Practice

Face development and defects

(face, jaws, nose, palate)

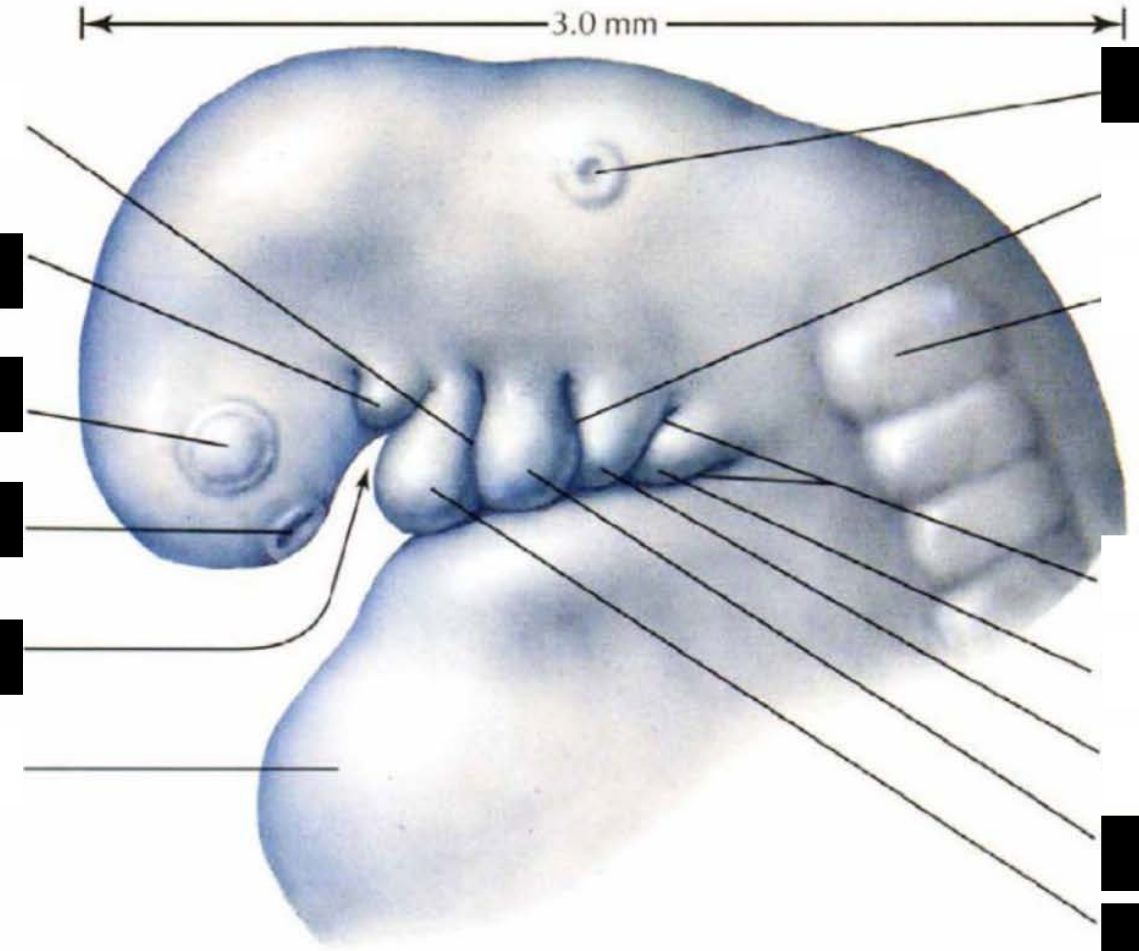
Congenital malformations

12.5.2021

Jan Křivánek

Test

28 days old embryo – lateral view



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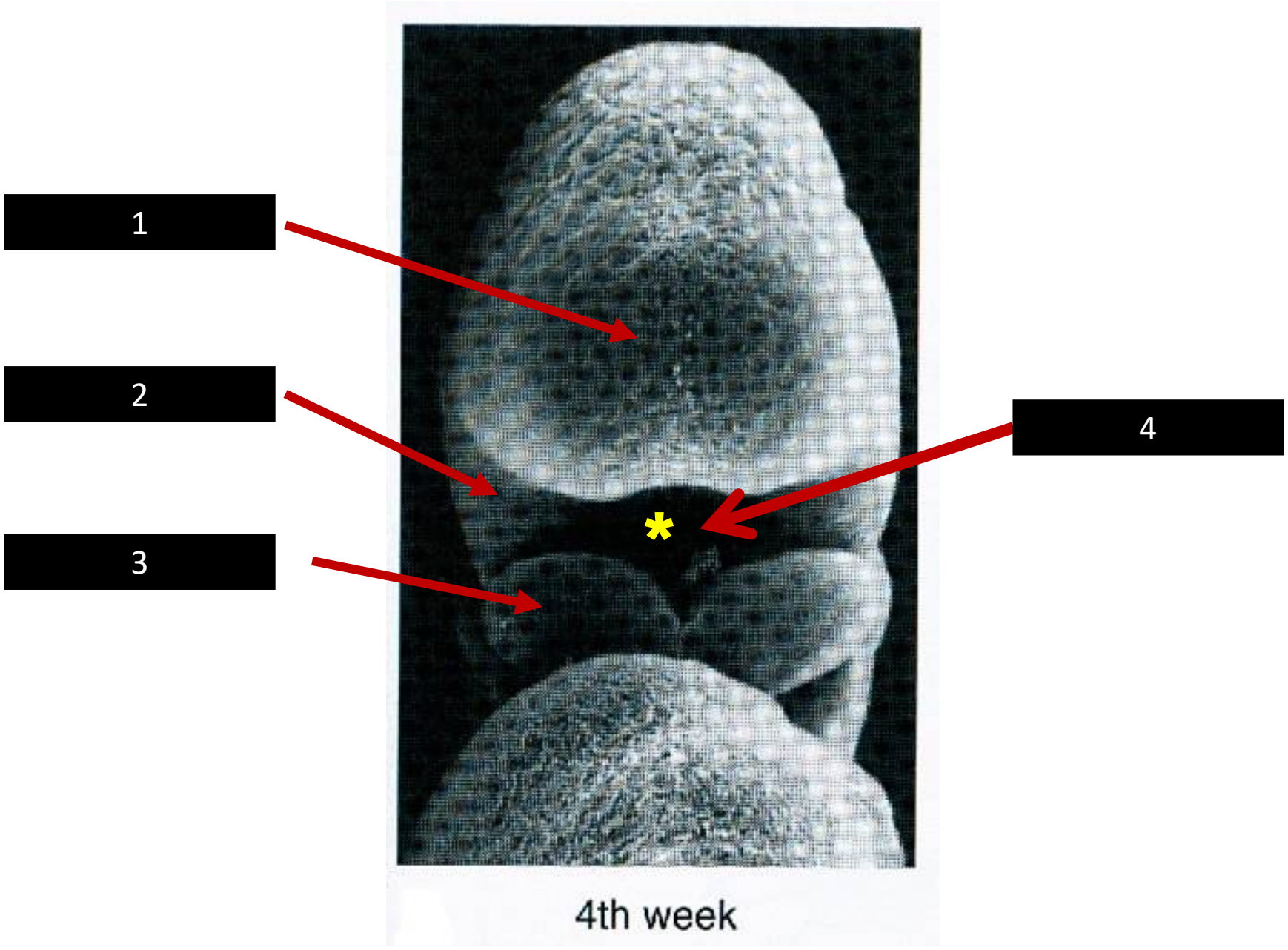
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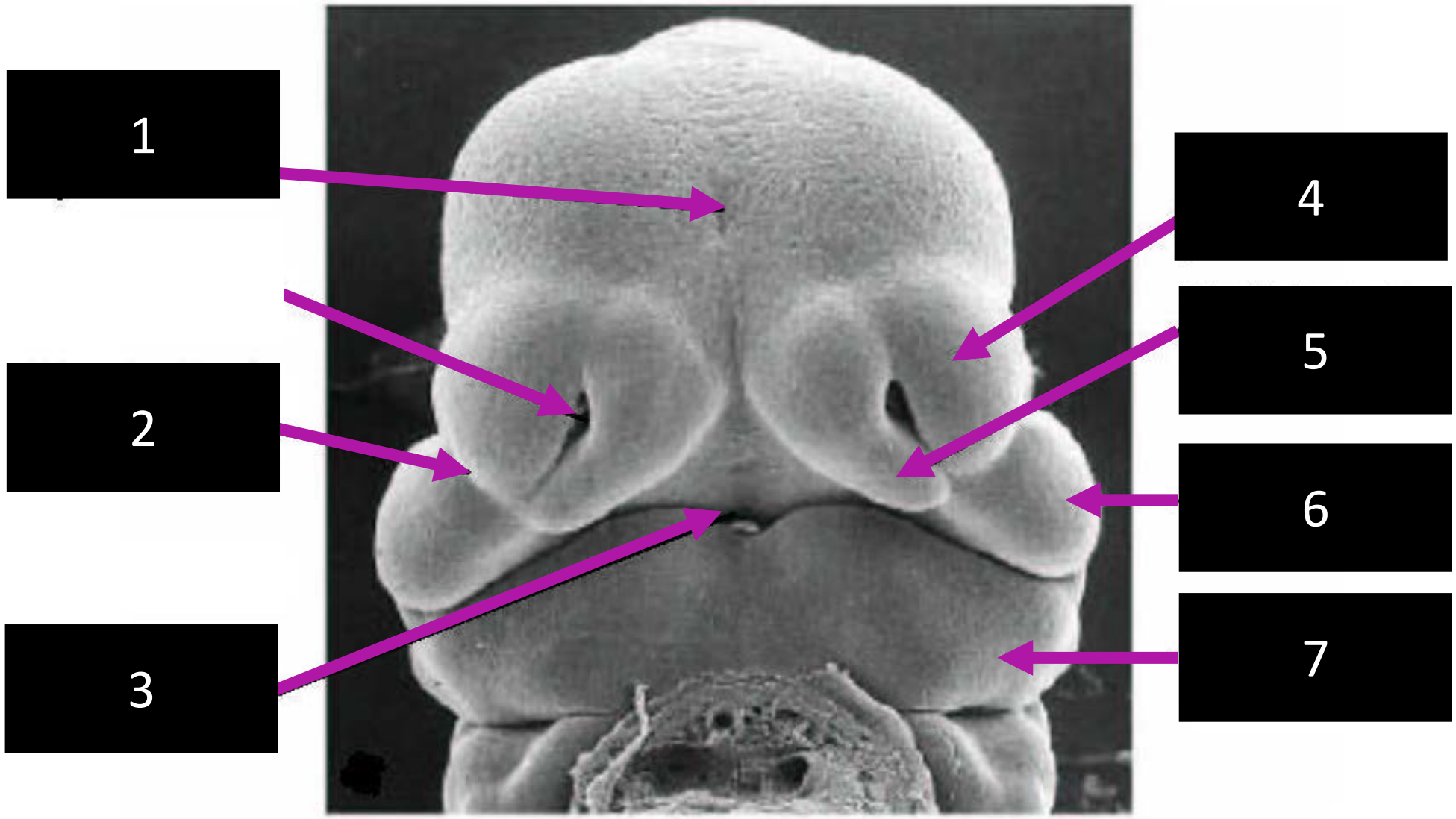
4th week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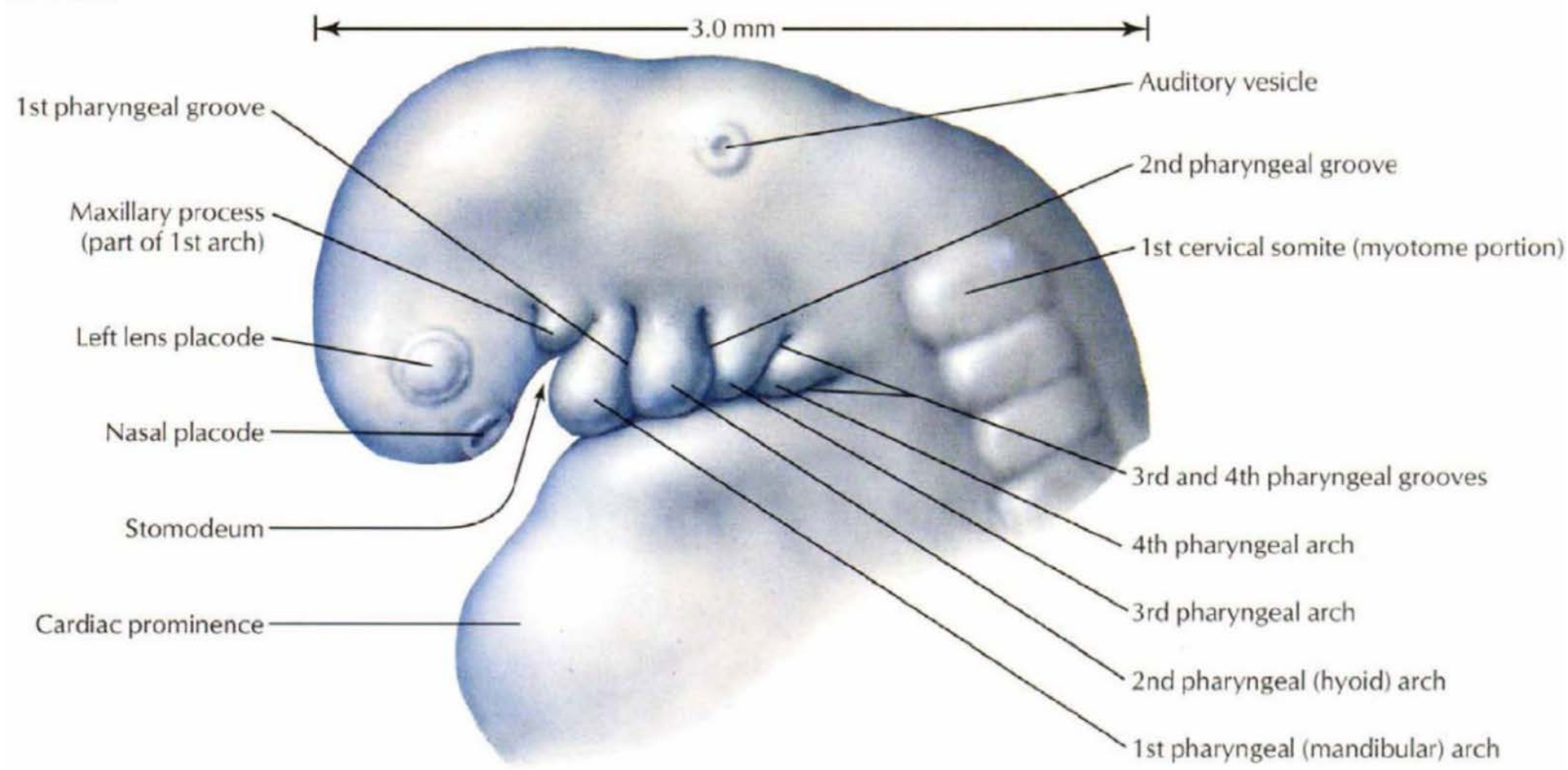
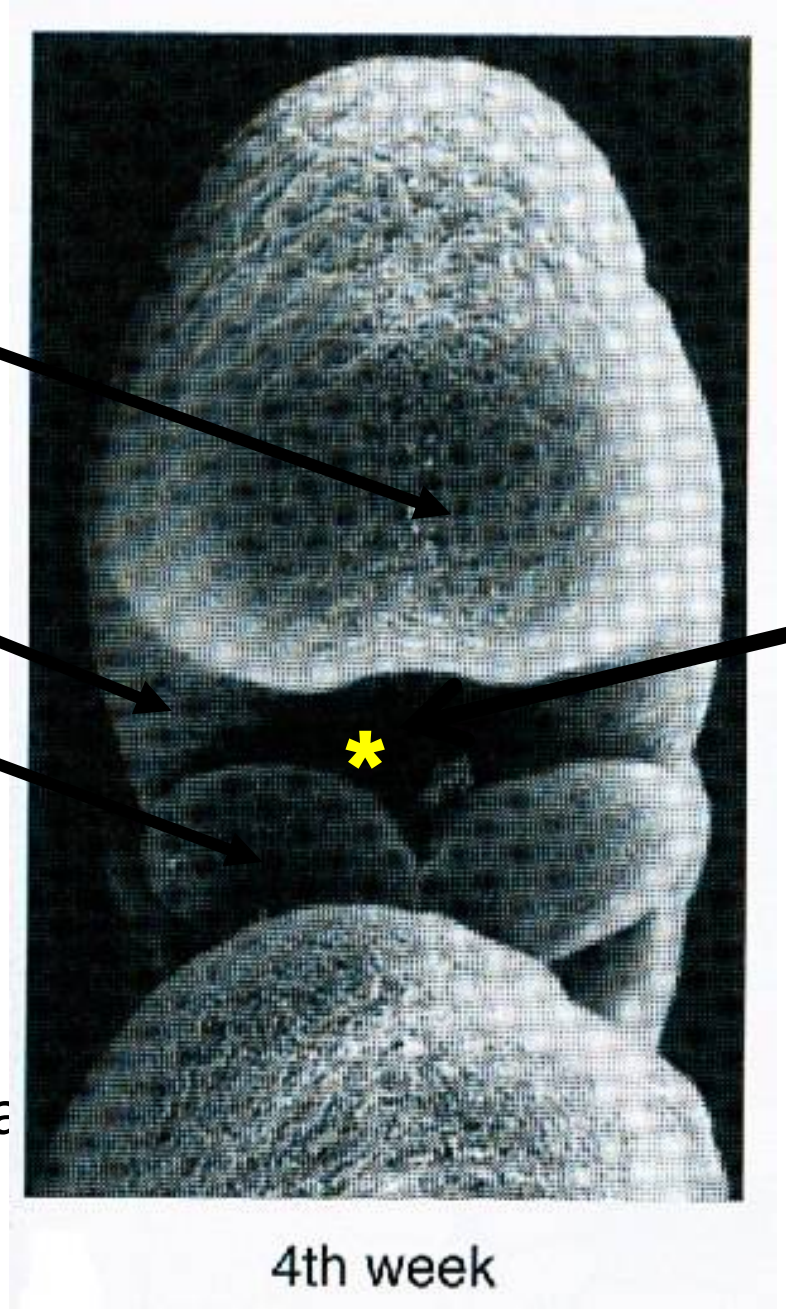
28 days old embryo – lateral view



front(onaz)ální výběžek
(processus front(onas)alis)

