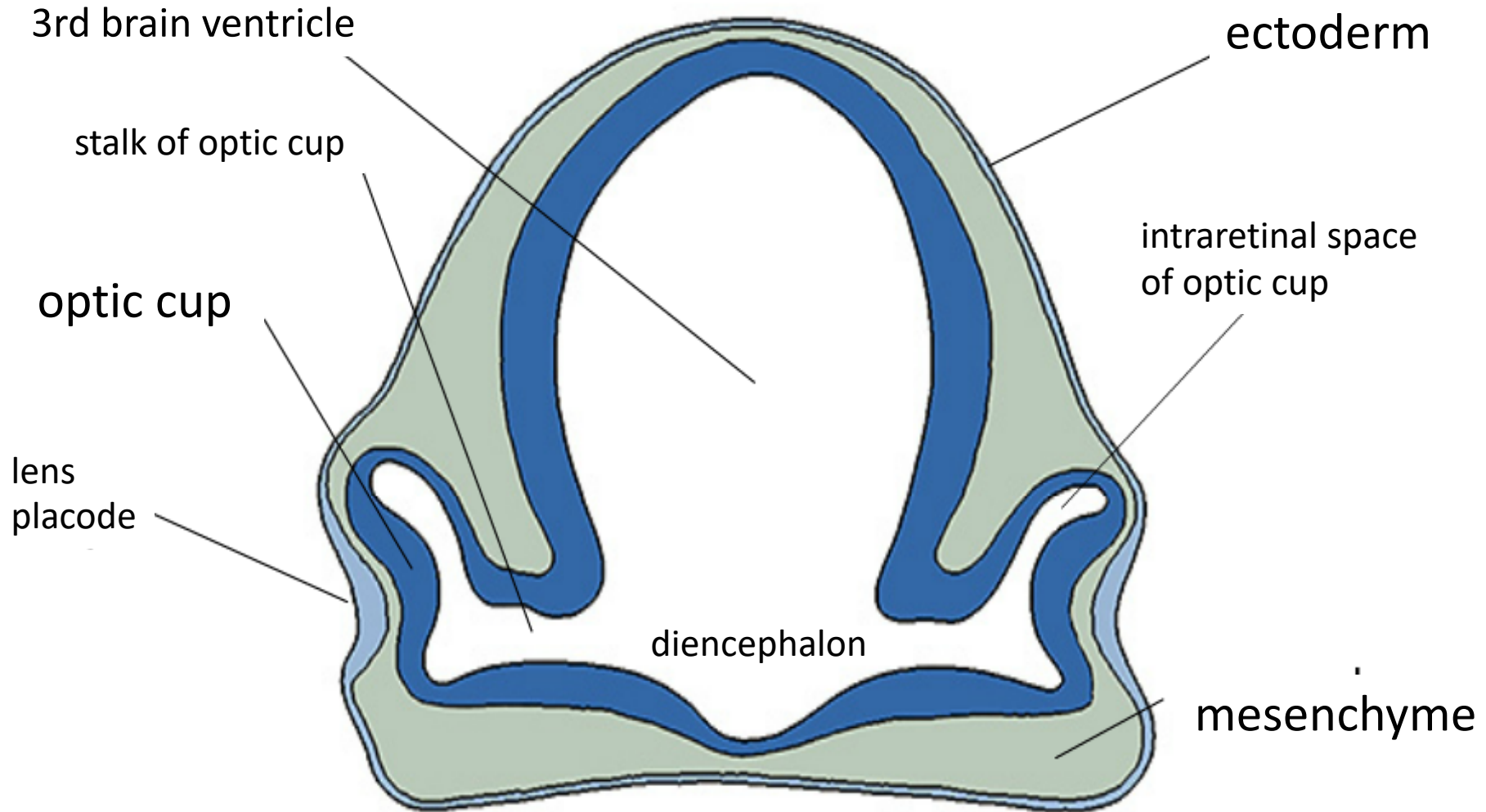


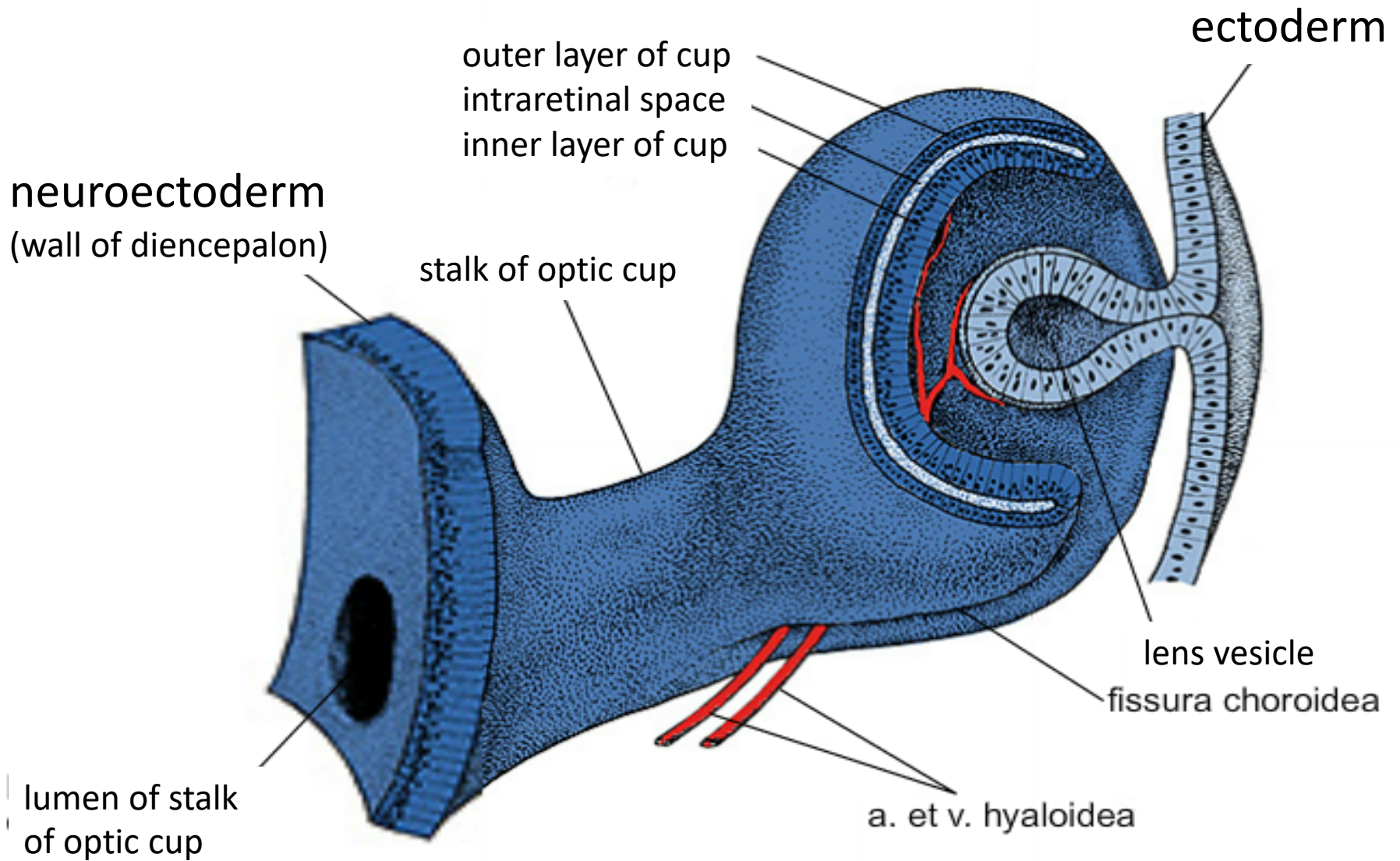
Development and teratology of sensory organs

