Development and teratology of sensory organs

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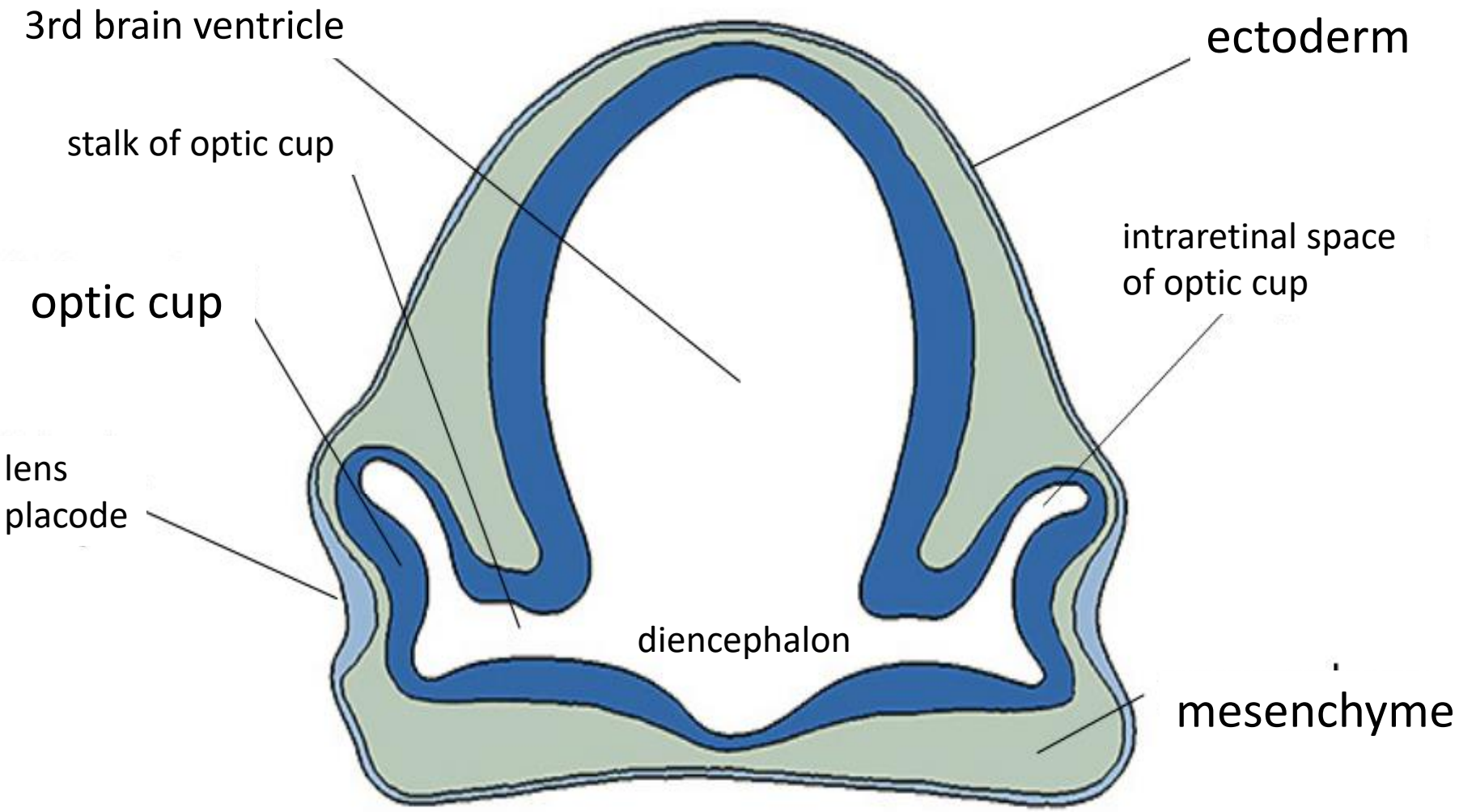
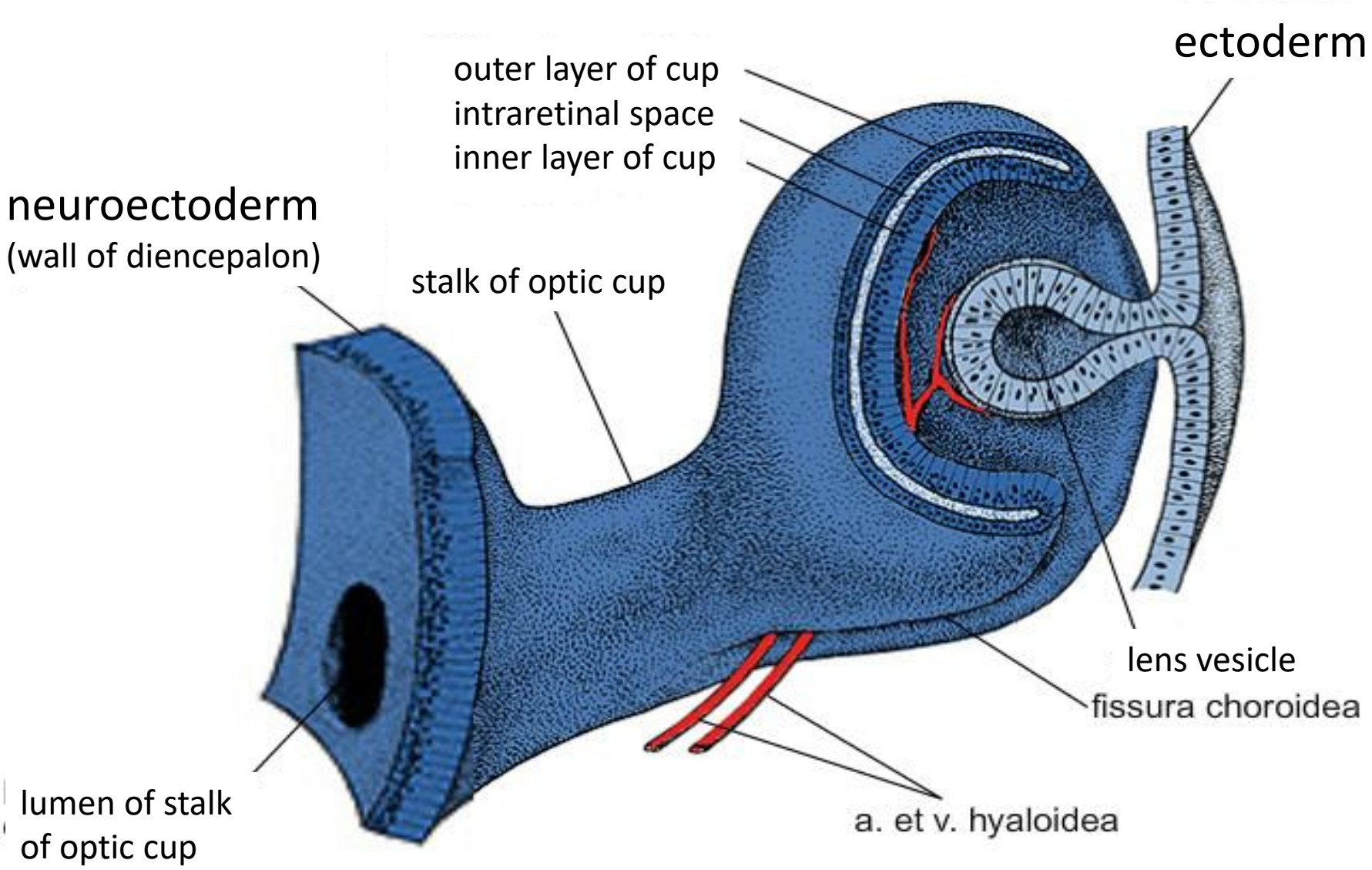
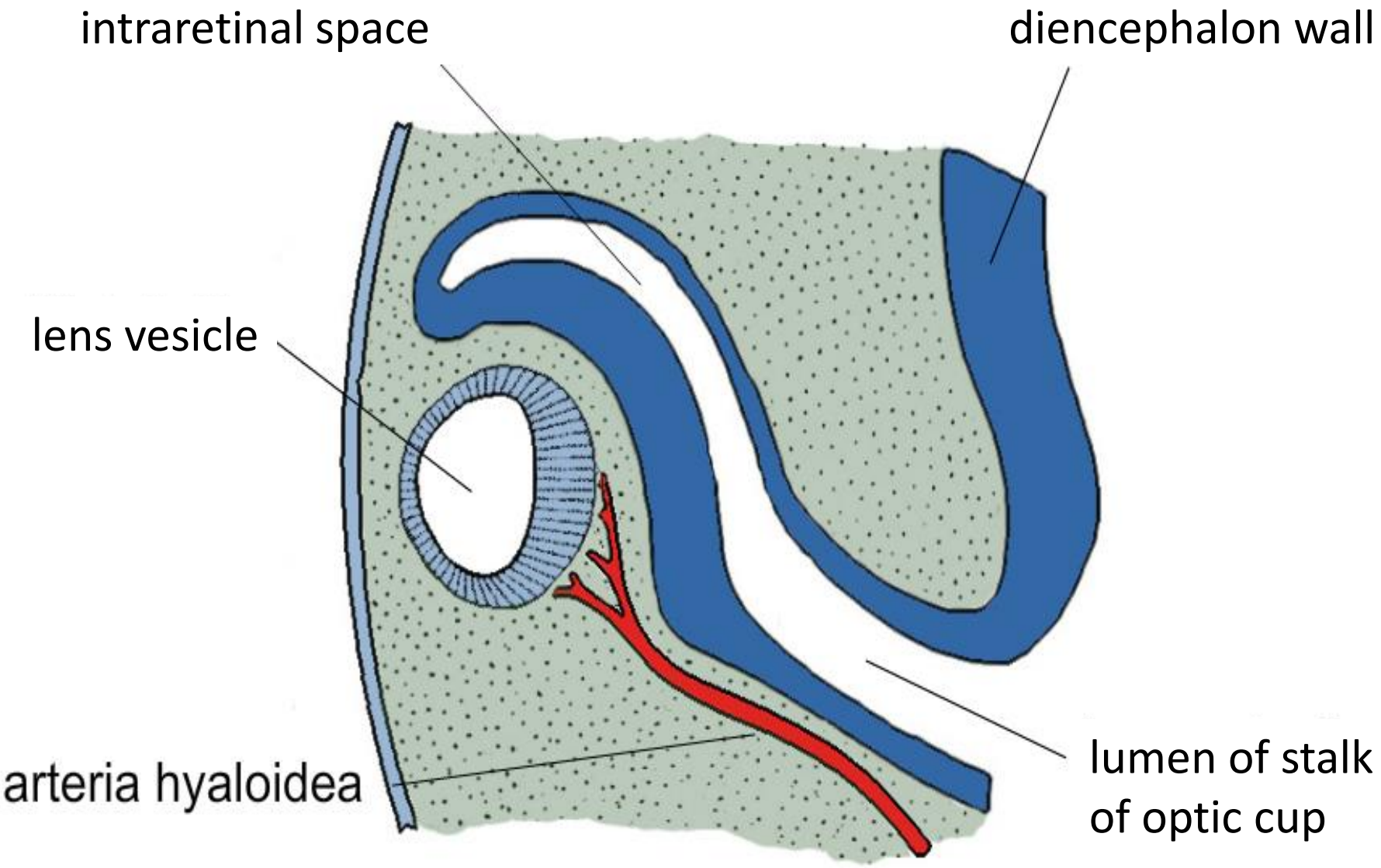
29.4.2023

