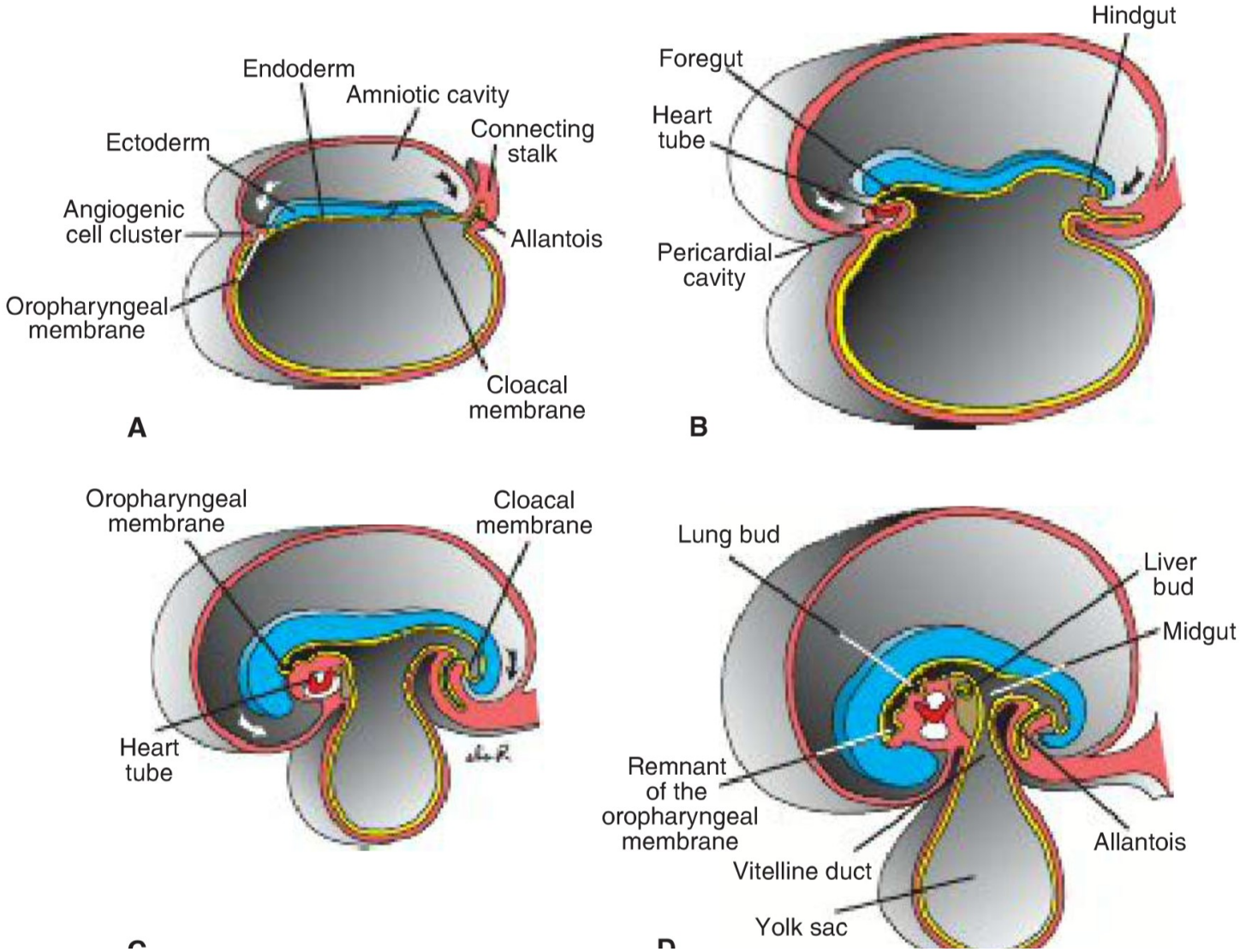
Development and teratology of digestive system.

Development of facial and cervical region, face clefts.

Anna Mac Gillavry

22.03.2021

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



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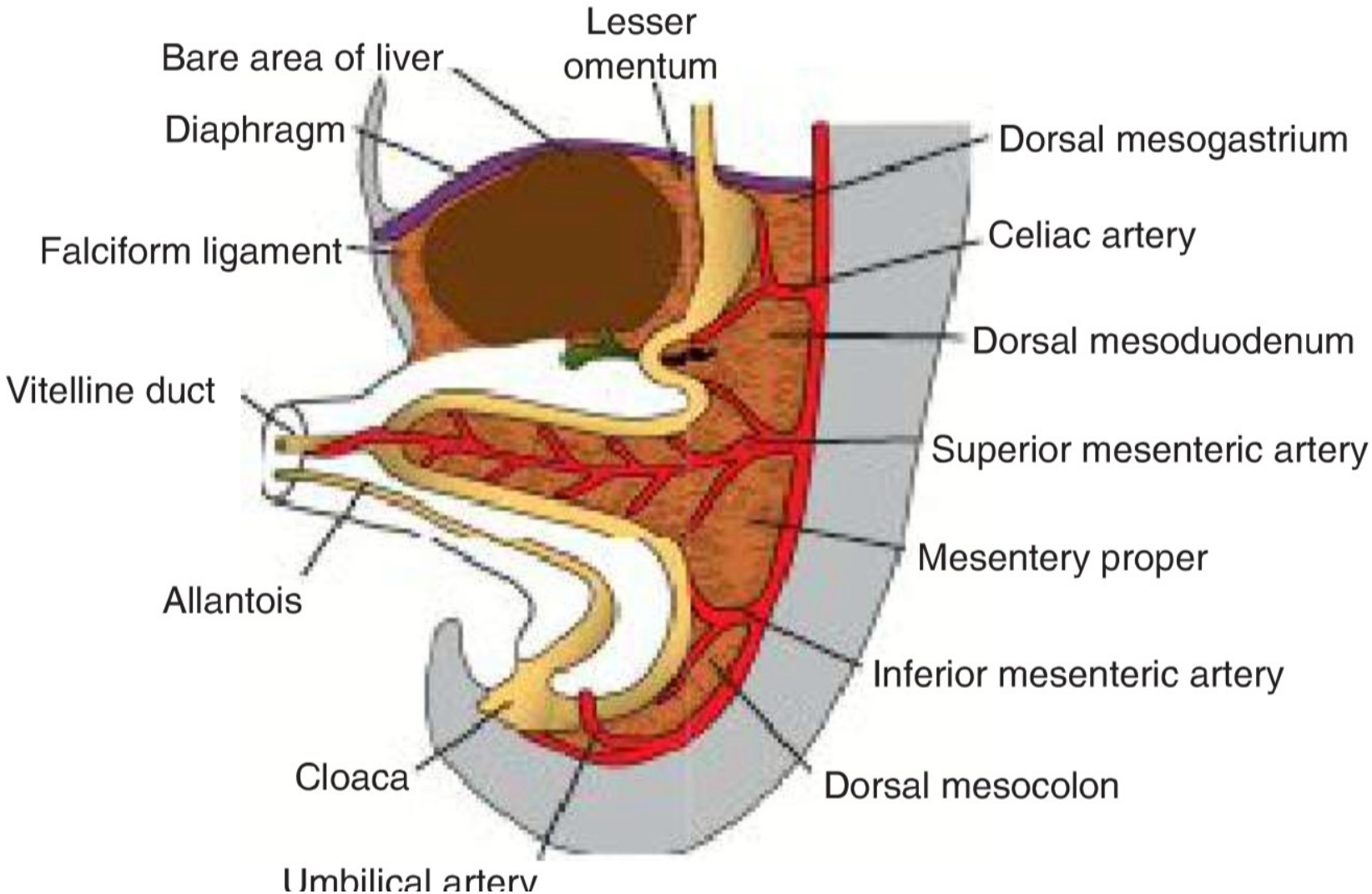

