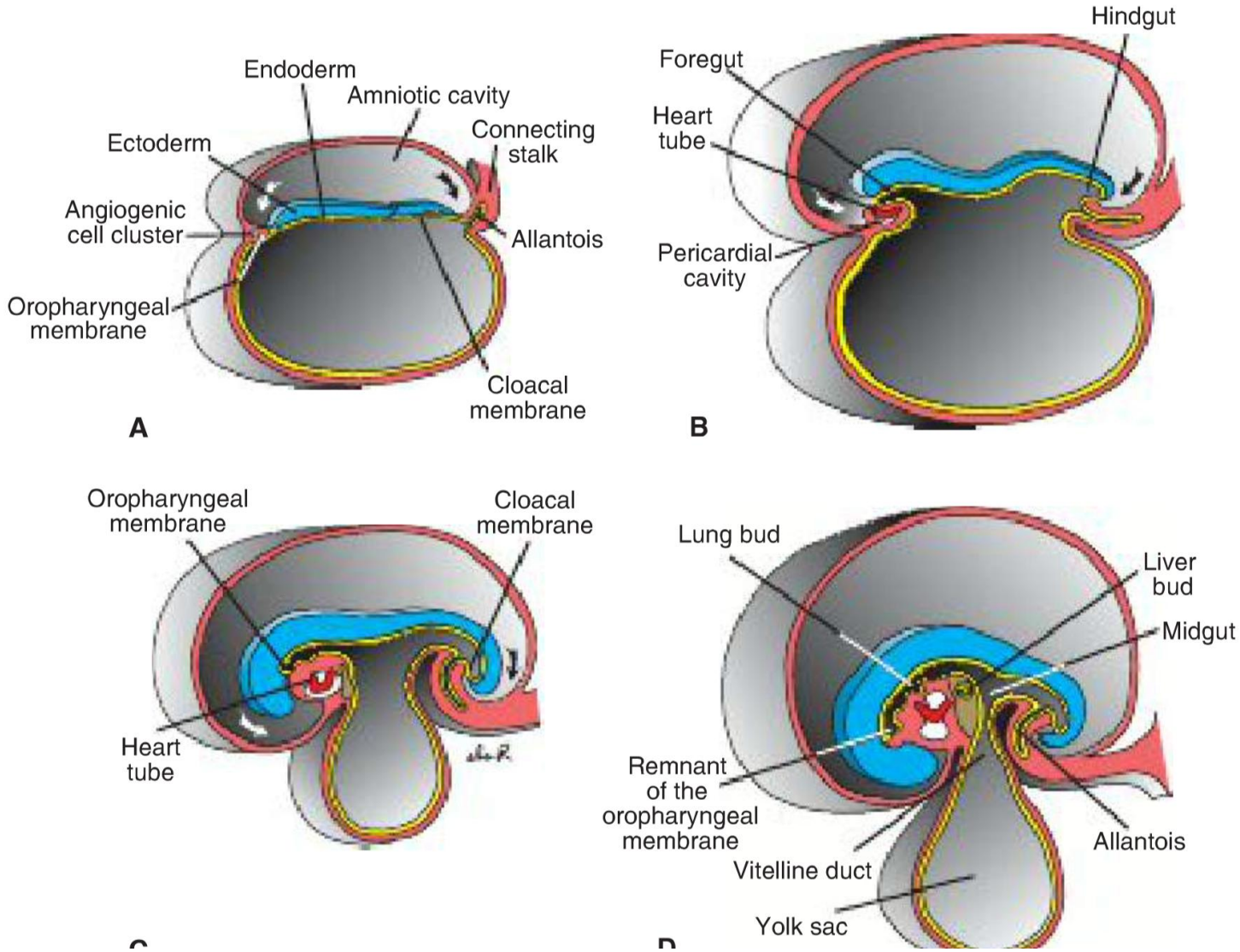
Development and teratology of digestive system.

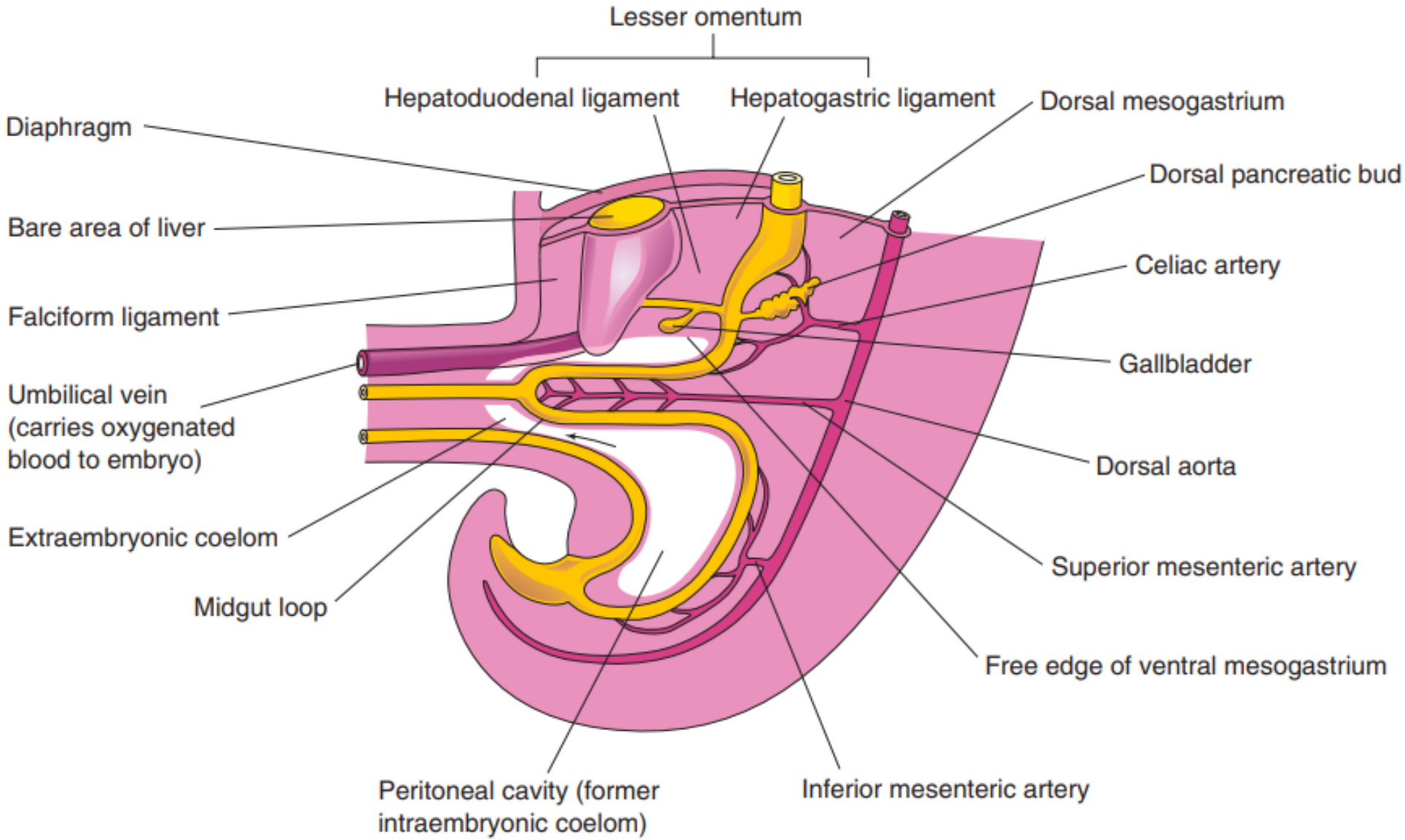
Anna Mac Gillavry

03.03.2025

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



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



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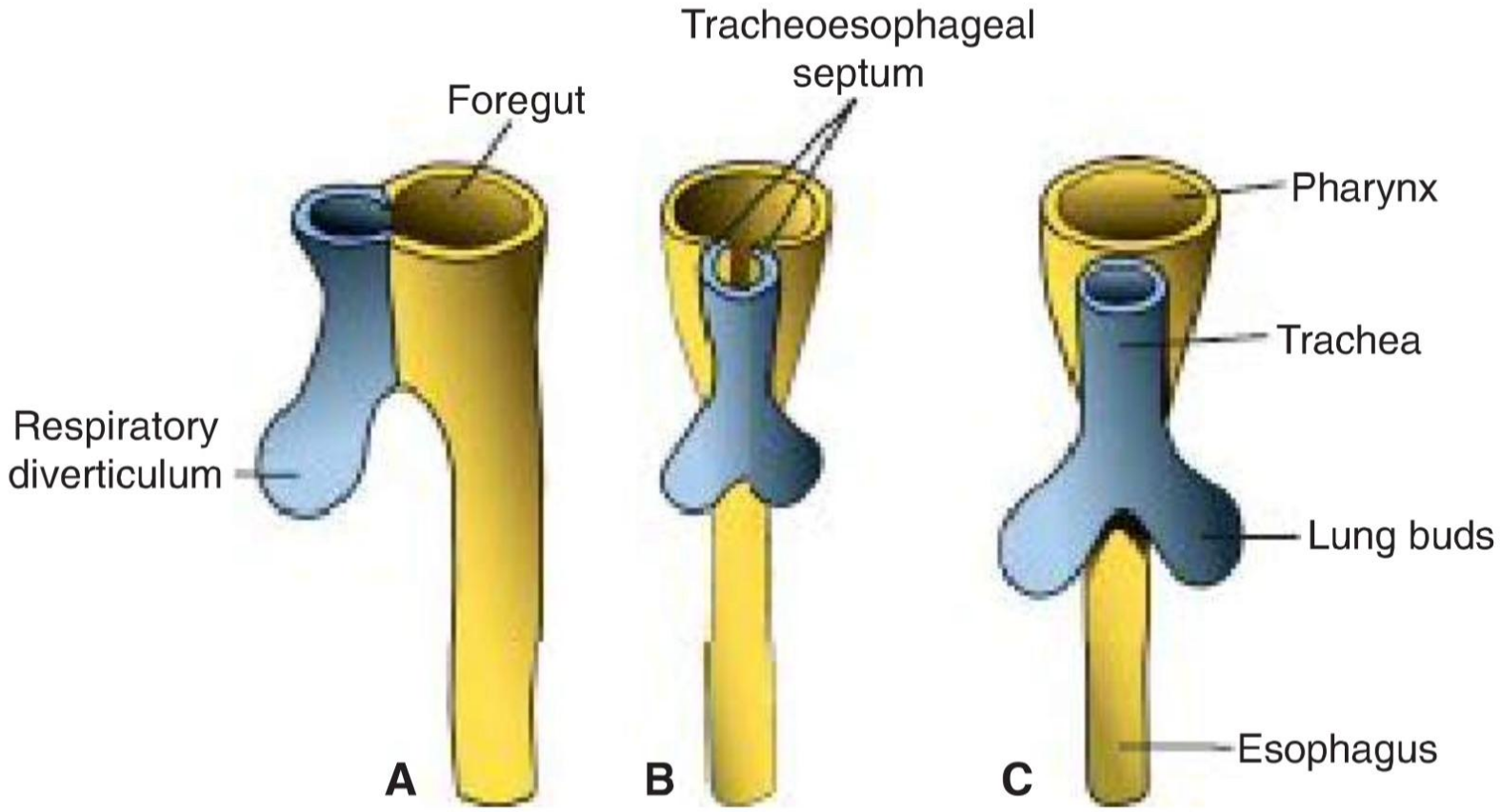
Esophagus