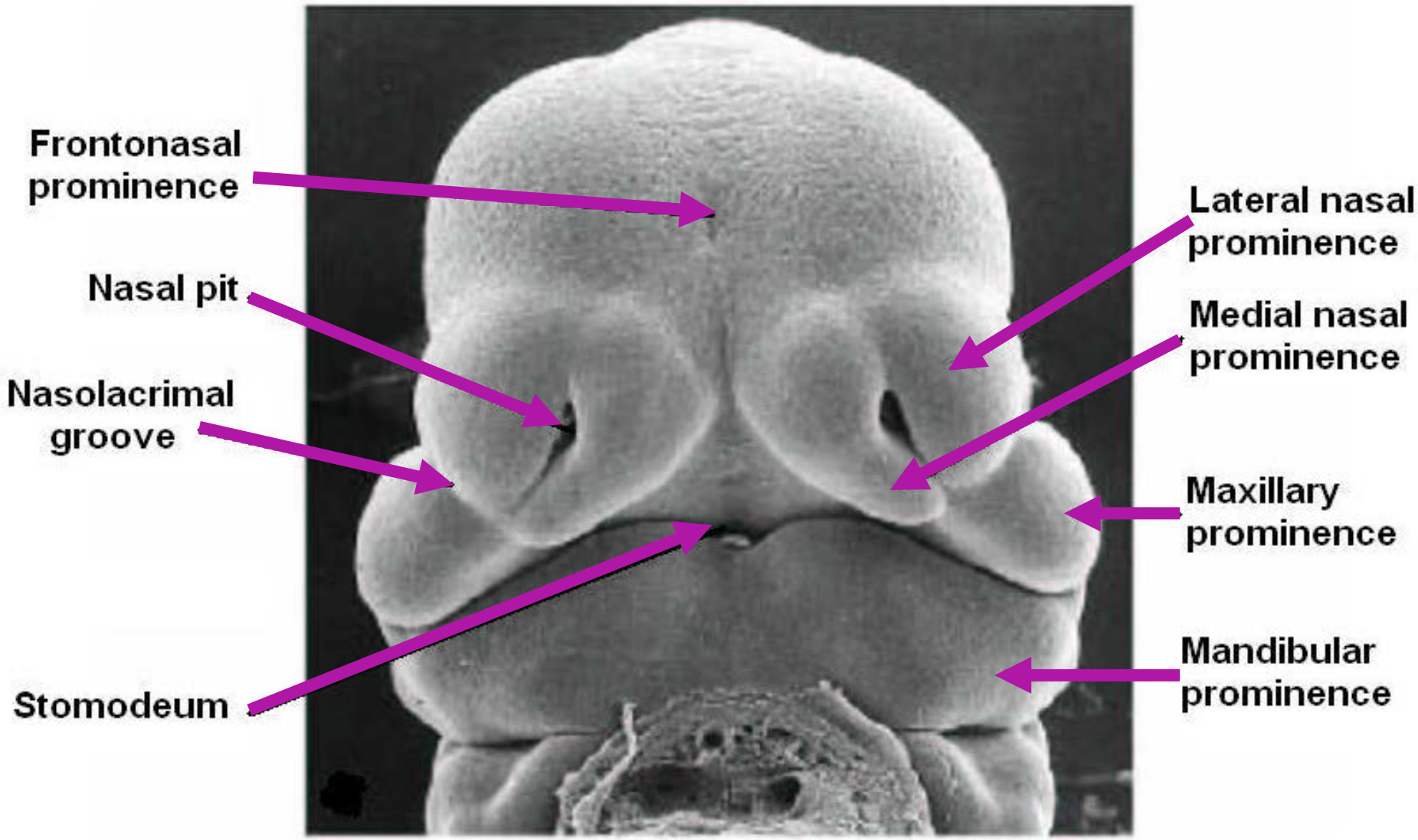
výběžek pro horní čelist
(processus maxillaris)

výběžek pro dolní čelist
(processus mandibularis)



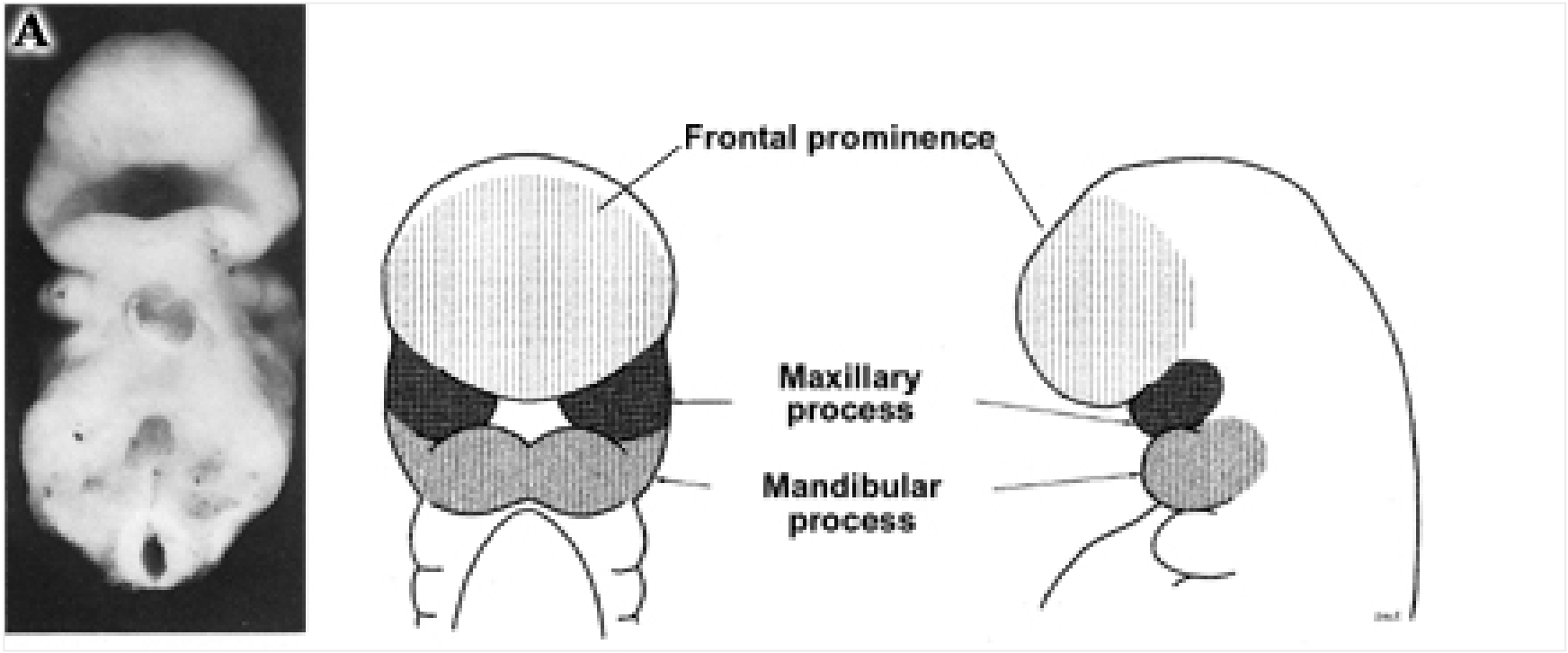
* faryngová membrána
(membrana oropharyngea

Face development - repetition

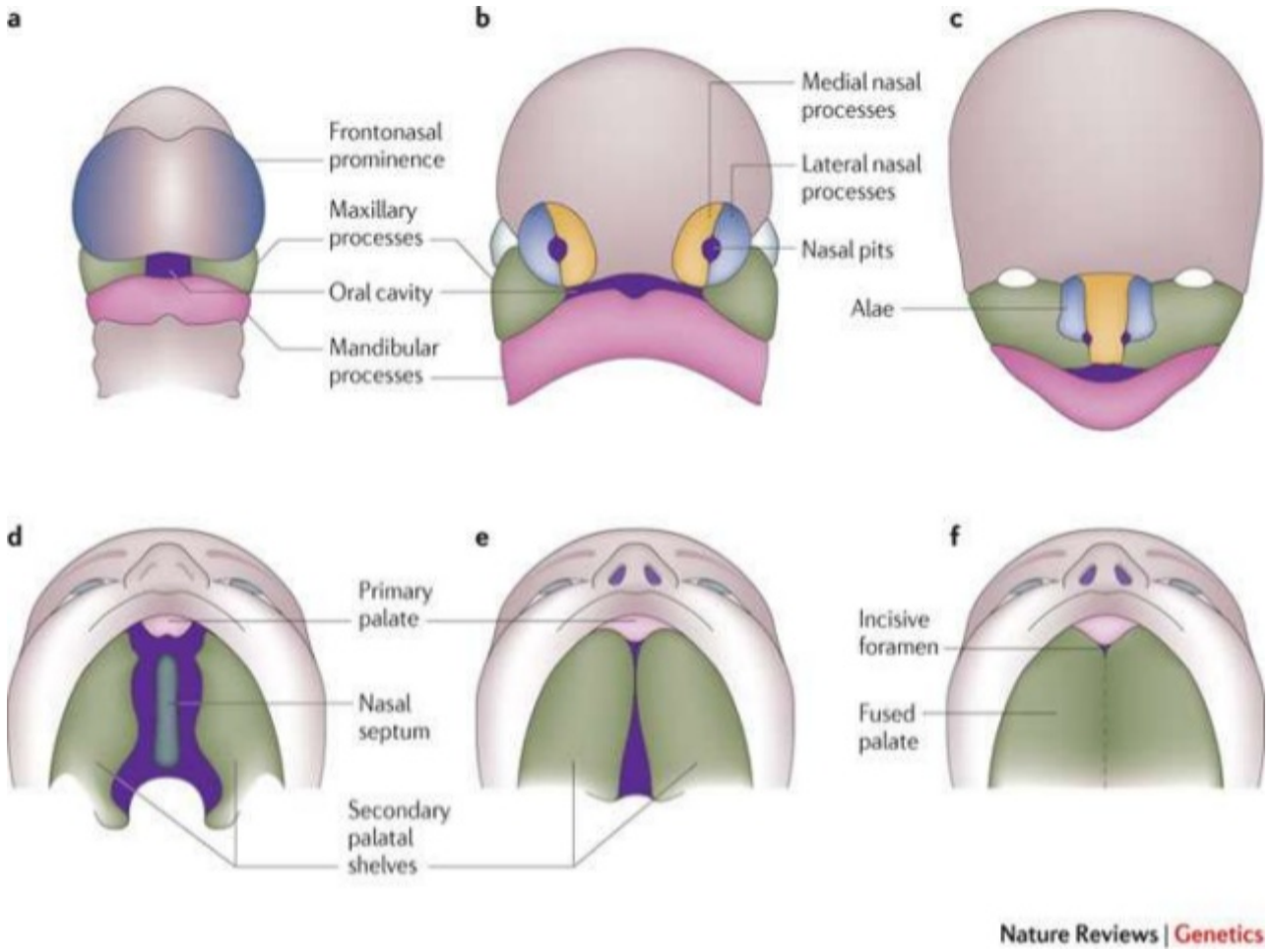

Frontonasal prominence
Nasal pit
Nasolacrimal groove
Stomodeum
Lateral nasal prominence
Medial nasal prominence
Maxillary prominence
Mandibular prominence

<u>1. Pharyngeal arch</u> (mandibular) is divided into :