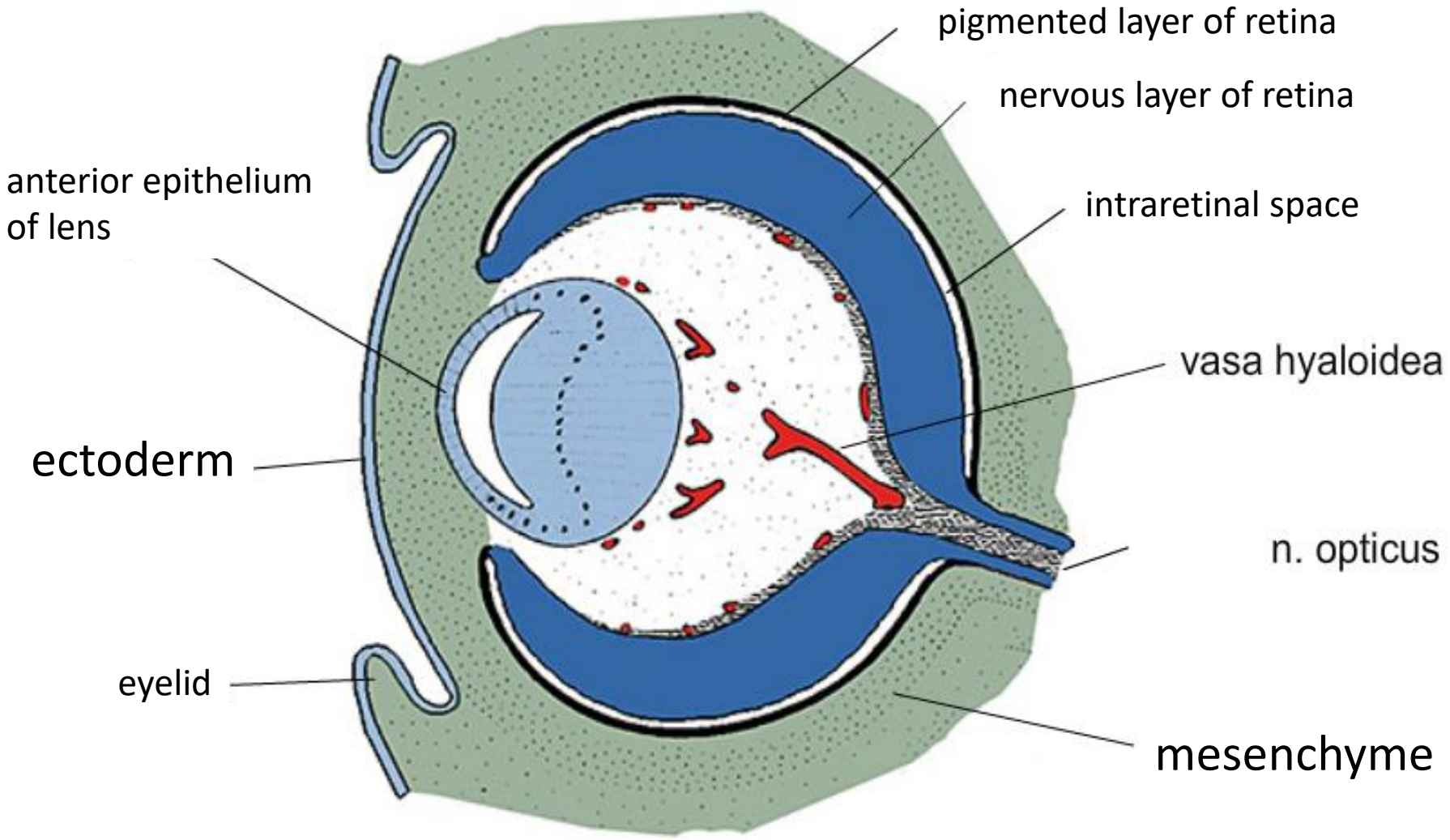
Development of the eye

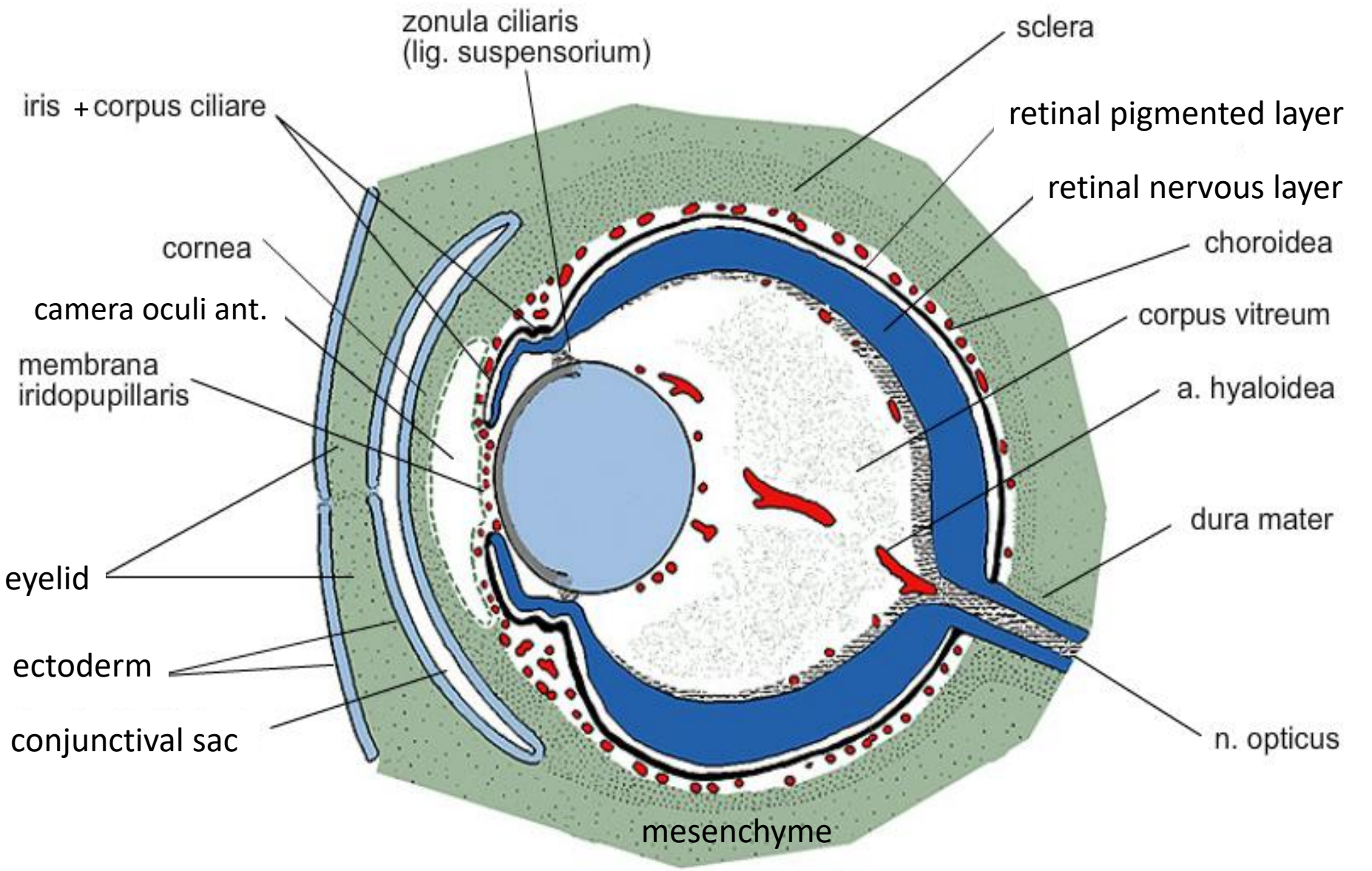


Development of the optic cup and the lens




intraretinal space
diencephalon wall
lens vesicle
arteria hyaloidea
lumen of stalk
of optic cup


pigmented layer of retina
nervous layer of retina
anterior epithelium of lens
intraretinal space
vasa hyaloidea
ectoderm
n. opticus
eyelid
mesenchyme

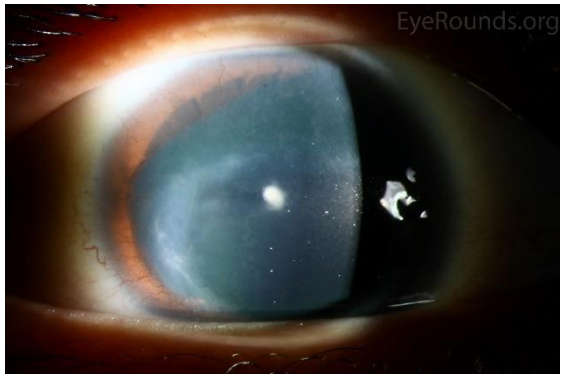

zonula ciliaris
(lig. suspensorium)
sclera
iris + corpus ciliare
retinal pigmented layer
retinal nervous layer
cornea
choroidea
camera oculi ant.
corpus vitreum
membrana
iridopupillaris
a. hyaloidea
dura mater
eyelid
ectoderm
conjunctival sac
n. opticus
mesenchyme