4th week



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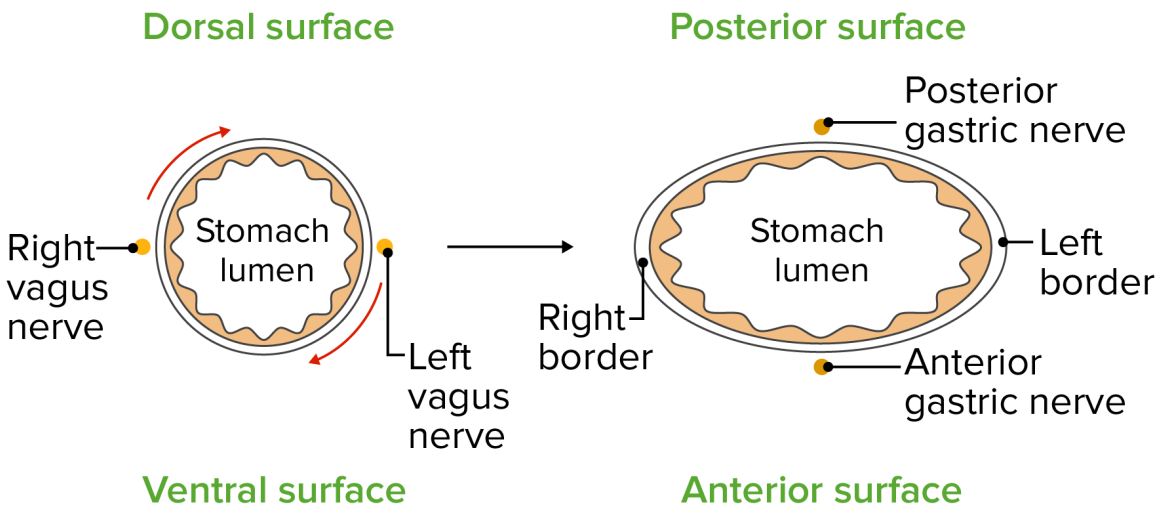
Stomach

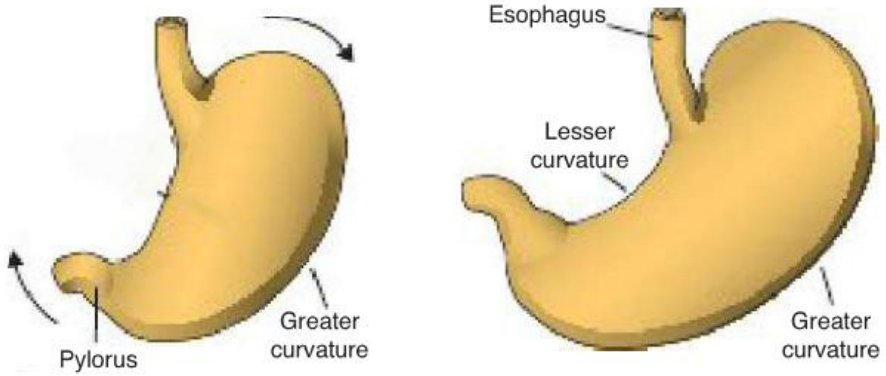
Two rounds of rotation:

1. 90° clockwise along the longitudinal axis



Desarrollo de los Órganos Abdominales | Concise Medical Knowledge (lecturio.com)

2. anteroposterior axis



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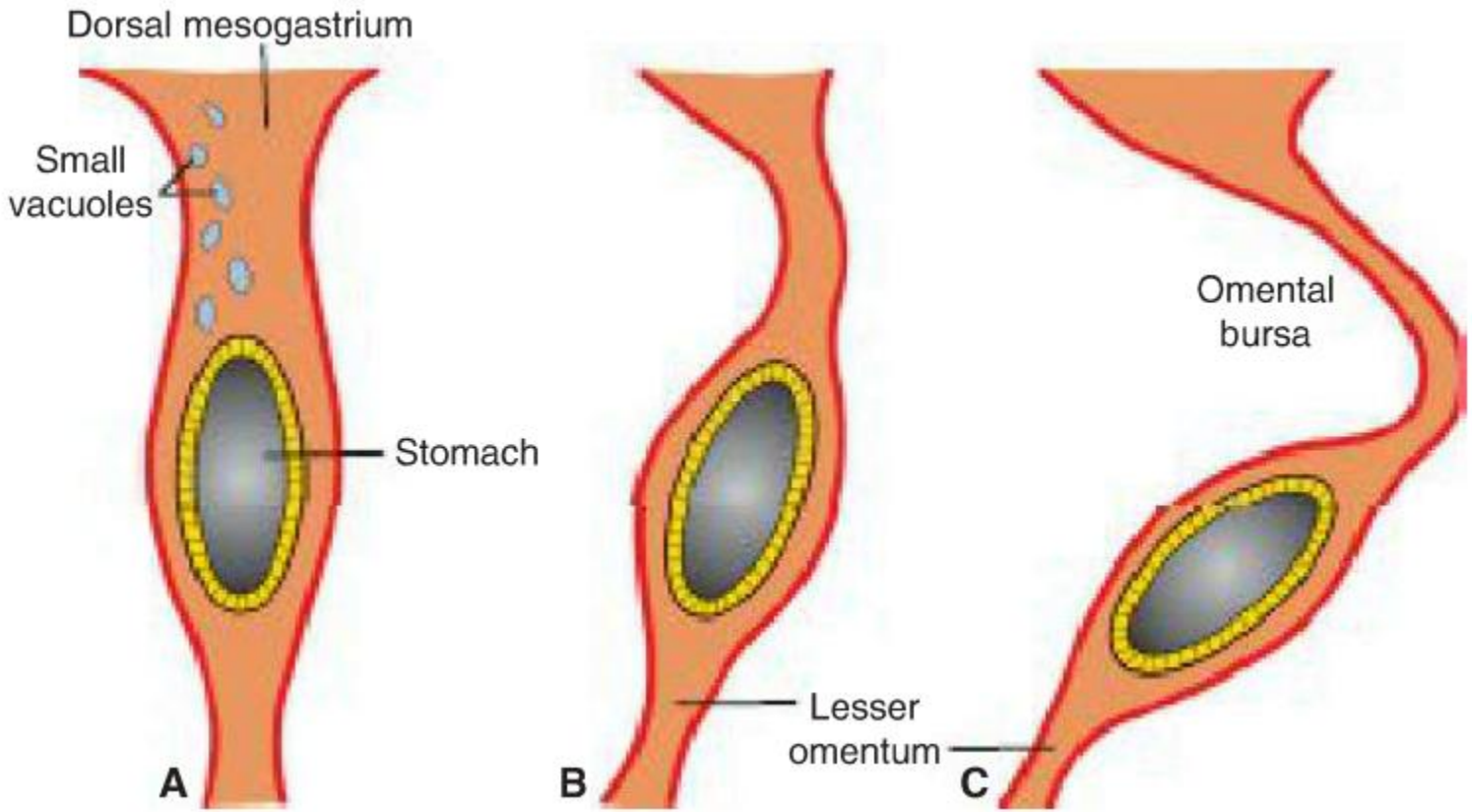

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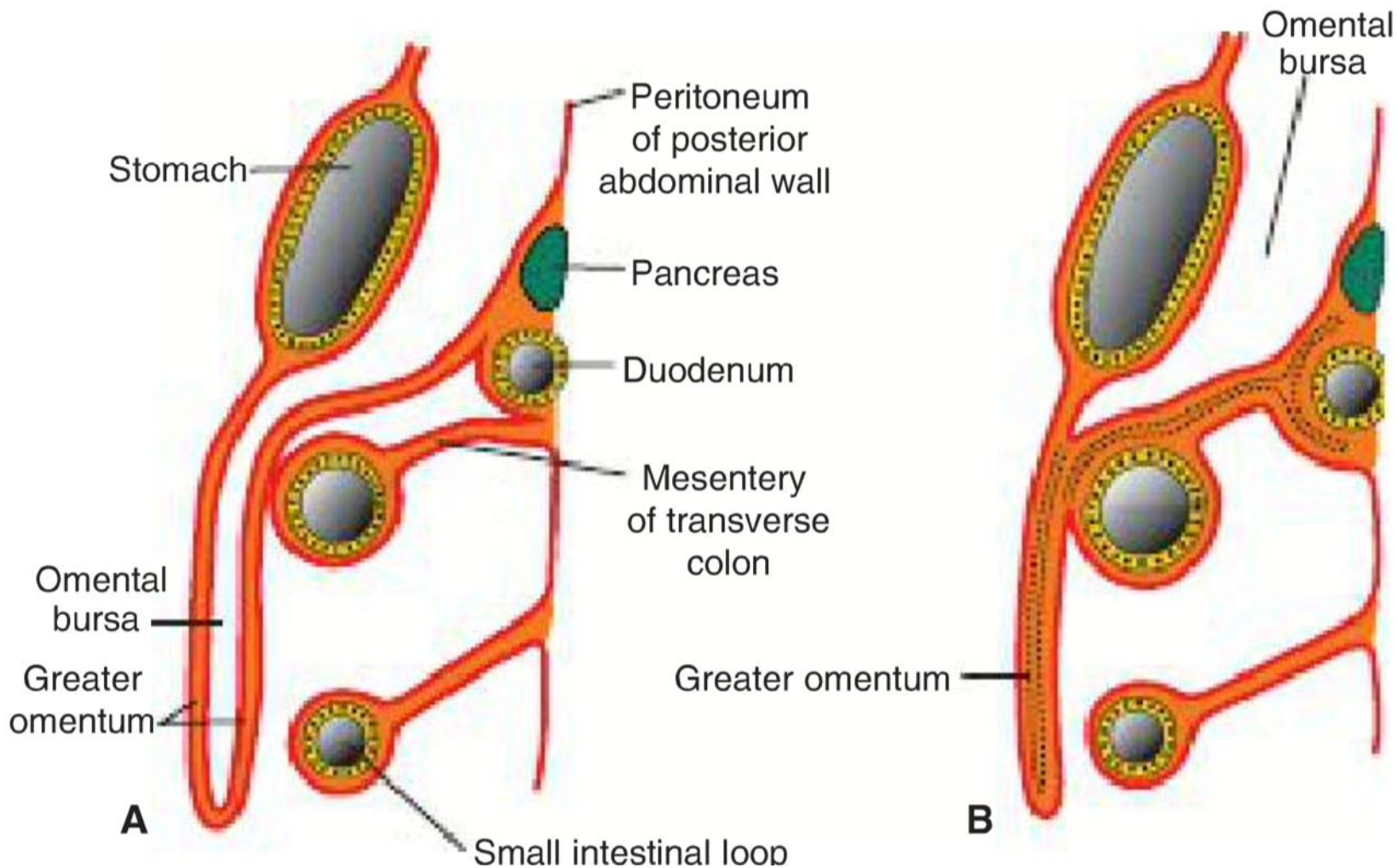