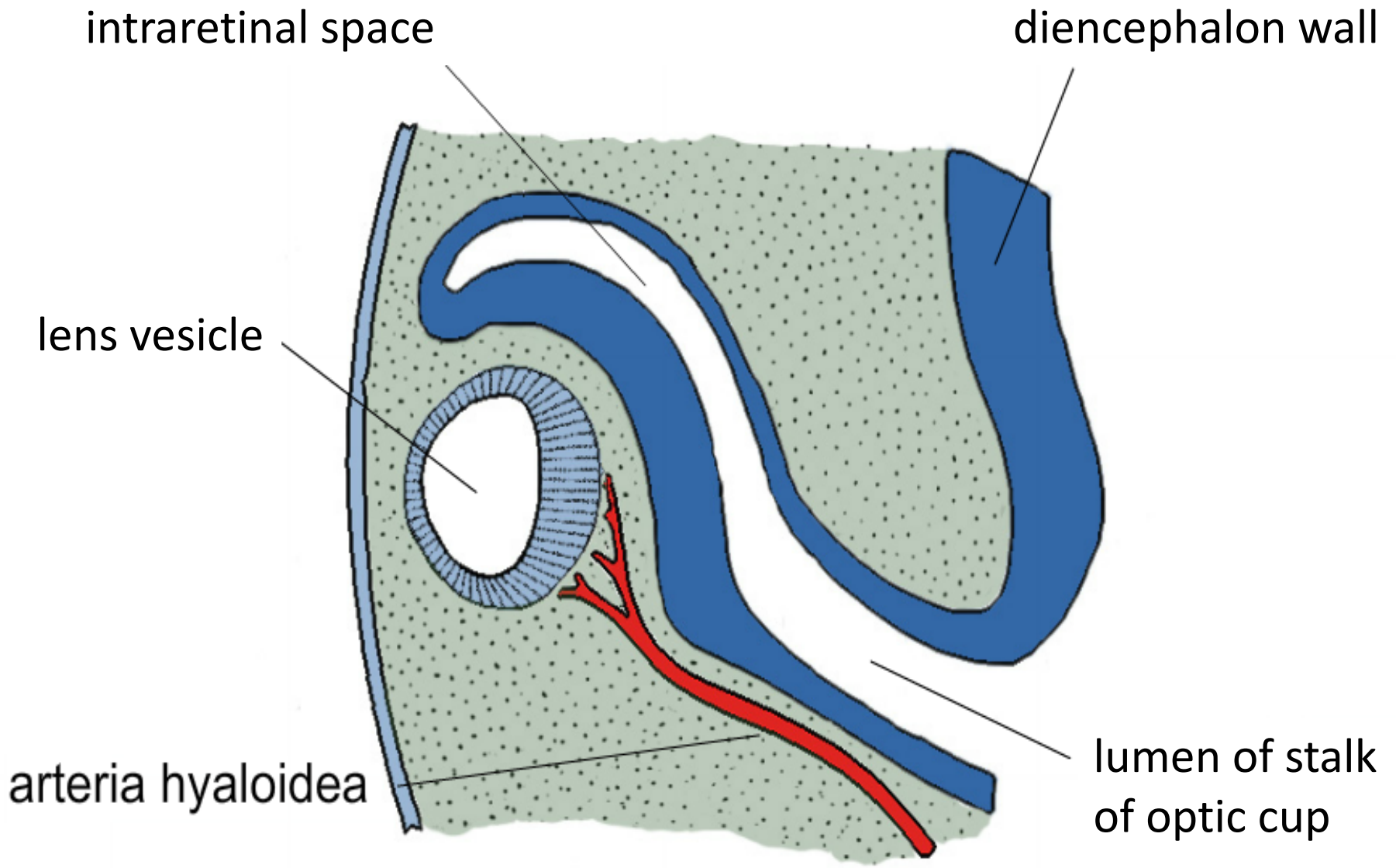
Anna Mac Gillavry Danylevska
22.5.2023

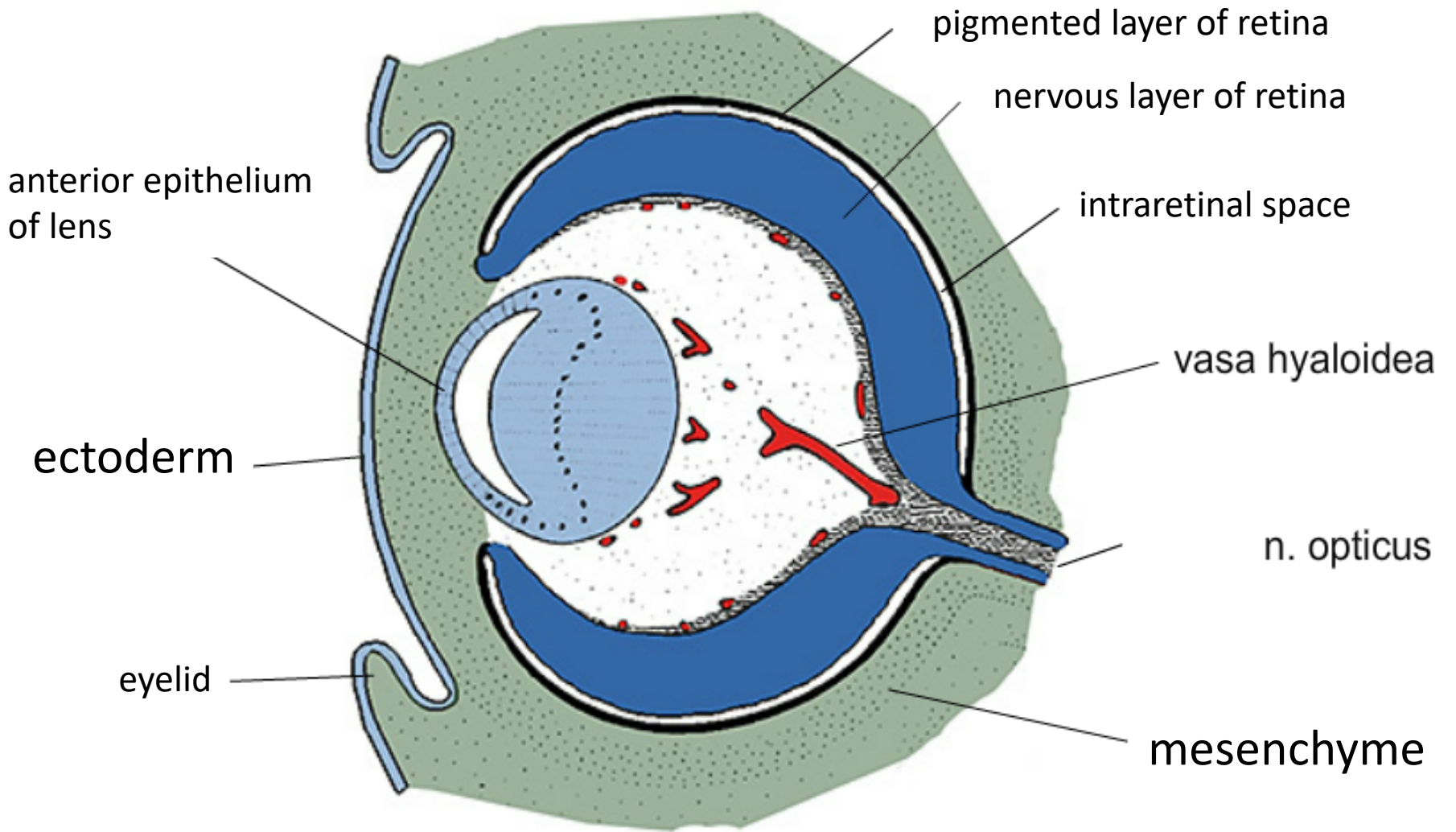
Development of the eye

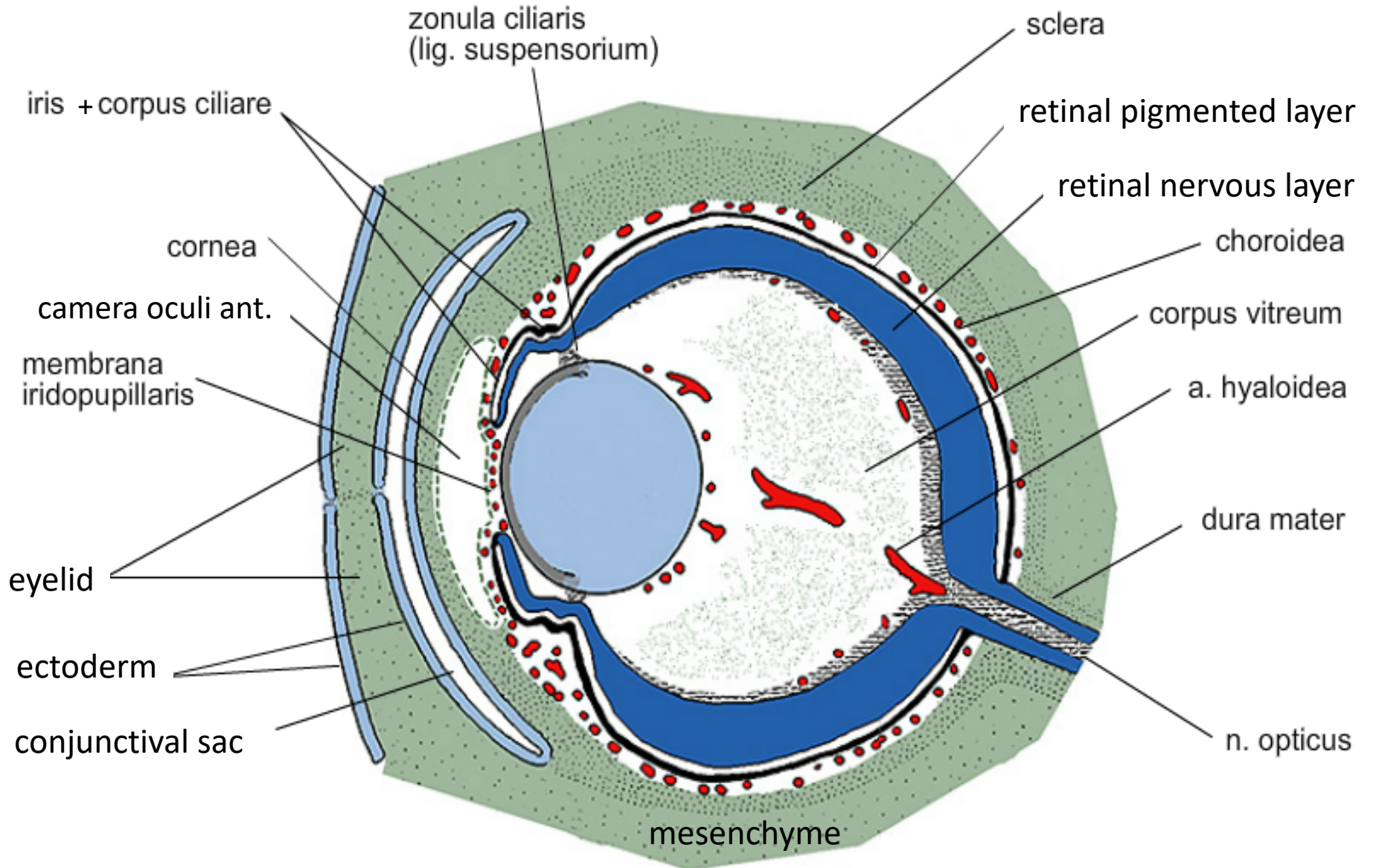