Eye abnormalities

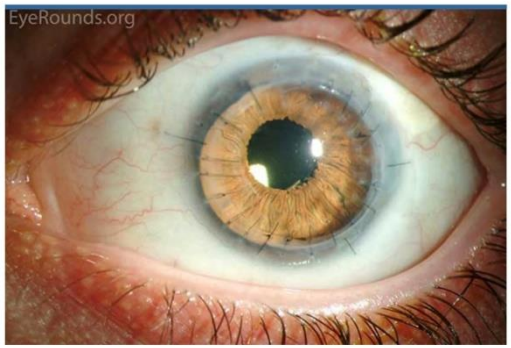
- Coloboma – in most cases coloboma iridis, frequently associated with other eye abnormalities; optic nerve coloboma is linked to PAX2 gene mutations – renal coloboma syndrome – autosomal dominant

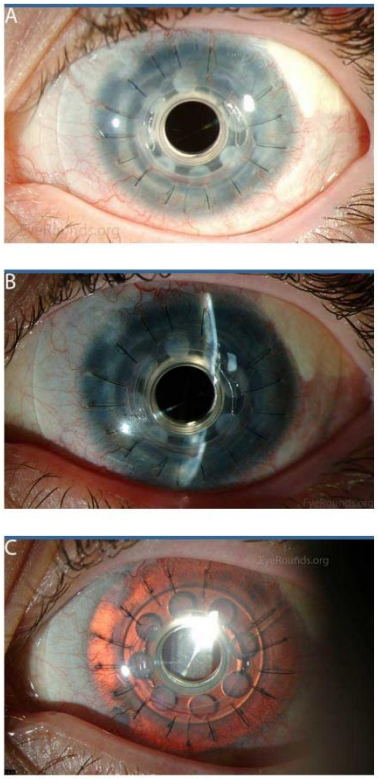
https://atrl.net/forums/topic/430624-girl-comes-forward-claiming-to-be-the-missing-madeleine-mccann/

- Congenital aphakia/aniridia – mutation in PAX6 results in aniridia; WAGR syndrome – microdeletion in chromosome 11 (PAX6 and WT1)



Fil:Aniridi2.jpg – JournalWiki (uio.no)



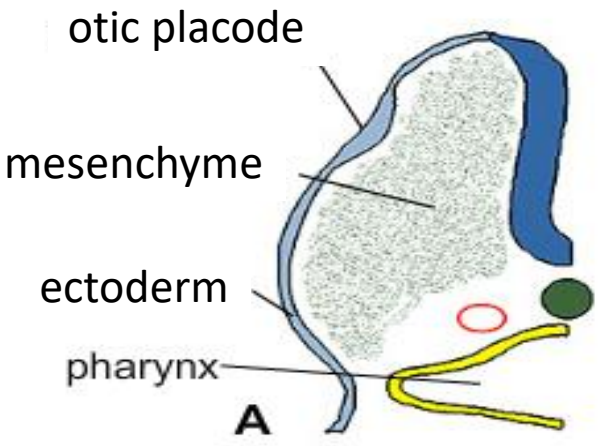


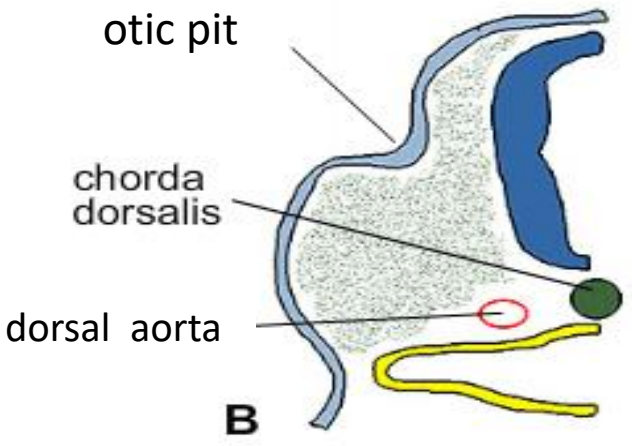
https://webeye.ophth.uiowa.edu/eyeforum/cases/211-Aniridia.htm#gsc.tab=0

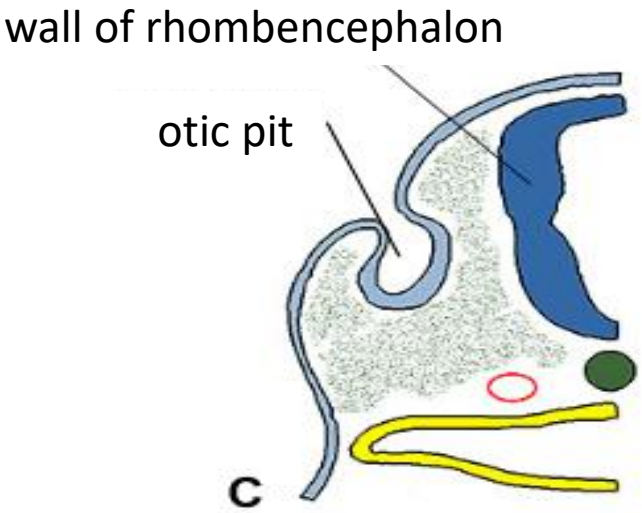
- Iridopupillary membrane
- Sclerocornea
- Peters anomaly - anterior segment dysgenesis
- Congenital cataracts – genetically determined or rubella virus!
- Persistent hyaloid artery (in 3 % of full term infants) – amblyopia, vitreous haemorrhage, retinal detachment