- **Processus maxillaris**
- **Processus mandibularis**

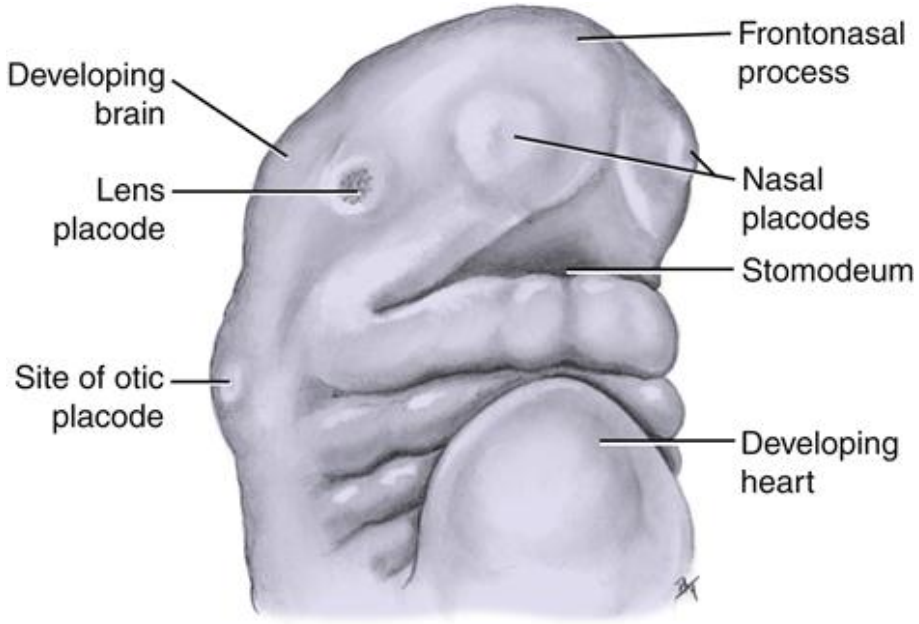


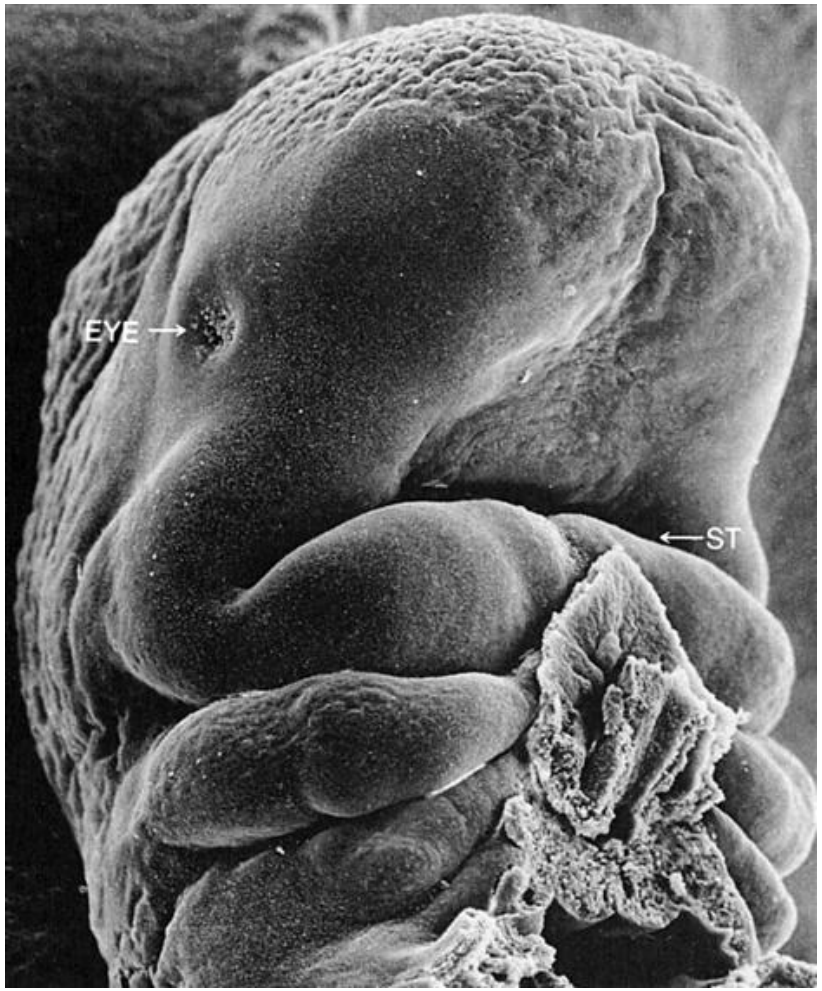
Face development



Face development

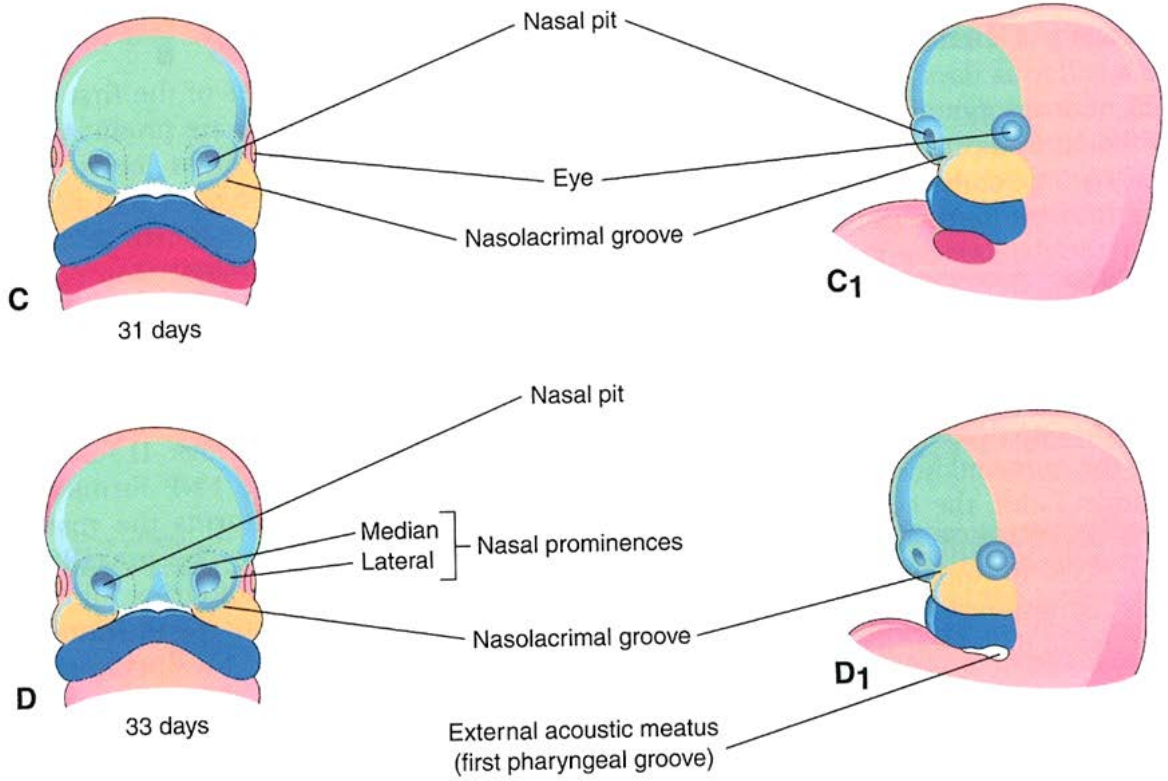
4th week

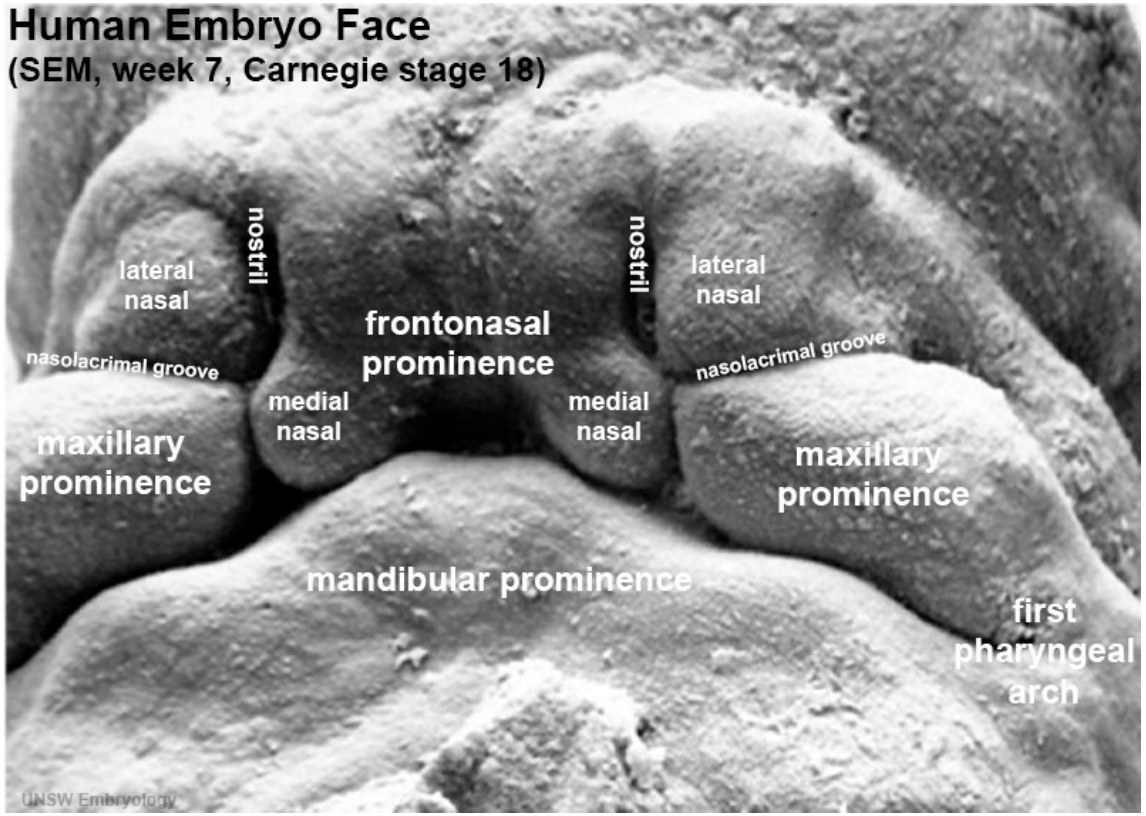


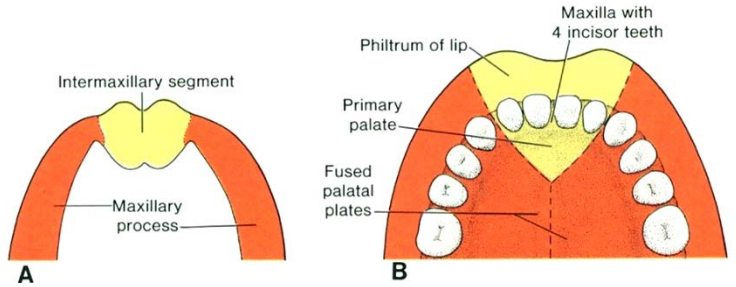


Doc. Petr Vaňhara

Face development





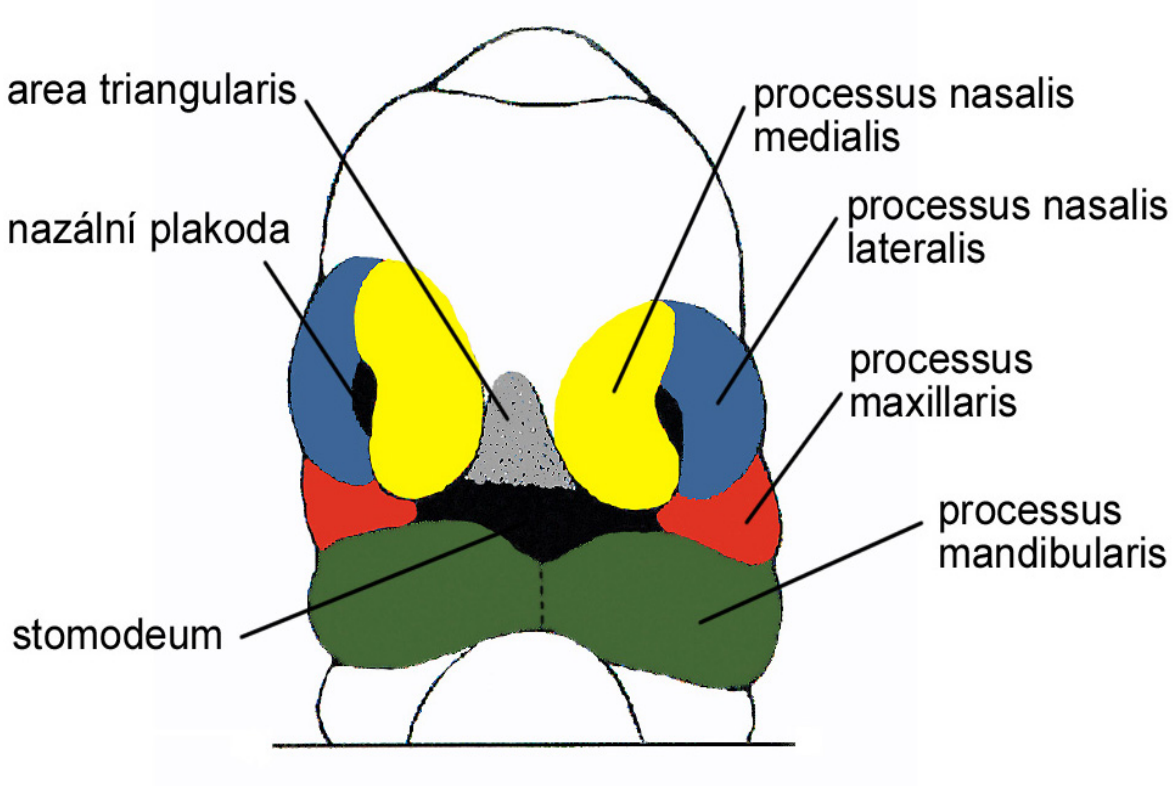


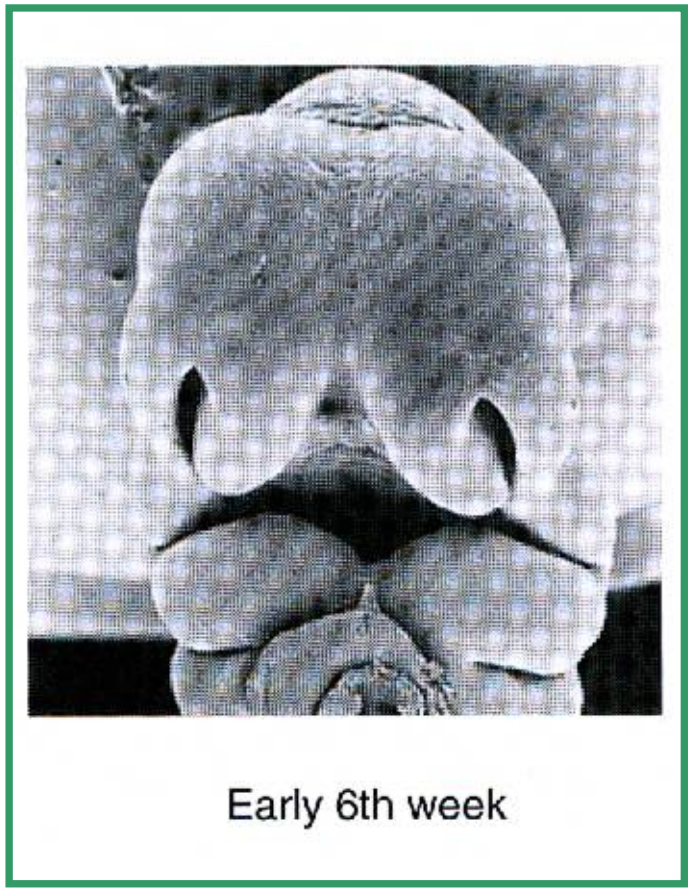
- Nasal pits surrrounded by paired prominences – **medial and lateral nasal prominence**
- **Area triangularis** (nose)
- **Intermaxilary segment** (medial part of upper lip, part of upper jaw, primary palate)

Doc. Petr Vaňhara

After the formation of nasal pits the ectomesenchyme is divided into parts:
Processus nasalis medialis
Processus nasalis lateralis

*Triangular area between medial nose processes is called **area triangularis***





Intermaxillary segment

By the end of the **5th week**, the **medial nasal prominences fuse with each other to form the intermaxillary segment**

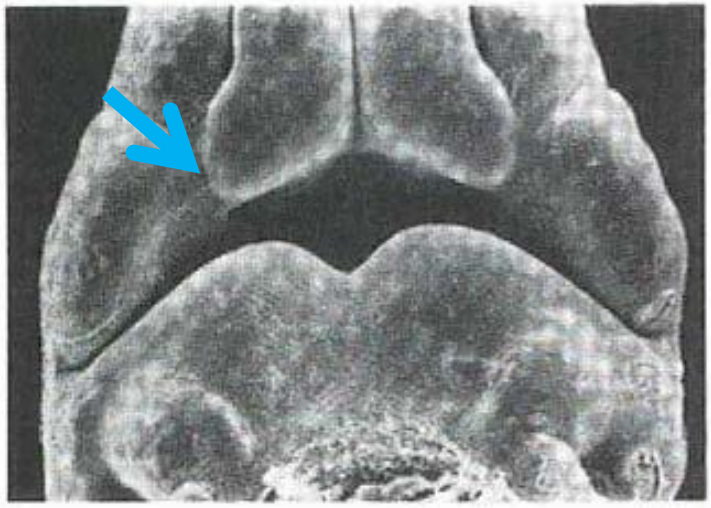
The segment proliferates caudally and inserts between ends of maxillary prominences which merge with it during the **6th week**

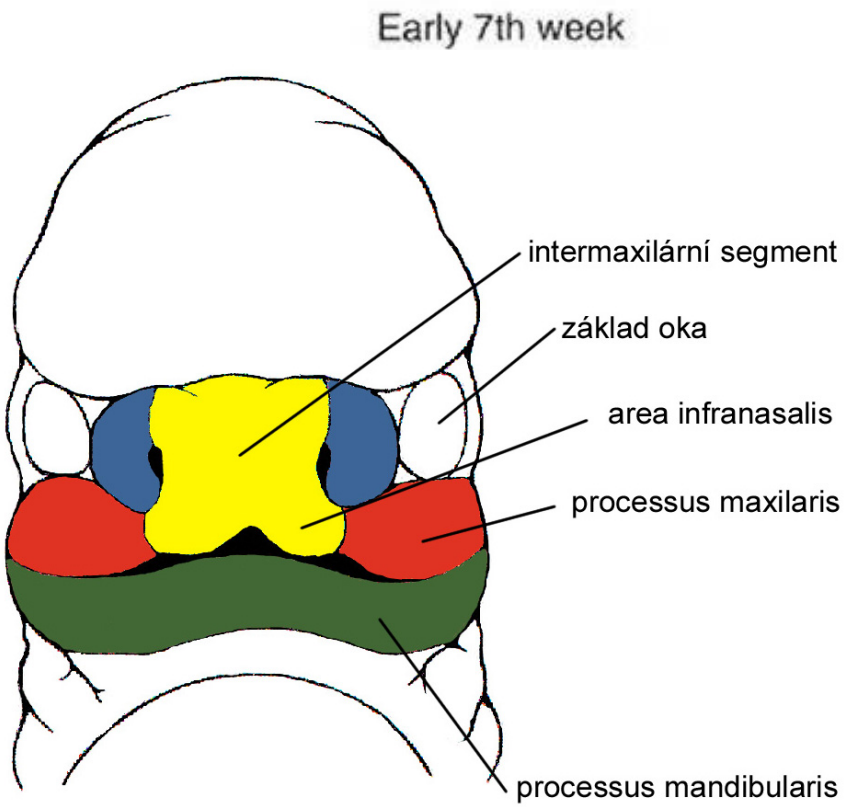
The intermaxillary segment gives rise to:

a) middle portion of the upper lip, or philtrum
b) the premaxillary part of the maxilla
c) the primary palate

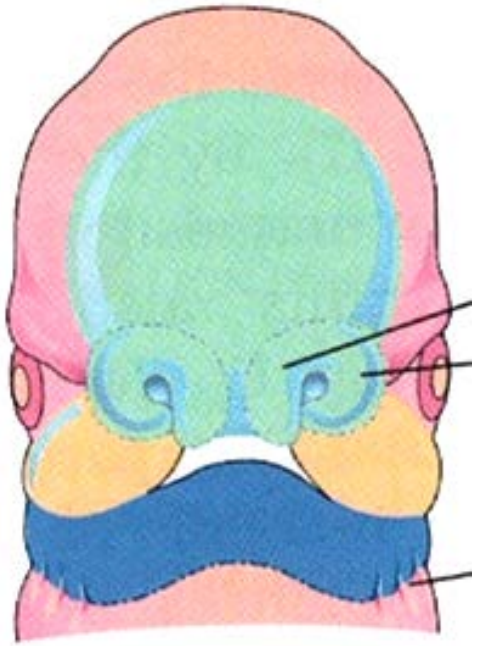
At first, lateral nasal prominences are separated from the maxillary prominences by a furrow, called the **nasolacrimal groove**

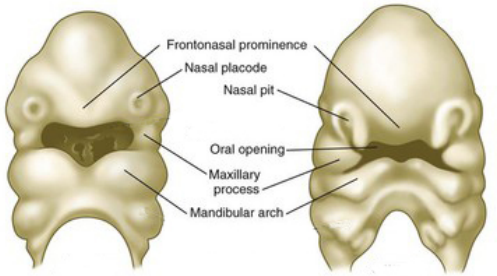
During next days, the maxillary prominences enlarge and fuse with lateral nasal prominences.