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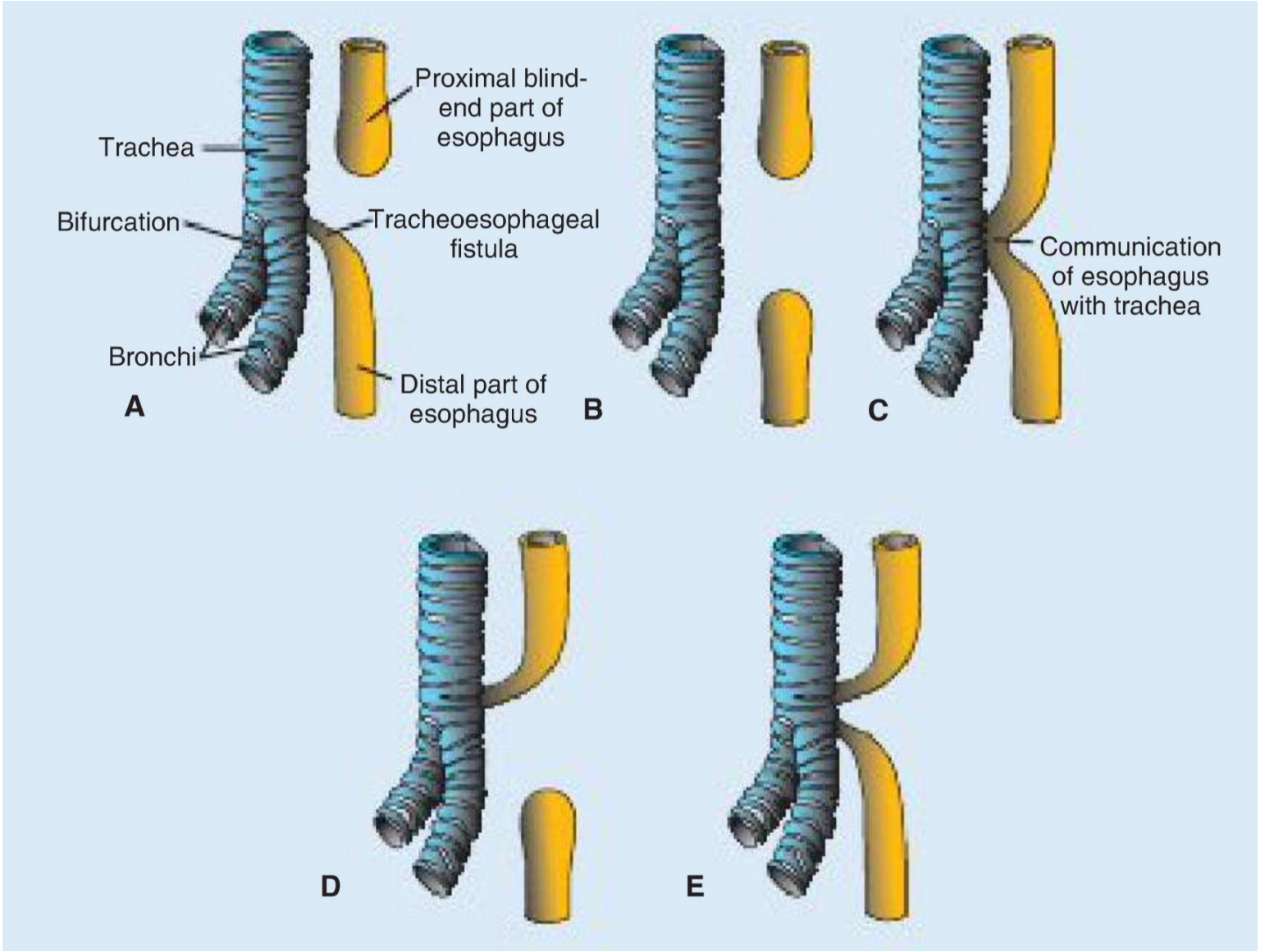
Esophagus:

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

Stomach:

- pyloric stenosis (1 in 150 males, 1 in 750 females) – develops during fetal life, however, can develop as a result of postnatal exposure (e.g. erythromycin)



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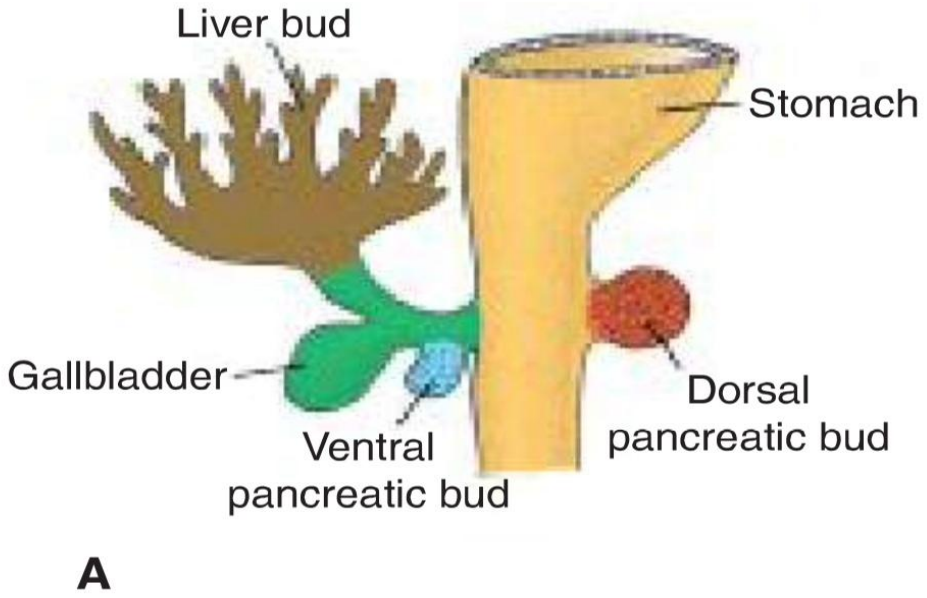
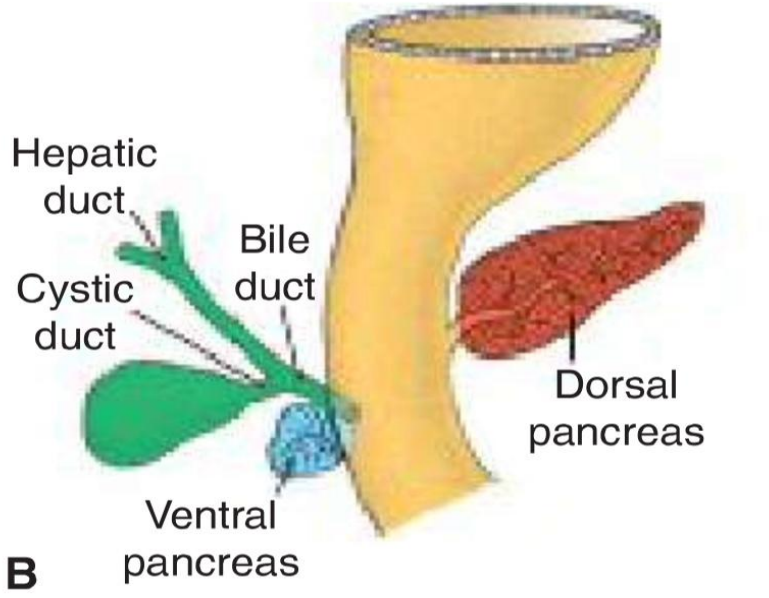
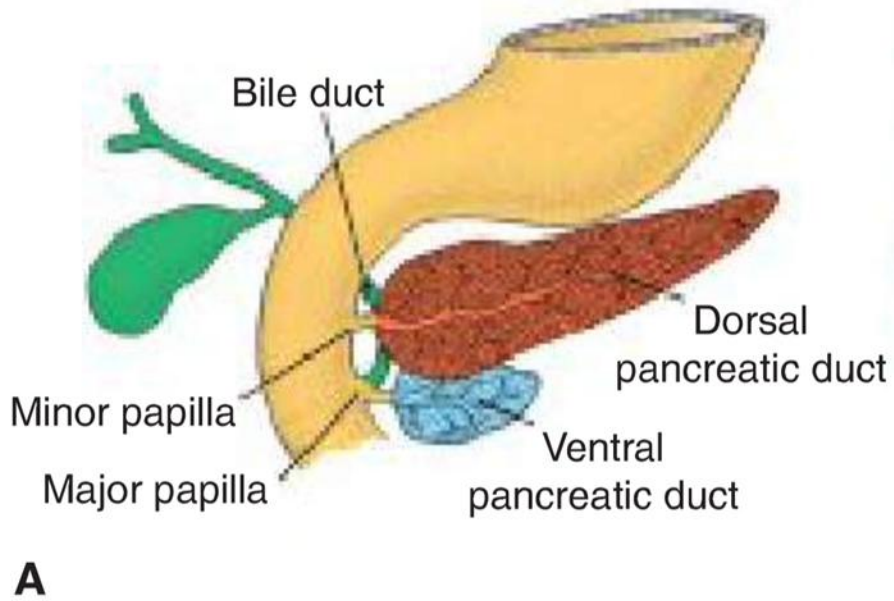
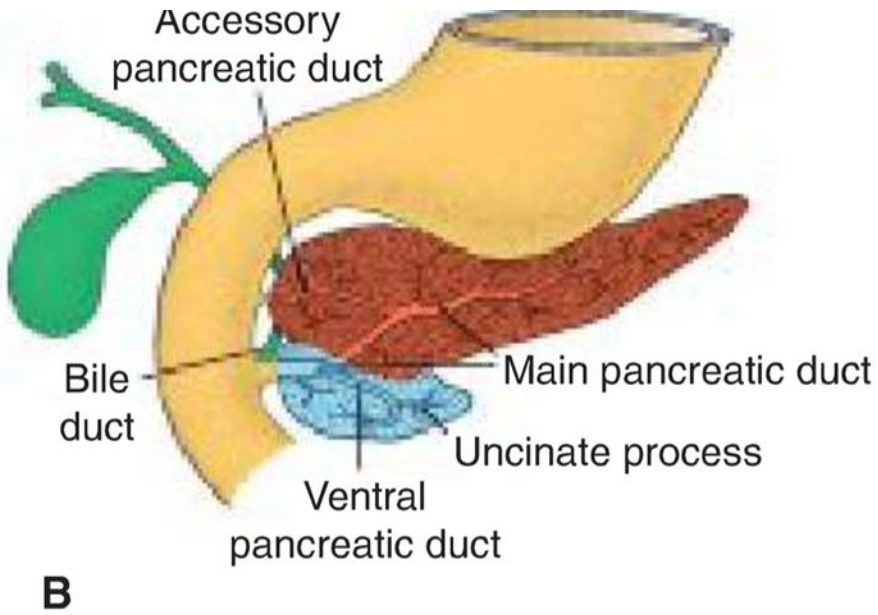
Development of the liver and pancreas









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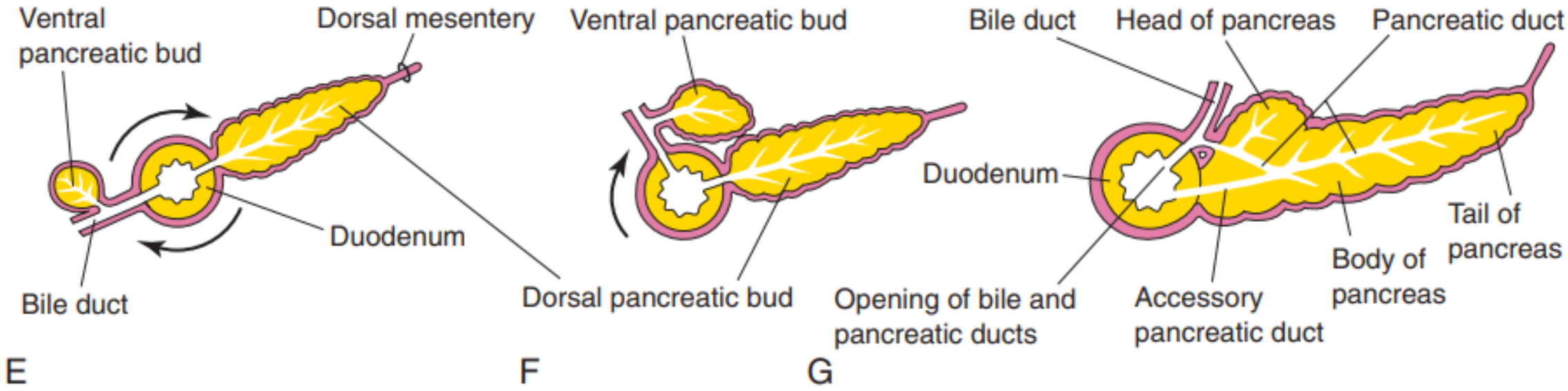
Development of the pancreas