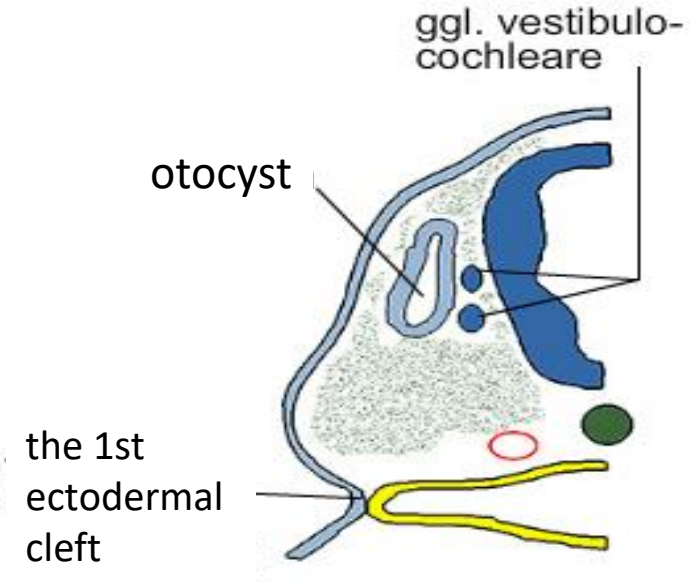
- Microphtalmia – 1,5-1,9 in 10000, cytomegalovirus, toxoplasmosis
- Anophtalmia – 0,18-0,4 in 10000
- Cyclopia and synophtalmia (loss of midline tissue - holoprosencephaly) – alcohol, maternal diabetes, mutations in SHH and abnormalities in cholesterol metabolism

