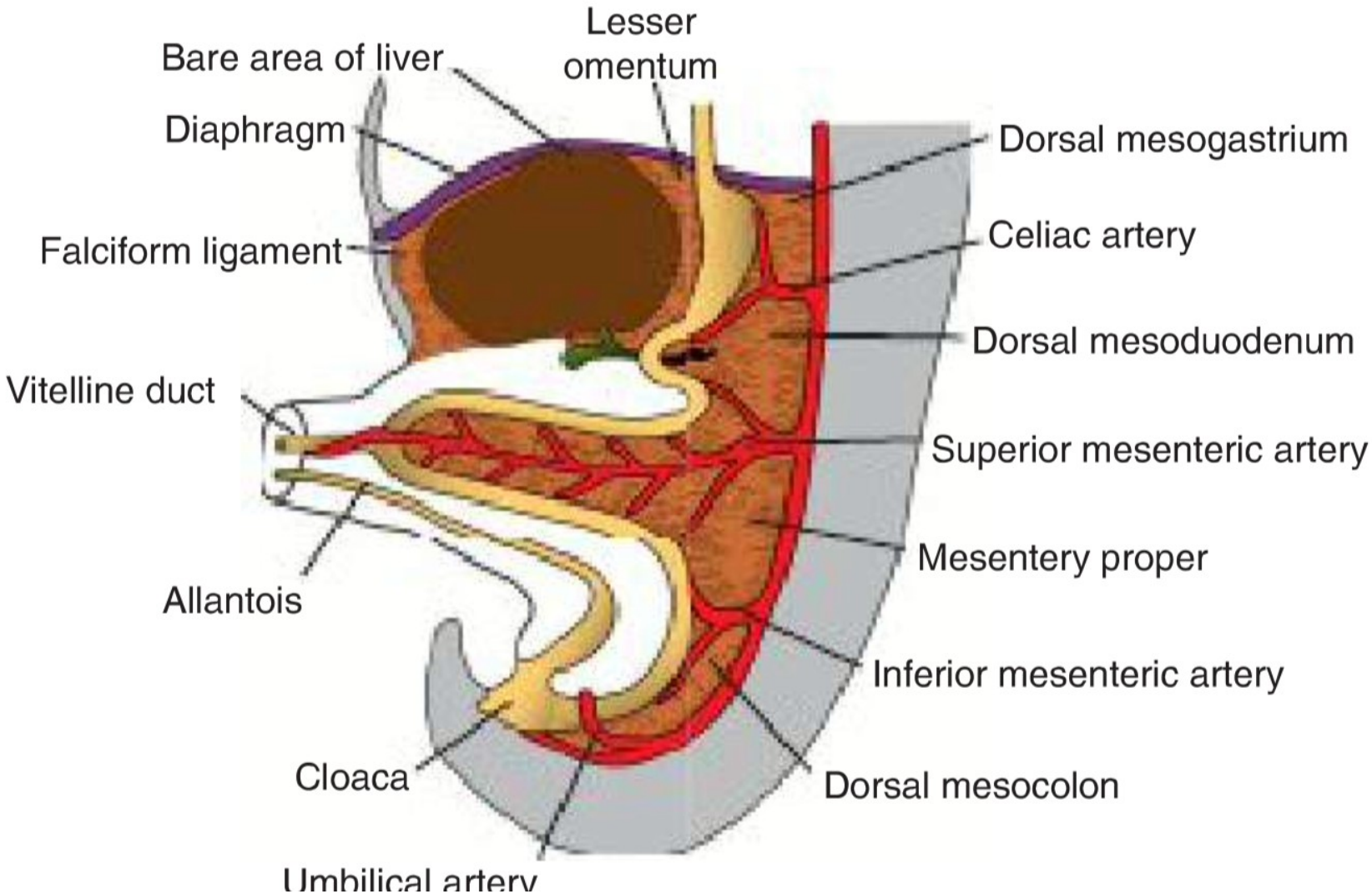
Anna Mac Gillavry

14.03.2022

- Primitive gut formation results from the lateral folding of the embrio
- Foregut, midgut and hindgut
- (Yolk sac, allantois)



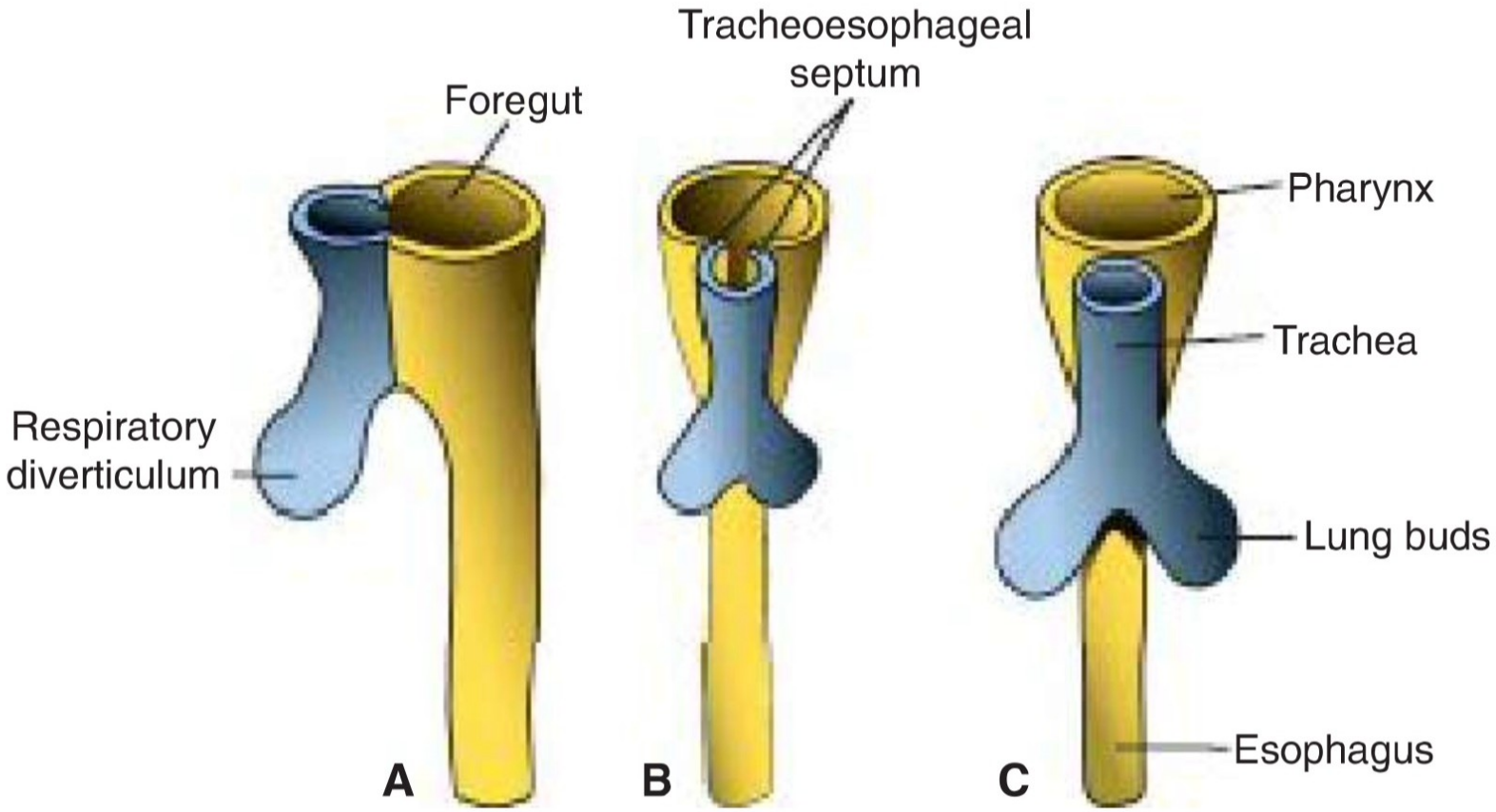
T.W. Sadler, Langman´s medical embryology, 14th edition