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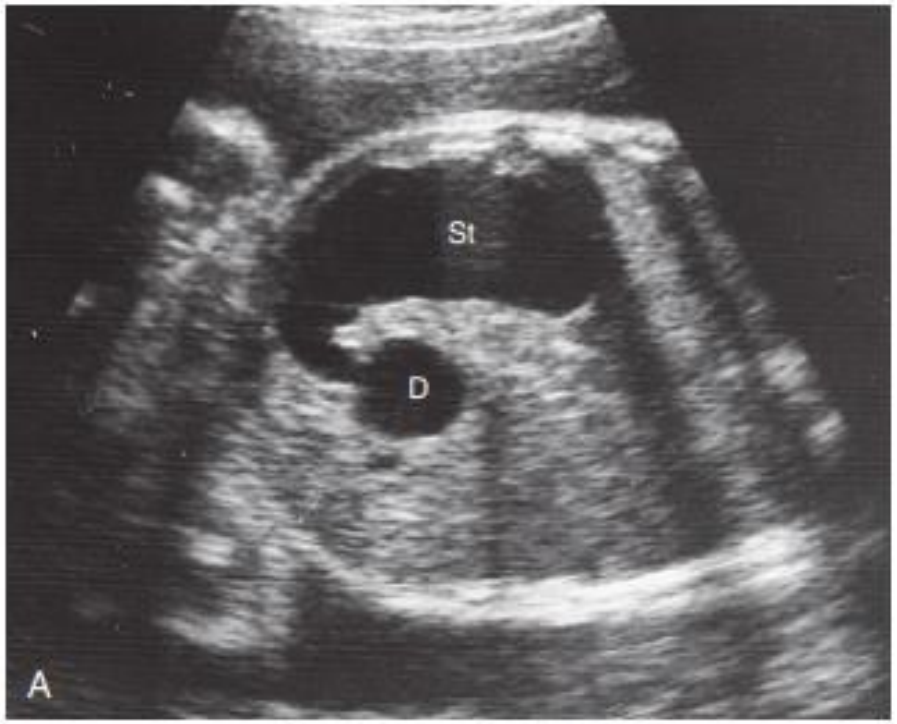

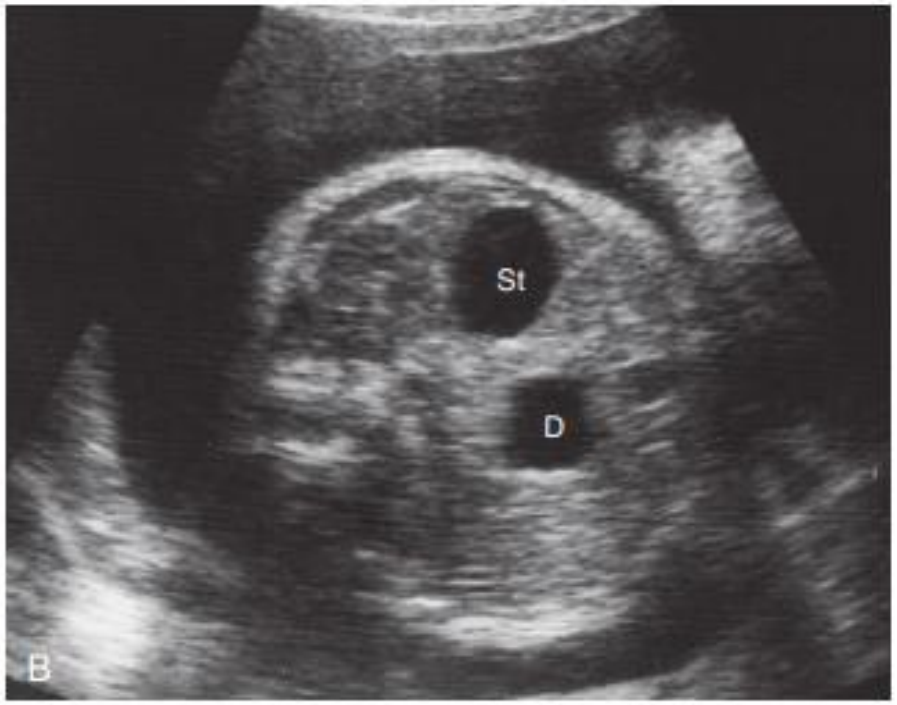

Duodenum:

- duodenal stenosis/atresia – results from incomplete recanalization; affects 20-30% of patients with Down syndrome, 20% of premature neonates

symptoms: polyhydramnios

„Doble-Bubble“ = stomach and proximal duodenum

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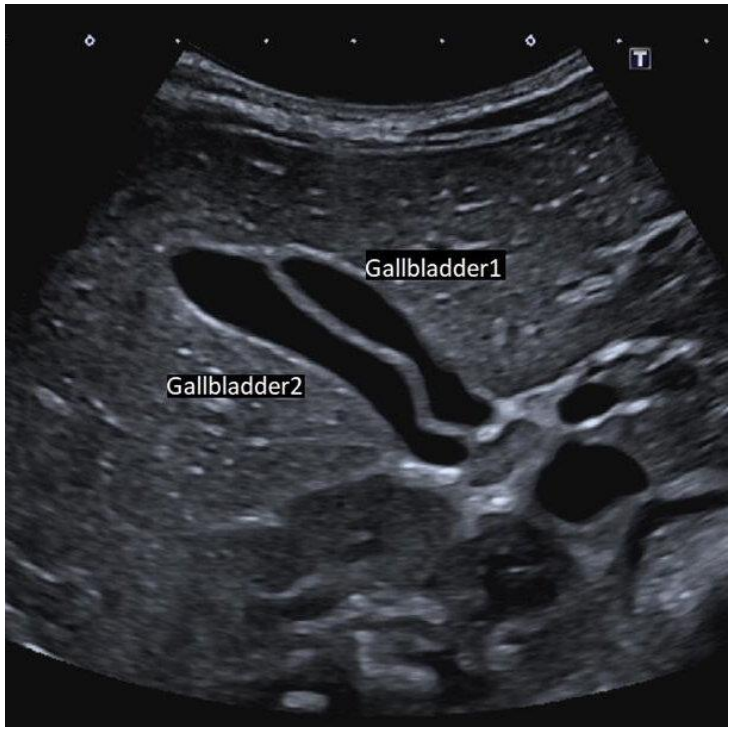
Liver - birth deffects are rare:

Accessory hepatic ducts – usually asymptomatic, in 5% of population

Gallbladder duplication - usually asymptomatic

Intrahepatic biliary duct atresia/hypoplasia (1/100000)

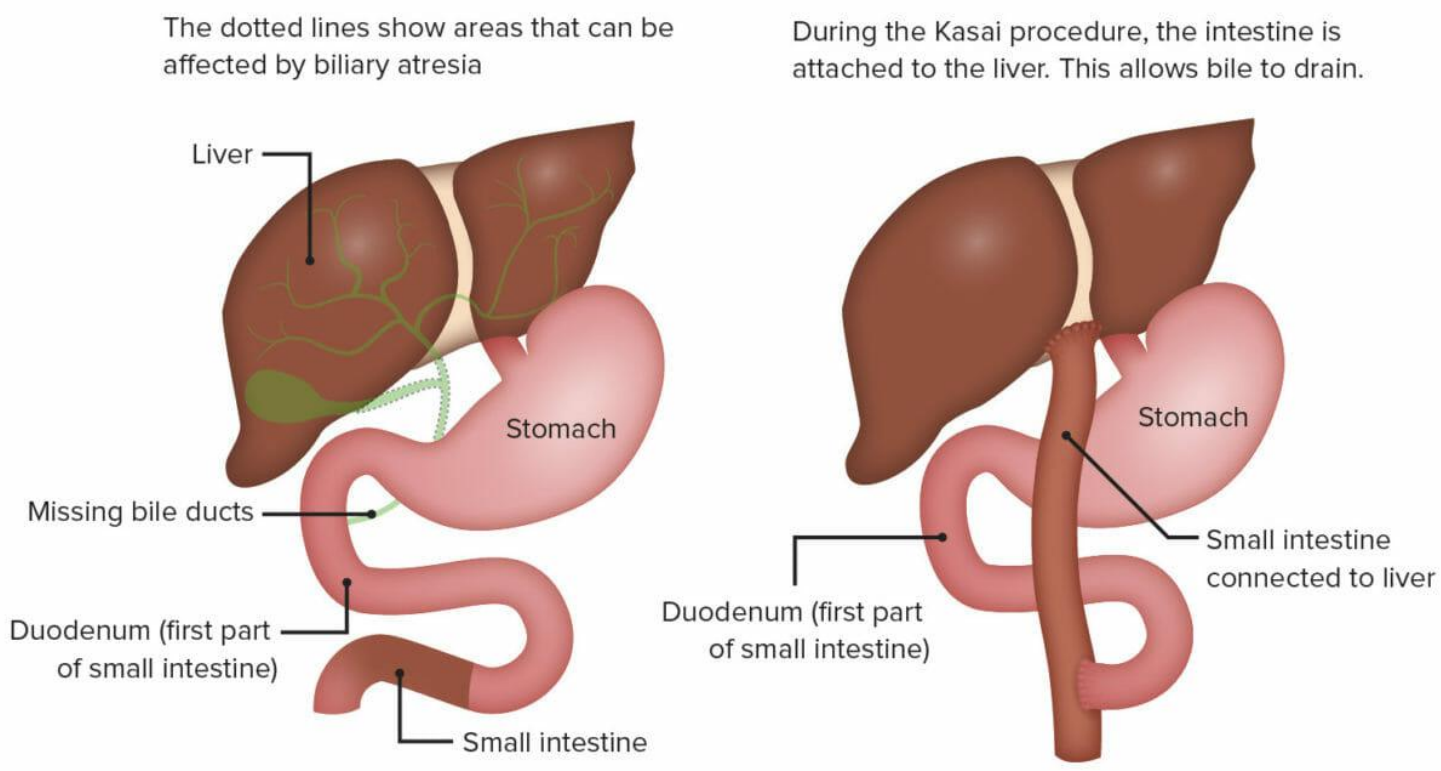


https://www.researchgate.net/publication/342134005_Prenatal_ultrasound_diagnosis_of_duplication_gallbladder_a_multicenter_study

Extrahepatic biliary atresia (1/15000 in US, however, higher rates in East Asia) –

15-20% has a potent proximal duct and fixable defect, the rest requires the liver transplant; *symptoms:* neonatal jaundice;

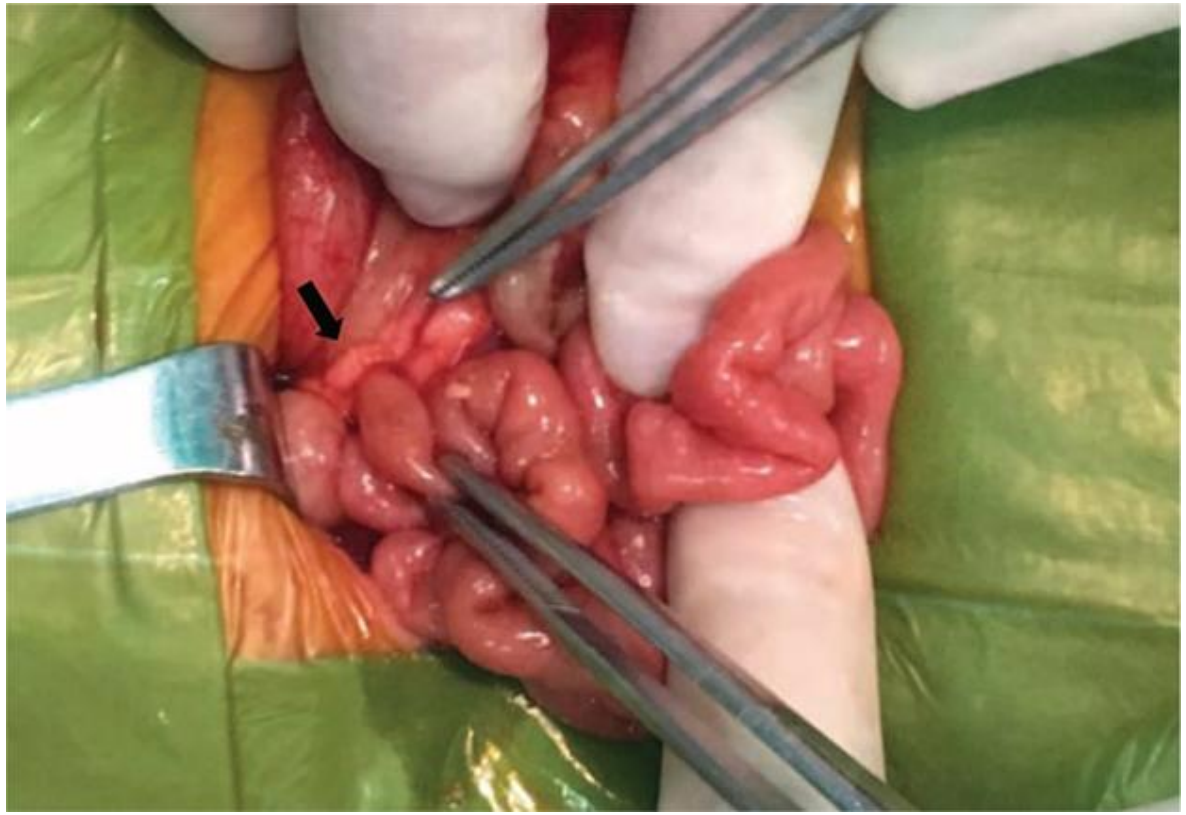
Kasai procedure (hepatoportoenterostomy) → liver transplant!!!