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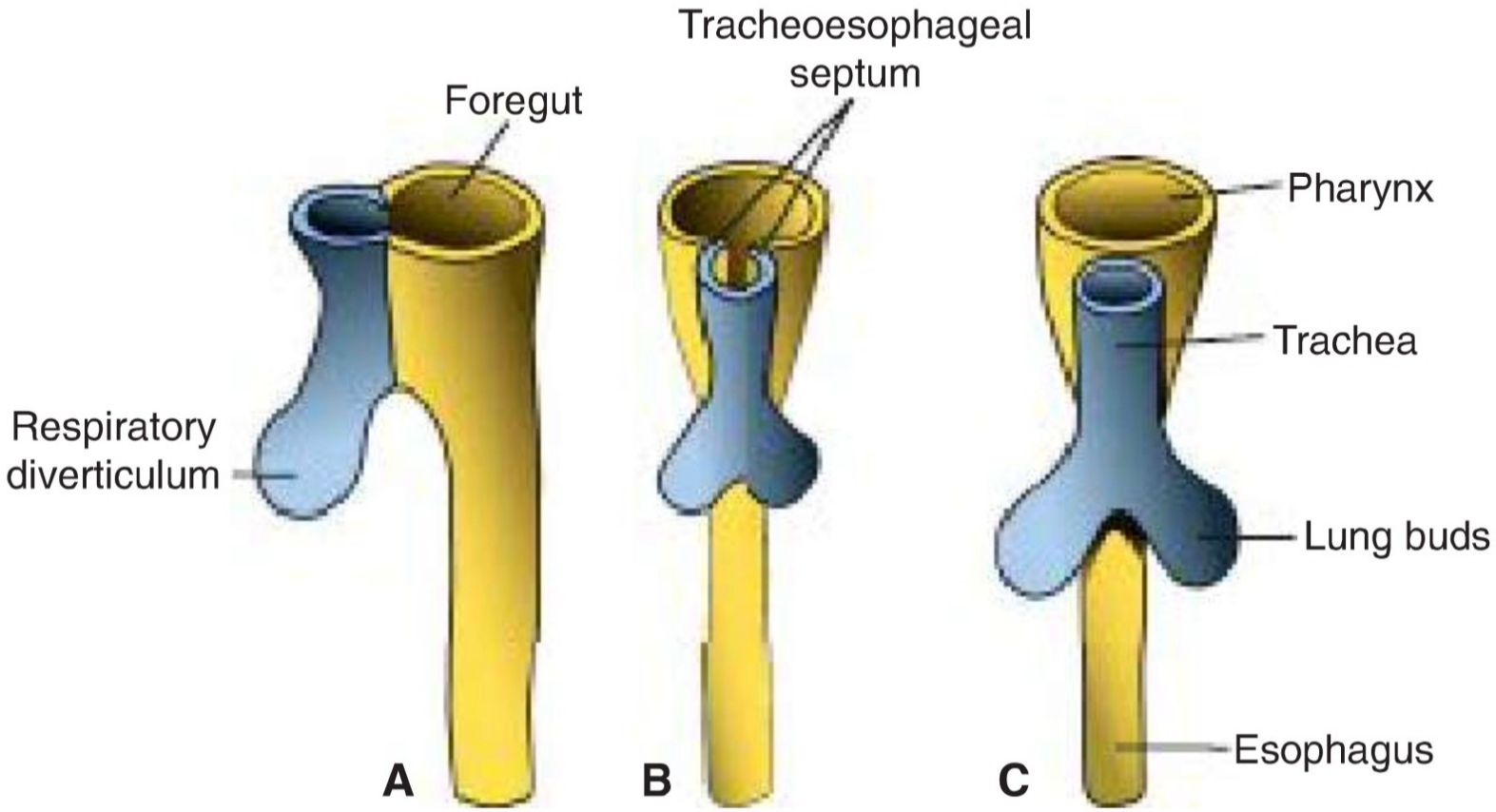
Esophagus

4 weeks



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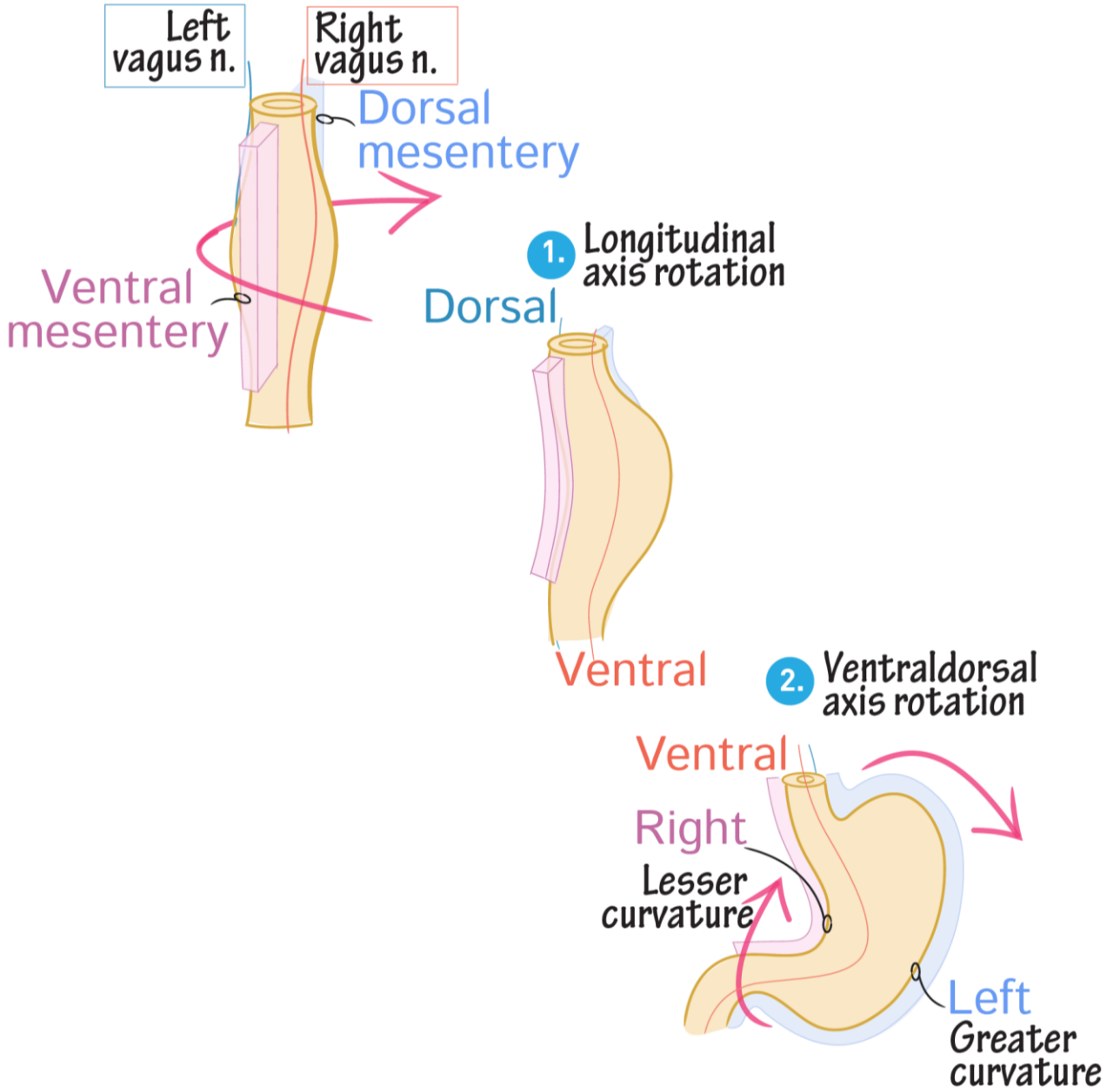
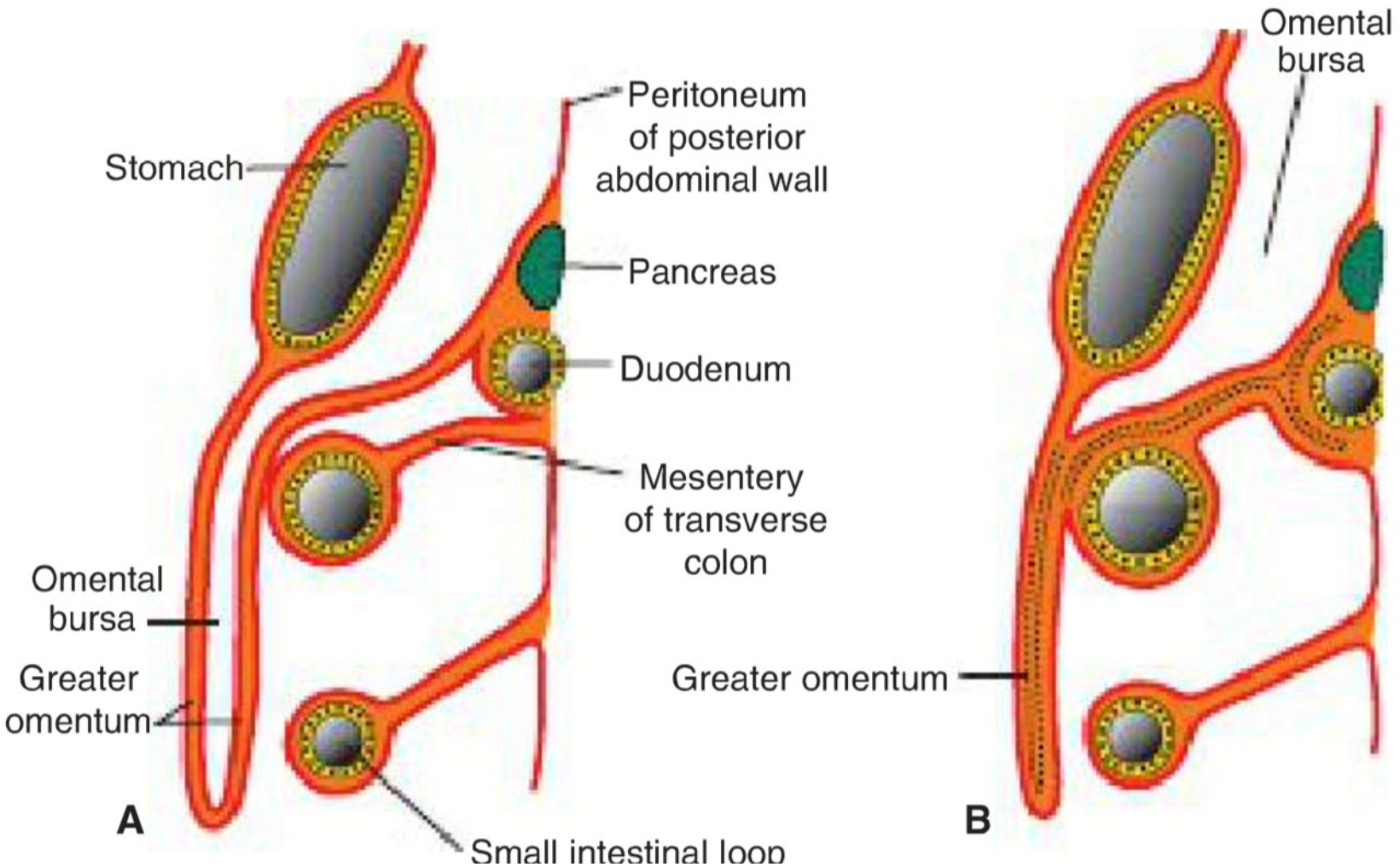
Stomach