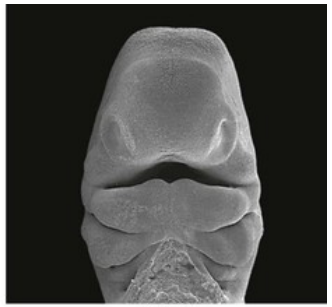


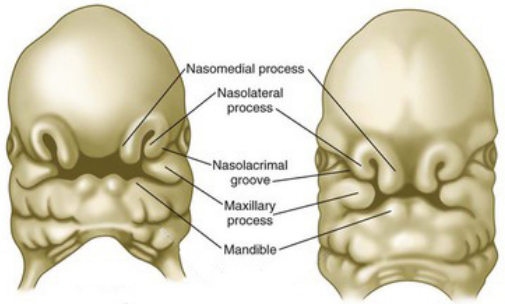


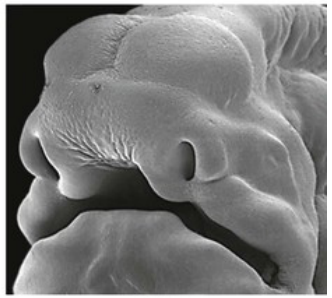
Face development

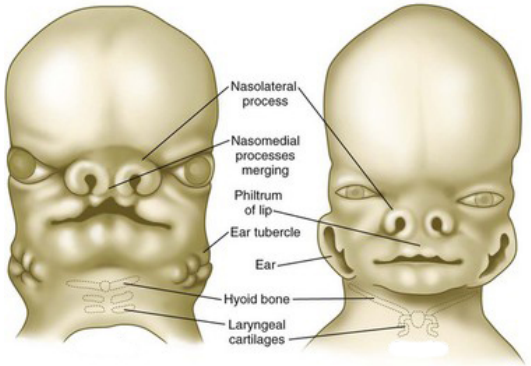


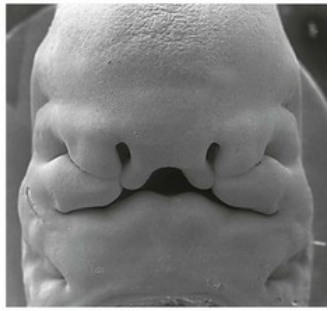










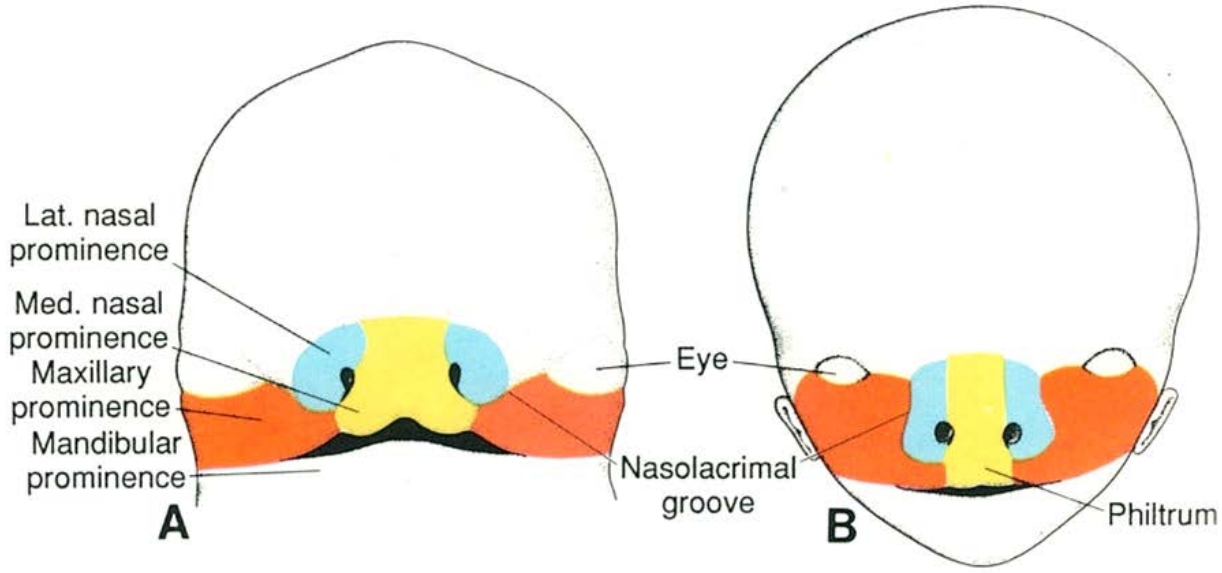


Maxillary prominences fuse with:

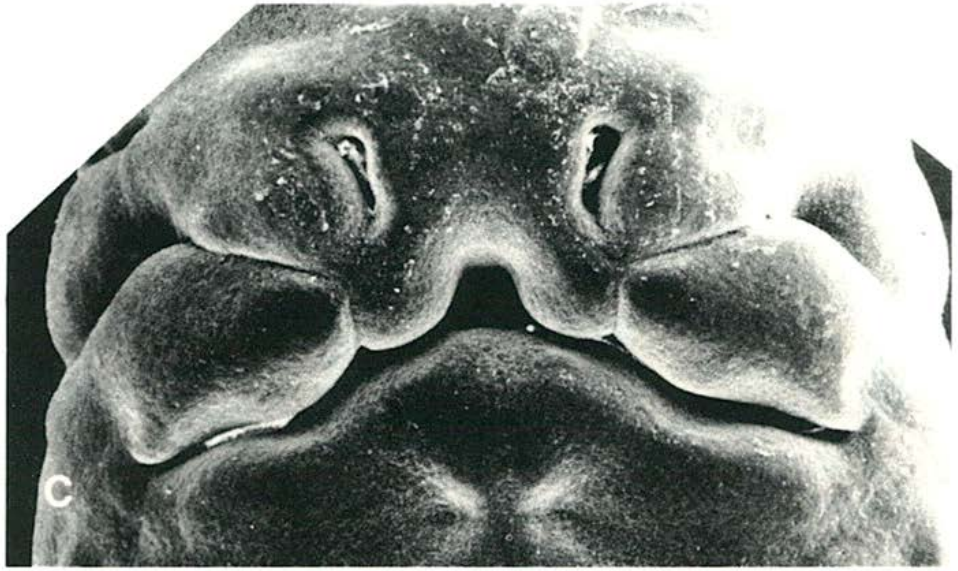
1. **Intermaxillar segment** (formation of upper lip and palate)
2. **Lateral nasal prominences** (the rest of upper lip and part of nose)

Lateral nasal prominences are divided from the maxillary prominences by - **sulcus nasolacrimalis**

Face development



7th week



Development of oral cavity

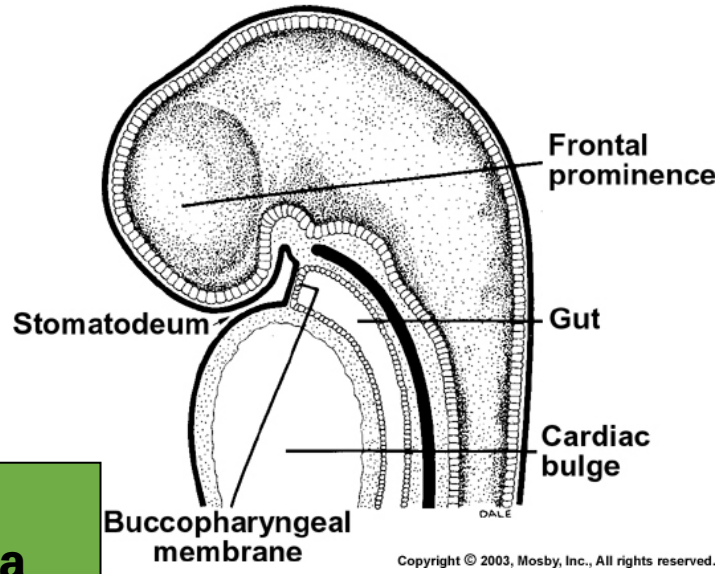
Oral cavity develops from the stomodeum or primitive mouth
5 processes limit the stomodeum:
frontonasal prominence
paired maxillary prominences (processus maxillares)
paired mandibular prominences (processus mandibulares) on sides

Stomodeum communicates with the body surface via primitive oral entrance

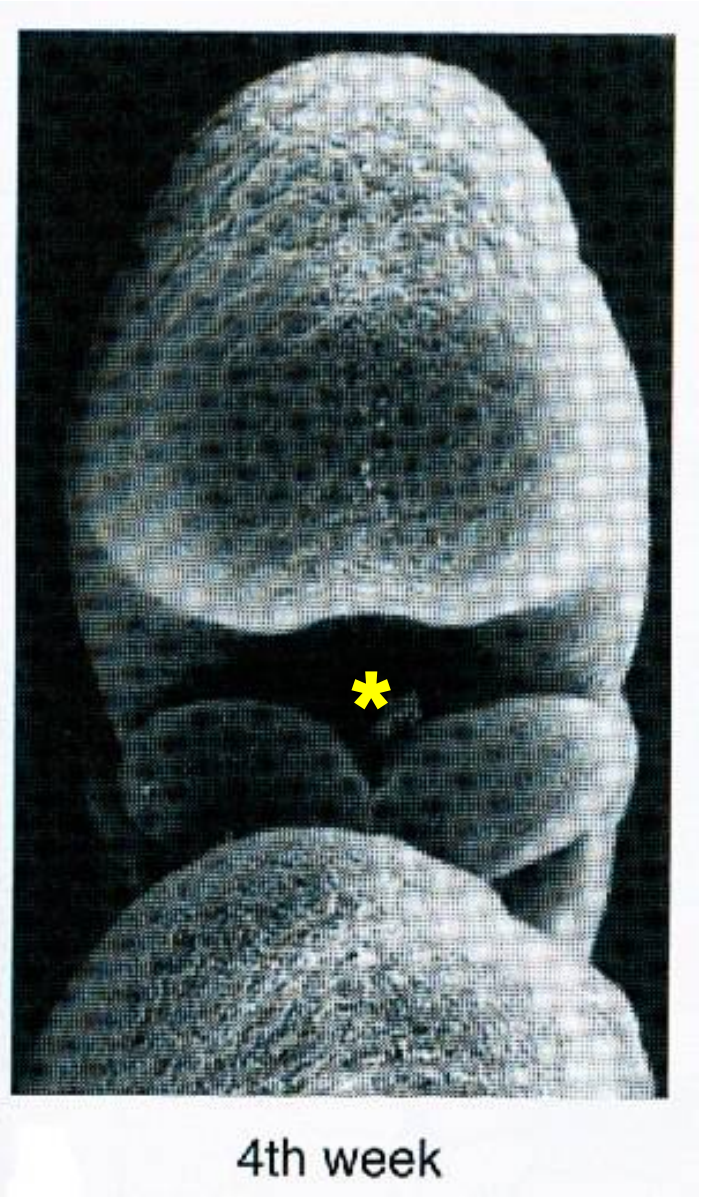
Bottom of the stomodeum - oropharyngeal membrane (membrana oropharyngea)

When the oropharyngeal membrane ruptures, the stomodeum becomes continuous with the foregut

Roof of the stomodeum consists of a mesenchyme and ectoderm of the frontonasal prominence



membrana oropharyngea



Palatogenesis

Begins at the 7th week

Completed by the end of the 12th week

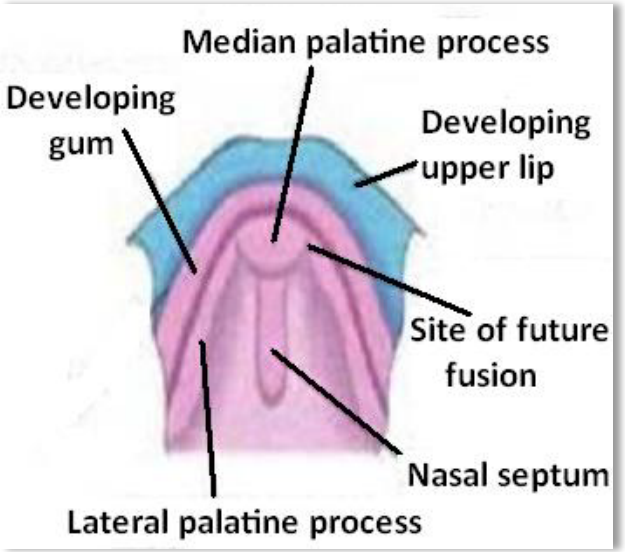
The most critical period for the development of palate is from the beginning of 7th week to the beginning of 9th week

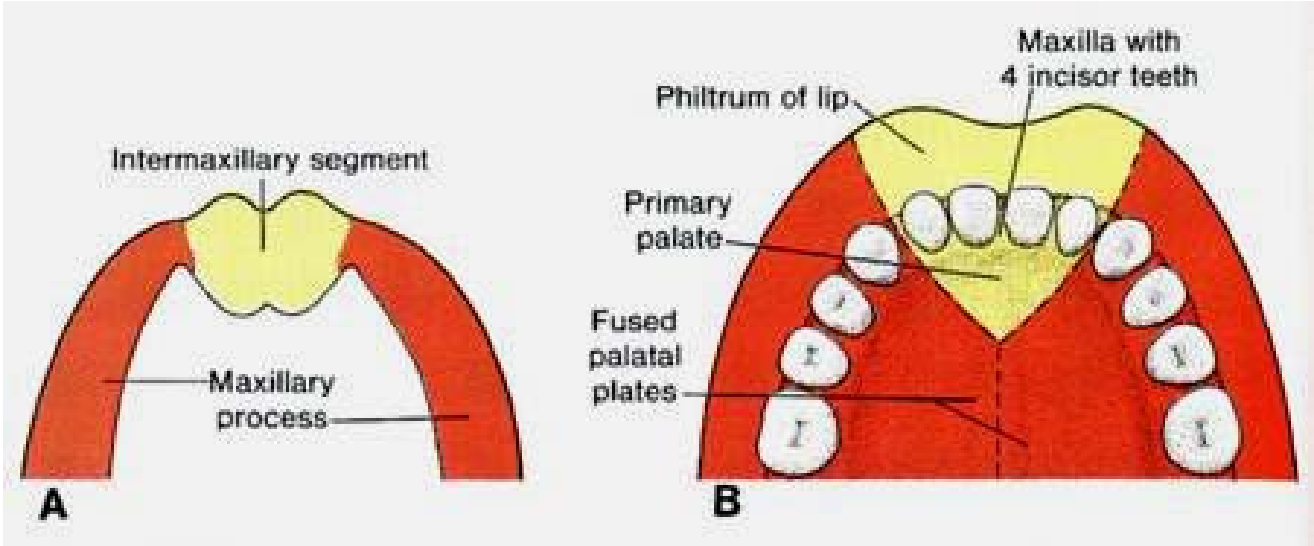
<u>3 primordia:</u>

Unpaired medial palate process and **paired lateral palate processes** (palatal shelves)

a) <u>The medial palate process</u>

Grows from the dorsal side of the intermaxillary segment at the end of the 5th week and gives rise to primary palate