Colestase em Recém-Nascidos e Pequenos Lactentes | Concise Medical Knowledge

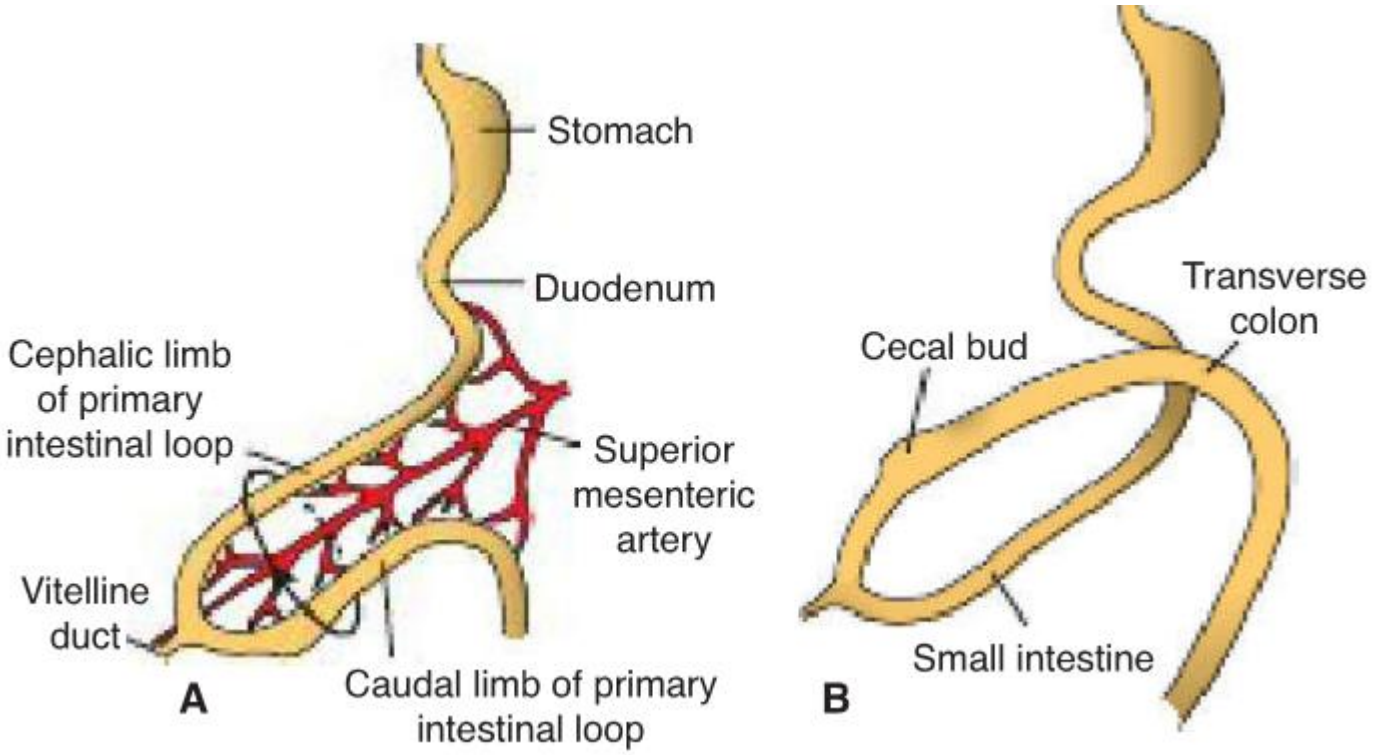
Pancreas:

Annular pancreas
Accessory pancreatic tissue
Accessory spleens – in 10 % of population



A newborn patient with both annular pancreas and Meckel's di... : Medicine (lww.com)

Midgut development. Physiological herniation.



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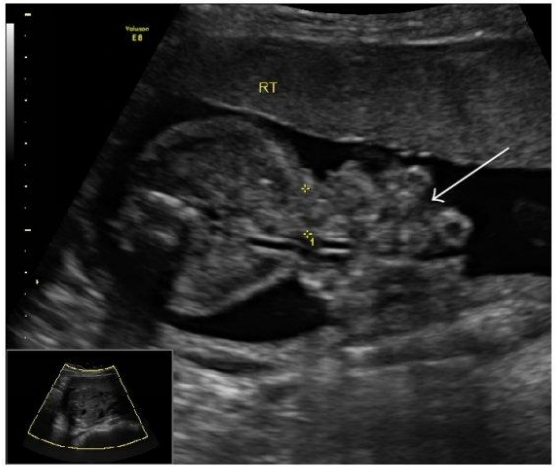
Midgut development. Physiological herniation.

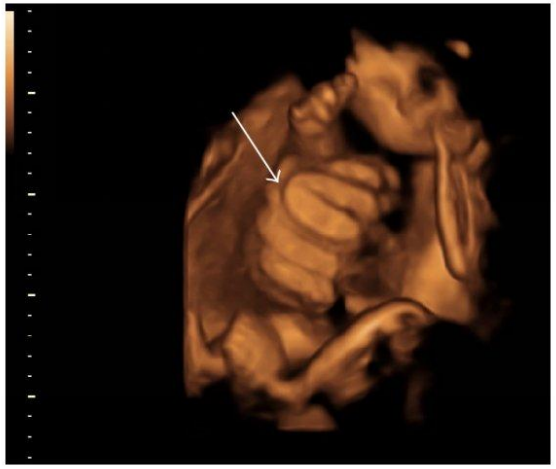
6th – 10th week

https://youtu.be/AscKR_cQExY

Body wall defects

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

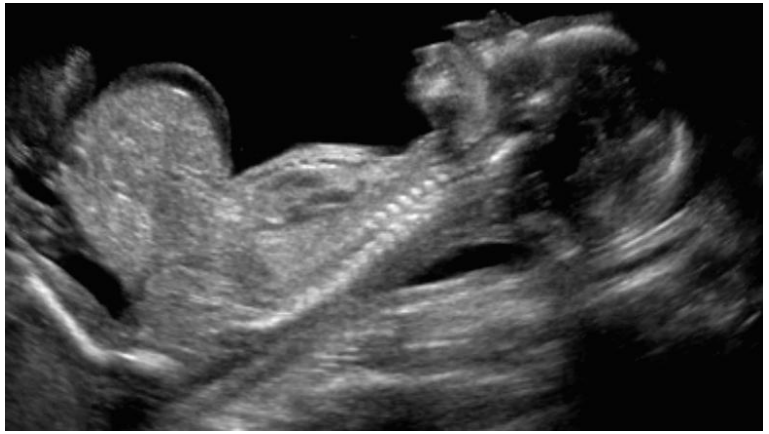


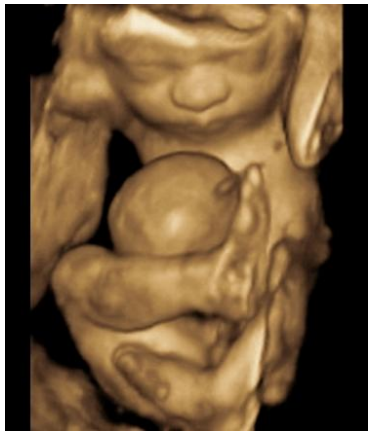


https://www.researchgate.net/publication/270909178_Gastroschisis_Antenatal_Sonographic_Predictors_of_Adverse_Neonatal_Outcome

Omphalocele (2,5/10000) –

up to 25 % mortality rate





Omphalocele | Children's Hospital of Philadelphia (chop.edu)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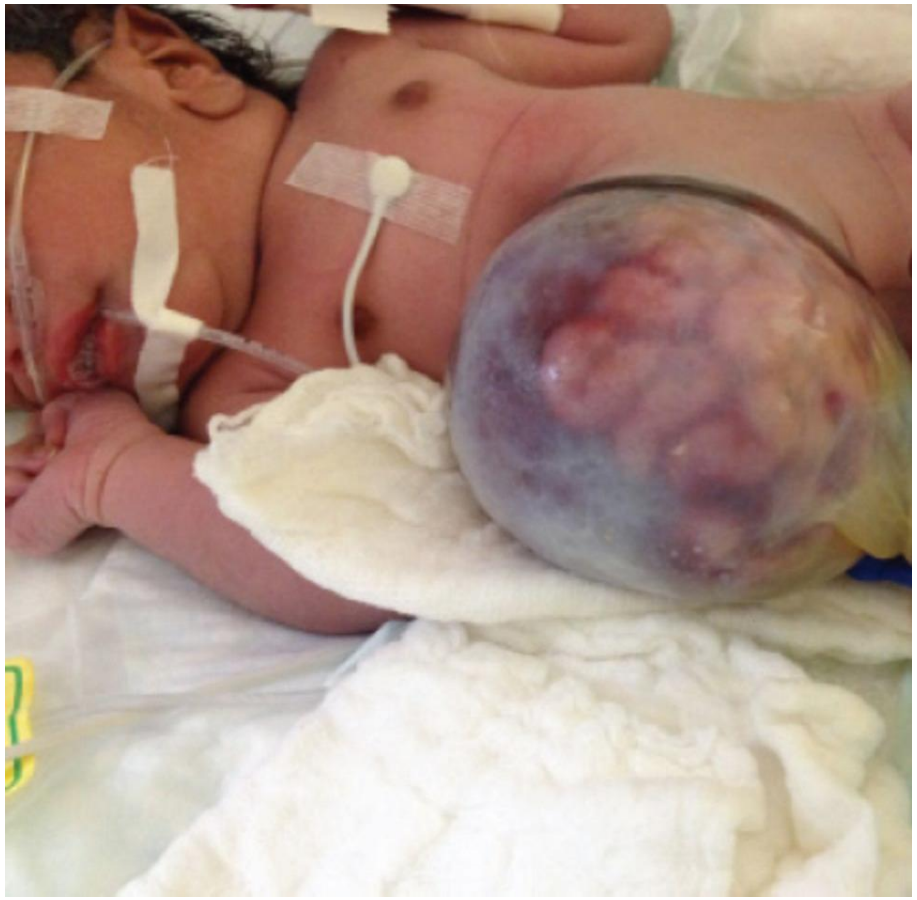
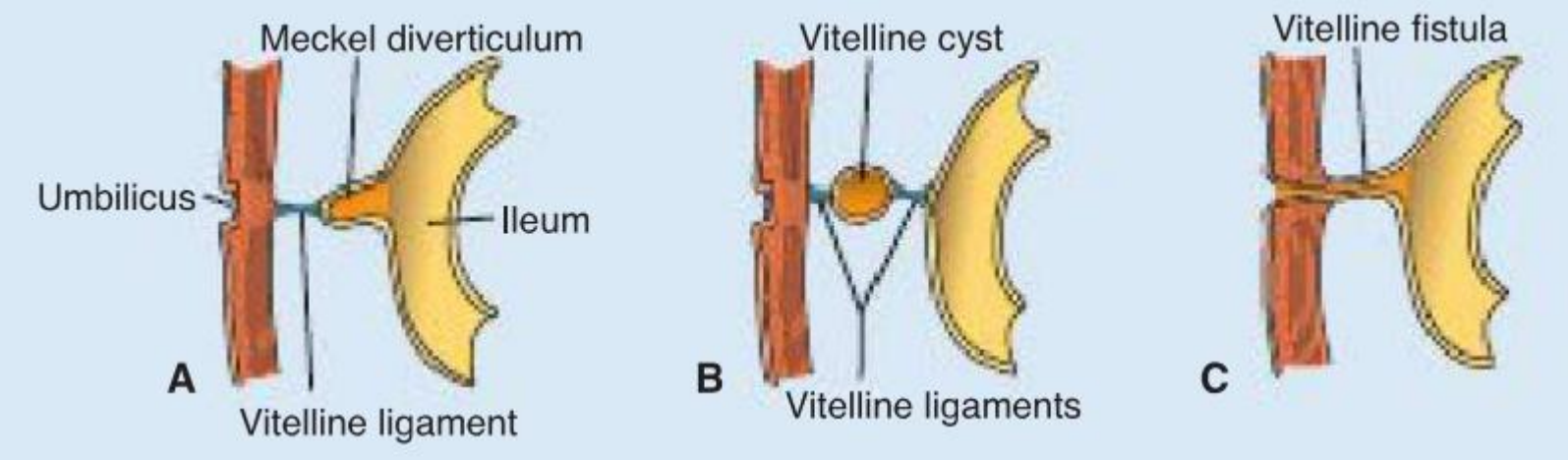