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



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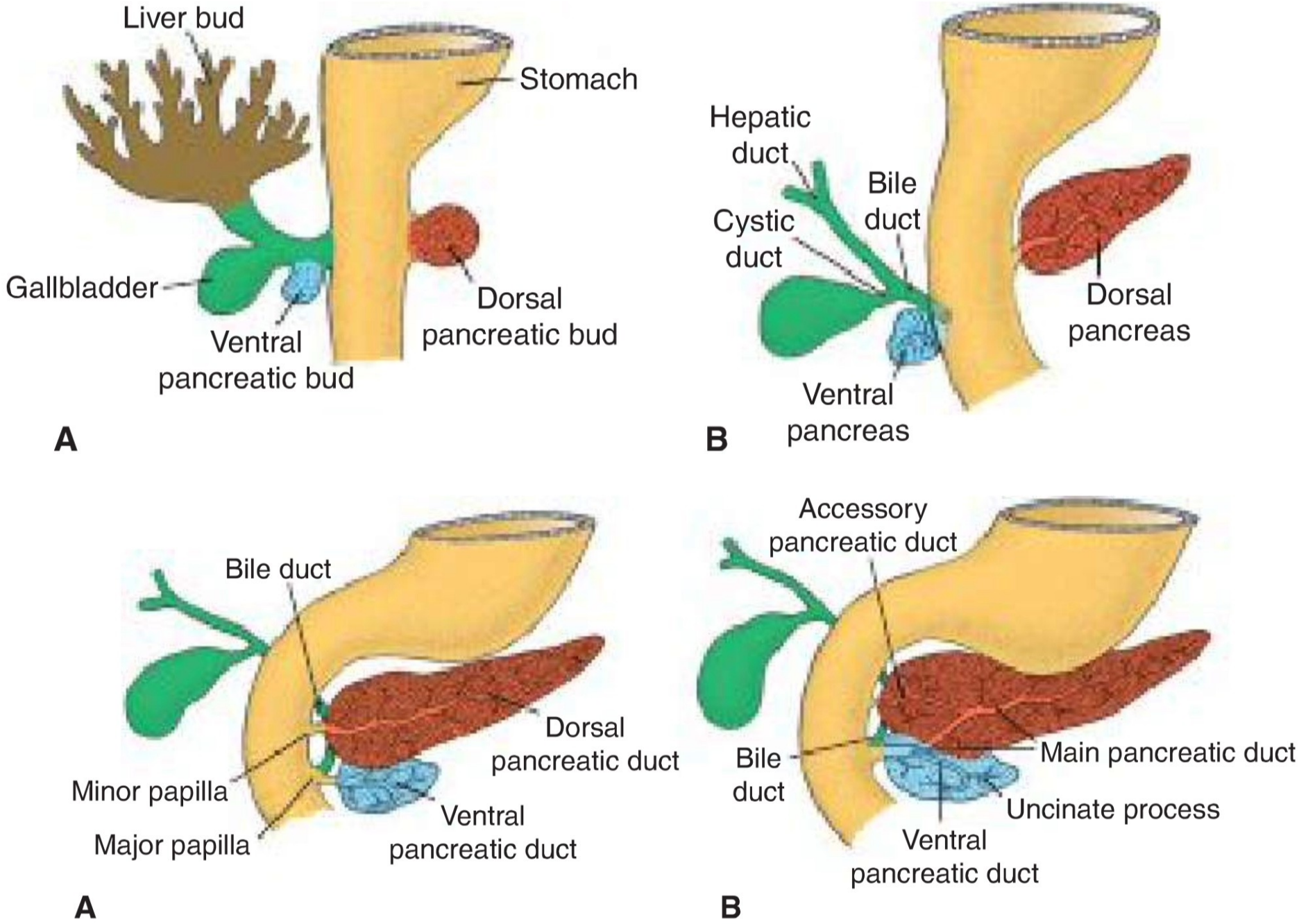

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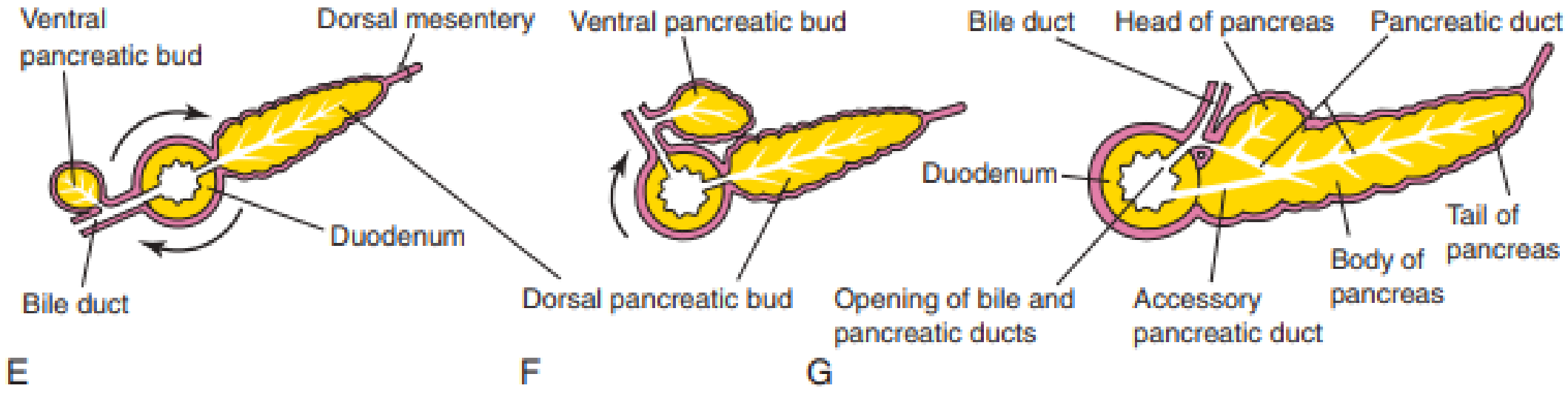