T.W. Sadler, Langman´s medical embryology, 14th edition

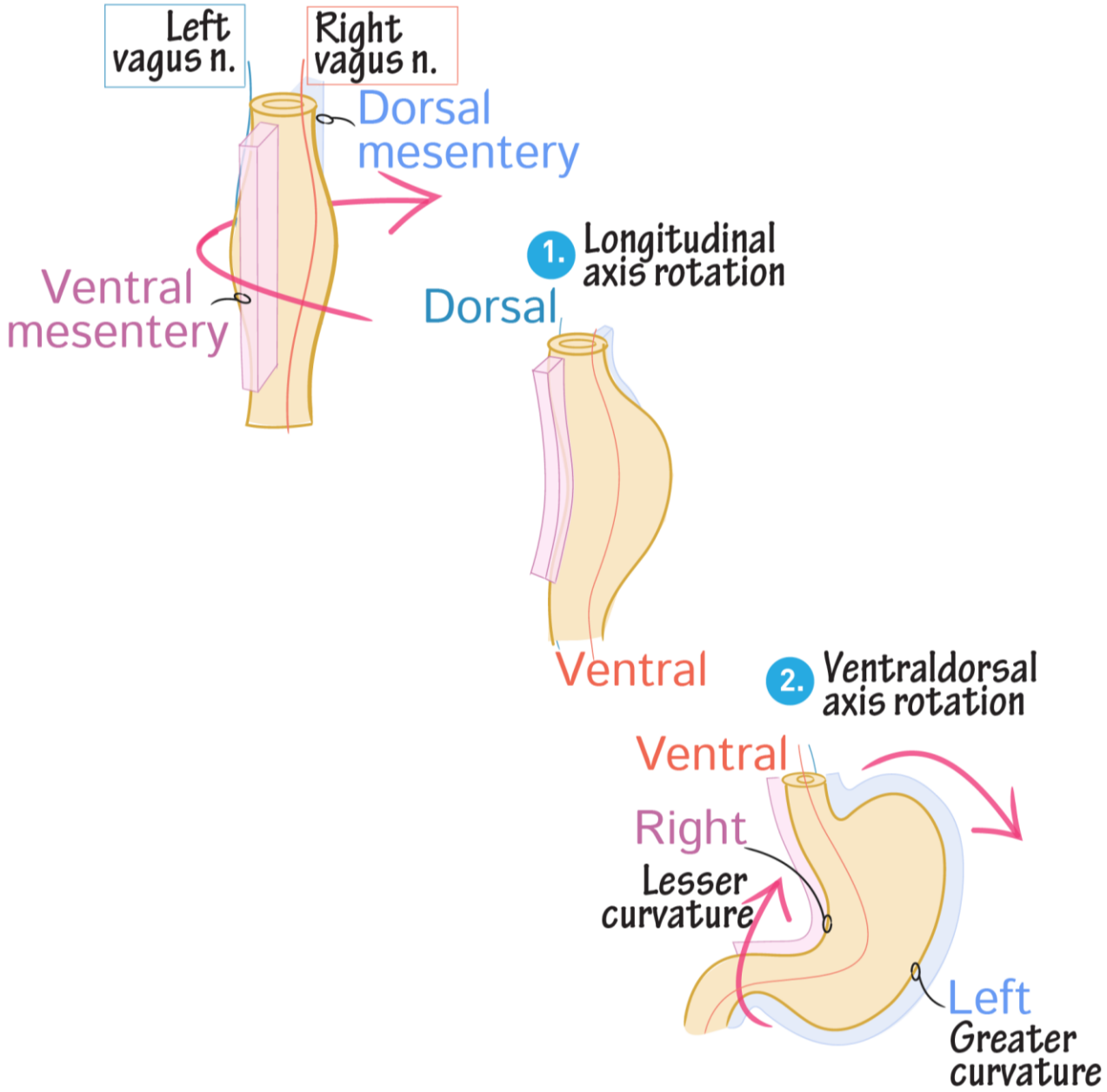
Esophagus

4 weeks

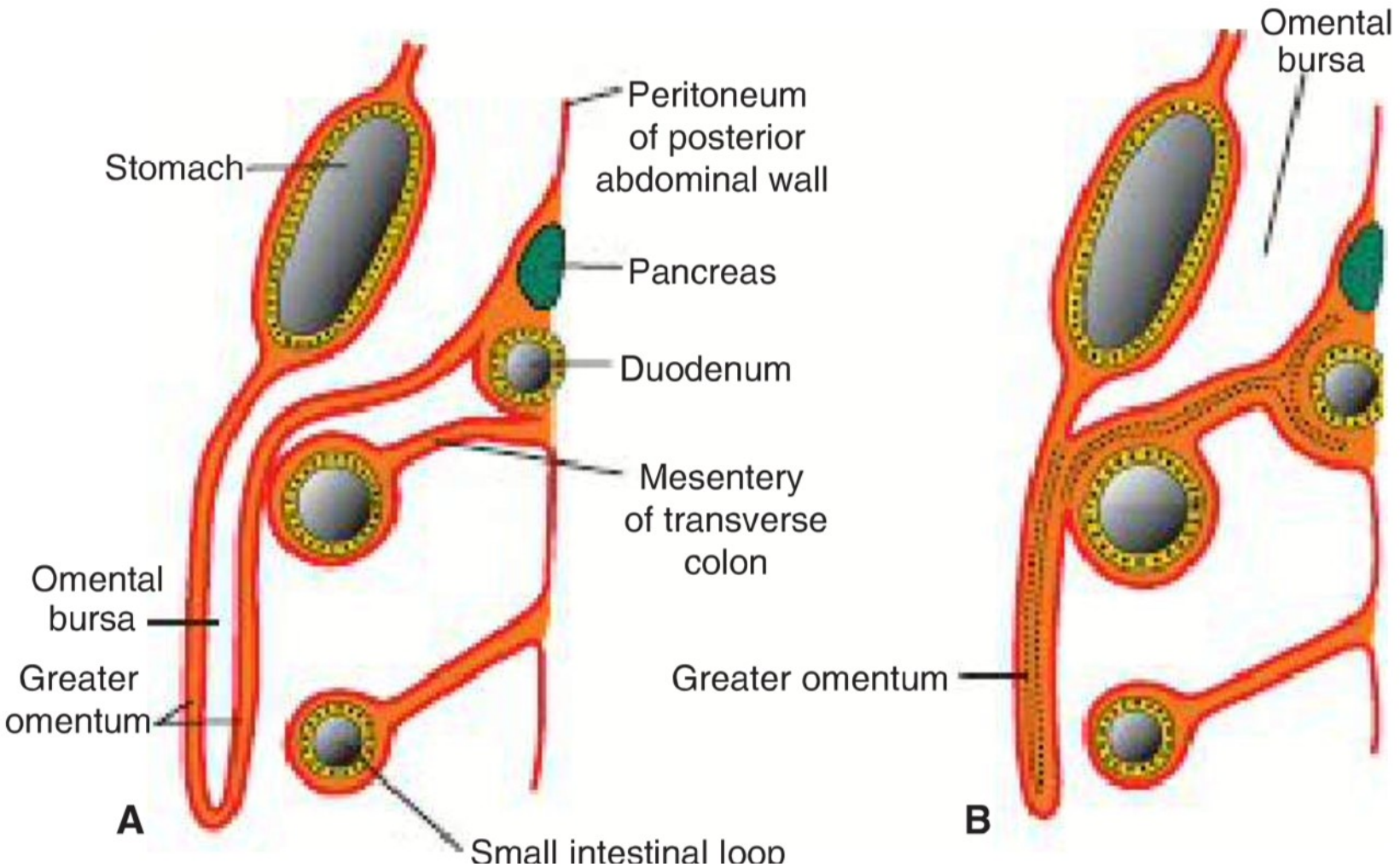


T.W. Sadler, Langman´s medical embryology, 14th edition

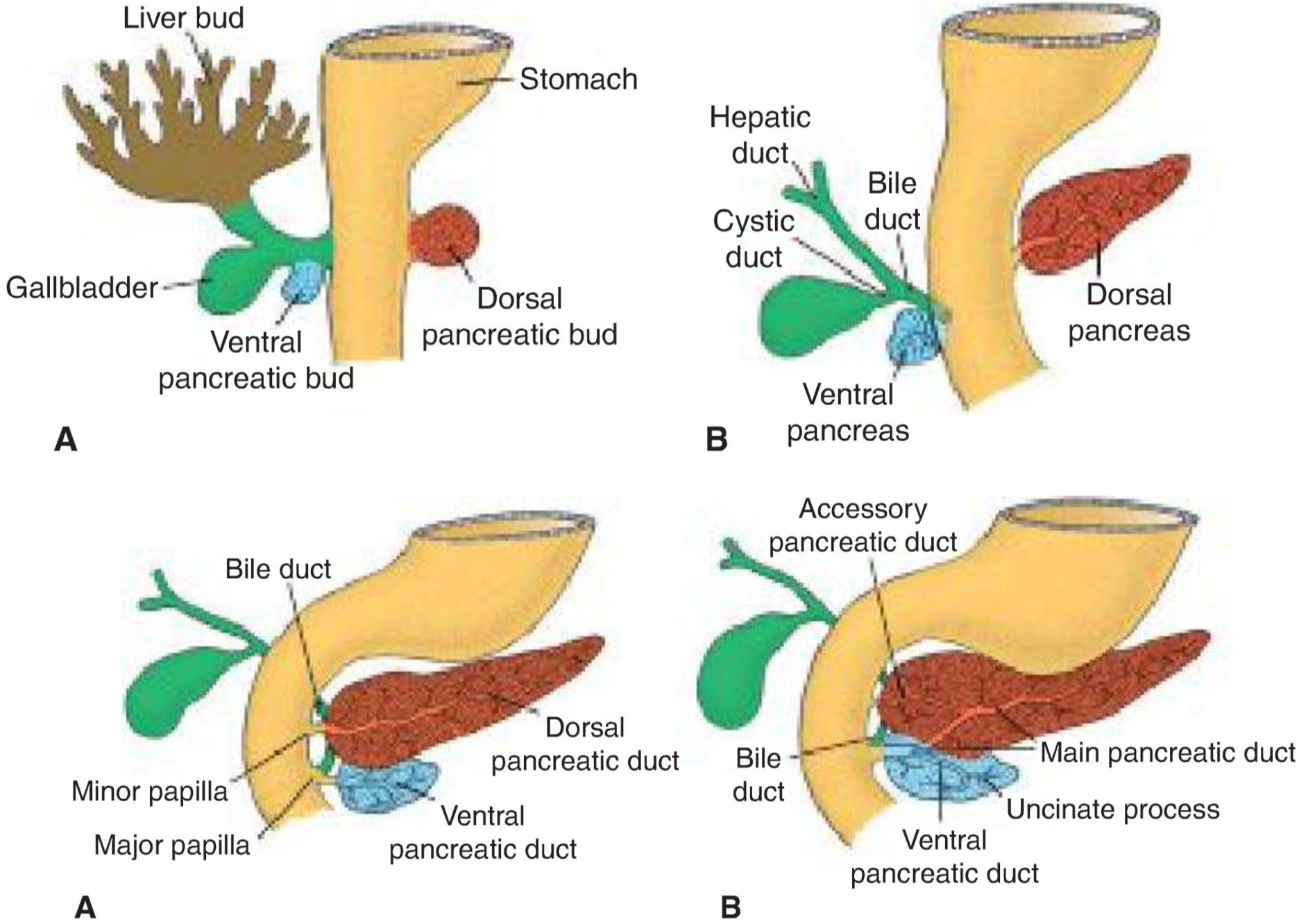
Stomach