Development of the optic cup and the lens









Eye abnormalities

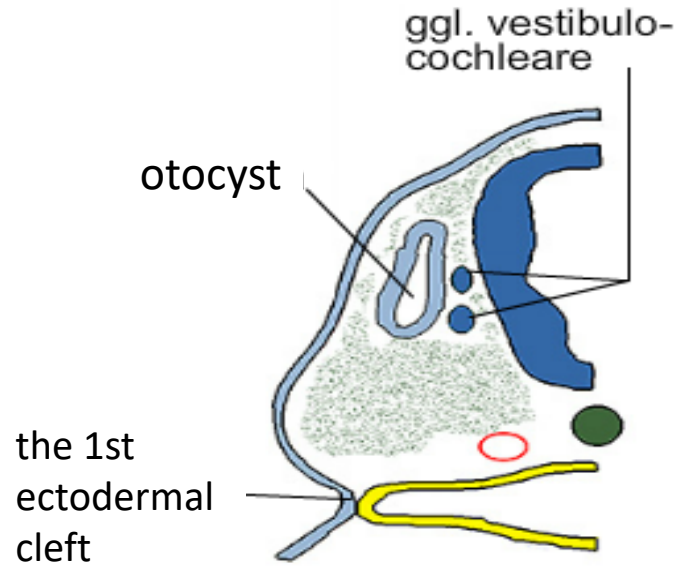
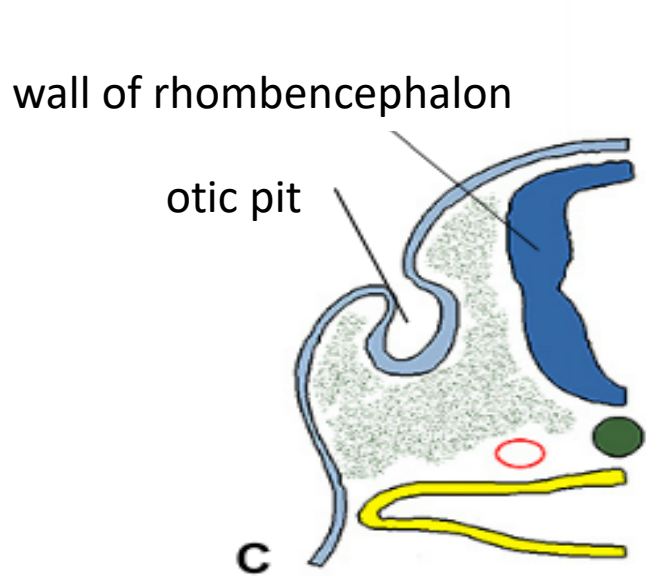
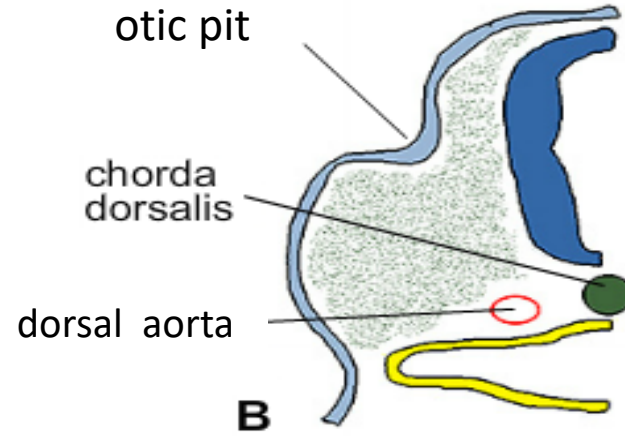
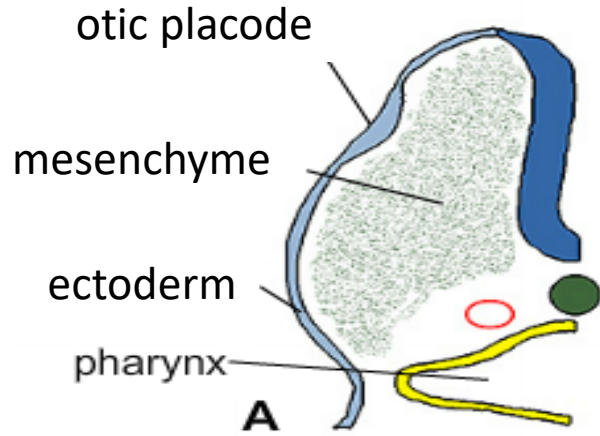
- Coloboma – in most cases coloboma iridis, frequently associated with other eye abnormalities; optic nerve coloboma is linked to PAX2 gene mutations – renal coloboma syndrome – autosomal dominant