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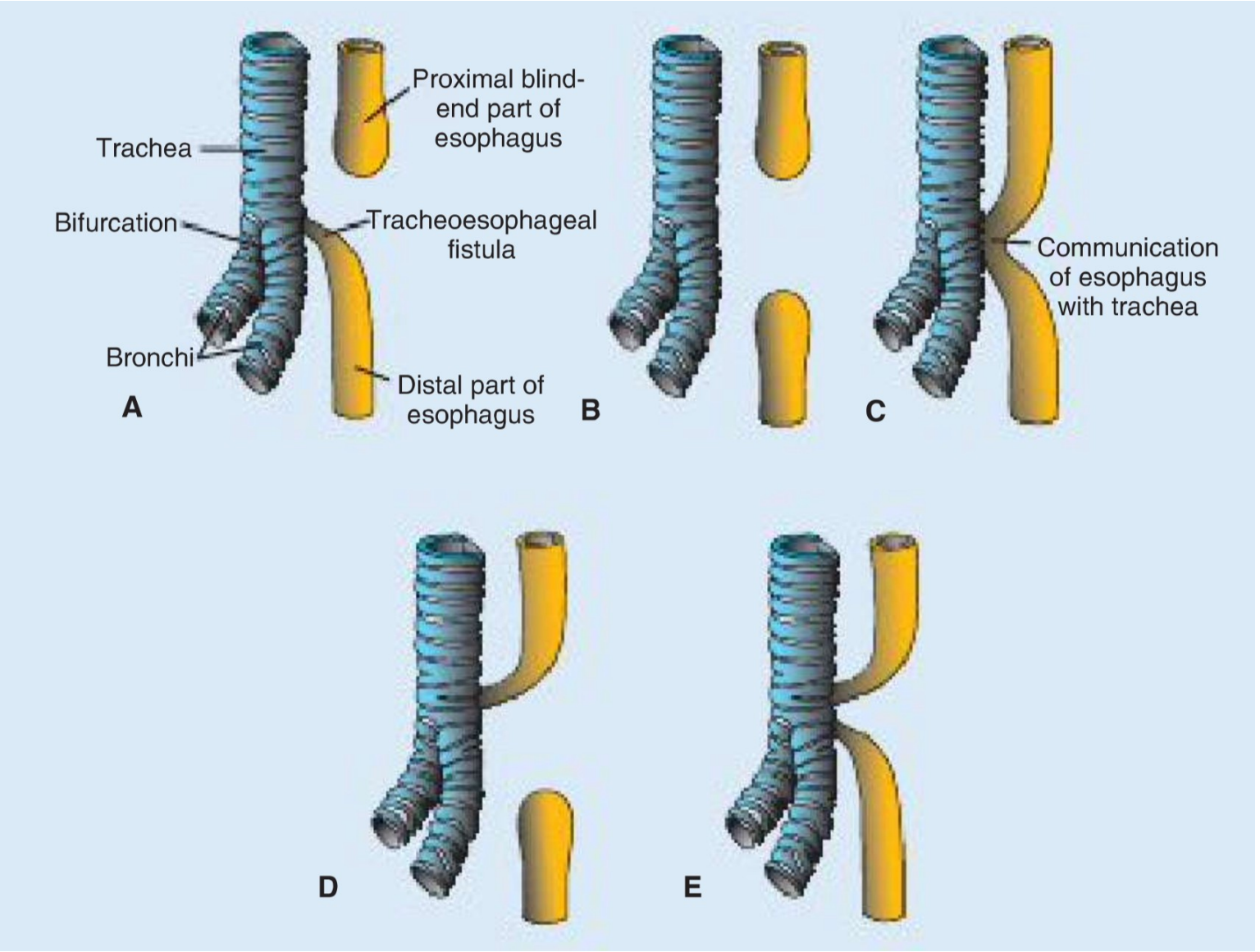
Esophagus:

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



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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

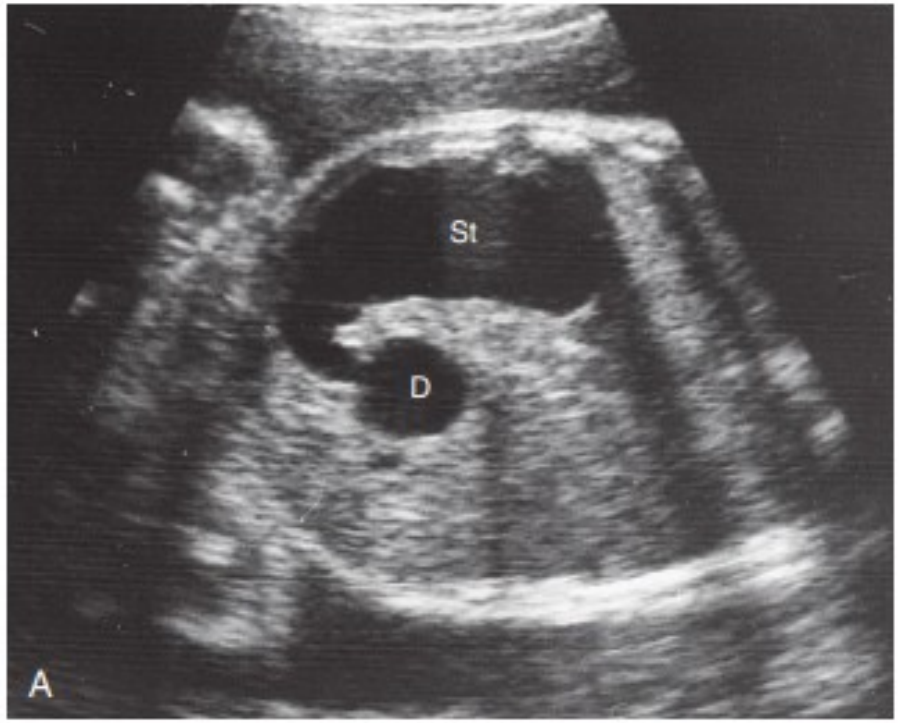
Extrahepatic biliary atresia (1/15000)

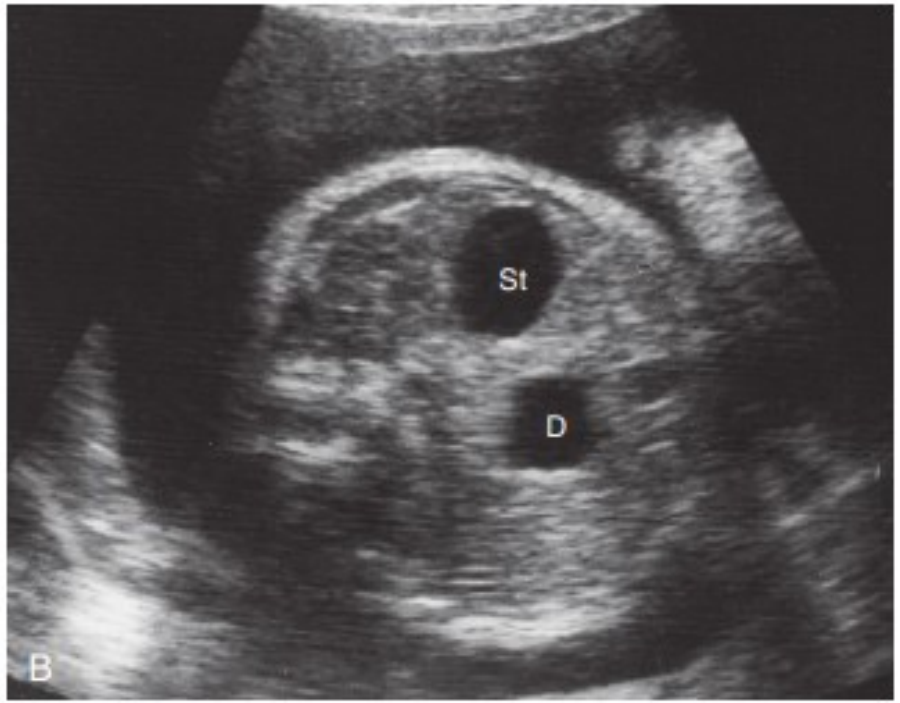
Intrahepatic biliary duct atresia/hypoplasia (1/100000)