Vitelline duct abnormalities

Meckel or ileal diverticulum – in 2 to 4 % of people, 3-5 times more prevalent in males

Enterocystoma or vitelline cyst

Umbilical or vitelline fistula



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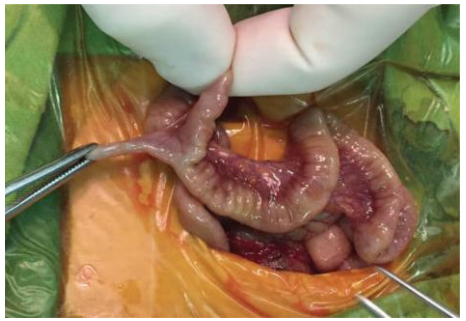


A newborn patient with both annular pancreas and Meckel's di... : Medicine (lww.com)

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

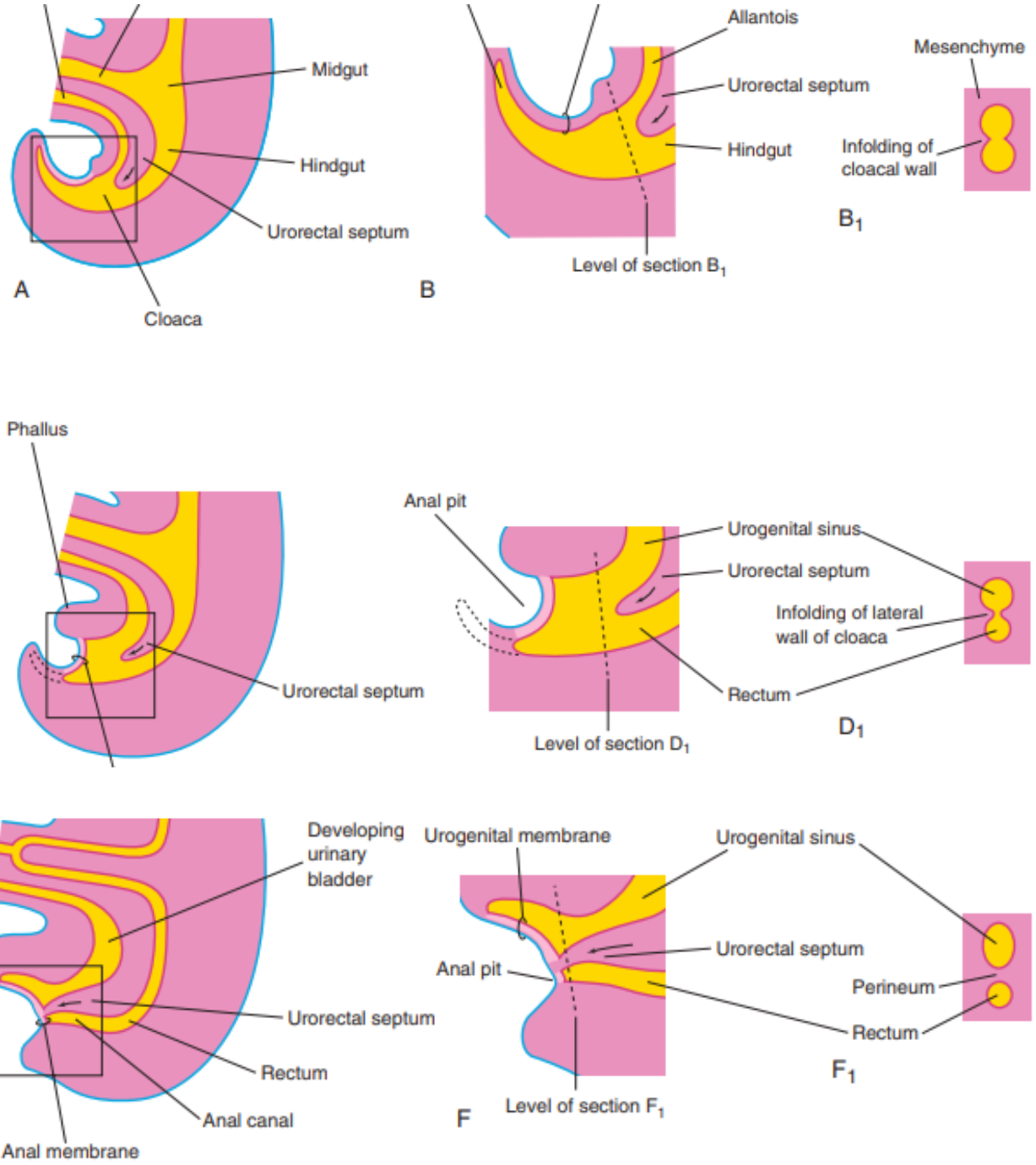
- Most occur in duodenum, fewest in the colon, equal number in jejunum and ileum; in 50 % of cases a region of bowel is missing completely, in 20 % cases the fibrous cord is present; stenoses represent only 5 % of cases

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!!!



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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 ends superior or inferior to the *puborectalis* muscle respectively)

Low: anal agenesis, with or without fistula

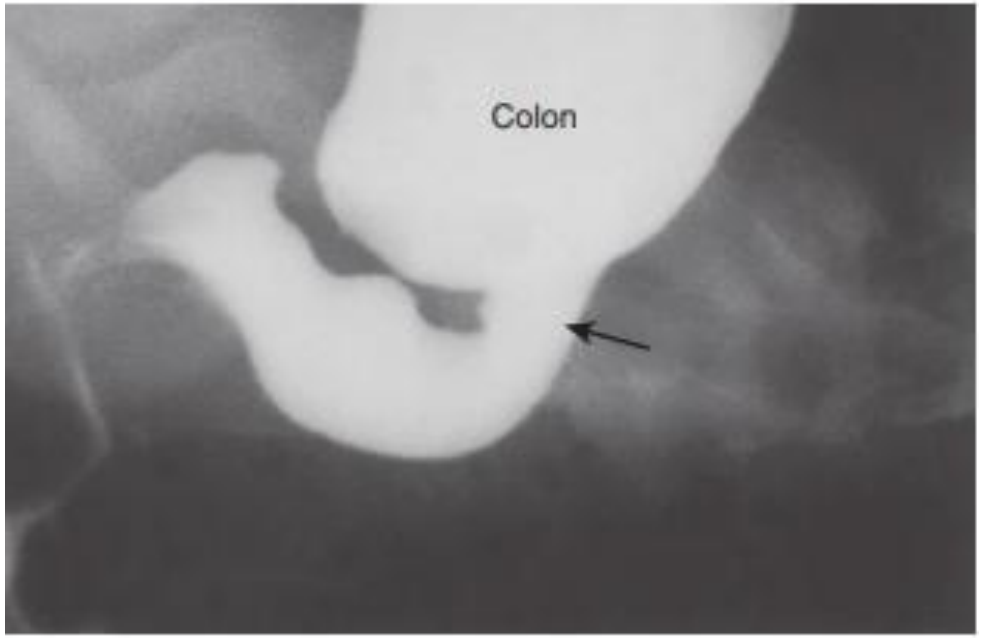
anal stenosis

membranous atresia of anus

High: anorectal agenesis, with or without

fistula (2/3 of anorectal defects)

rectal atresia



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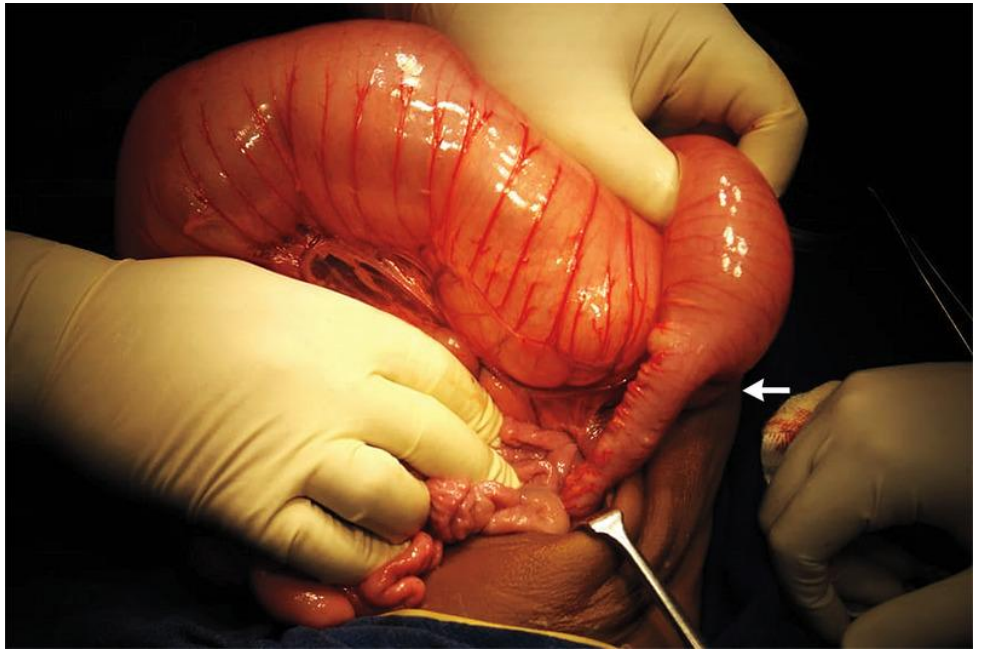
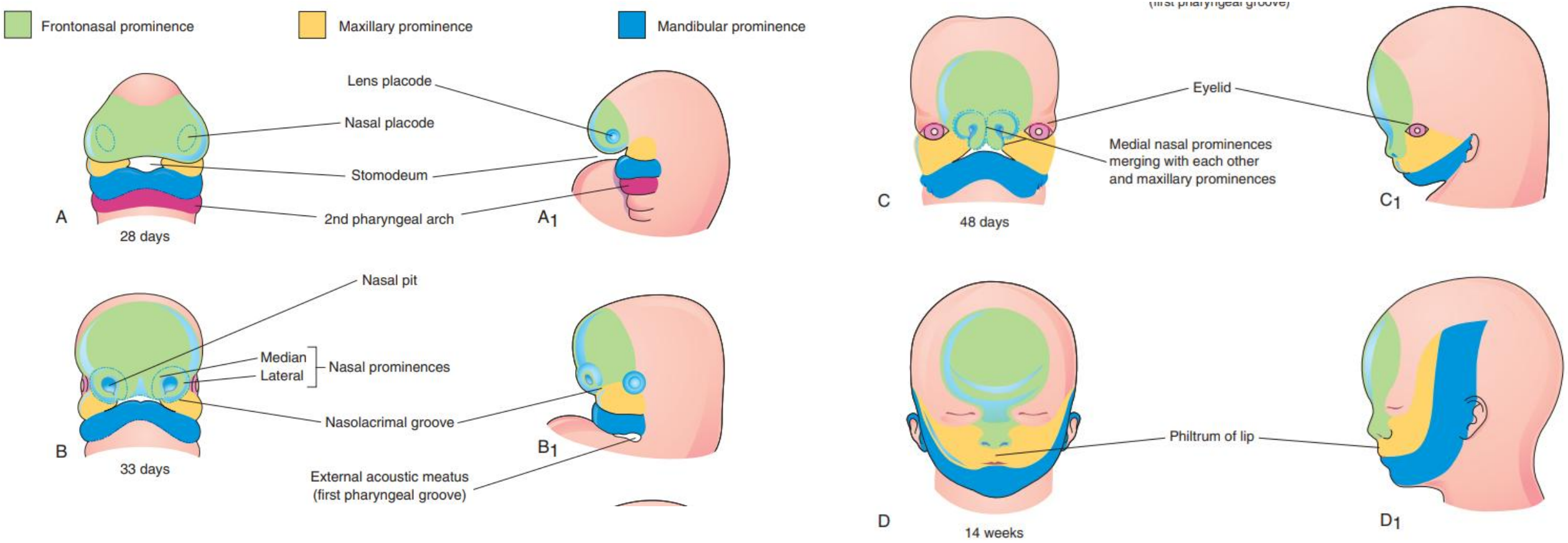