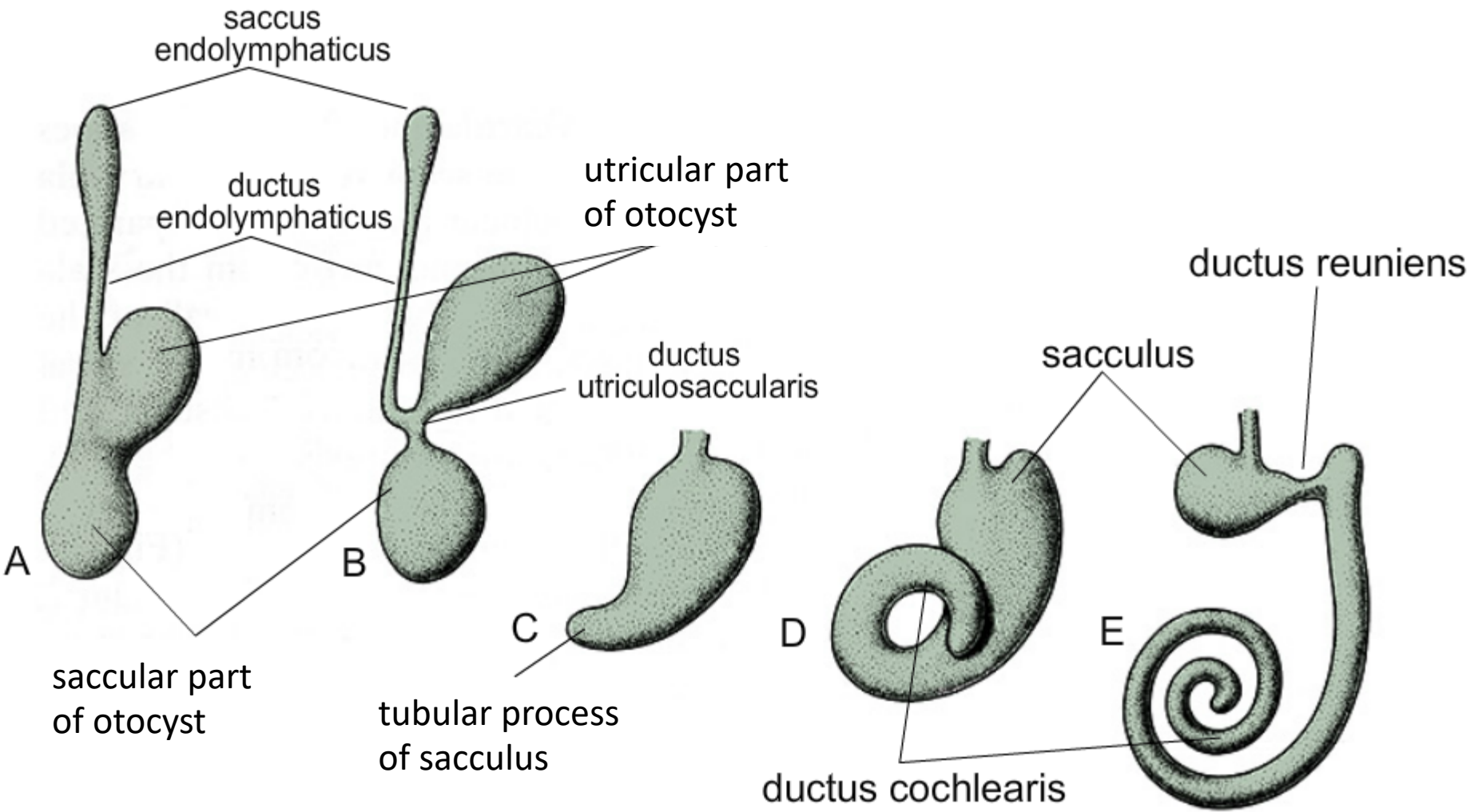
- Iridopupillary membrane
- Sclerocornea
- Peters anomaly
- Congenital cataracts – genetically determined or rubella virus!
- Persistent hyaloid artery (in 3 % of full term infants) – amblyopia, vitreous haemorrhage, retinal detachment