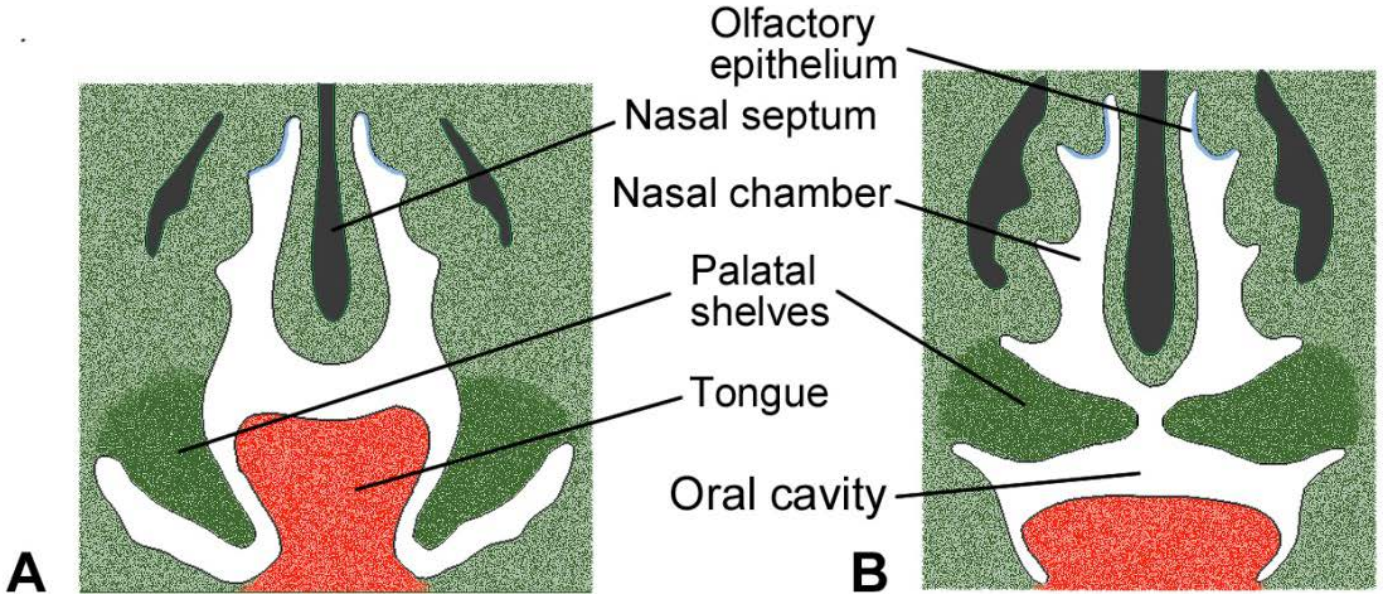


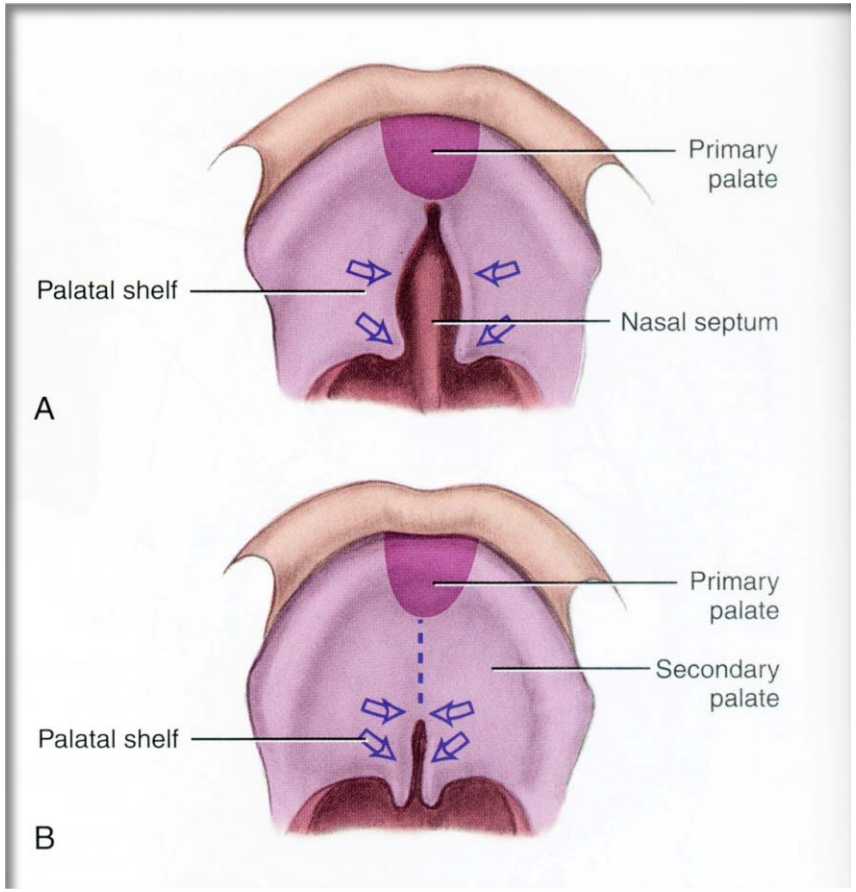


Palatogenesis

b) Lateral palate processes - grow out from medial aspects of the maxillary prominences and give rise to the secondary palate

lateral palate processes are formed by mesenchyme, are covered by ectoderm a have shelf-like form (palatal shelves)





Palatal shelves initially grow in caudal direction and laterally along to primordium of the tongue later, due more rapid vertical growth of mandibular processes the tongue descends caudally

Palatogenesis

During the 10th week shelves meet in the midline to finally fuse

The site of fusion of both lateral palate processes is known as **raphe palati**

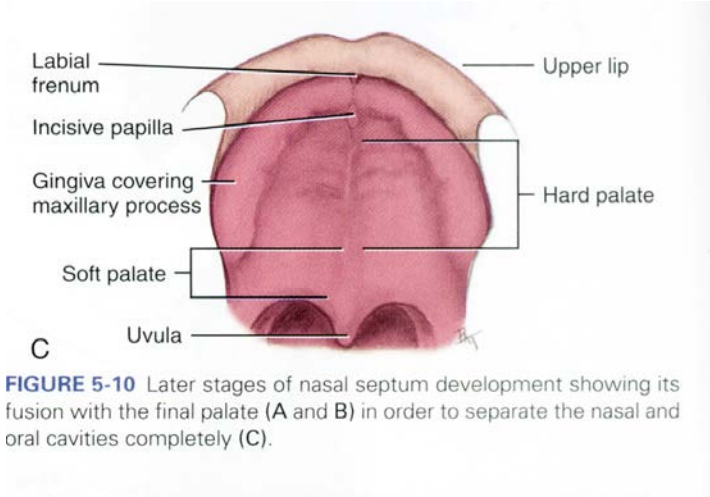
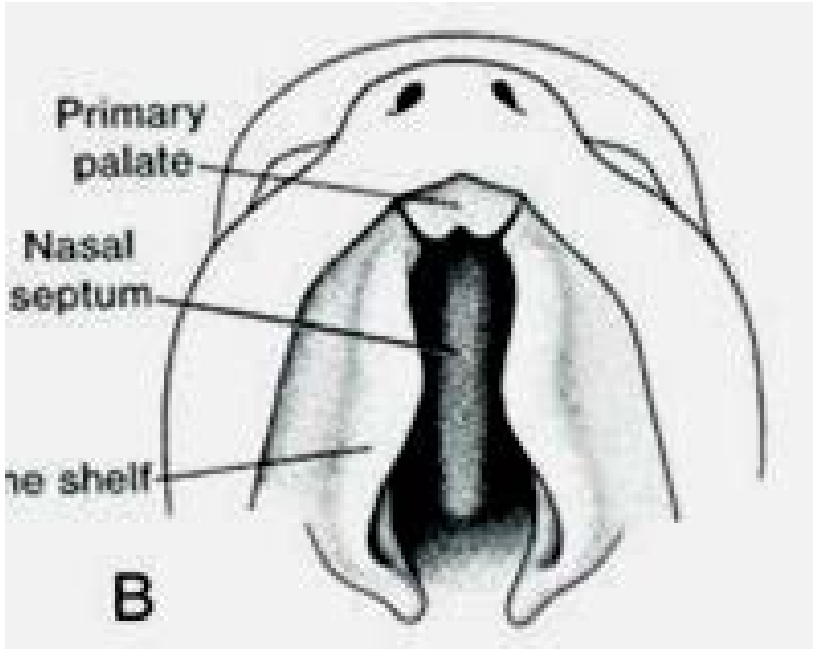
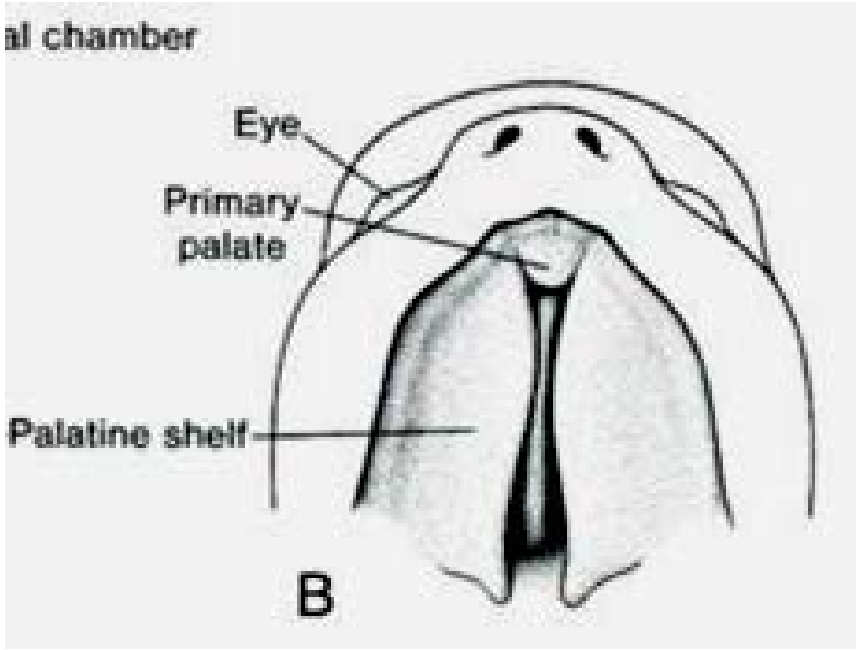
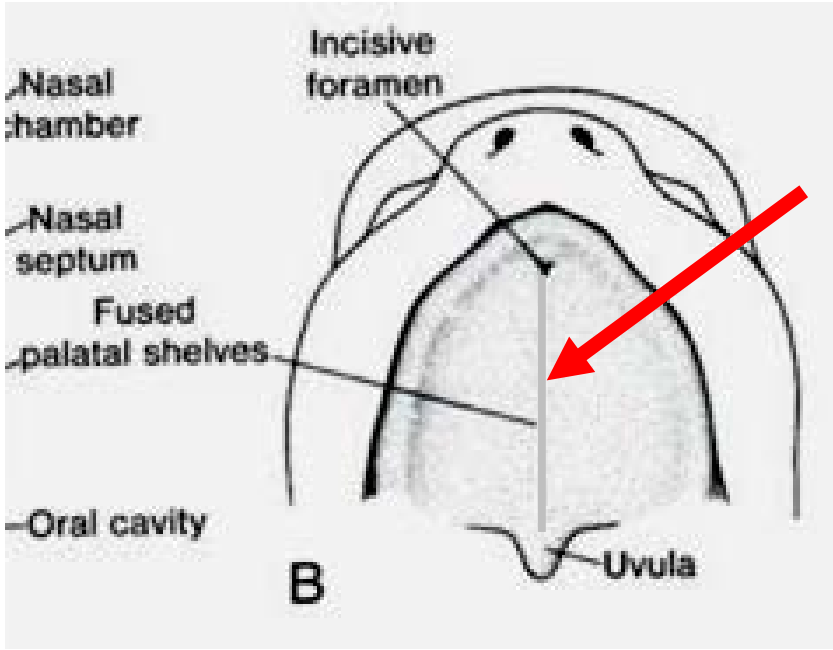


FIGURE 5-10 Later stages of nasal septum development showing its fusion with the final palate (A and B) in order to separate the nasal and oral cavities completely (C).







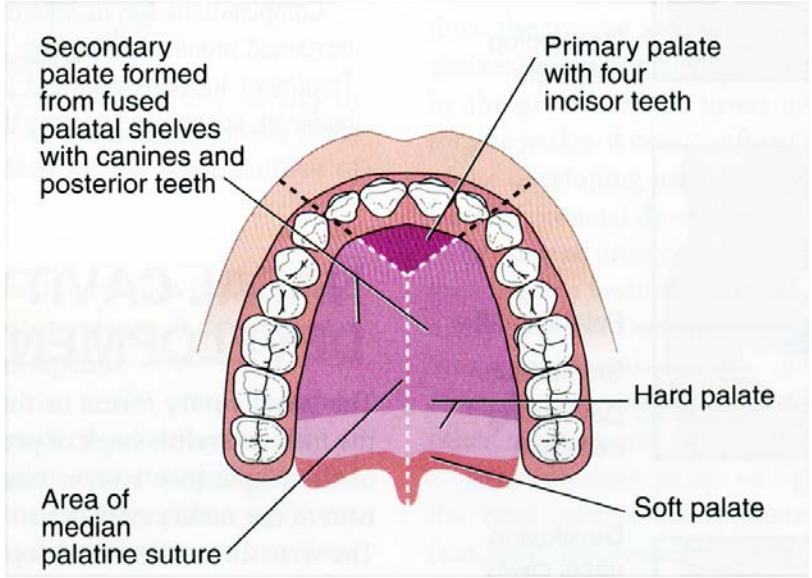
Palatogenesis

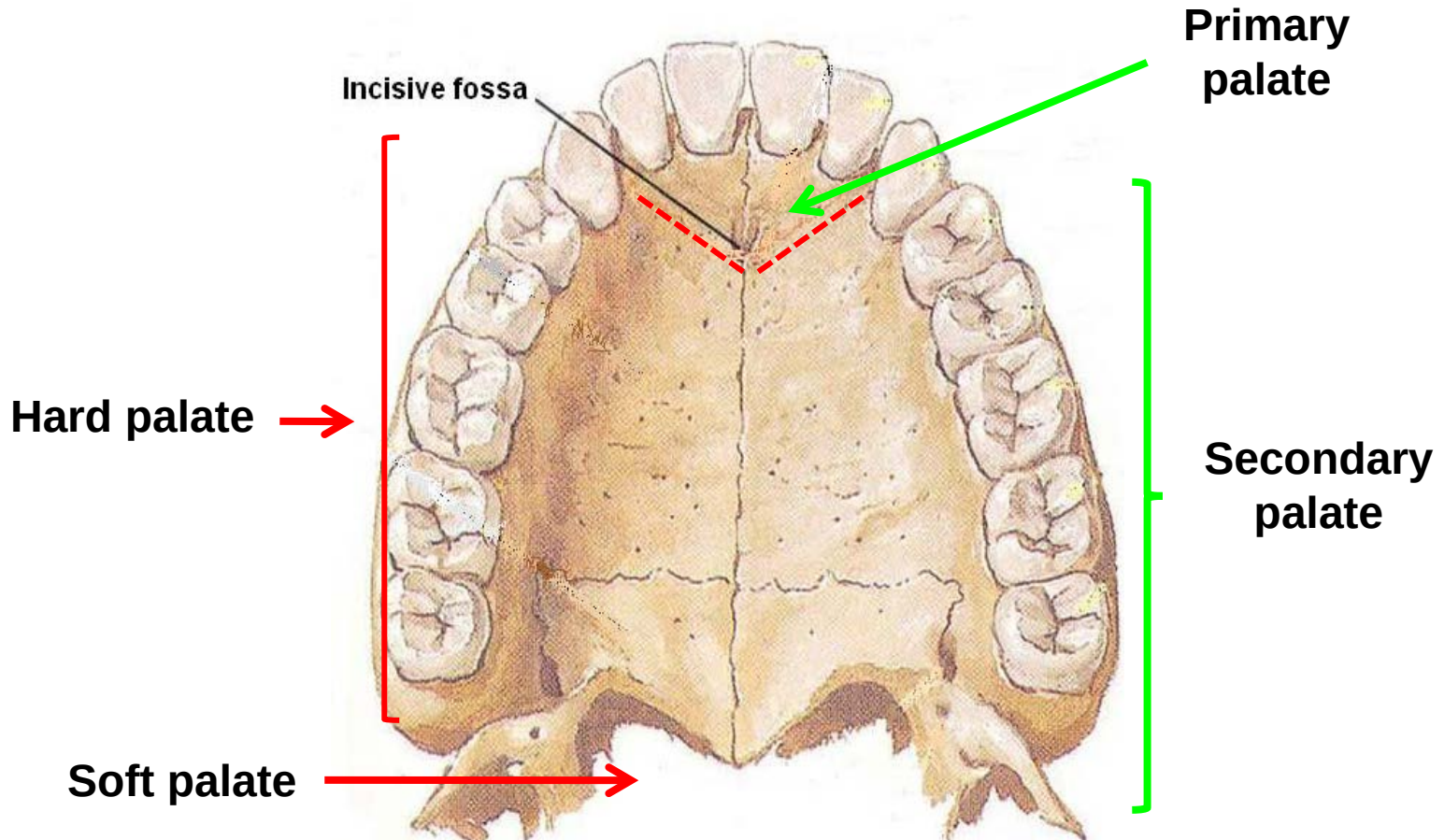
definitive palate originates by fusion of ventral edges of both lateral palatal shelves with the medial palate process