Transition Zone in Hirschsprung's Disease | NEJM

Development of the face



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

Anterior cleft deformities

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

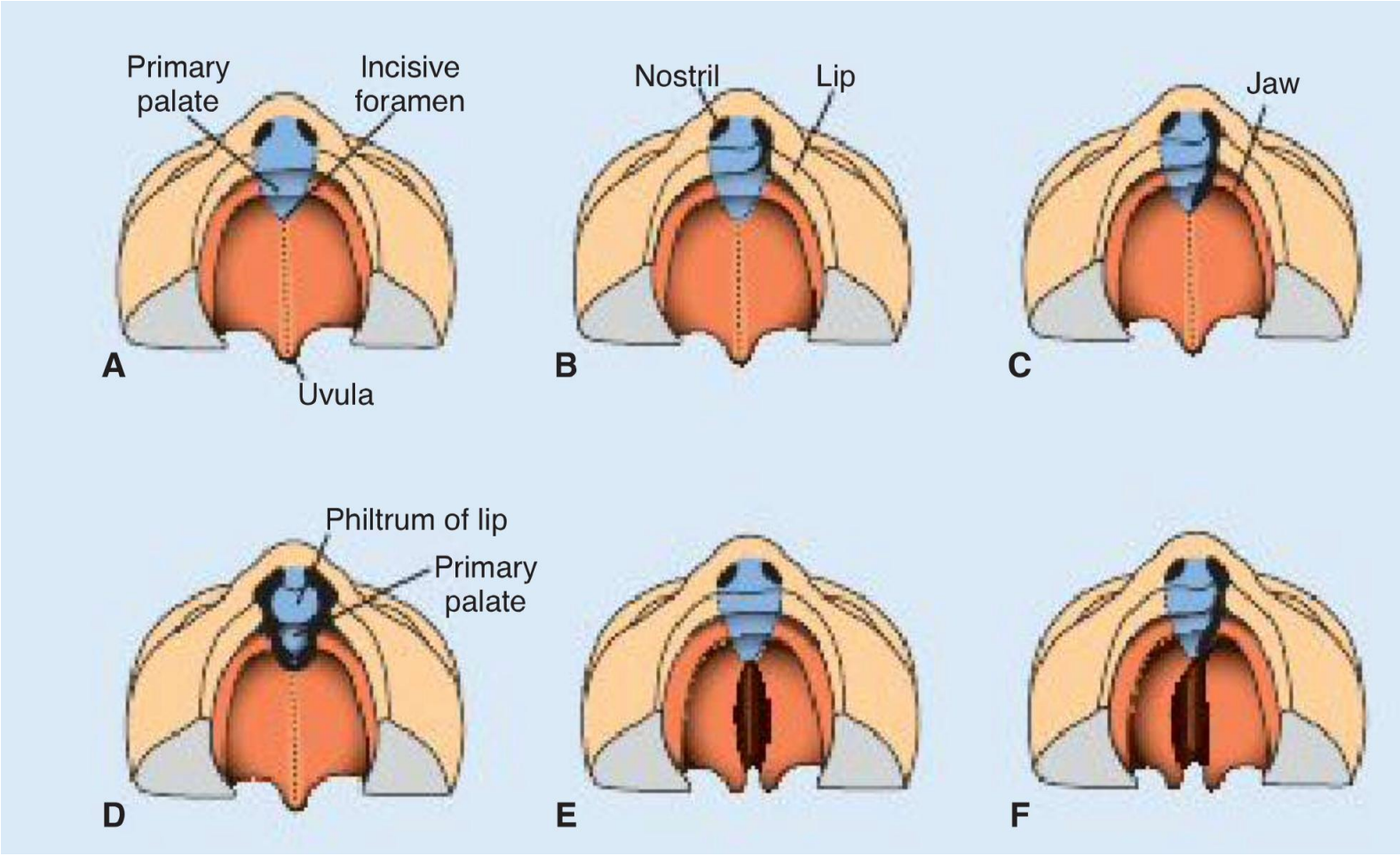
Cleft upper jaw

Cleft between the primary and the secondary palates

Posterior cleft deformities

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

Cleft uvula



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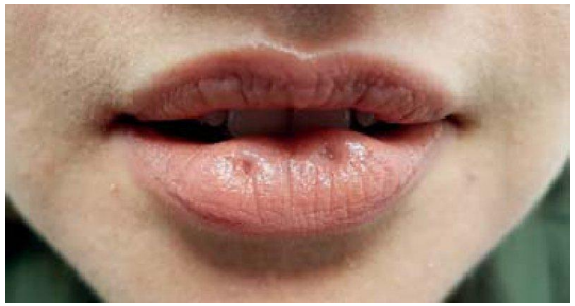
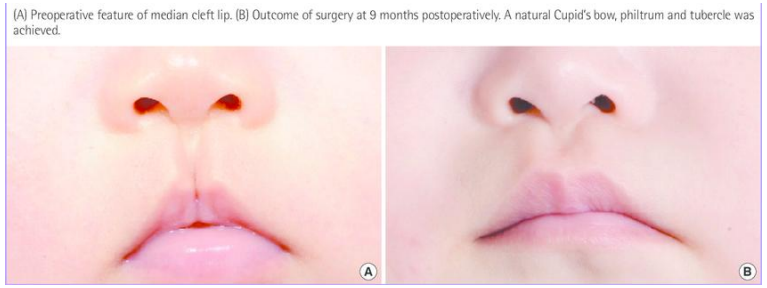
Van der Woude syndrome – pits in the lower lip in 88 % of patients

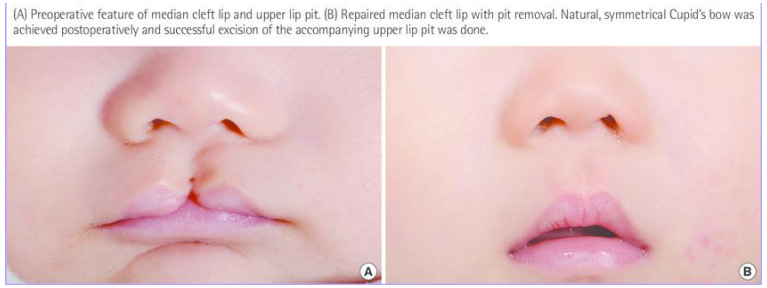


Van der Woude Syndrome (30.10.2020) (aerzteblatt.de)

Oblique facial cleft

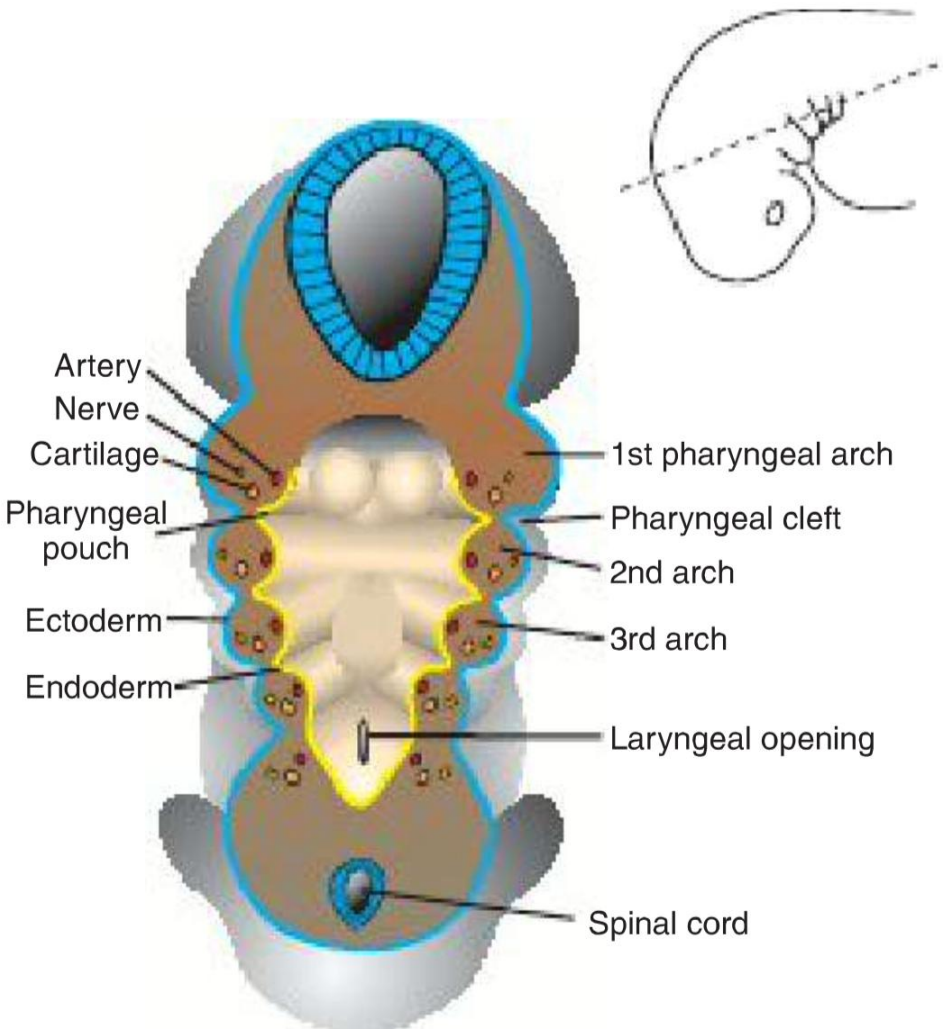
Median cleft lip – incomplete merging of the two medial nasal prominences; different degrees of midline structures loss --------- holoprosencephaly – fusion of lateral ventricles, synophtalmia