Pancreas:

Annular pankreas

Accessory spleens – in 10 % of population





Duodenum:
Duodenal stenosis/atresia – results from incomplete recanalization

Polyhydramnios

„Doble-Bubble“

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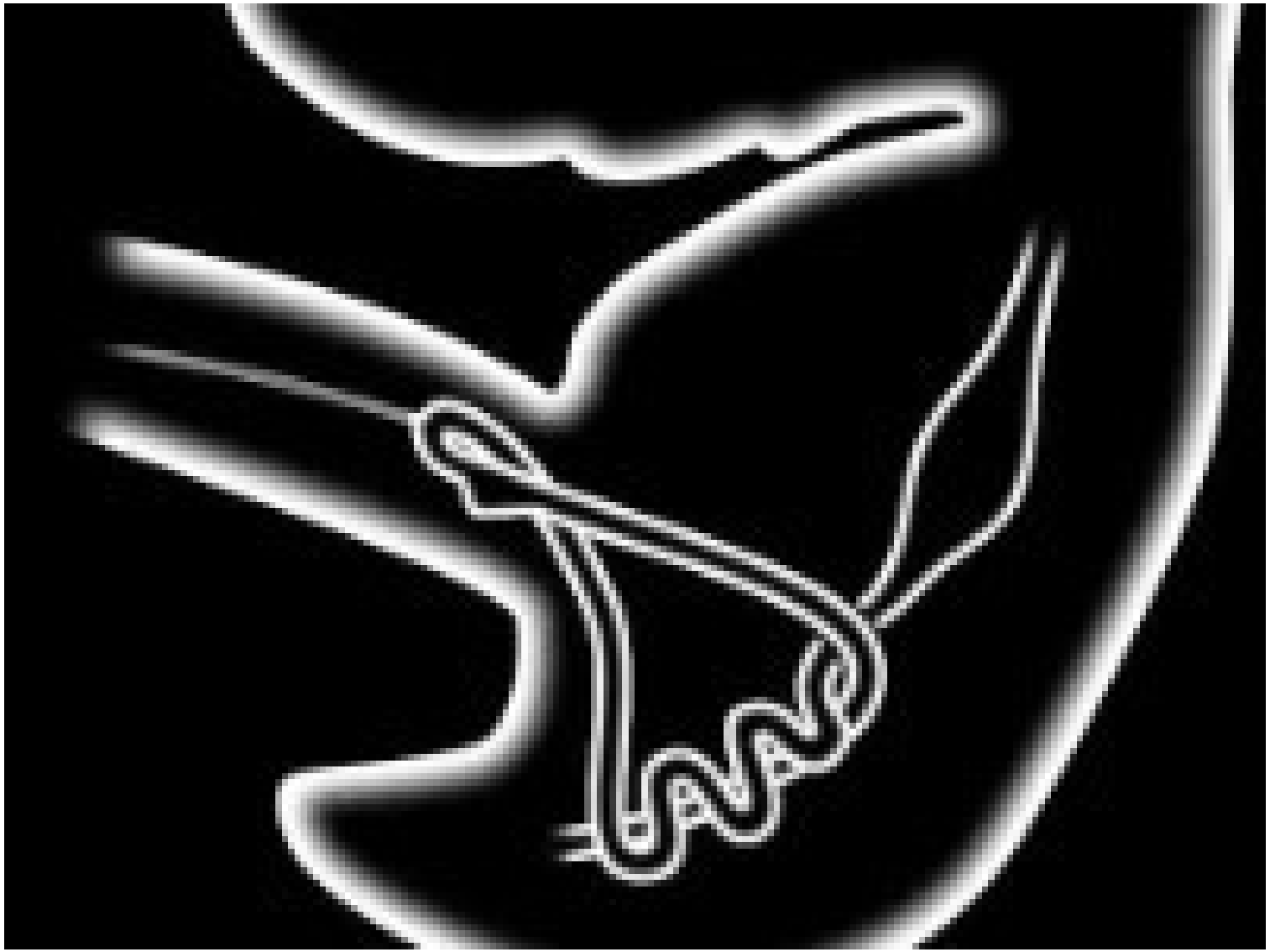
Midgut development. Physiological herniation.

6th – 10th week



https://youtu.be/AscKR_cQExY

Body wall defects

Gastroschisis (3,5/10000) – most common in infants from white thin women under 20; usually not assotiated 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 if the intestinal loop

Duplications of intestinal loops and cysts

Gut atresias and stenoses

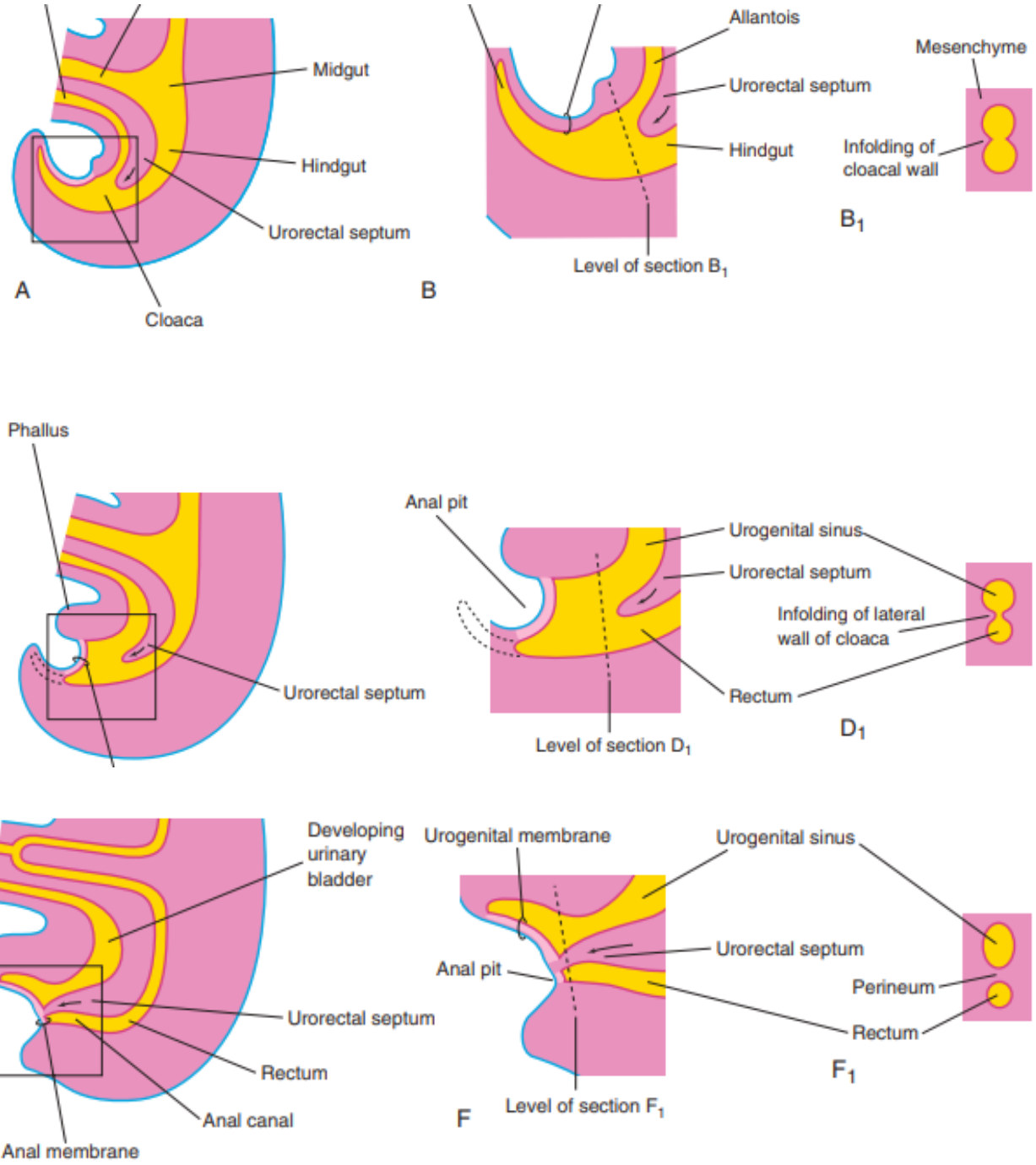
- Most occur in duodenum, fewest in the colon, equal number in jejunum and ileum; in 50 % of cases a region of bowel is missing completely, in 20 % cases the fibrous cord is present; stenoses represent only 5 % of cases