Line of fusion corresponds to the incisive canal (canalis incisivus)

The region of medial palate process (primary palate) and ventral parts of lateral palate processes undergo endesmal ossification

The posterior portions of the lateral palate processes do not undergo ossification and give rise to the soft palate and uvula





Vestibular and Dental lamina

Oral vestibule development

Oral vestibule develops from the **labiogingival lamina** (vestibular lamina)

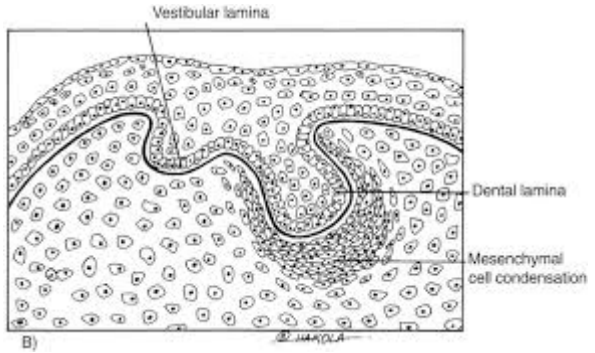
Emerges during the **6th week**

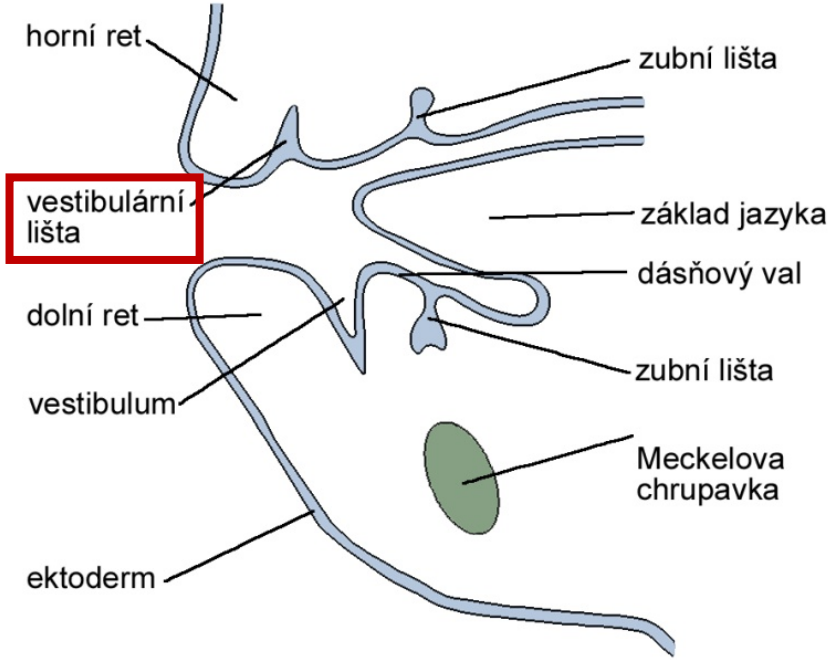
Thickened area of the ectoderm, fast proliferation of ectoderm against mesenchymal core of prominences that delineate the stomodeum

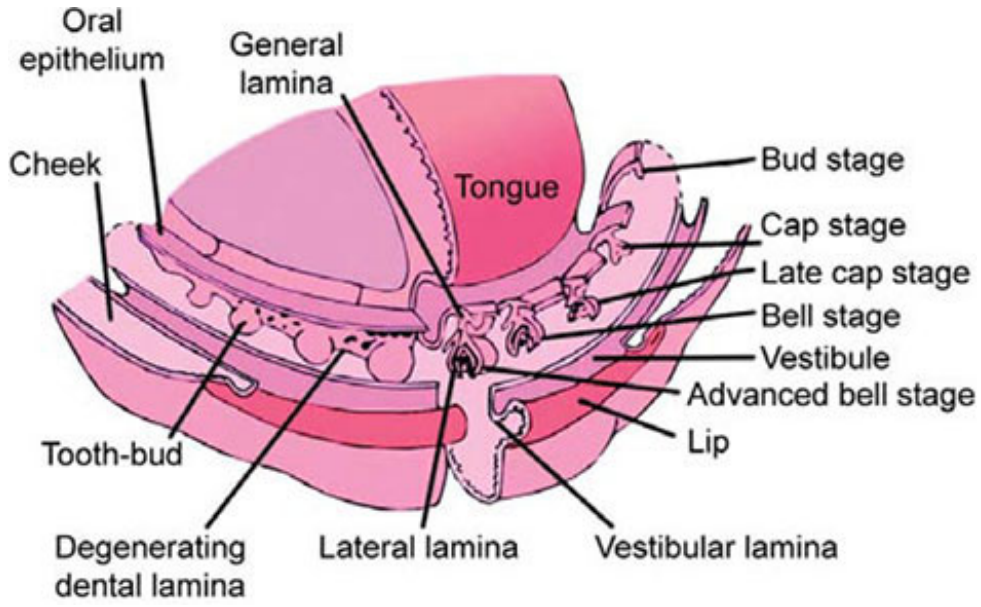
Cells in the center of lamina then undergo apoptosis - **labiogingival groove** is established

Ventral section - the definitive lip

Dorsal section - the gingival ridge (torus)







Development of maxilla and mandible

Maxilla

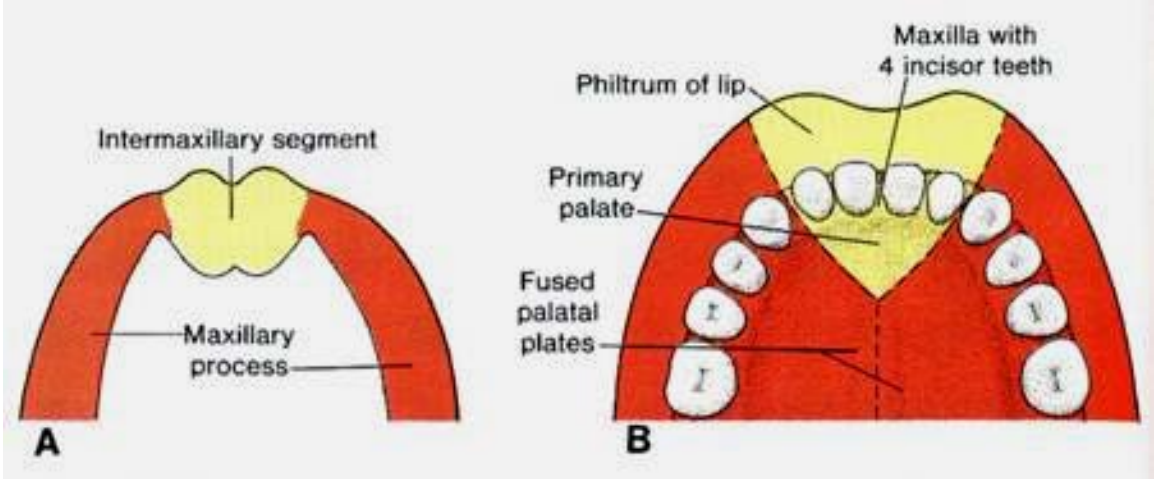
Paired bone, intramembranous ossification

Fusion of 3 parts:

Frontal part of the bone with incisors (intermaxilla) - intermaxillary segment

Lateral parts of the bone - both maxillary prominences (processus maxillares)

Lateral parts fuse to the frontal segment in incisive suture (sutura incisiva) on both sides
Ossification begins between 6 - 8 week



maxilla in newborns is shallow because has not formed alveolar processes yet (developed during the eruption of deciduos dentition)

Development of maxilla and mandible

Mandible

develops partly by intramembranous, partly by intracartilaginous ossification

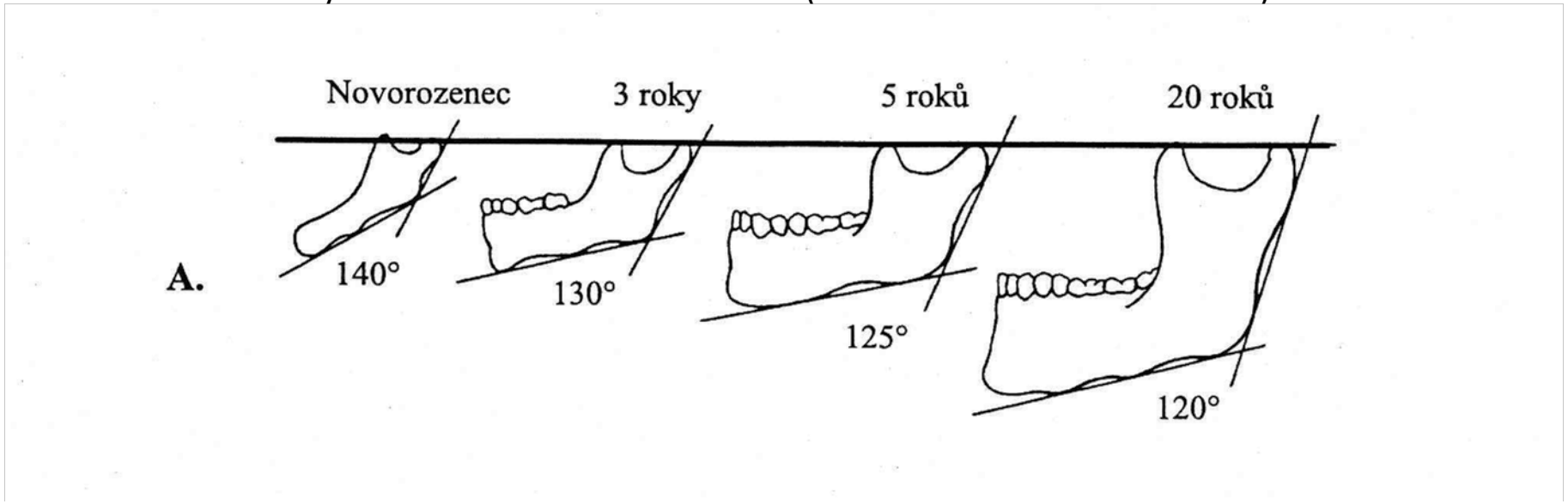
Body of mandible and both ramus of mandible are of intramembranous origin (for ossification is used mesenchyme located anterolateral to the Meckel cartilage that support the mandibular prominences

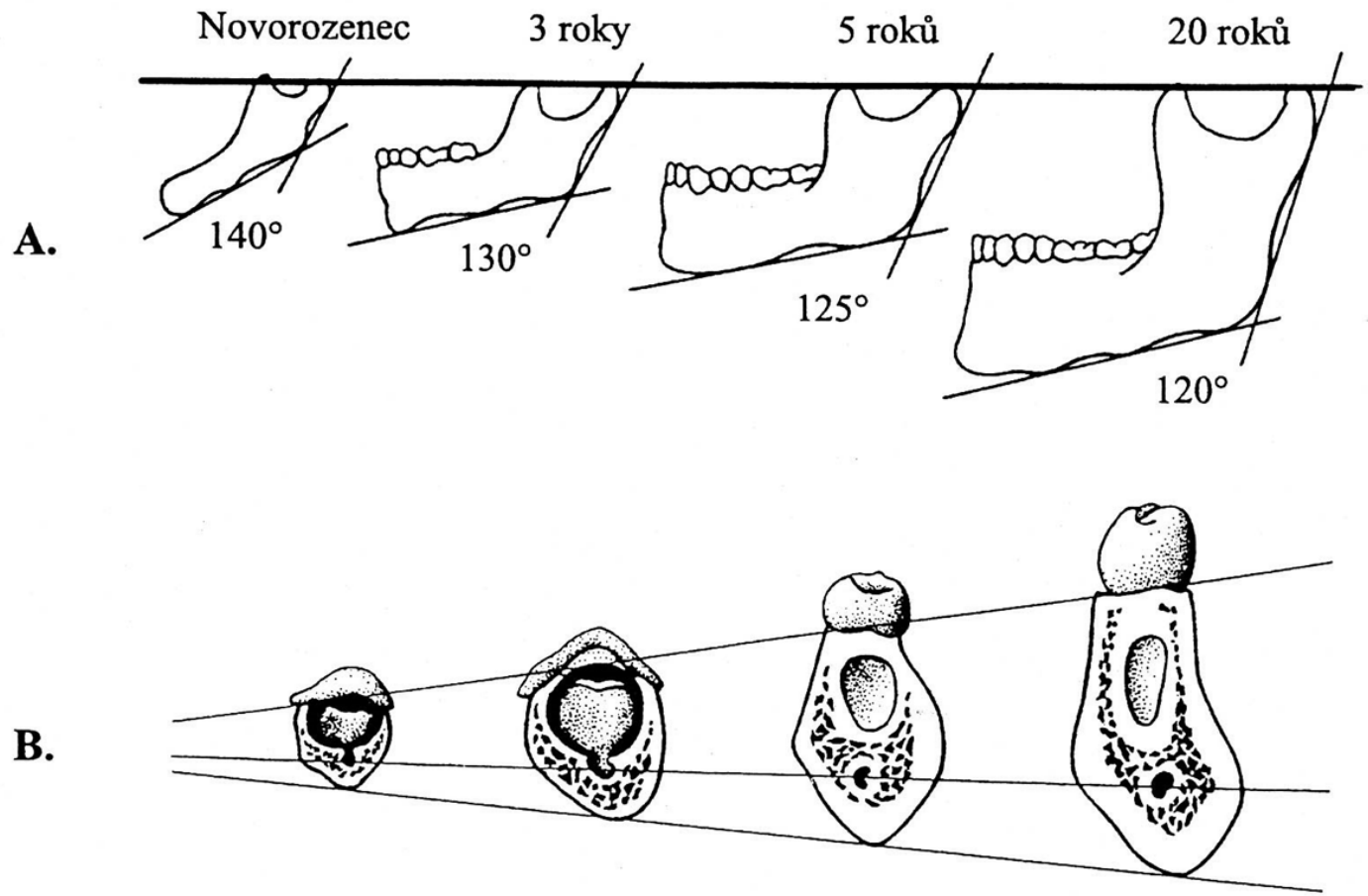
Ossification begins in the 6th week.

Condyle and coronoid process develop by intracartilaginous ossification (condyle between 12 - to 20 weeks, coronoid process yet later)

Lower jaw of neonates is low and its development continues in postnatal period

The angle between ramus and body of mandible continual reduces (from 140-150 to 120 for adult)