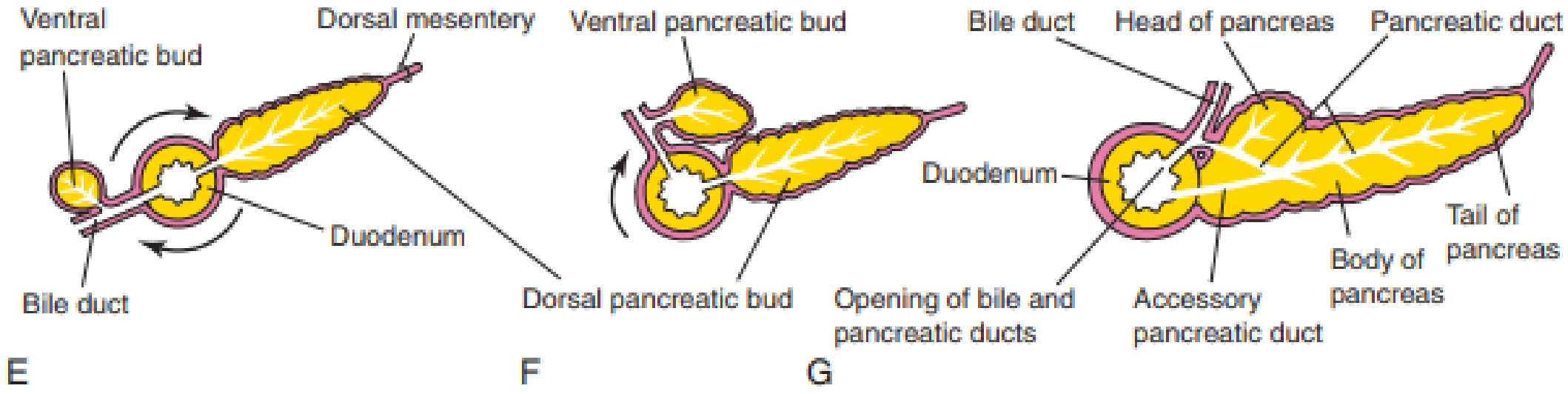
https://www.drawittoknowit.com/course/embryology/glossary/developmental-process/rotation-of-the-stomach



T.W. Sadler, Langman´s medical embryology, 14th edition



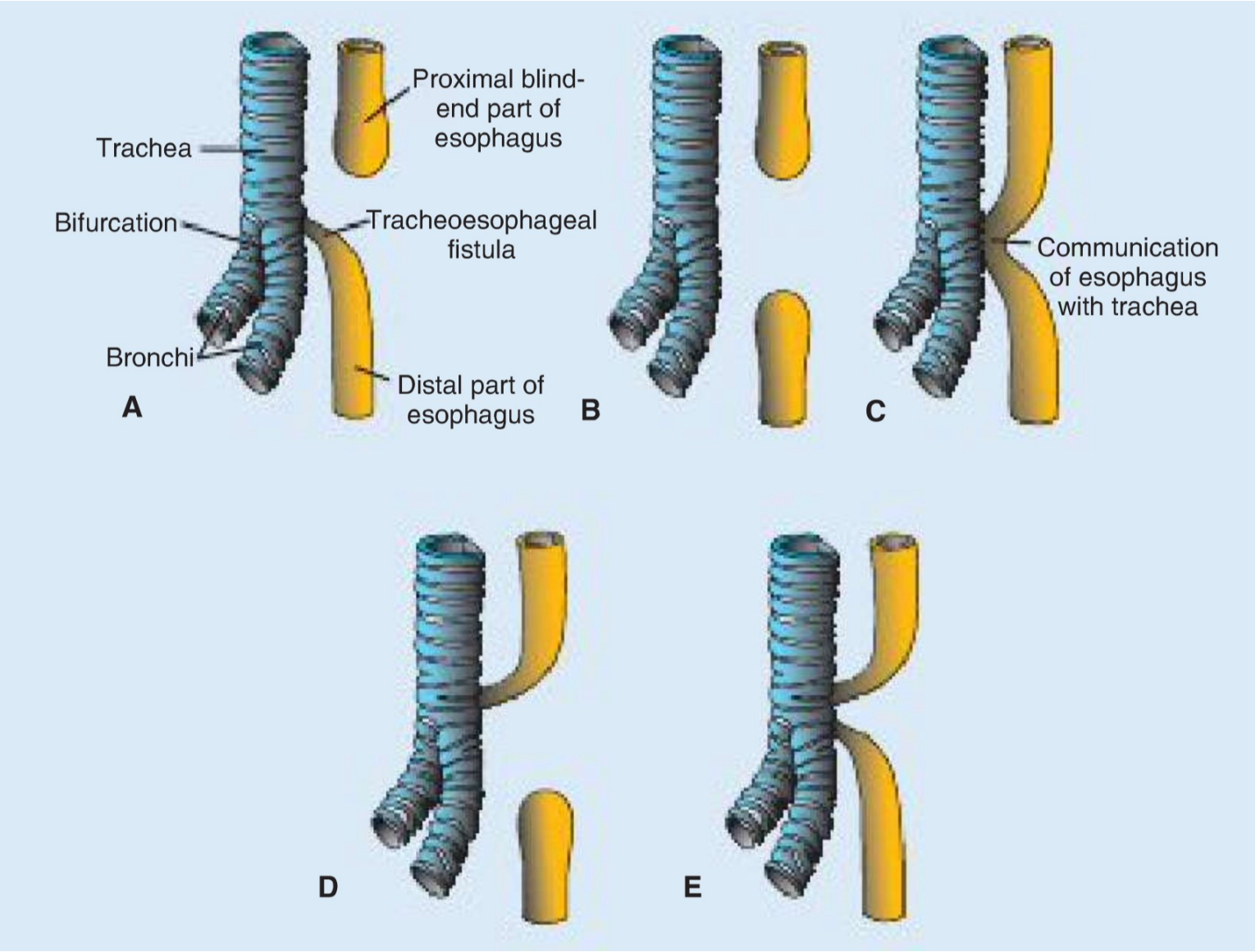
T.W. Sadler, Langman´s medical embryology, 14th edition