- Microphthalmia – 1,5-1,9 in 10000, cytomegalovirus, toxoplasmosis
- Anophthalmia – 0,18-0,4 in 10000
- Congenital aphakia/aniridia – mutation in PAX6 results in aniridia; WAGR syndrome – microdeletion in chromosome 11 (PAX6 and WT1)
- Cyclopia and synophthalmia (loss of midline tissue - holoprosencephaly) – alcohol, maternal diabetes, mutations in SHH and abnormalities in cholesterol metabolism

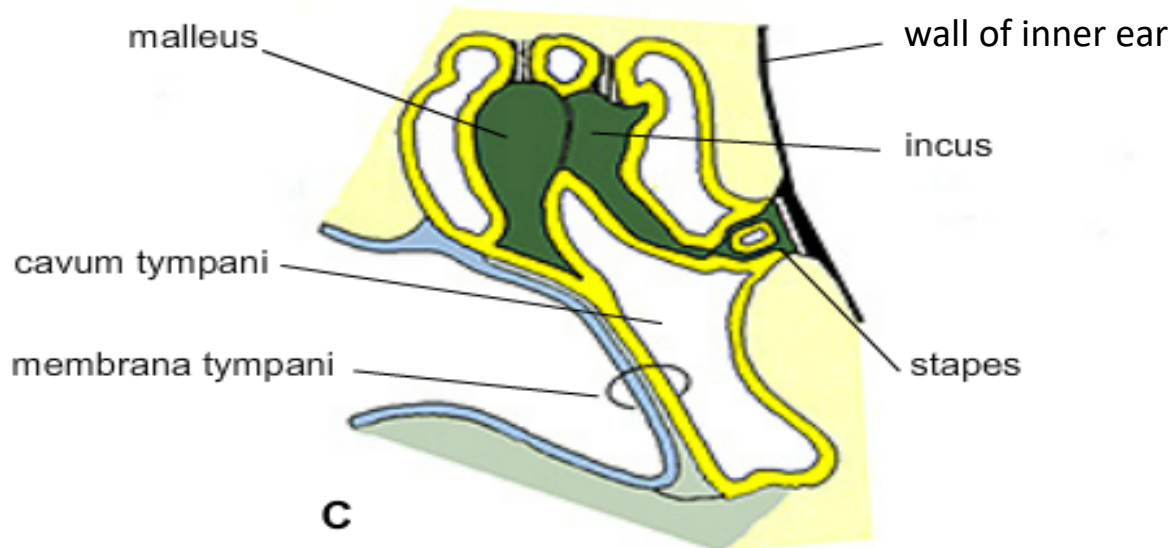
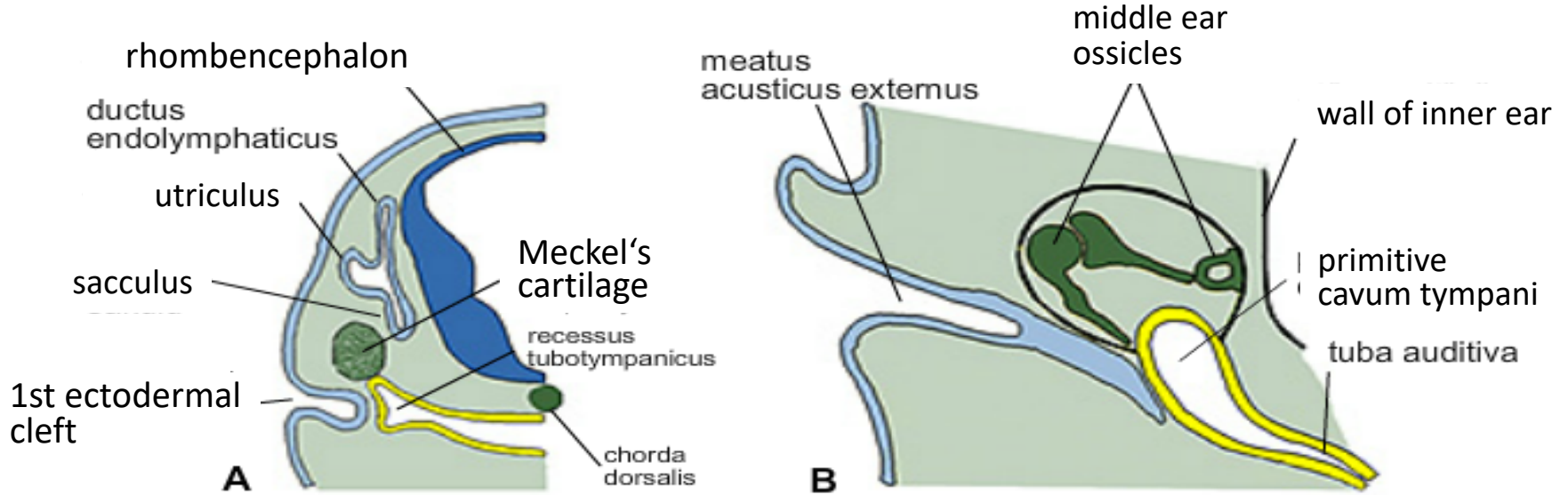
Development of the inner ear