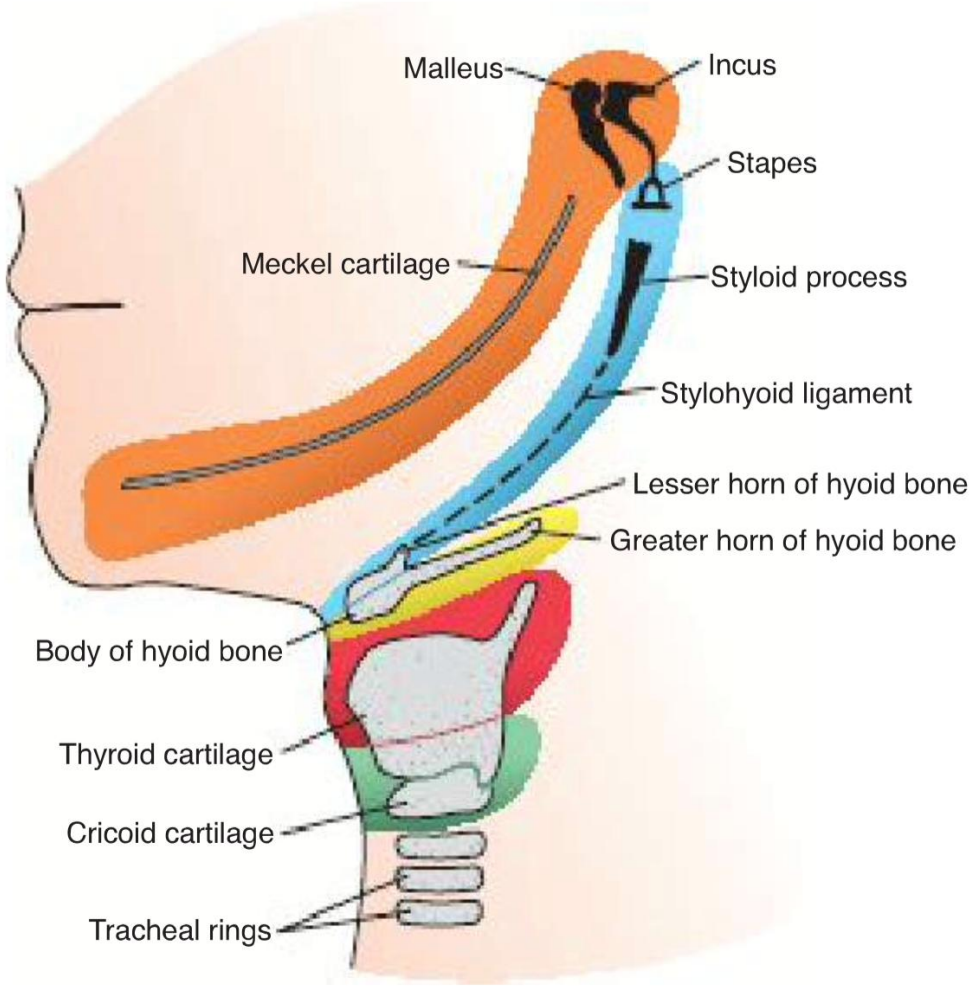




https://www.researchgate.net/publication/303392213_Clinical_Features_and_Management_of_a_Median_Cleft_Lip

Neck region





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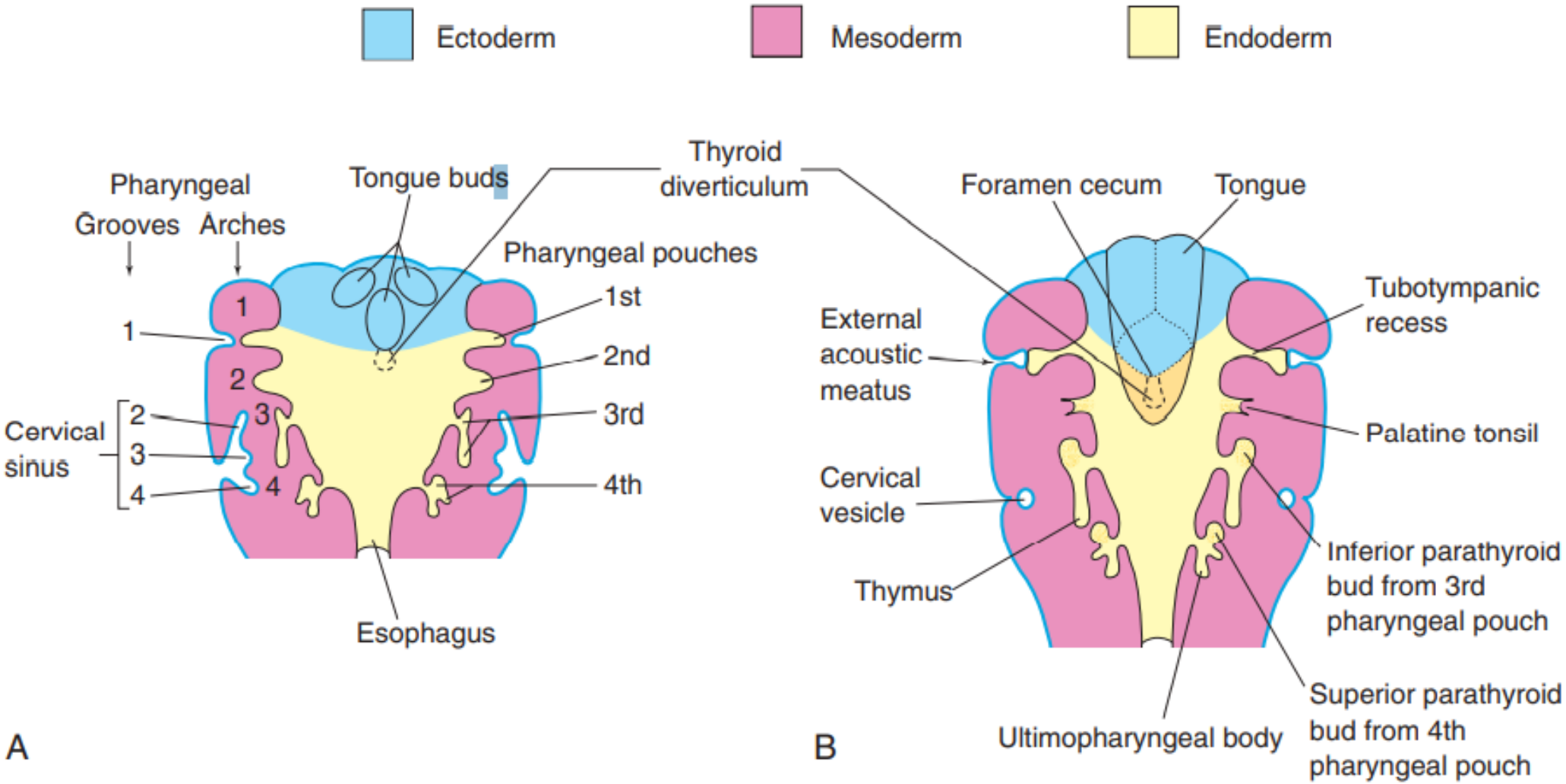
Neck region – pharyngeal apparatus



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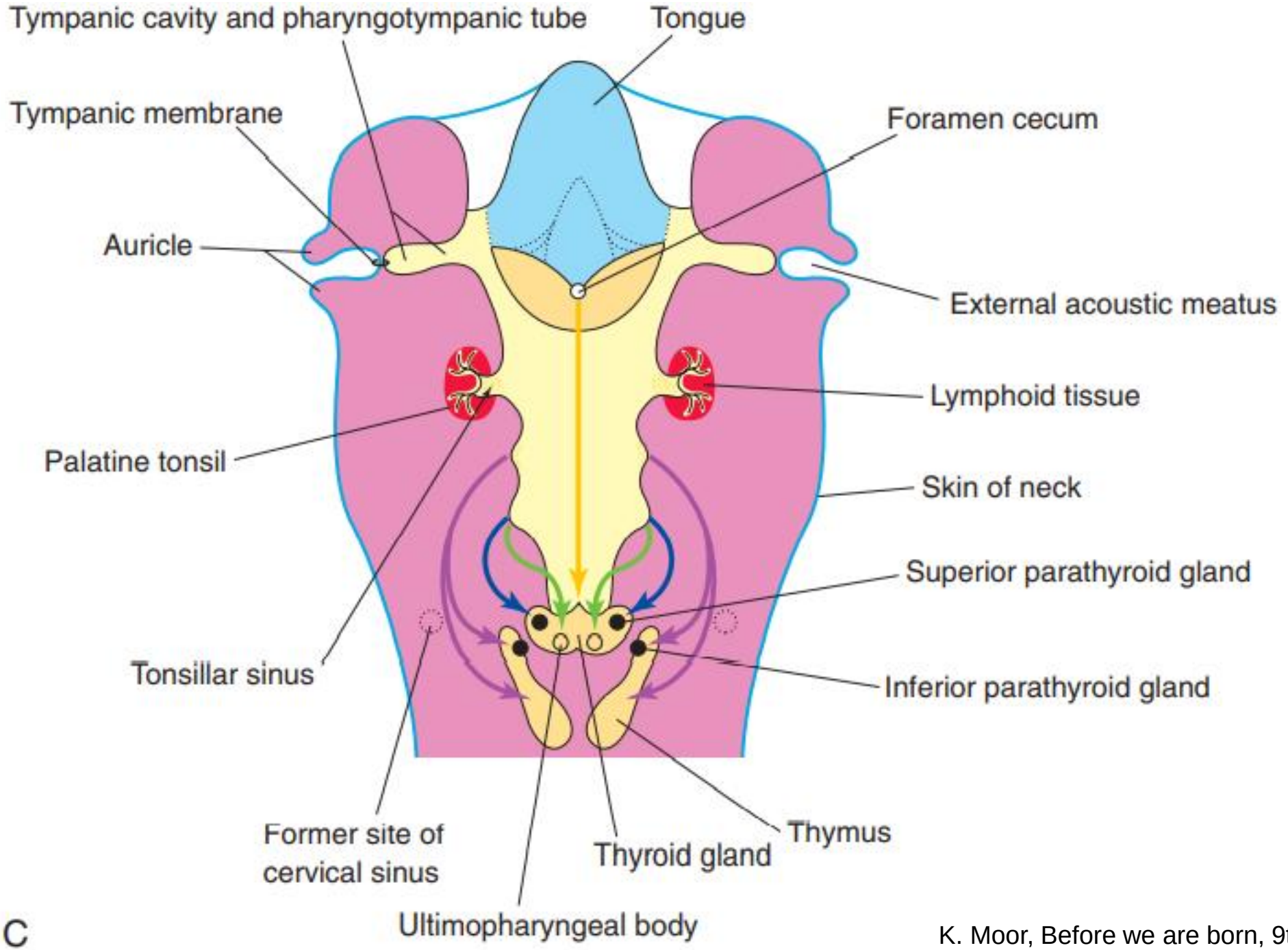
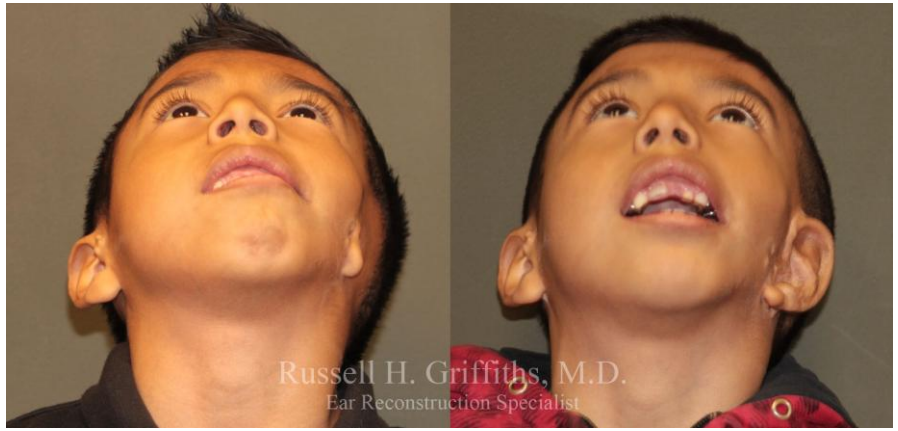


K. Moor, Before we are born, 9th edition

Craniofacial defects associated with neural crest cells

- Mandibulofacial dysostosis – Treacher Collins syndrome: autosomal dominant, 1/50000
- Robin sequence: first-arch structures, 1/8500 – micrognatia, cleft palate and glossoptosis
- 22q11.2 deletion syndromes: DiGeorge syndrome, DiGeorge anomaly, velocardiofacial syndrome etc. - 1/4000
- Hemifacial microsomia (oculoauriculovertebral spectrum – Goldenhar syndrome) – 1/5600, assymetry in 65% cases; involves maxillary, temporal, zygomatic bones, ears, eyes, vertebrae. Cardiac deffects in 50% cases.

https://economictimes.indiatimes.com/magazines/panache/man-born-with-treacher-collins-syndrome-was-rejected-by-biological-parents-36-hrs-after-birth-for-his-face-but-his-adoptive-mom-didnt-care/articleshow/93541374.cms?from=mdr



Case 1 - One Stage Microtia Reconstruction for Hemifacial Microsomia - Before and After Gallery