K. Moor, Before we are born, 10th edition

Esophagus:

- esophageal atresia and/or tracheoesophageal fistula
- esophageal stenosis
- congenital hiatal hernia



T.W. Sadler, Langman´s medical embryology, 14th edition

Stomach:

Pyloric stenosis (1 in 150 males, 1 in 750 females) – developes during fetal life, however, can develop as a result of postnatal exposure

Liver - birth deffects are rare:

Accessory hepatic ducts

Gallbladder duplication

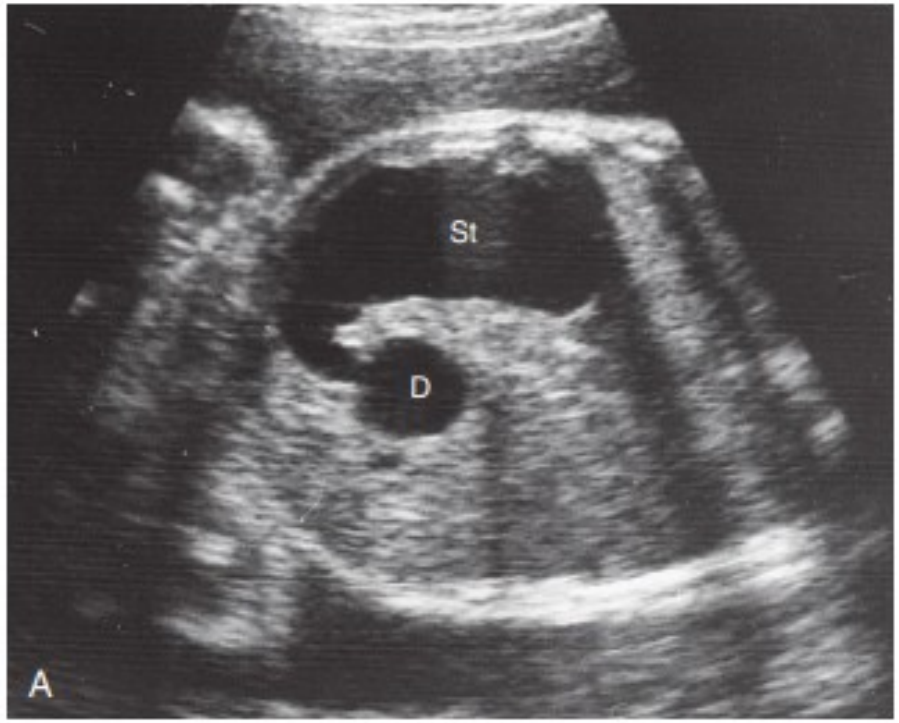
Extrahepatic biliary atresia (1/15000)

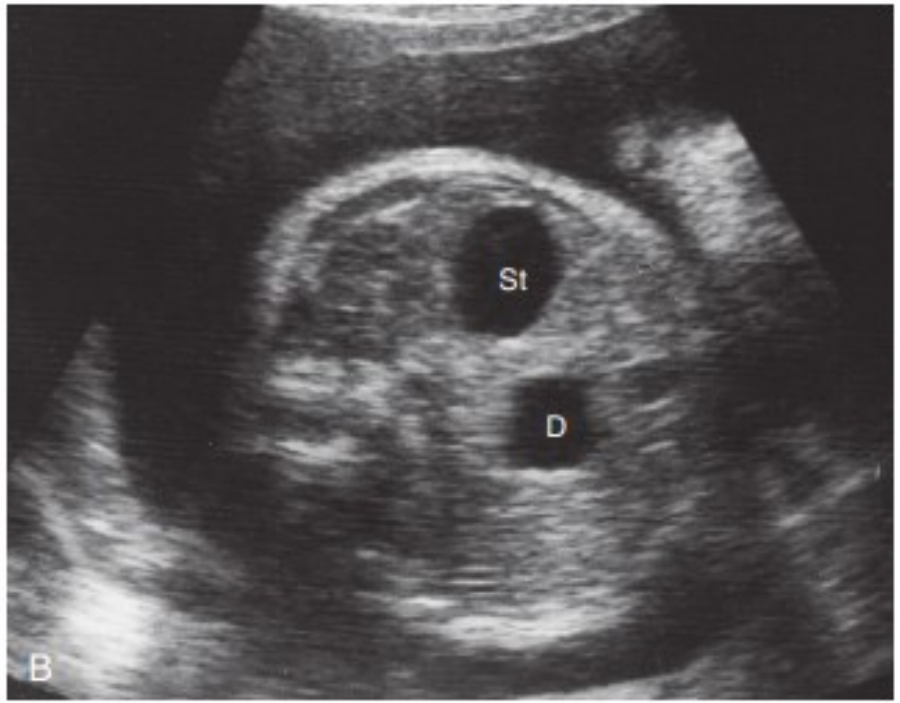
Intrahepatic biliary duct atresia/hypoplasia (1/100000)

Pancreas:

Annular pankreas

Accessory spleens – in 10 % of population





Duodenum:
Duodenal stenosis/atresia – results from incomplete recanalization

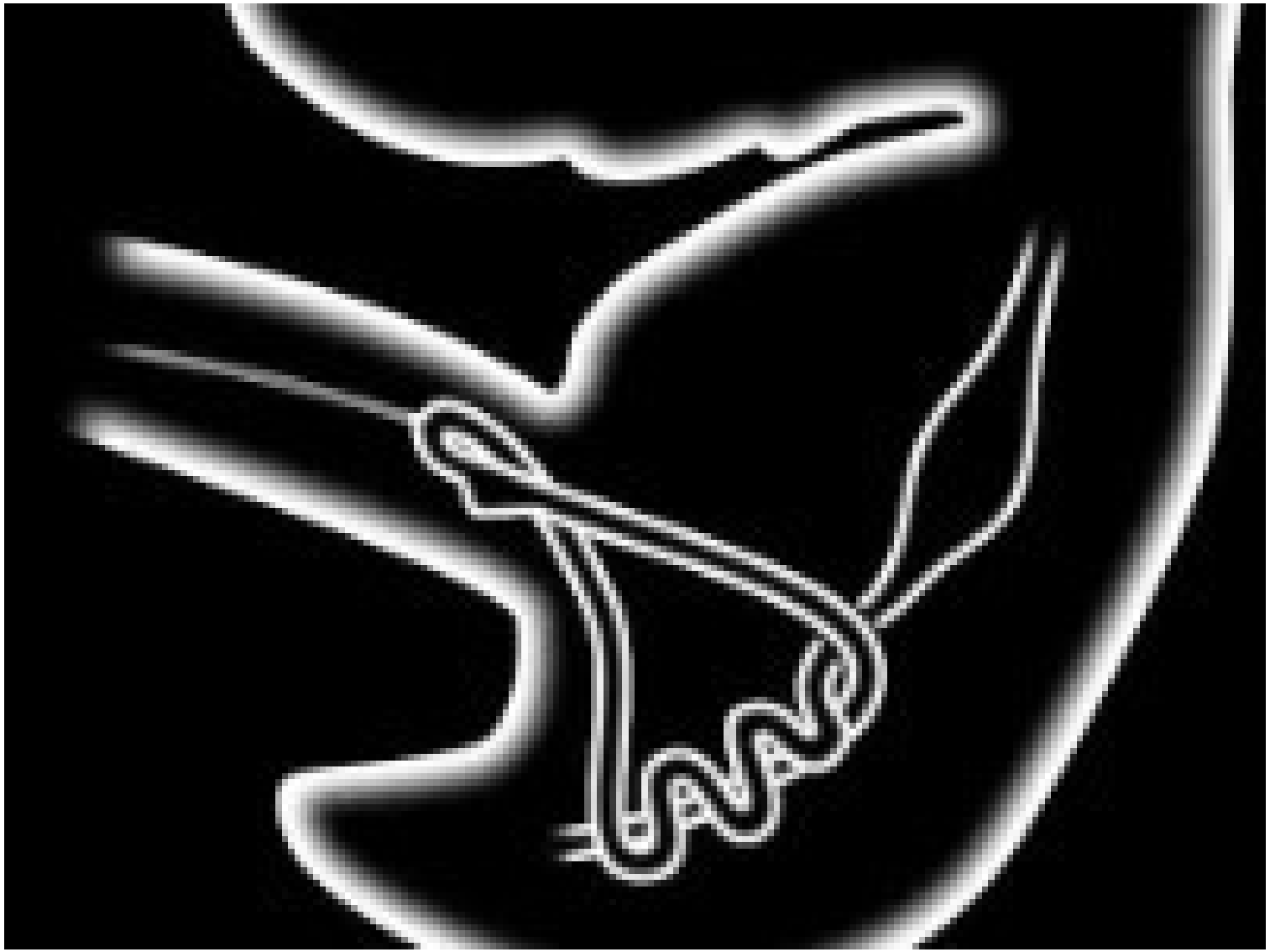
Polyhydramnios

„Doble-Bubble“

K. Moor, Before we are born, 10th edition

Midgut development. Physiological herniation.

6th – 10th week



https://youtu.be/AscKR_cQExY

Body wall defects

Gastroschisis (3,5/10000) – most common in infants from thin women under 20; usually not associated with chromosomal abnormalities and other severe deffects, thus the mortality rate is low (unless associated with volvulus)

X

Omphalocele (2,5/10000) – up to 25 % mortality rate

Vitelline duct abnormalities

Meckel or ileal diverticulum – in 2 to 4 % of people, 3-5 times more prevalent in males (inflamation symptoms mimic those of appendicites)

Enterocystoma or vitelline cyst

Umbilical or vitelline fistula

Gut rotation defects

Left-sided colon – colon and cecum are the first to return from the umbilical cord cavity as the result of only 90° rotation

Reversed rotation of the intestinal loop

Duplications of intestinal loops and cysts

Gut atresias and stenoses

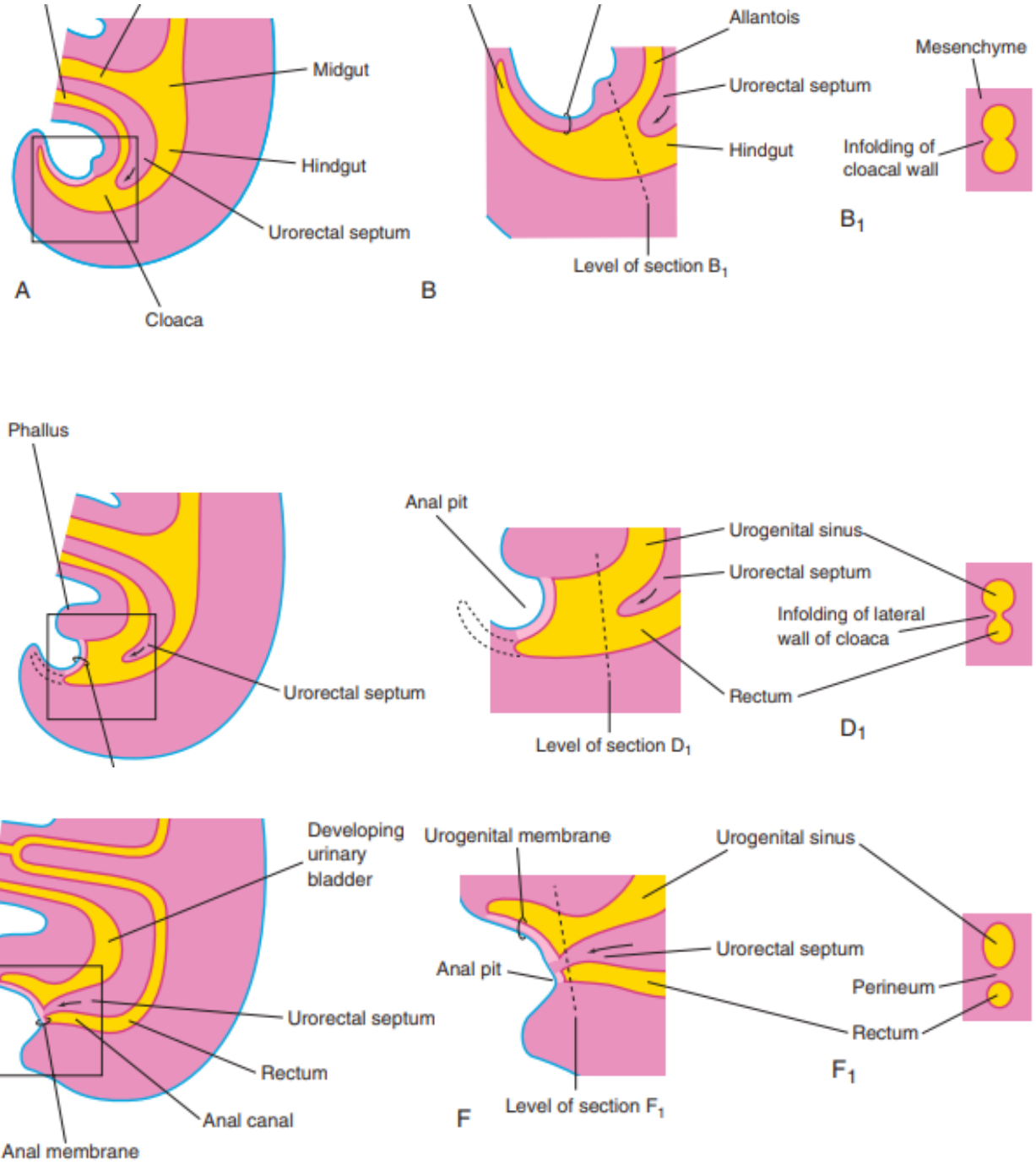
- Most occur in duodenum, fewest in the colon, equal number in jejunum and ileum; in 50 % of cases a region of bowel is missing completely, in 20 % cases the fibrous cord is present; stenoses represent only 5 % of cases

Apple peel atresia - 10 % of atresias: in the proximal jejunum, intestine is short, portion distal to the lesion coiled around remanant of mesenteries

Hindgut

Hindgut derivatives:

- Left third of the transverse colon, descending colon, sigmoid colon, rectum, superior part of the anal canal
- The epithelium of the urinary bladder and most of the urethra!!!



K. Moor, Before we are born, 10th edition

Congenital megacolon

(Hirschsprung disease) – 1/5000, males are affected 4 times more often than females.