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!!!



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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

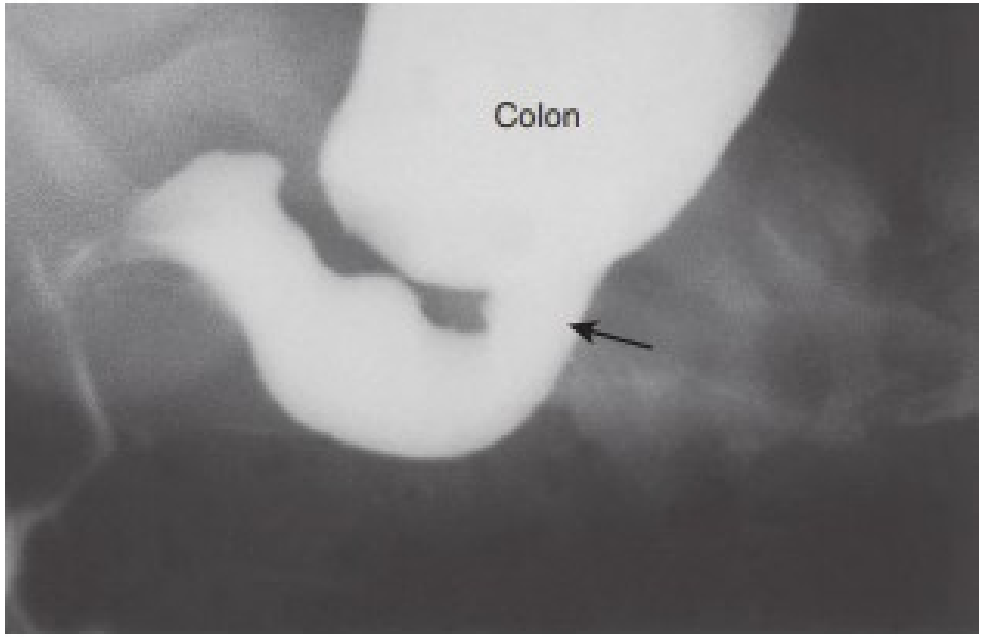
anal stenosis

membranous atresia of anus

High: anorectal agenesis, with or without

fistula (2/3 of anorectal defects)

rectal atresia



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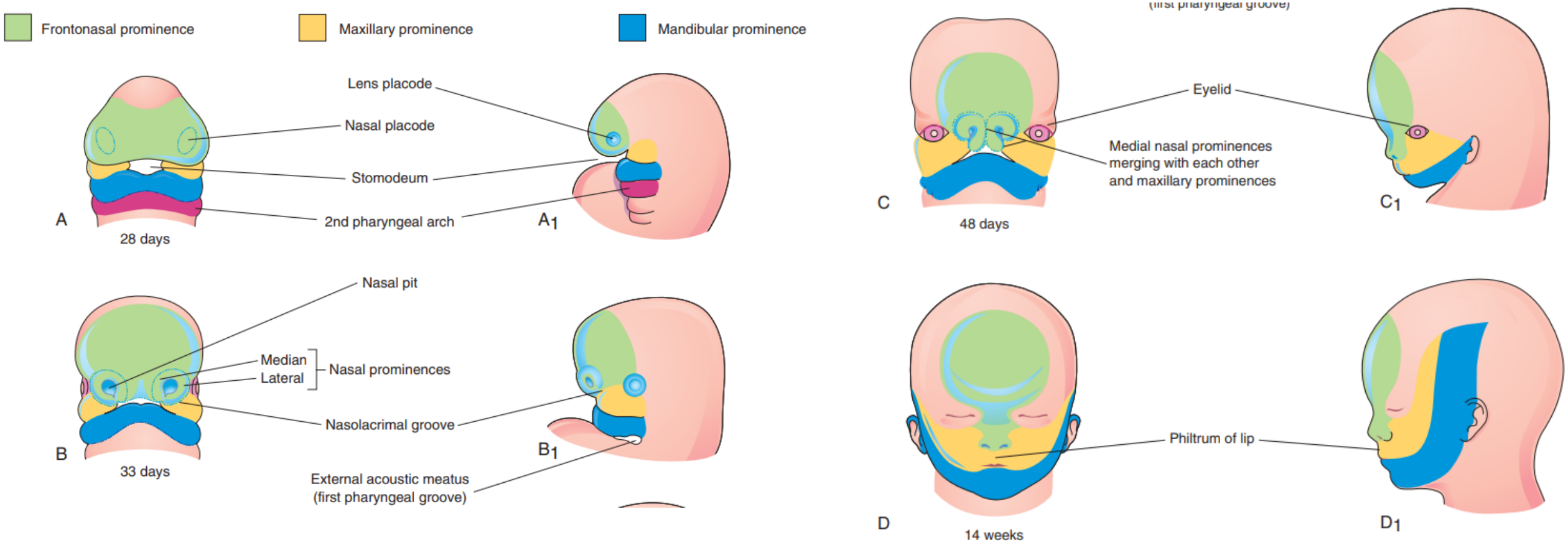
Development of the face