Development of the inner ear

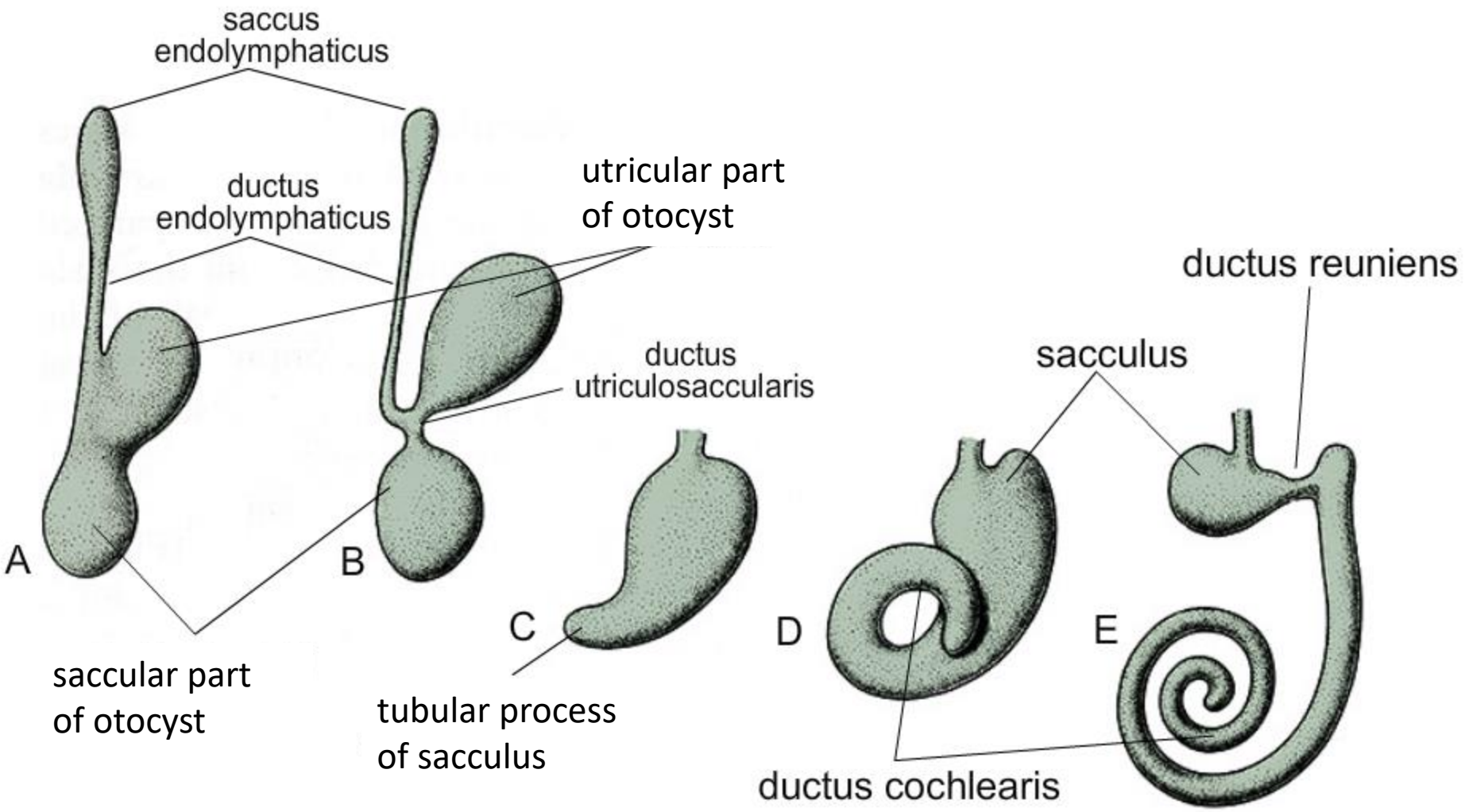






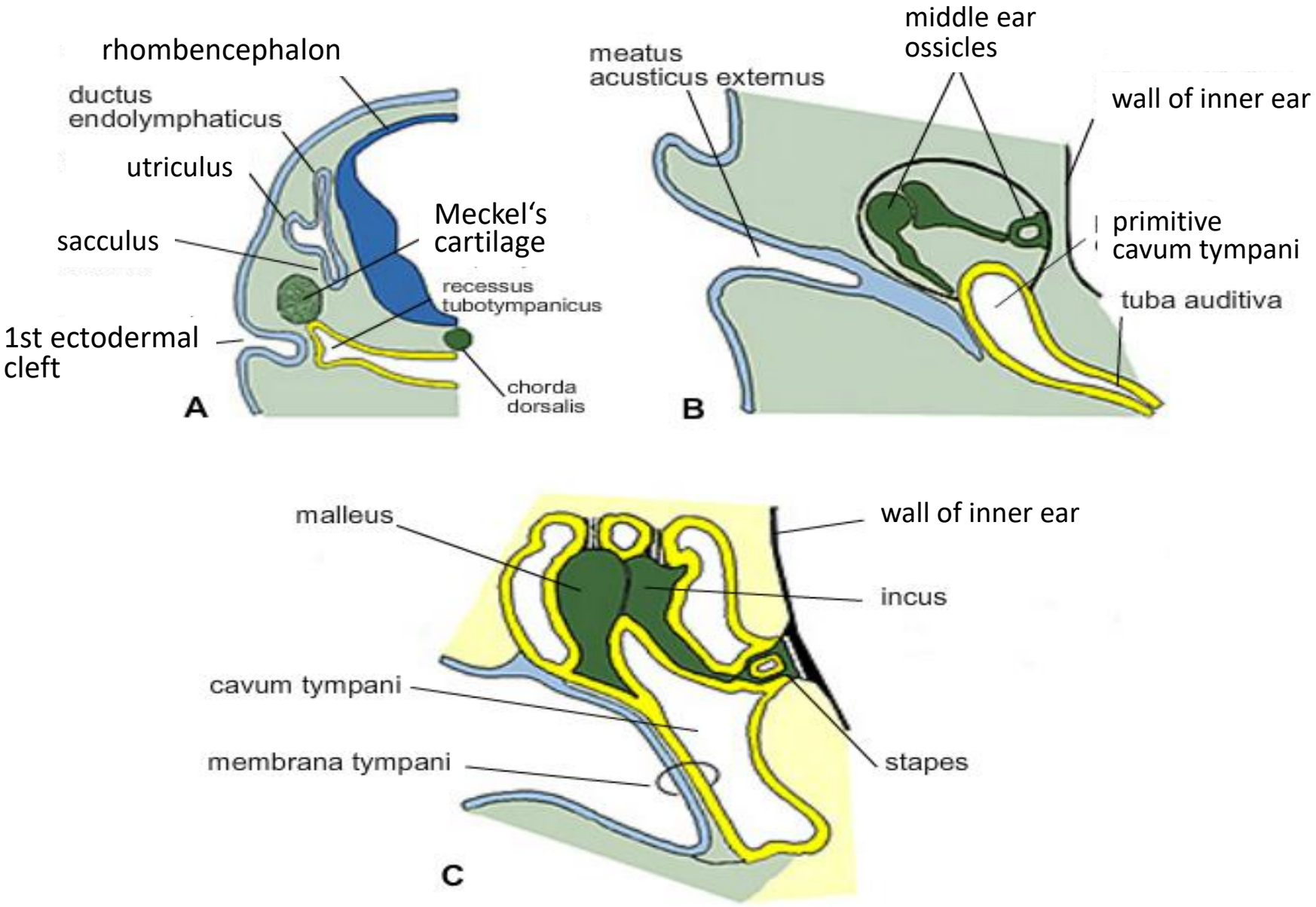


Development of the membranous labyrinth

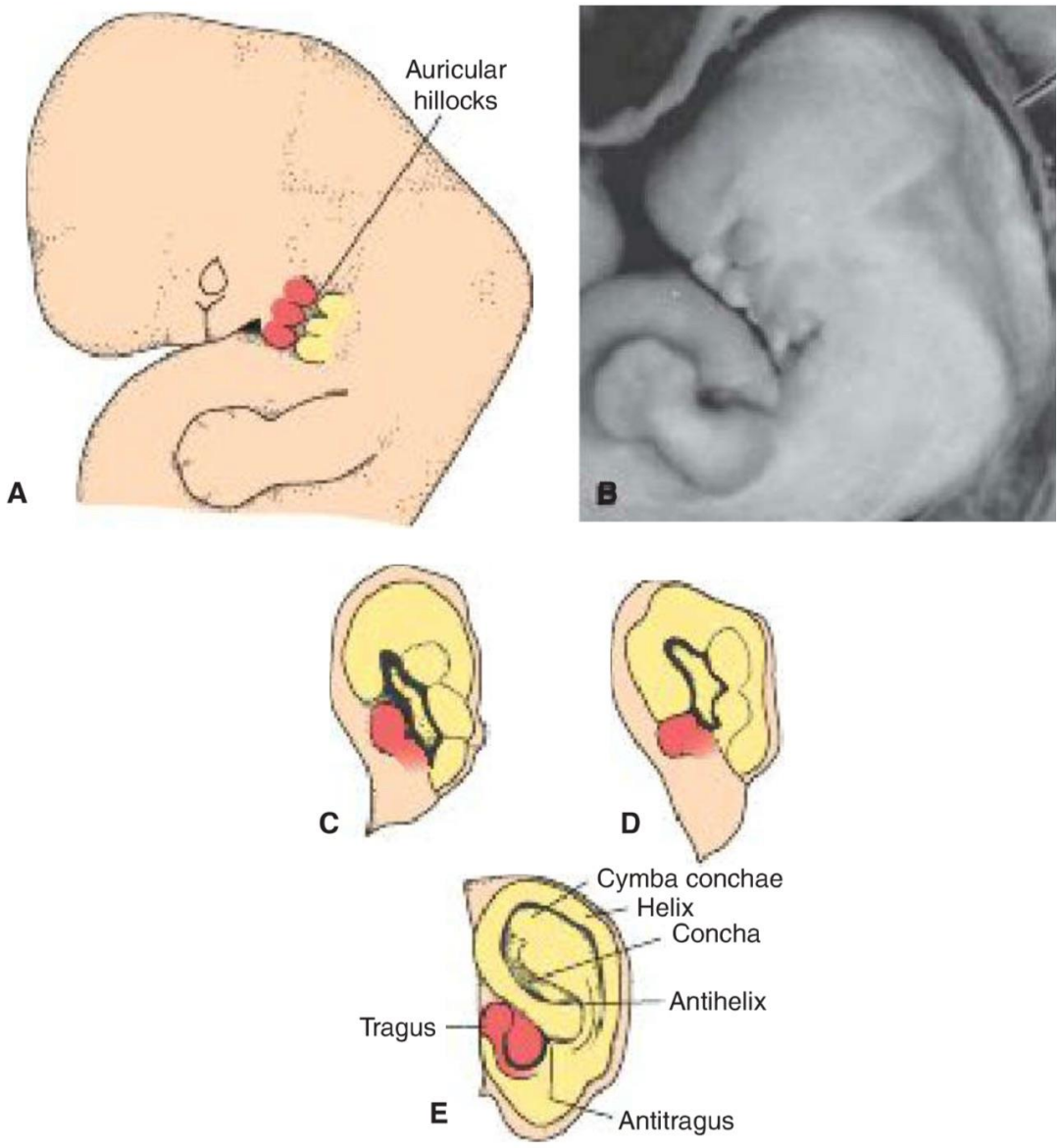


T. W. Sadler, Langman's medical embryology, 12th edition

Development of the middle ear



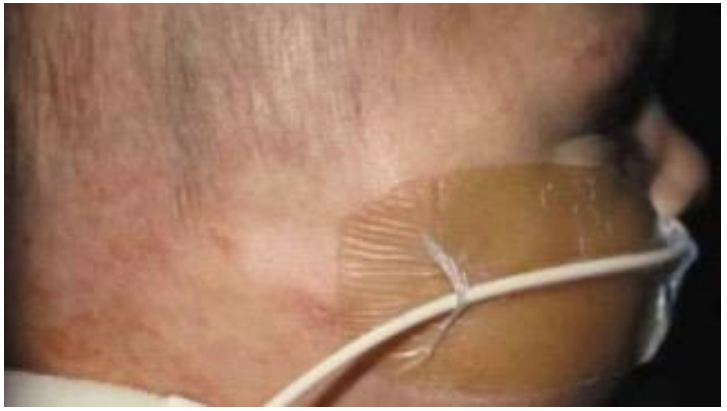
Development of the external ear



T. W. Sadler, Langman's medical embryology, 14th edition

External ear defects

- Anotia
- Microtia



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

- (1 in 6000-12000) usually one ear is affected, the external accustic meatus is absent or narrowed – conductive hearing loss

- Preauricular appendages and pits

All of the frequently occurring chromosomal syndromes and most of the less common have ear anomalies as one of their characteristics!!! (Langman's medical embryology, T. W. Sadler, 14th edition, p. 359)

Congenital hearing loss

Sensorineural (hair cells or auditory nerve ganglia) vs.

conductive (most commonly stapes)

- genetic factors: 50% cases (e.g. Treacher Collins syndrome, Down syndrome)
- perinatal infections: rubella virus, cytomegalovirus, herpes simplex
- prematurity
- maternal diabetes
- isotretinoin!