Imperforate anus - 1/5000 more common in males than females

Anorectal birth defects

- High vs. Low (rectum end superior or inferior to the puborectalis muscle respectively)

Low: anal agenesis, with or without fistula

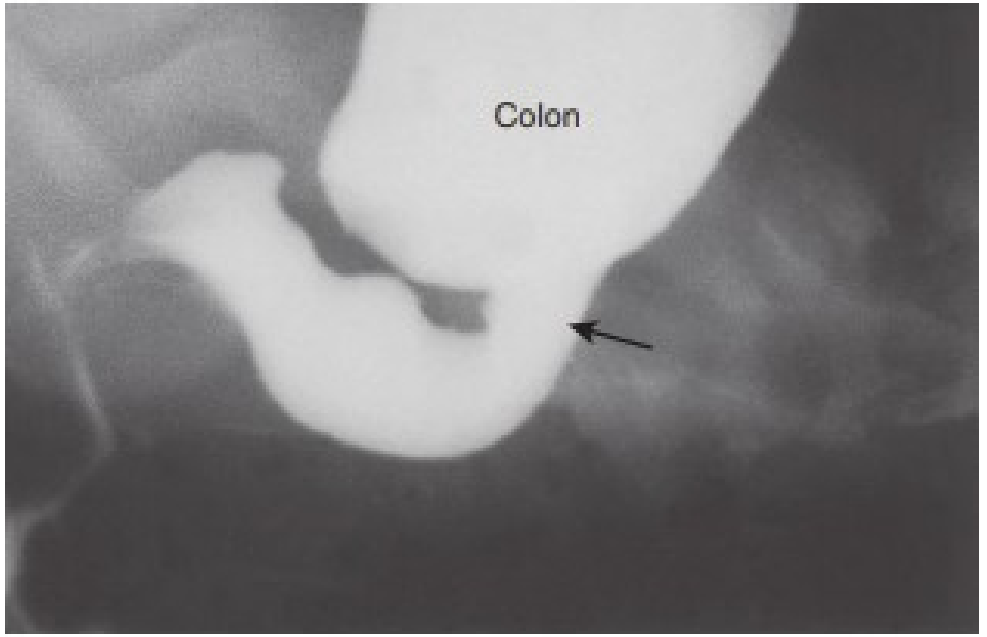
anal stenosis

membranous atresia of anus

High: anorectal agenesis, with or without

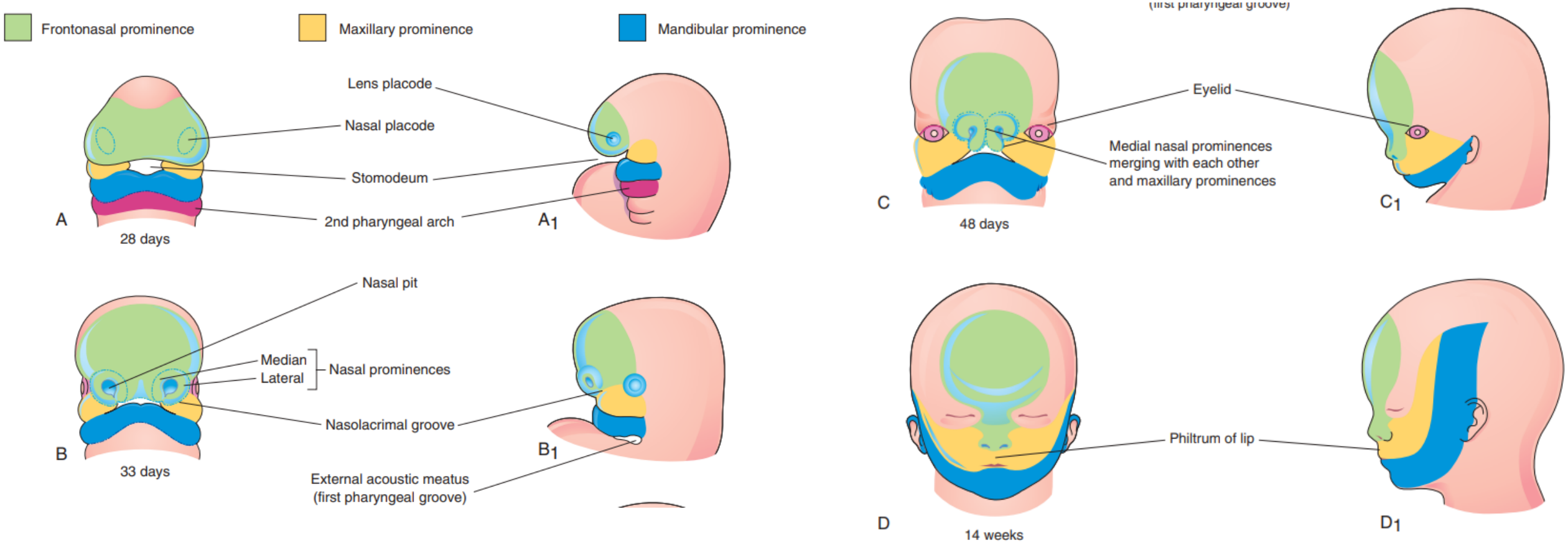
fistula (2/3 of anorectal defects)

rectal atresia



K. Moor, Before we are born, 10th edition

Development of the face



K. Moor, Before we are born, 9th edition

Anterior cleft deformities

Lateral cleft lip (1/700, 65 % - male infants)

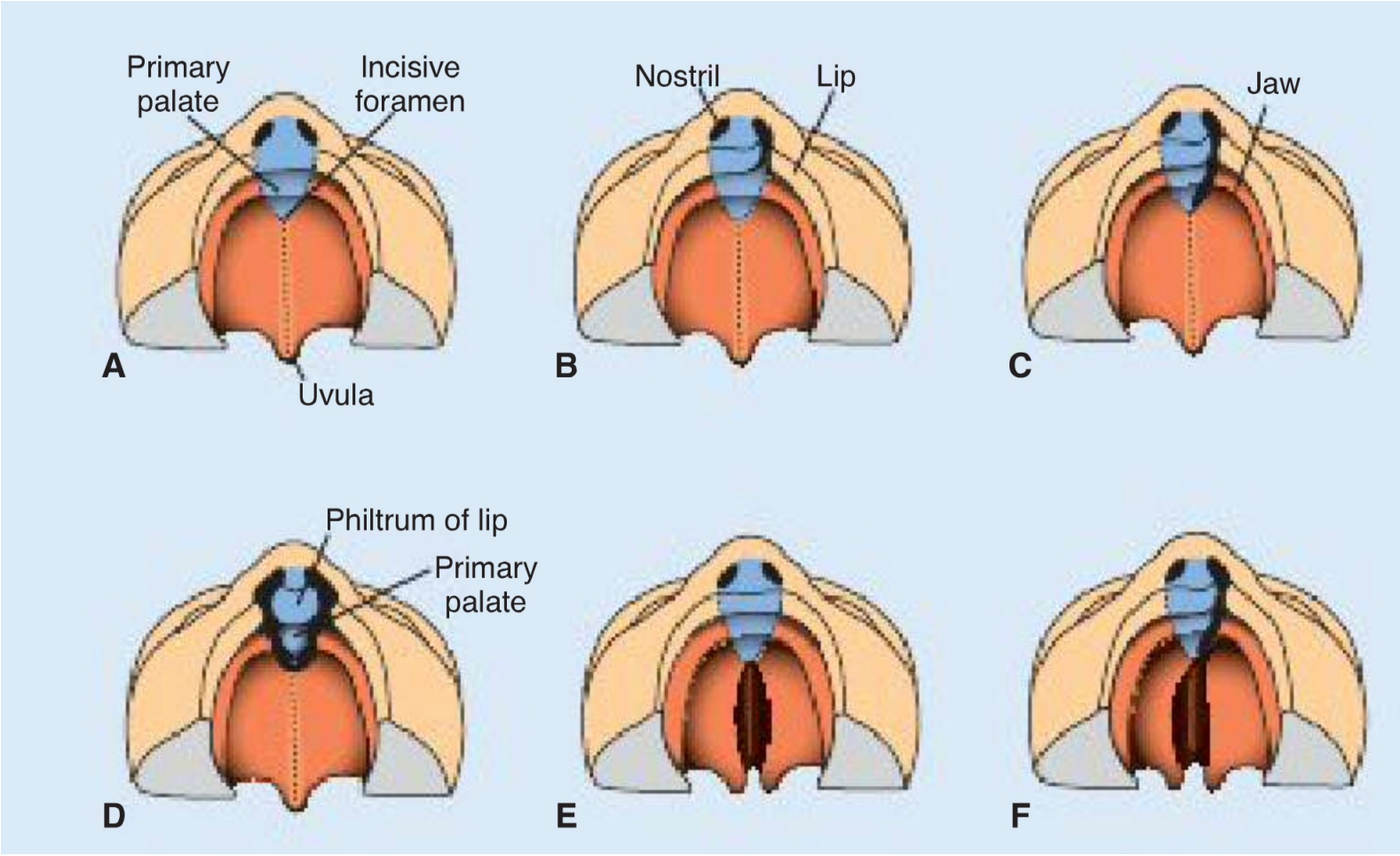
Cleft upper jaw

Cleft between the primary and the secondary palates

Posterior cleft deformities

Cleft secondary palate (1/1500, 55 % female infants)