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Anterior cleft deformities

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

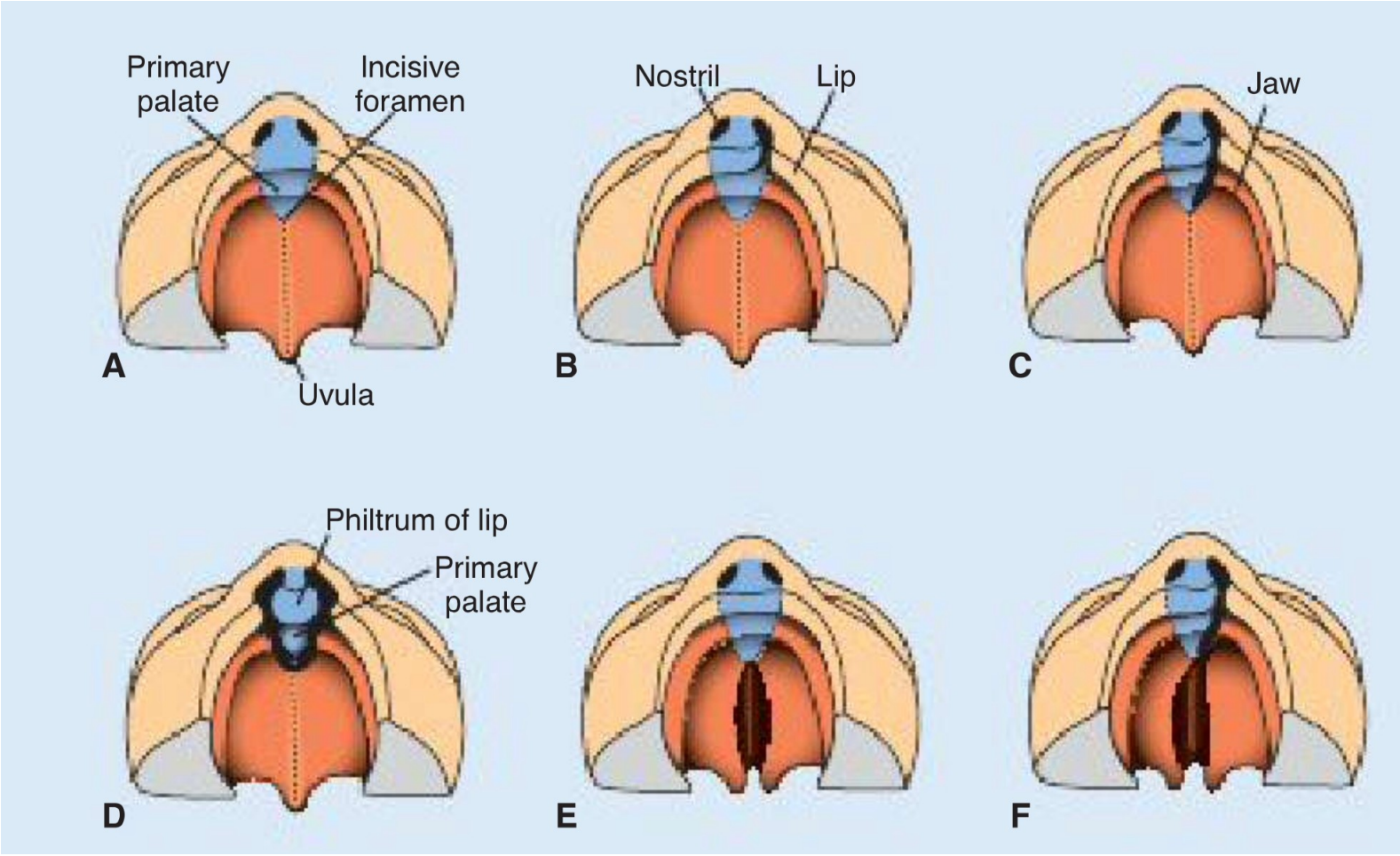
Cleft upper jaw

Cleft between the primary and the secondary palates

Posterior cleft deformities

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

Cleft uvula



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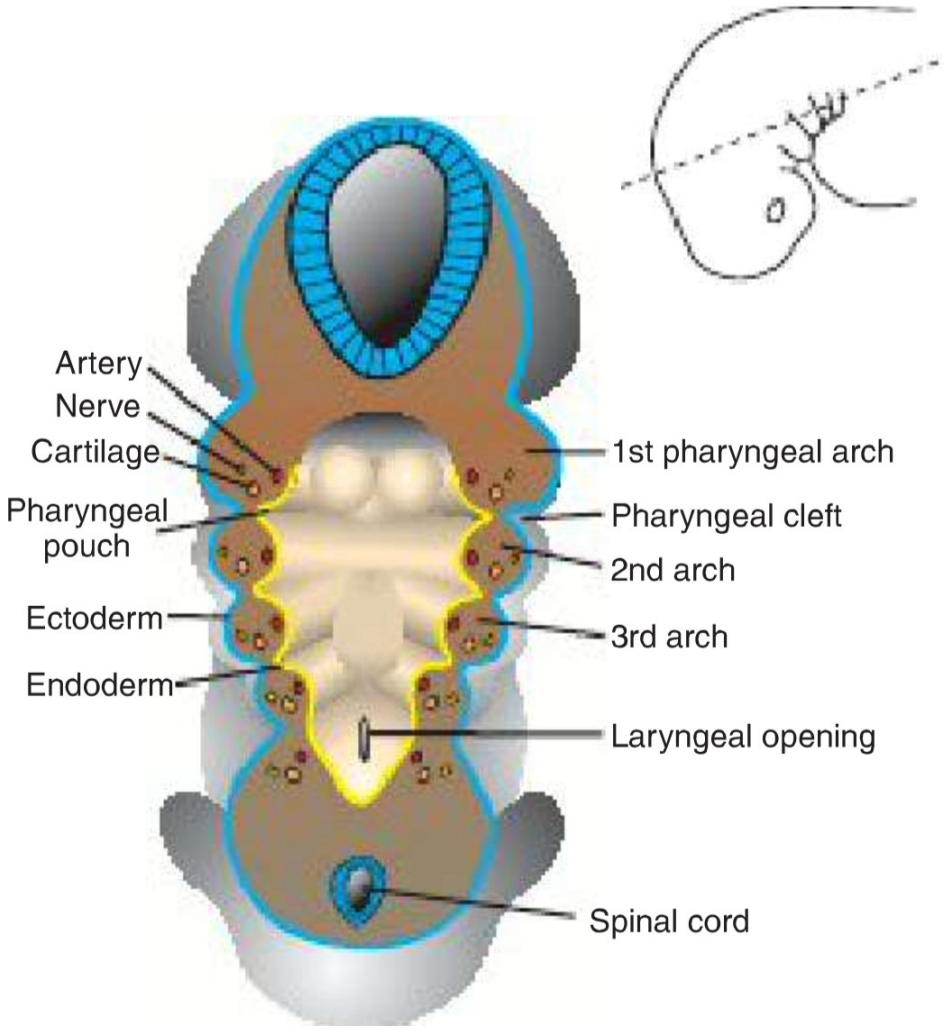
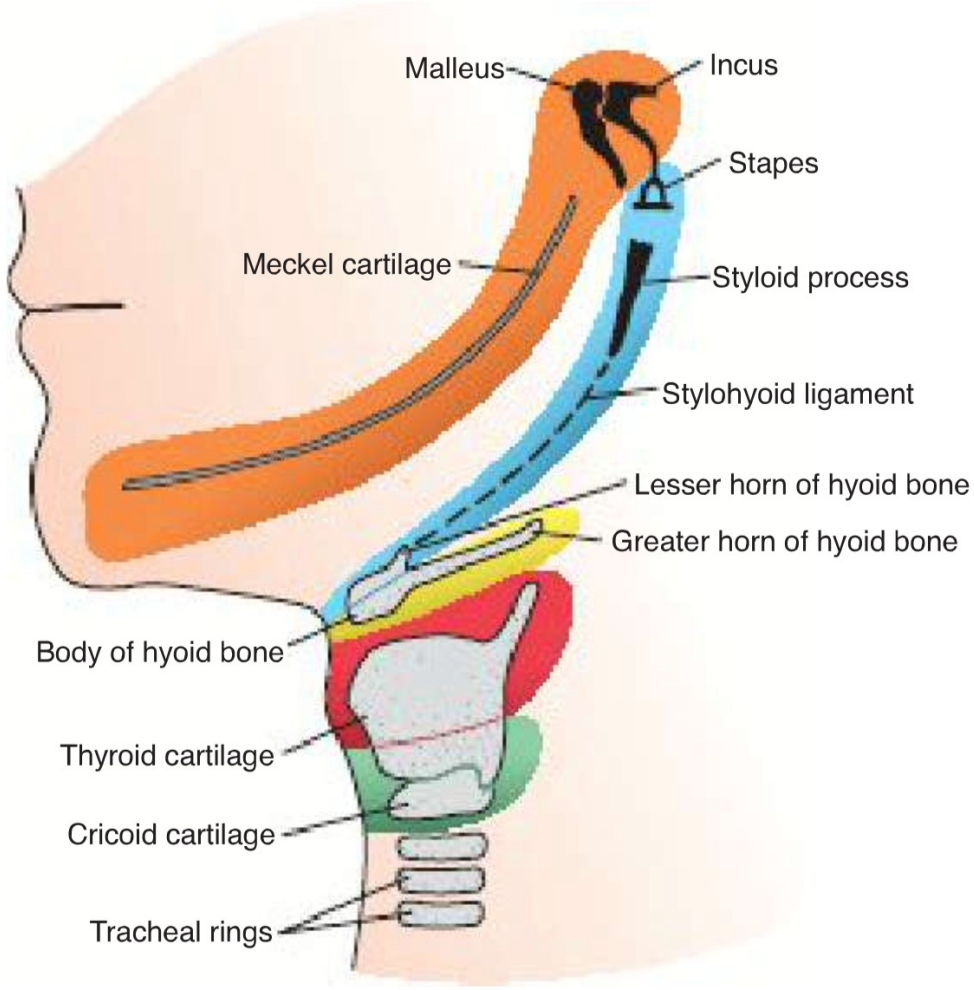
Van der Woude syndrome – pits in the lower lip in 88 % of patients