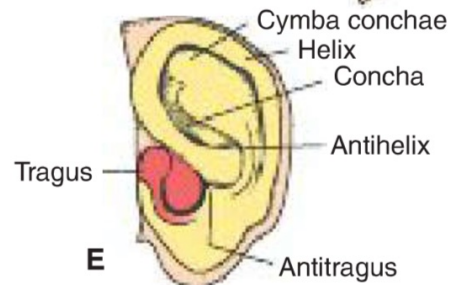
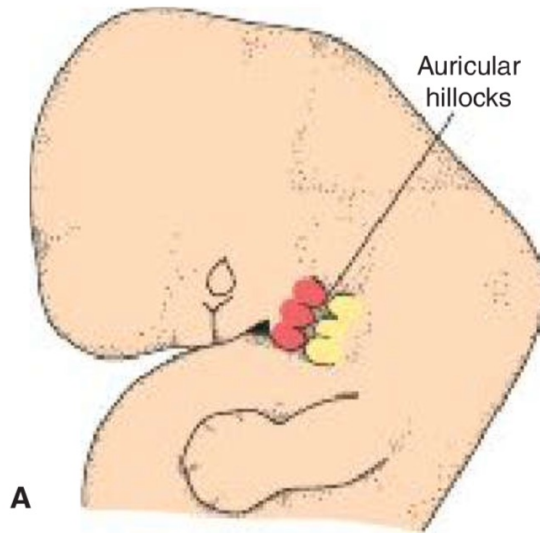
Development of the membranous labyrinth



Development of the middle ear

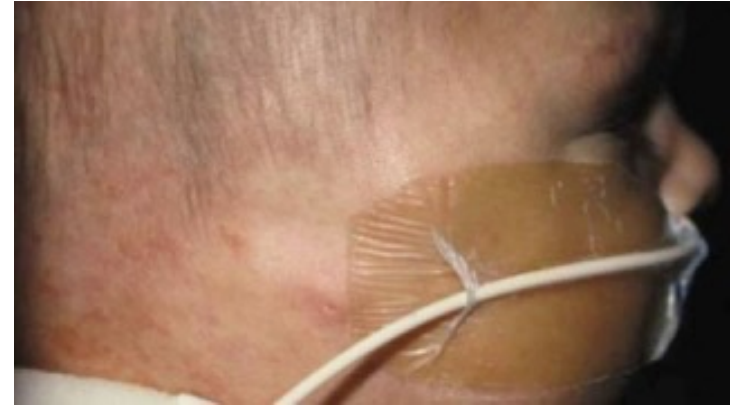


Development of the external ear



External ear defects

- Anotia
- Microtia



[Anotia | Children's Hospital of Philadelphia \(chop.edu\)](http://chop.edu)

- (1 in 6000-12000) usually one ear is affected, the external acoustic 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!