Cleft uvula



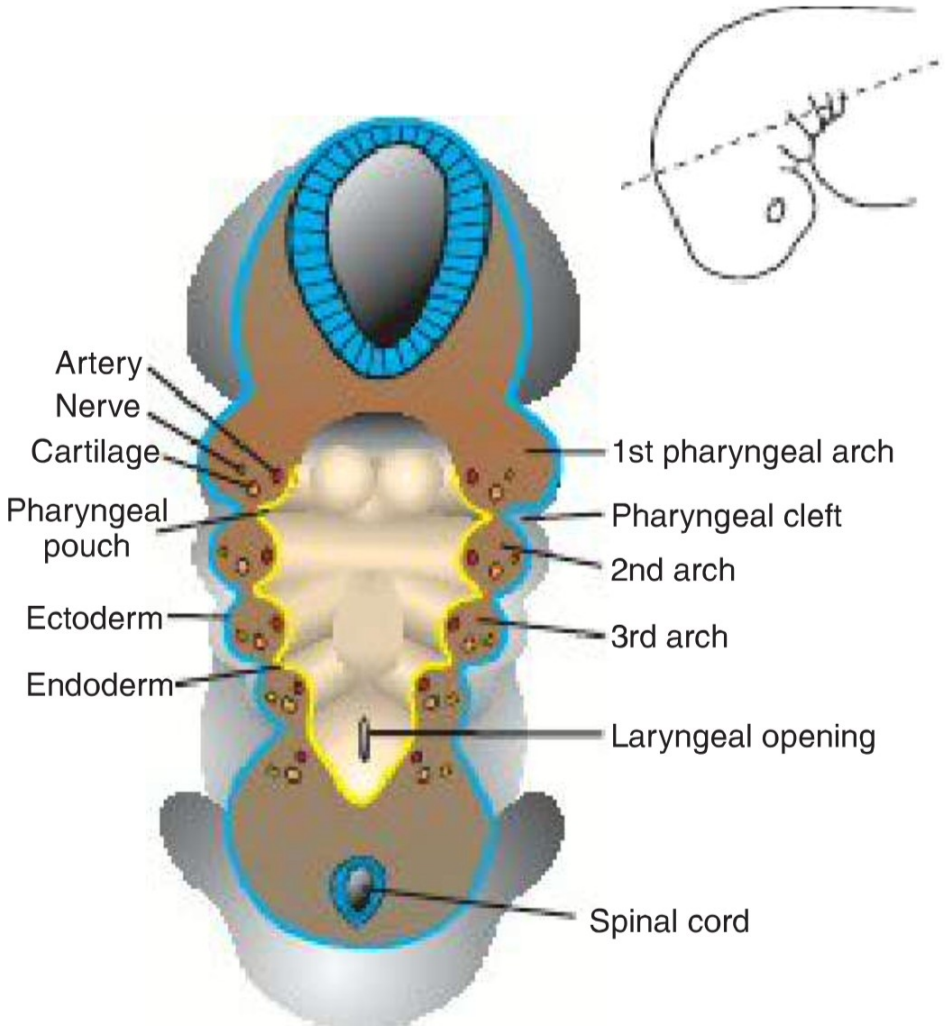
T.W. Sadler, Langman´s medical embryology, 14th edition

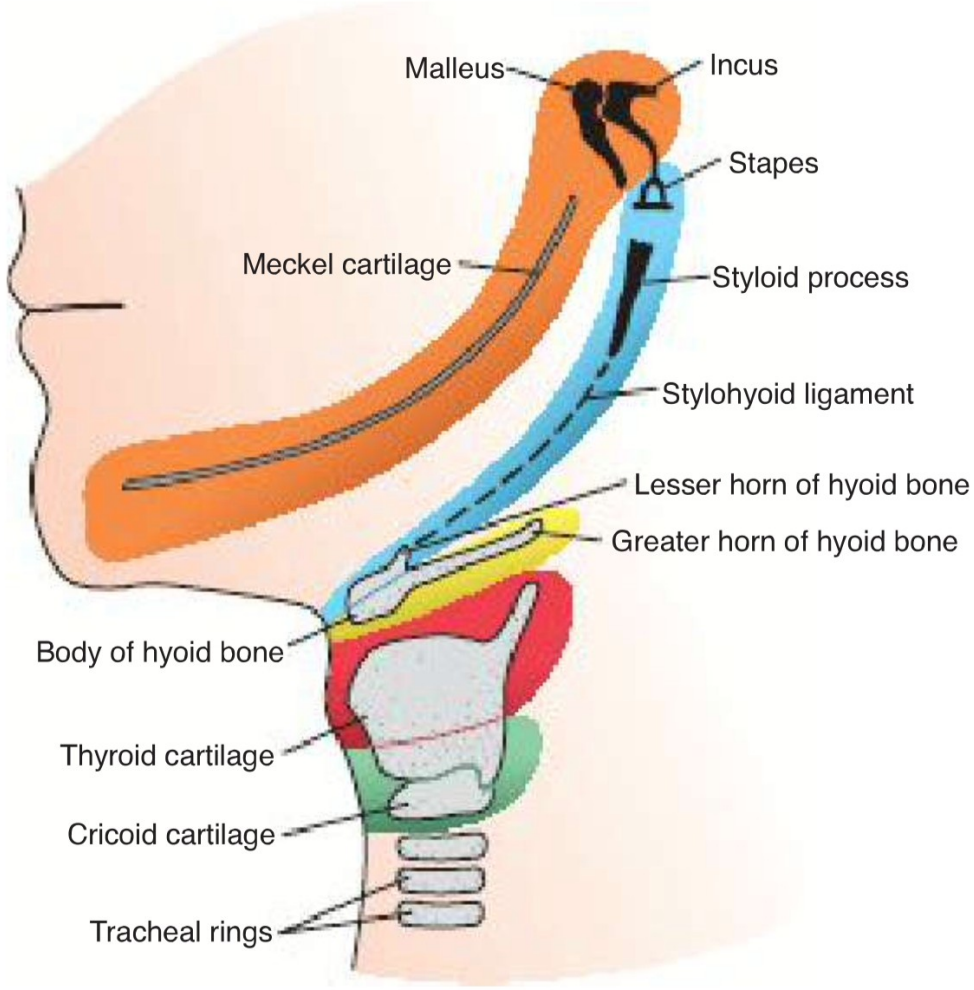
Van der Woude syndrome – pits in the lower lip in 88 % of patients

Oblique facial cleft

Median cleft lip – incomplete merging of the two medial nasal prominences; different degrees of midline structures loss ---------
holoprosencephaly – fusion of lateral ventricles, synophtalmia