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





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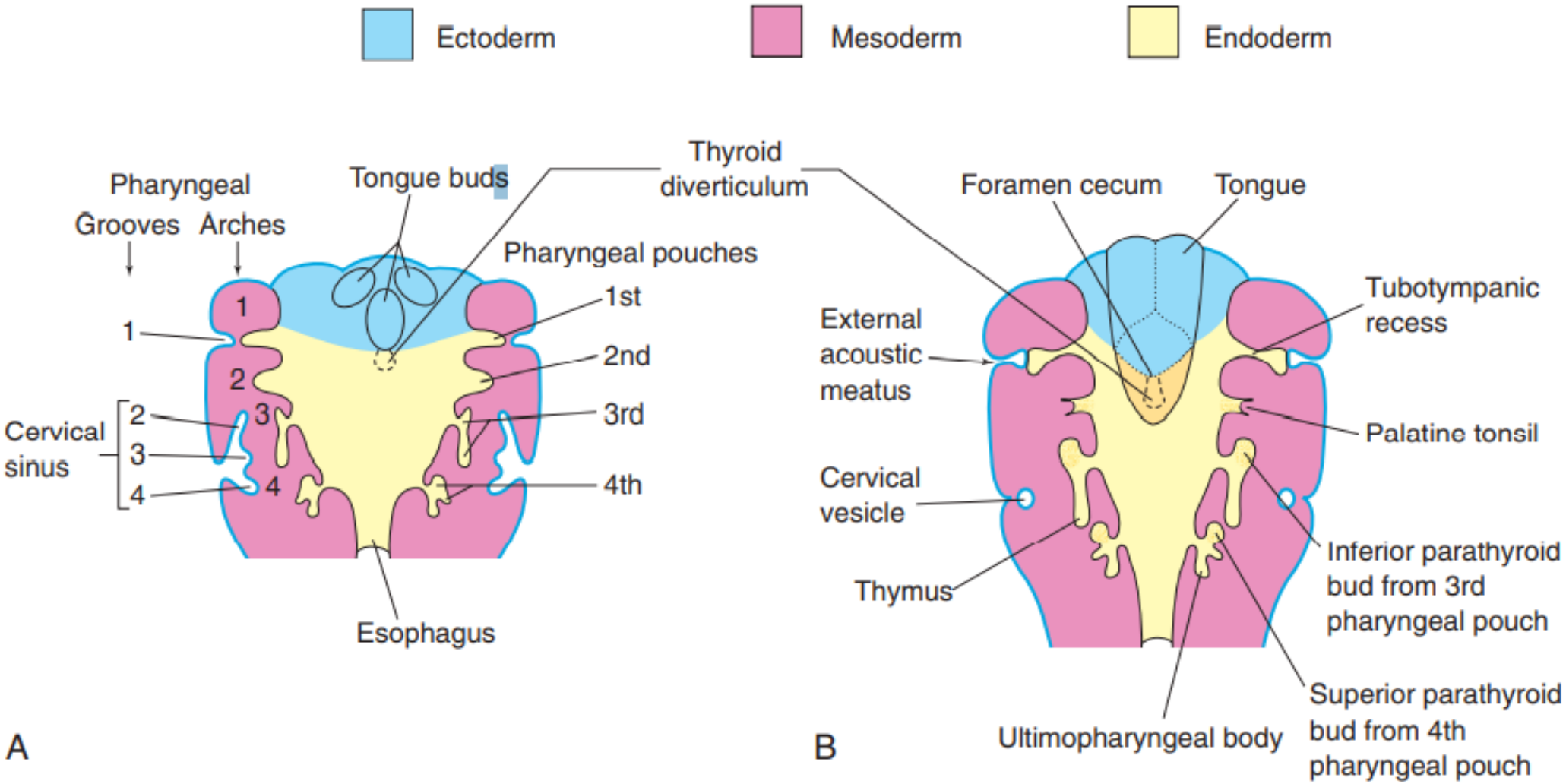
Neck region – pharyngeal apparatus



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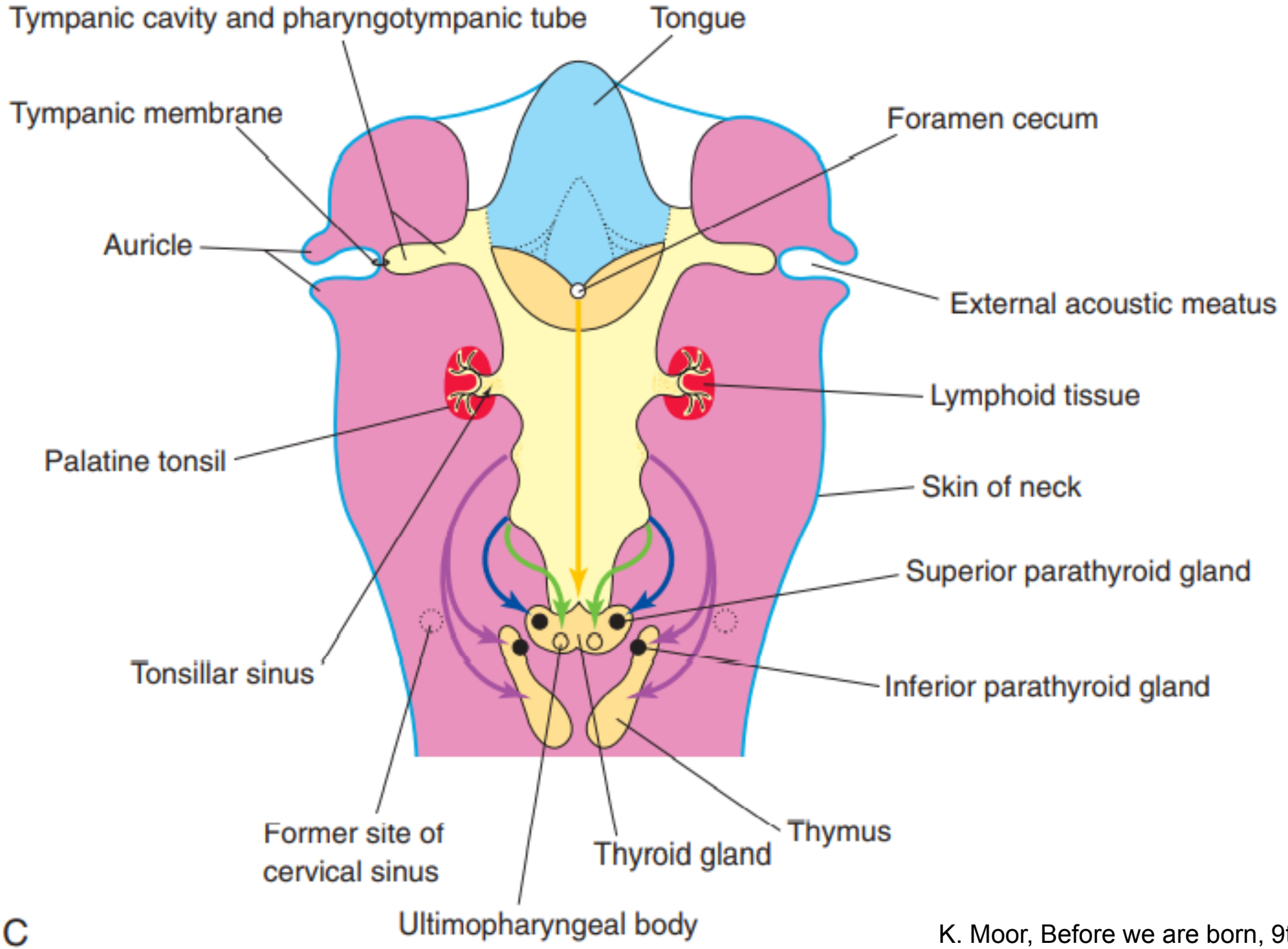

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Ectopic thymic and parathyroid tissue

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)