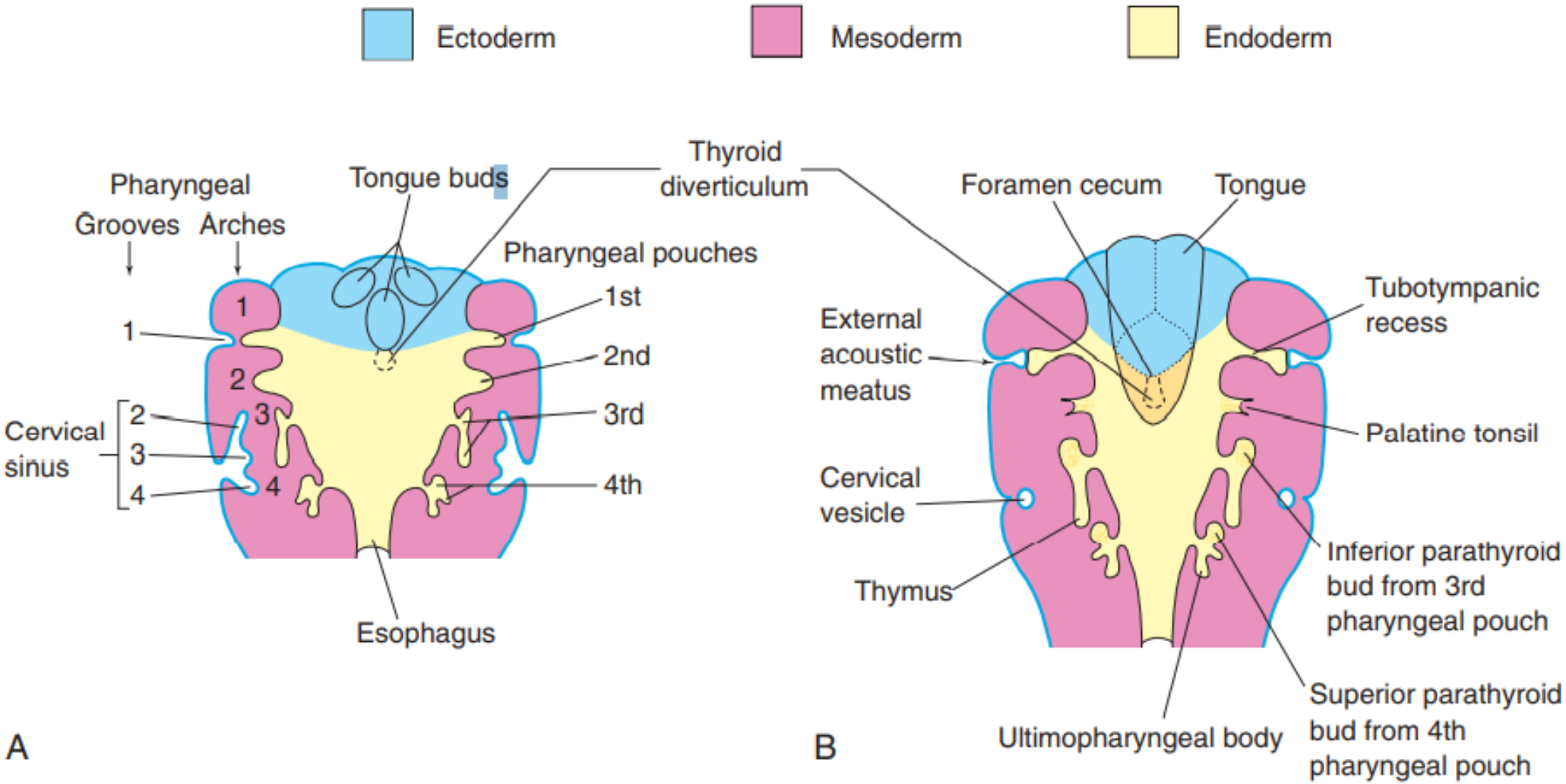
Neck region



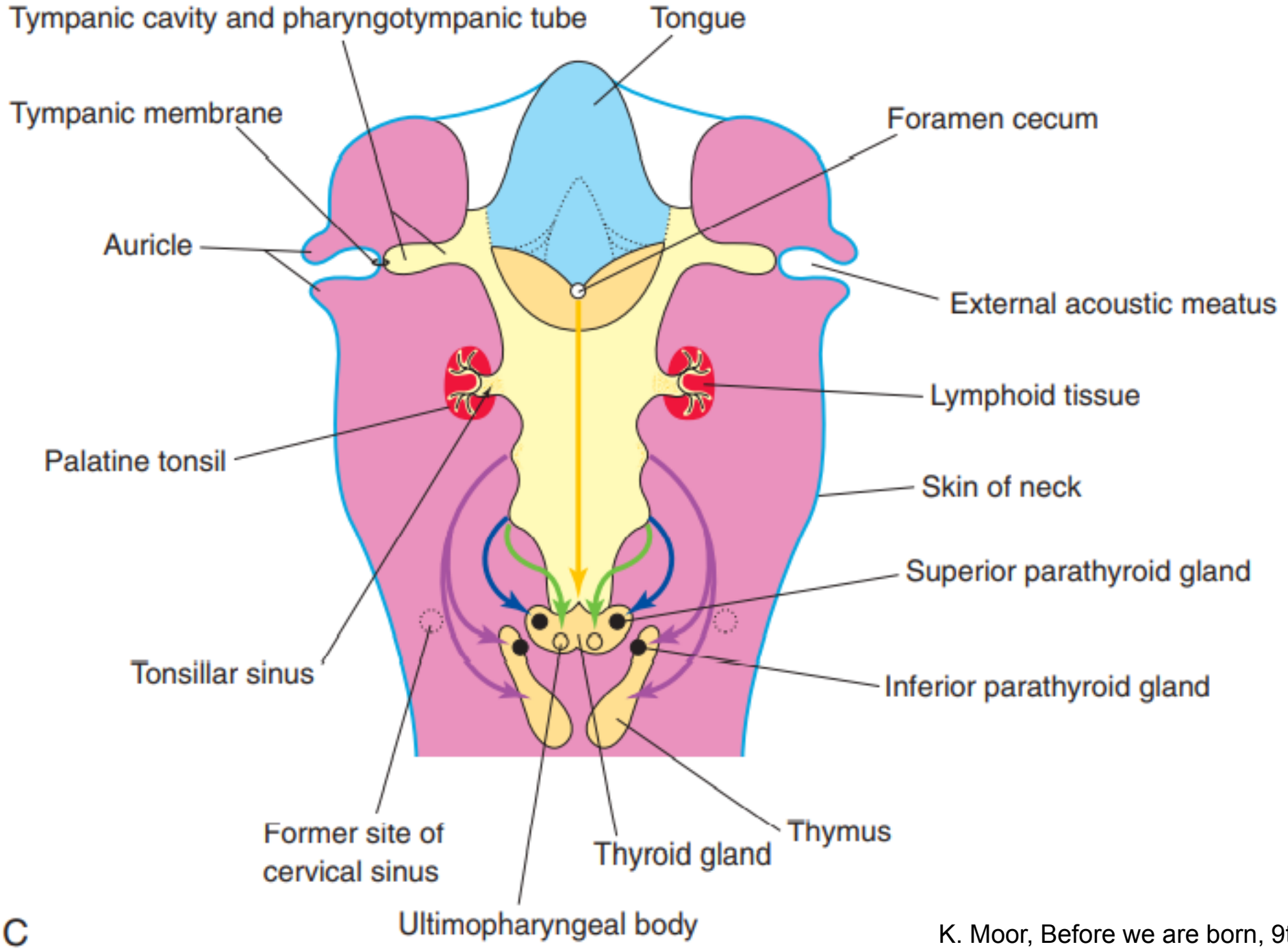


T.W. Sadler, Langman´s medical embryology, 14th edition

Neck region – pharyngeal apparatus



K. Moor, Before we are born, 9th edition



K. Moor, Before we are born, 9th edition

Ectopic thymic and parathyroid tissue

Branchial fistulas: external and internal

Cervical cysts

Craniofacial defects associated with neural crest cells:

- Mandibulofacial dysostosis – Treacher Collins syndrome
- Robin sequence
- DiGeorge syndrome, DiGeorge anomaly, velocardiofacial syndrome etc. - deletion on 22q11.2 (1/4000)
- Hemifacial microsomia (oculoauriculovertebral spectrum – Goldenhar syndrome)