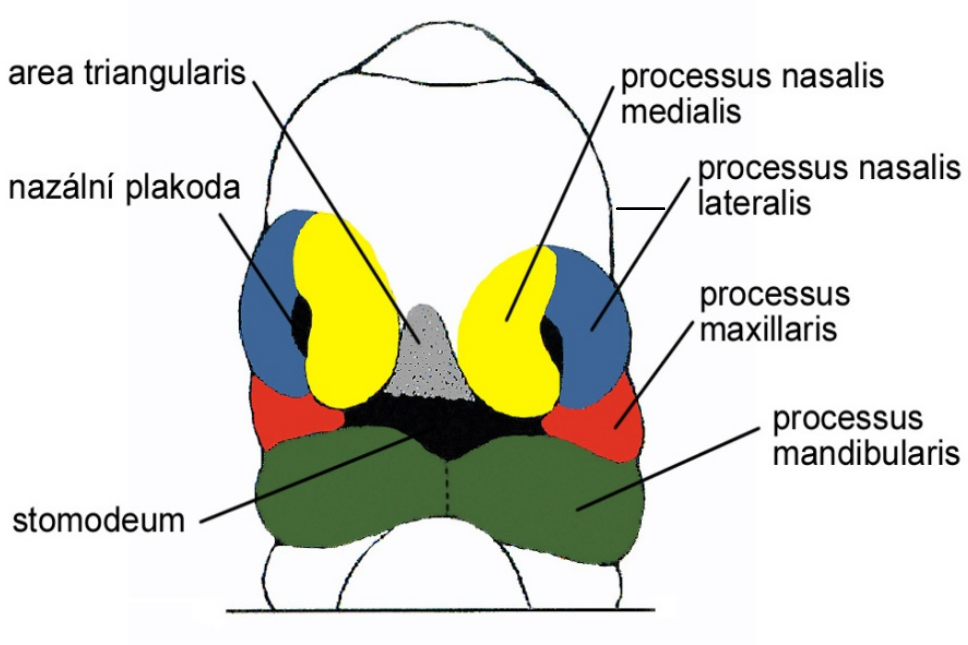


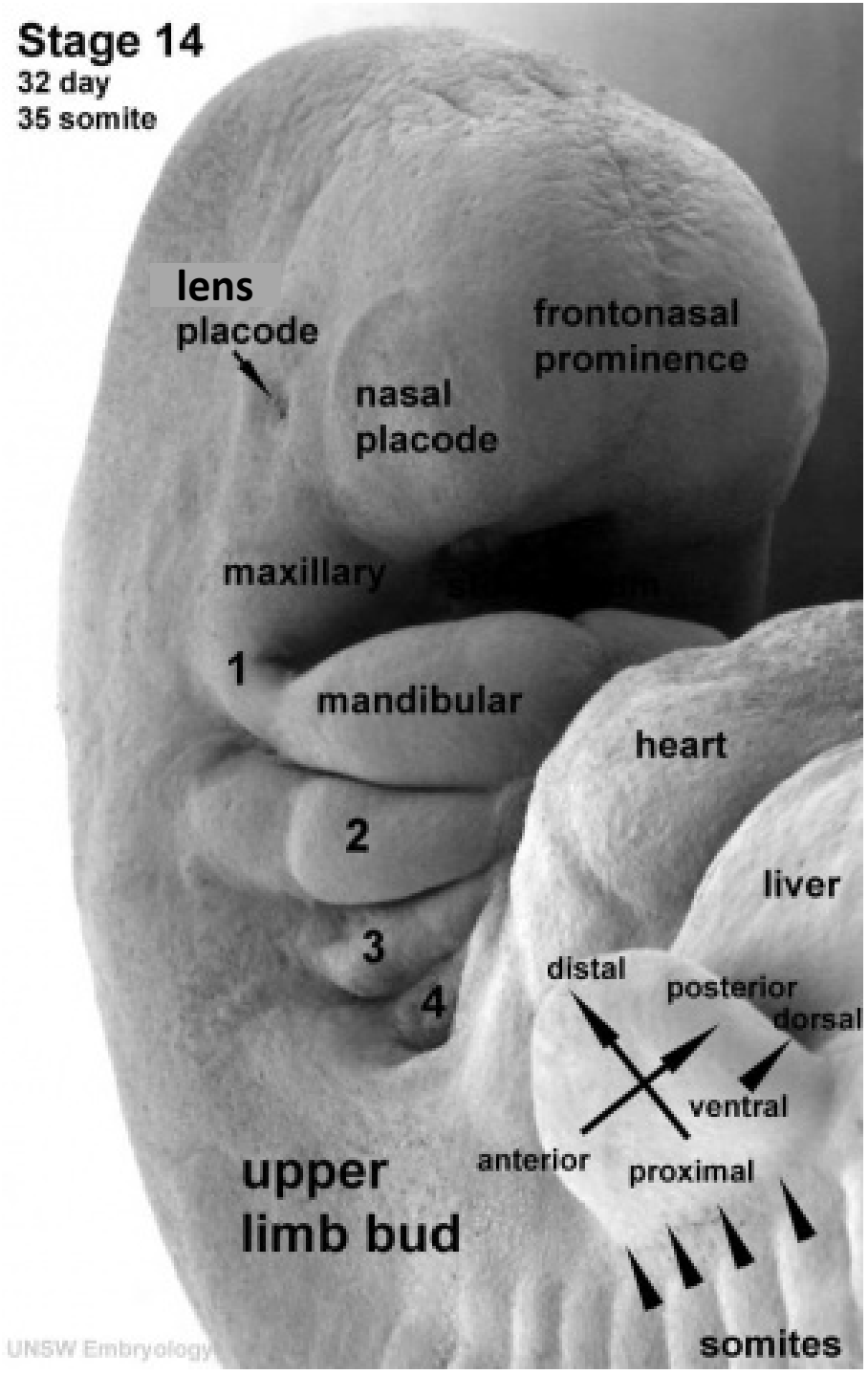


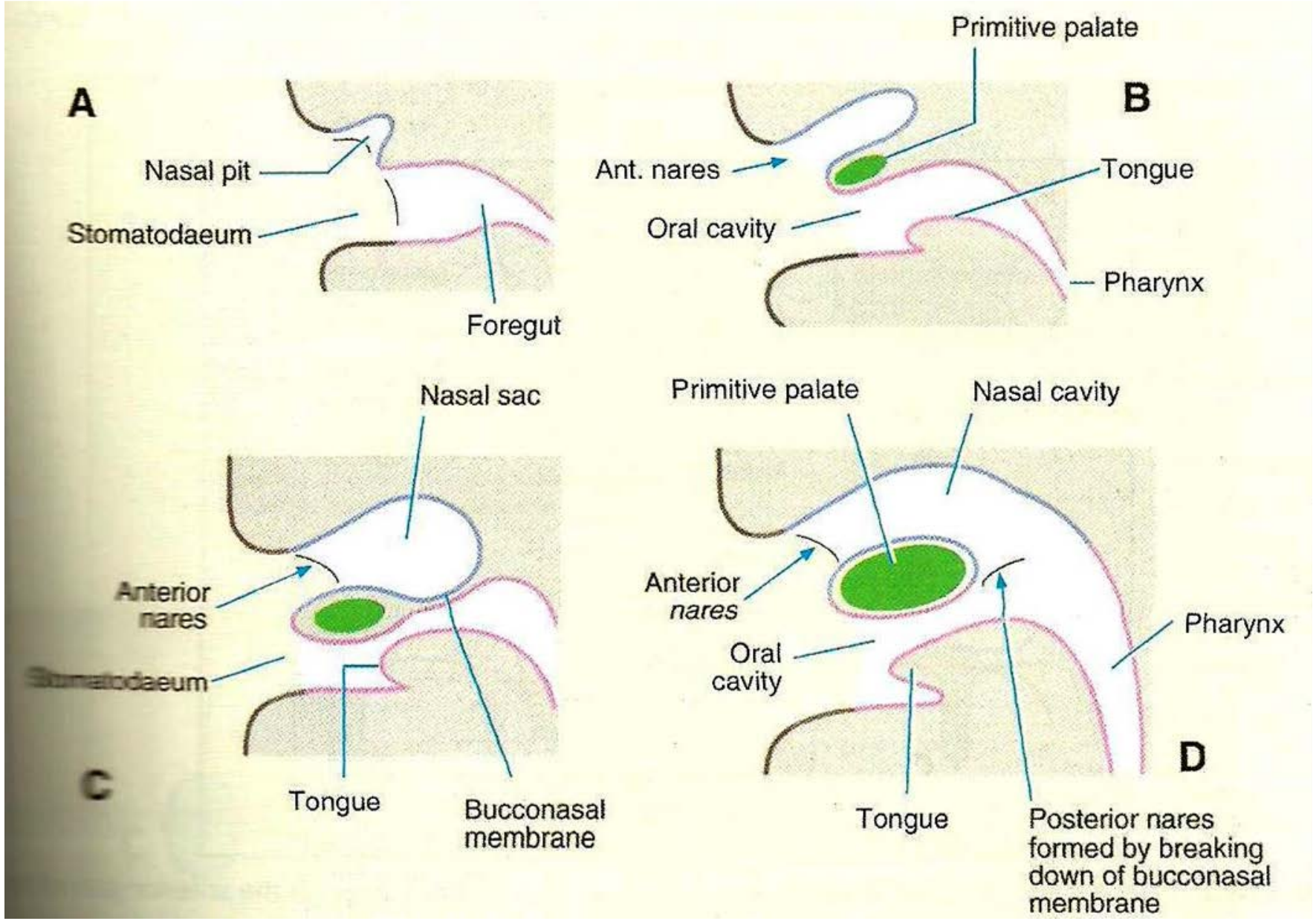
Obr. 16–3. Změny mandibuly v postnatálním vývoji. **A.** Zvětšuje se délka ramus mandibulae a zmenšuje se úhel mandibuly. **B.** Vývoj alveolární části vede k celkovému zvětšení tloušťky. Horizontální linie na obrázku prochází přes canalis mandibulae.

Nasal cavity

Nosal placodes → Nasal pits → Nasal sacs, grows dorsocaudally to roof of stomodeum, from which are initially separated by the oronasal membrane





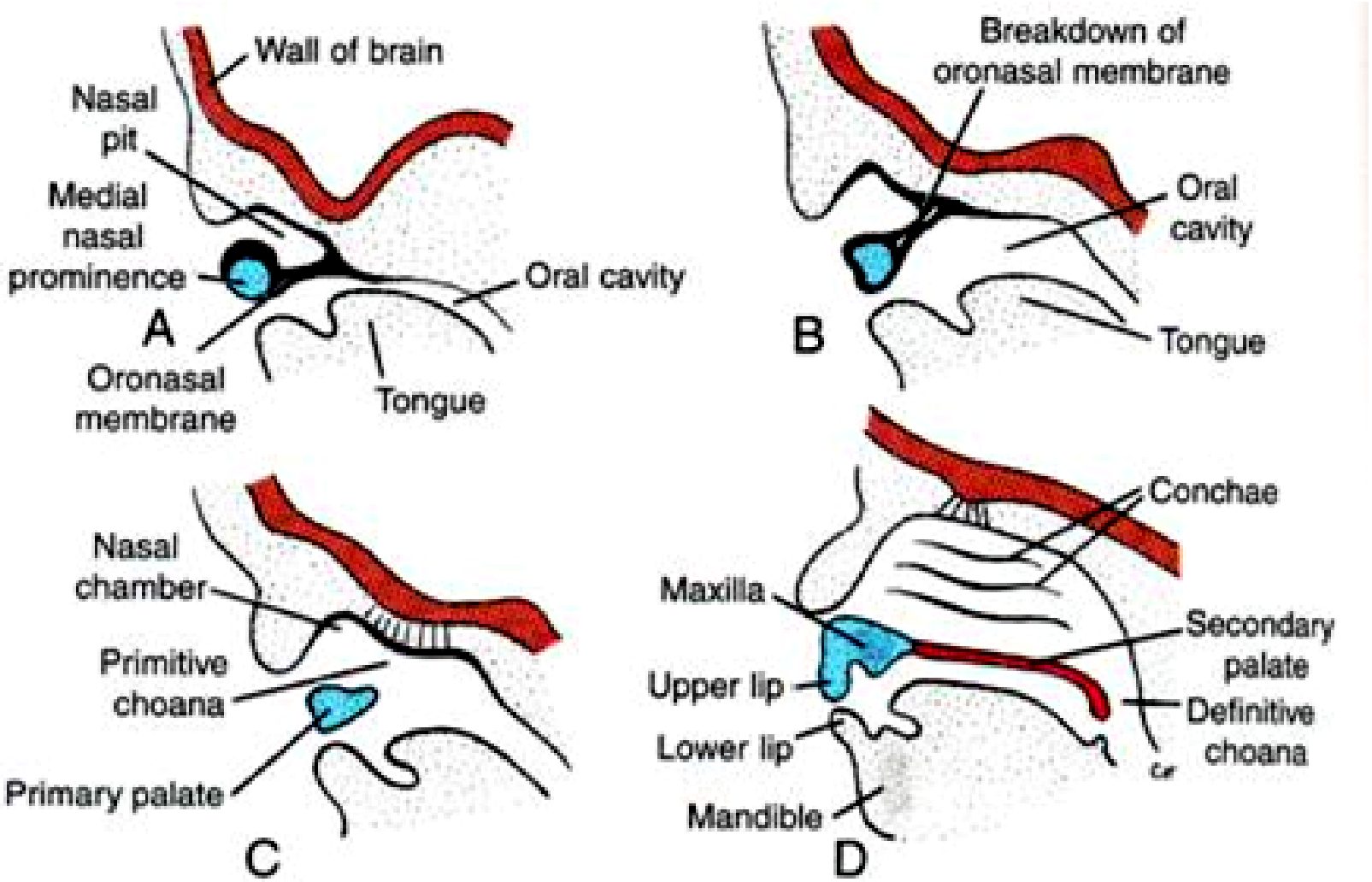


Connection of nasal and oral cavities

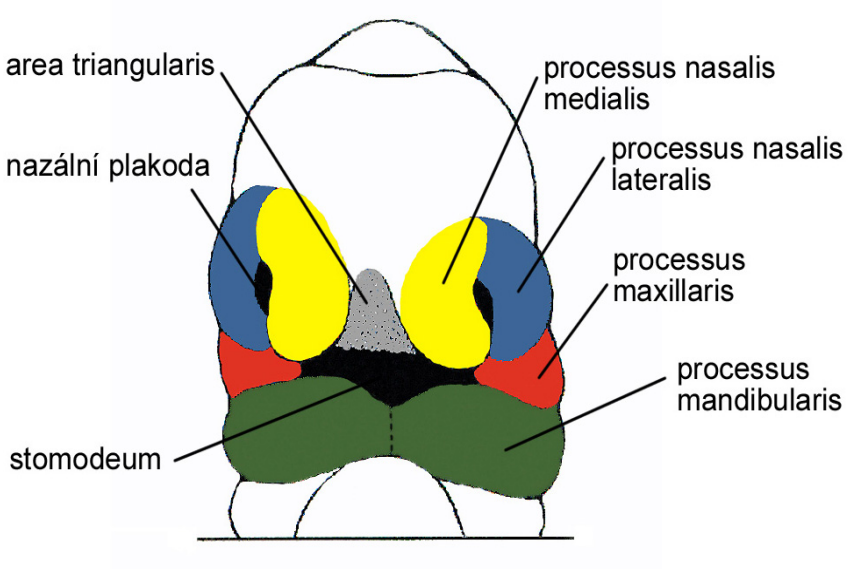
During the 5th week the oronasal membrane perforates via openings - the primitive choanae and both nasal sacs communicate to the stomodeum to form common mouth and nasal cavity (**oronasal cavity, only for +-7 days**) - see C

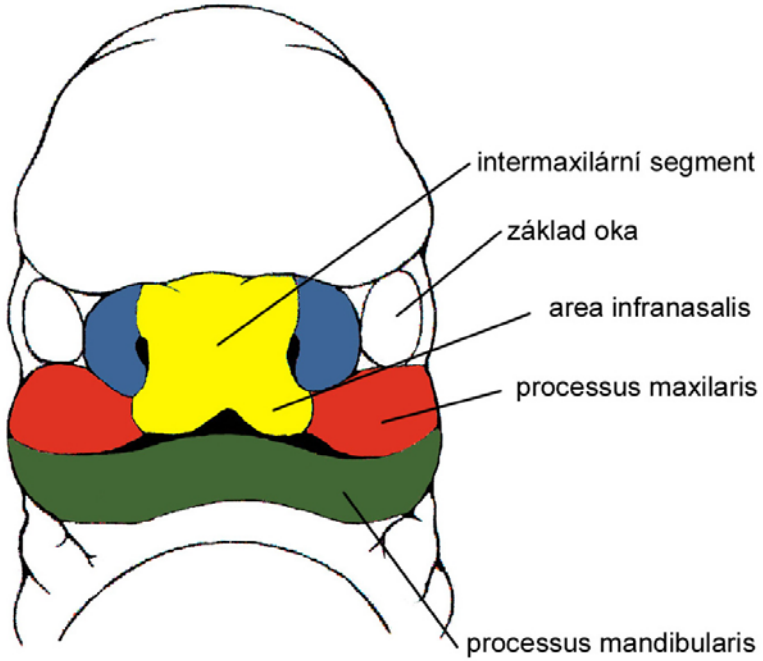
Sagittal sections through nasal pit and stomodeum:

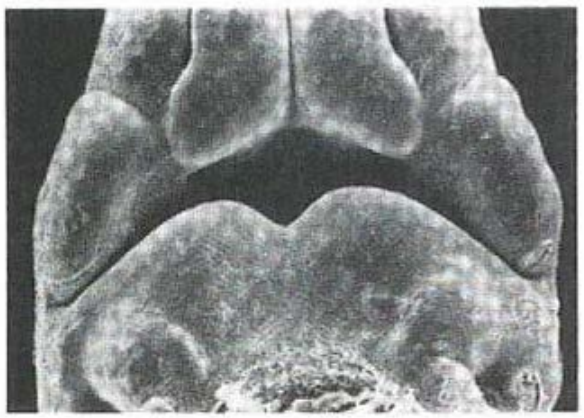
Double-layered **oronasal membrane** (ectoderm of nasal cavity and stomodeum)



Nose development







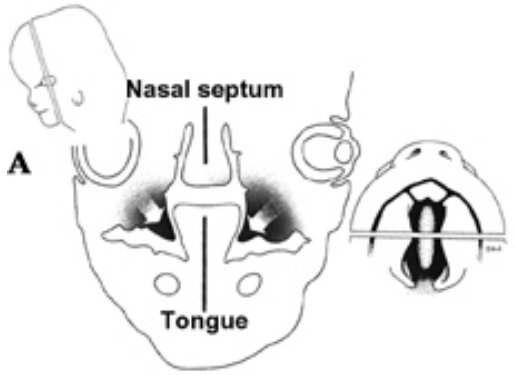
Early 7th week

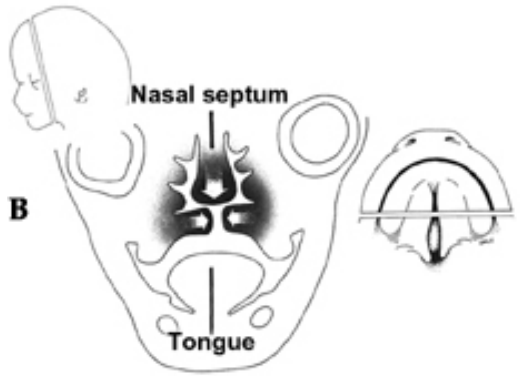
Nose develops from 3 primordia simultaneously with development of face:

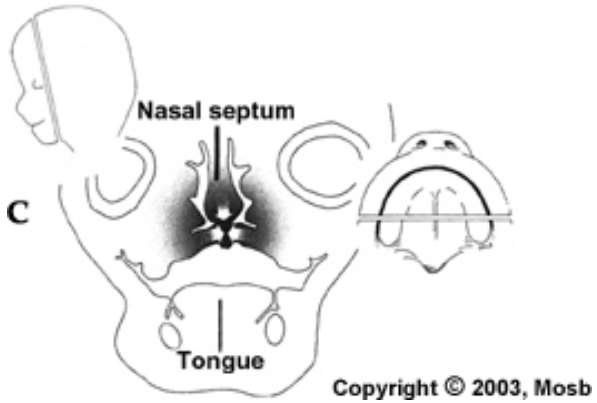
Middle and upper part of the intermaxillary segment - gives rise to the **apex**
Lateral nasal processes give rise to **Dorsum et radix nasi, alae nasi**
All primordia rapidly proliferate ventrally and nose protrudes (firstly flattened structure)

Lower part of intermaxillar segment – **philtrum**

Vývoj nosu a nosních dutin





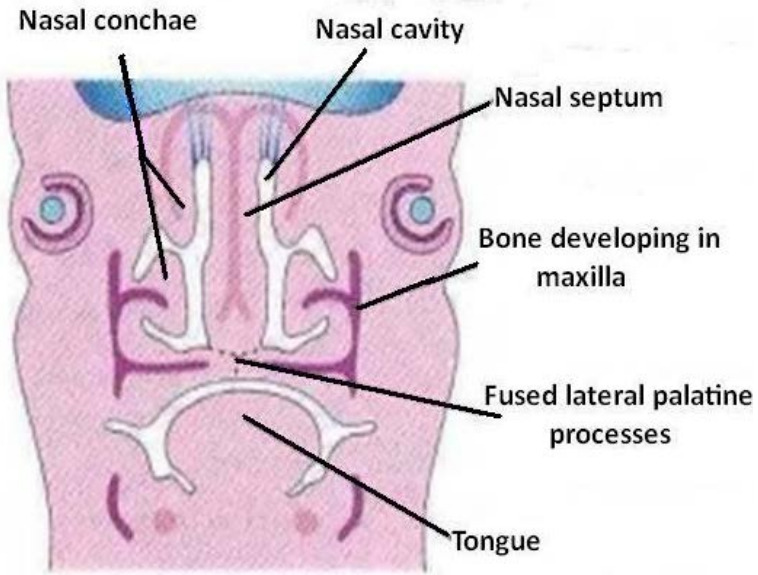


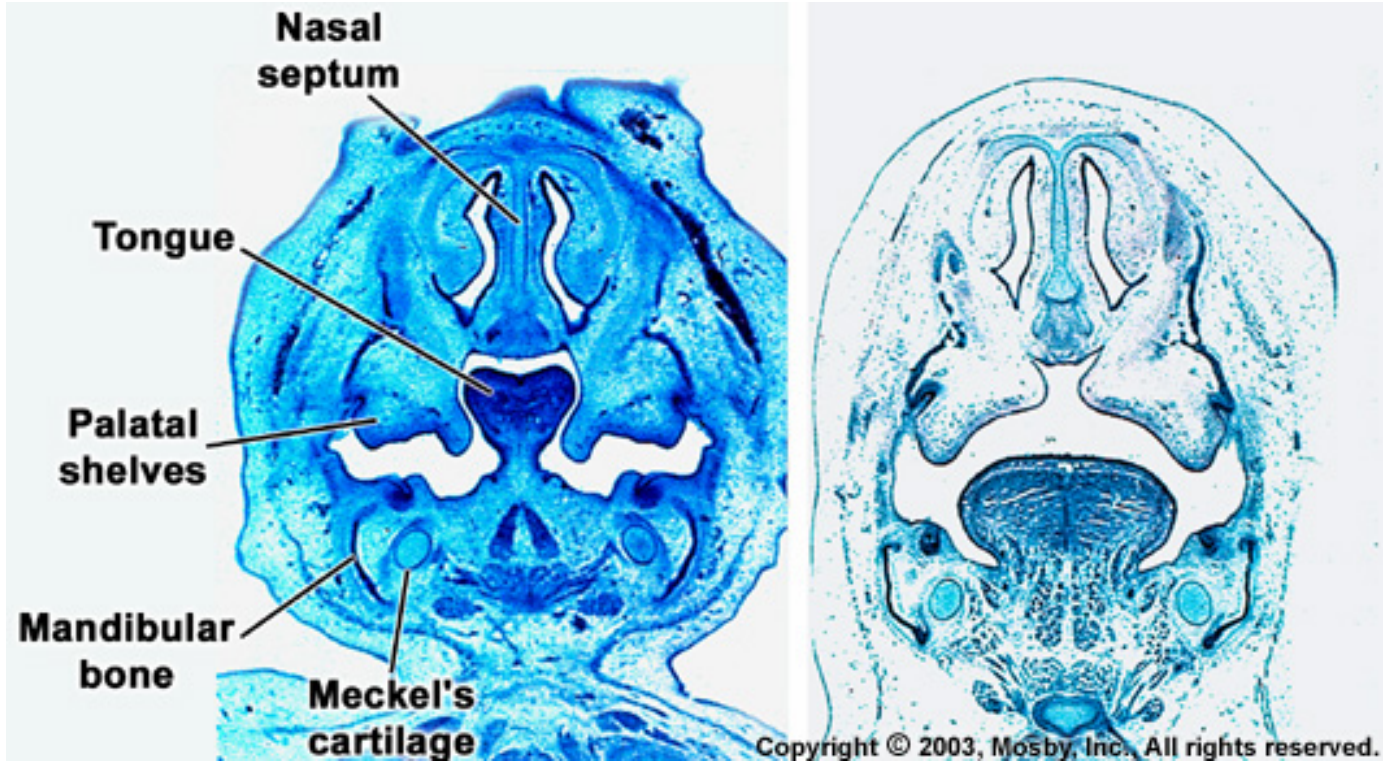
Septum nasi - grows from the intermaxilary segment in form of vertical plate, which fuses with lateral palate processes in the middle line (during 9-10th week)

At the time of development of the septum, bases of conchae develop on the lateral wall of each nasal fossa (lower, middle and upper)

After 13 week, the ectoderm covering roof of both nasal fossae transforms in the olfactory epithelium consisting of olfactory cells (unipolar neurons), whose axons constitute **fila olfactoria**

The epithelium of sinuses is of the ectodermal origin





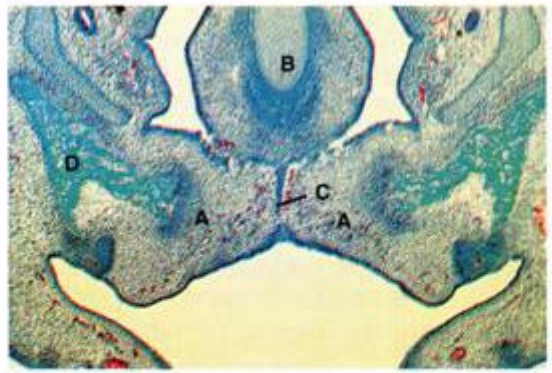


Fig. 18.5 Coronal section through developing oronasal regions following contact of the palatal shelves (A) and secondary nasal septum (B). C = Midline epithelial seam; D = developing bone of maxilla (Masson's trichrome; × 30).

Developmental defects of the nose

Defects are of rare occurrence

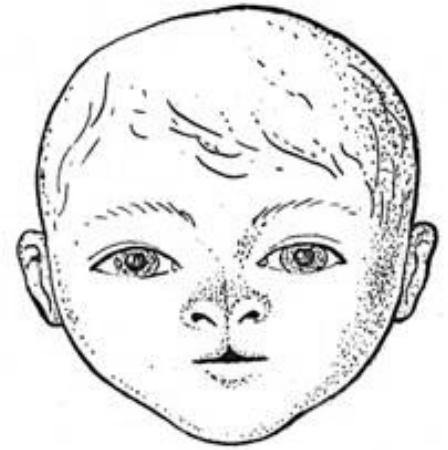
Occur separated or in association with anomalies of the upper lip and jaw or whole face

Aplasia (agenesis) of the nose - *caused by lack of nasal placodes*

Hypoplasia of the nose - a small nose with a single cavity

combined with micrognathy

Nasoschisis (nares bifides) - median cleft of the nose - caused by non-fused medial nasal prominences
The extent of cleft is variable - from shallow groove on the nose apex to the complete duplication of the nasal septum



Atresia introitus nasi (vestibuli nasi) - vestibulum nasi is closed by thin funnel shaped membrane (caused of persistence of epithelial plugs, which obturate nostrils of the fetus in the 3rd month)

Atresia choanarum – choana is closed with connective tissue membrane or bone plate

persistence of the oronasal (buconasal) membrane

1: 10 000

autosomal dominant inheritance

Other defects: nasus duplex (rhinodynia), **proboscis**

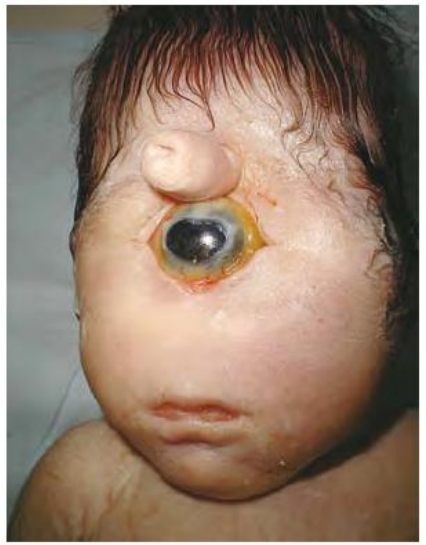
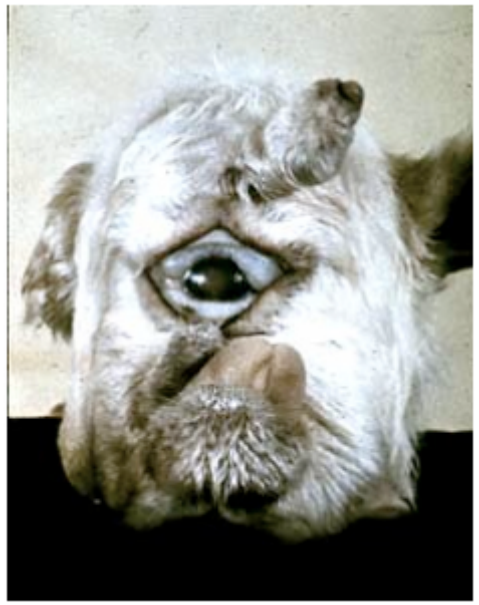
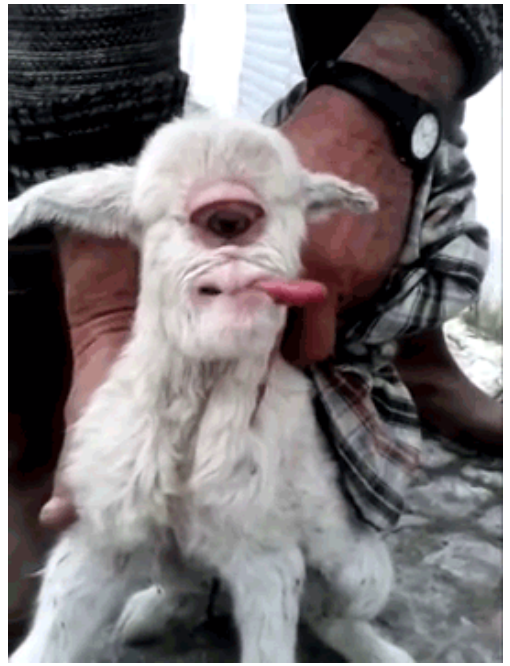


Figura 3. Foto del recién nacido. Se observa ojo único central, con probóscide, confirmando la etmocefalia.





Veratrum californicum

Congenital malformations - definition, terminology, frequency, causes

Congenital malformations are the main cause of child mortality - they account for about **1/5 of deaths**

Congenital malformation = a disorder of health (organ or system) of a structural, functional, metabolic, immunological or behavioral nature that has arisen in the prenatal phase of an individual's development

Types:

Major defects - the second half of the 3rd week to the 8th week after fertilization

Minor defects - do not cause damage to health. They occur in a later stage of development (microtia, short eye slits, pigment spots) - the presence of small defects is associated with large defects

A child with 1 small defect has a 3% probability of a large defect, 2 small defects: 10%, 3 small defects: 20%

They occur in all populations with a predictable incidence.

Many of them are inherited

Congenital malformations - definition, terminology, frequency, causes

Teratology – have a broader scope - morphological manifestations, general causes of developmental defects, classification of defects, prenatal manifestations of developmental defects, diagnosis of defects in pregnancy, registering of developmental defects, monitoring the frequency of developmental defects on an international scale

Clinical genetics – genetic risk assessment, performs diagnostics (in some cases also treatment), but mainly prevention of genetically determined birth defects

In individuals with a risk history, prevention begins preconception, continues prenatally and then postnatally (in children and adults).

Terminology of congenital malformations

Congenital malformation - any structural, functional, metabolic, or immunological disorder of an organ or part of the body

Disruption - structural or functional defects of organs, their parts (possibly certain parts of the body), caused by *environmental factors* during the originally normally started development

Teratogens (disruptors), Disruptions are not hereditary, (1940 - Gregg - rubella virus; 1961 - Lenz - thalidomide)

Deformation (regardless of whether severe or light) - shape or positional defects of an organ or part of the body caused by mechanical causes – e.g. pes equinovarus (clubfoot) - feet in plantar flexion - in oligohydramnios

Dysplasia - abnormal arrangement of cells in a tissue or organ without its obvious size or shape changes

Mutilation - distortion, mutilation (used to name congenital skeletal defects, usually of a mild nature)

Anomalies - irregularity, deviation from the rule - used to indicate the shape (numerical) deviation

Vitium – defect - is used for some congenital defects of the heart and blood vessels

Frequency in population

Statistical data:

2-3 % of live-borned babies after the 28th week have some developmental defect

In another 2-3 % of children, the defect is detected or manifested in the first years of life (up to the 5th year)

This means that 4-6% of children of the same age are affected by one or more developmental defects

Preimplantation losses that may not be recognized as pregnancy are not included here

<u>According to the incidence (frequency of occurrence), congenital malformations can be divided into:</u>

Congenital malformations with high frequency (1: 200 to 1: 400 births)

Congenital heart defects, minor skeletal mutilation (e.g. absence of a finger joint)

Congenital malformations with medium frequency (1: 500 to 1: 3000 births)

Cleft lip and oral defects, stenosis and atresia of the esophagus and intestine, rhachischisis, anencephaly, hydrocehalus, congenital malformations of the urogenital system, Down's syndrome

Congenital malformations with low frequency (1: 10,000 births)

Congenital malformations of respiratory and skin system, combined skeletal defects

Individual presence of congenital malformations

There are 3 types of occurrence in an individual:

Occurrence of only **one defect**

Occurrence of **sets of defects** for which the cause, type of inheritance and risk are known = **syndromes**

(e.g. Down's syndrome - trisomy of chromosome 21 (1: 800 births)

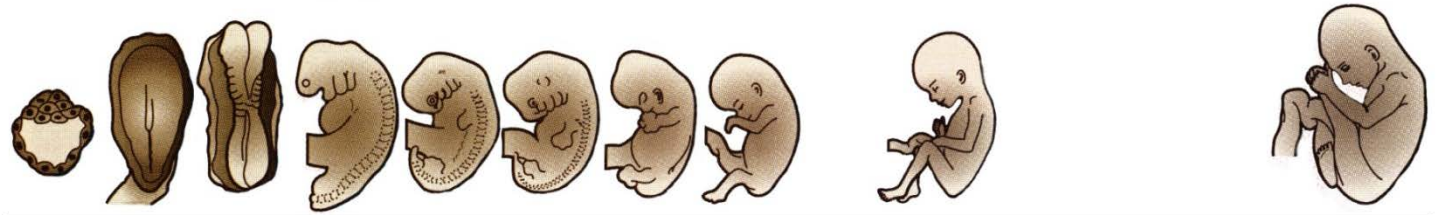
Body malformations: brachycephaly (smaller flattened head) flat nose root, oblique eye slits with skin fold in the inner corner of the eye (epicanthus), short and wide neck, protrusion of the tongue, clinodactyly of the little finger (malformation of the middle finger joint), large gap between the thumb on the feet and other toes, uninterrupted transverse furrow on the palm (so-called monkey furrow), congenital heart defects + retardation of mental development

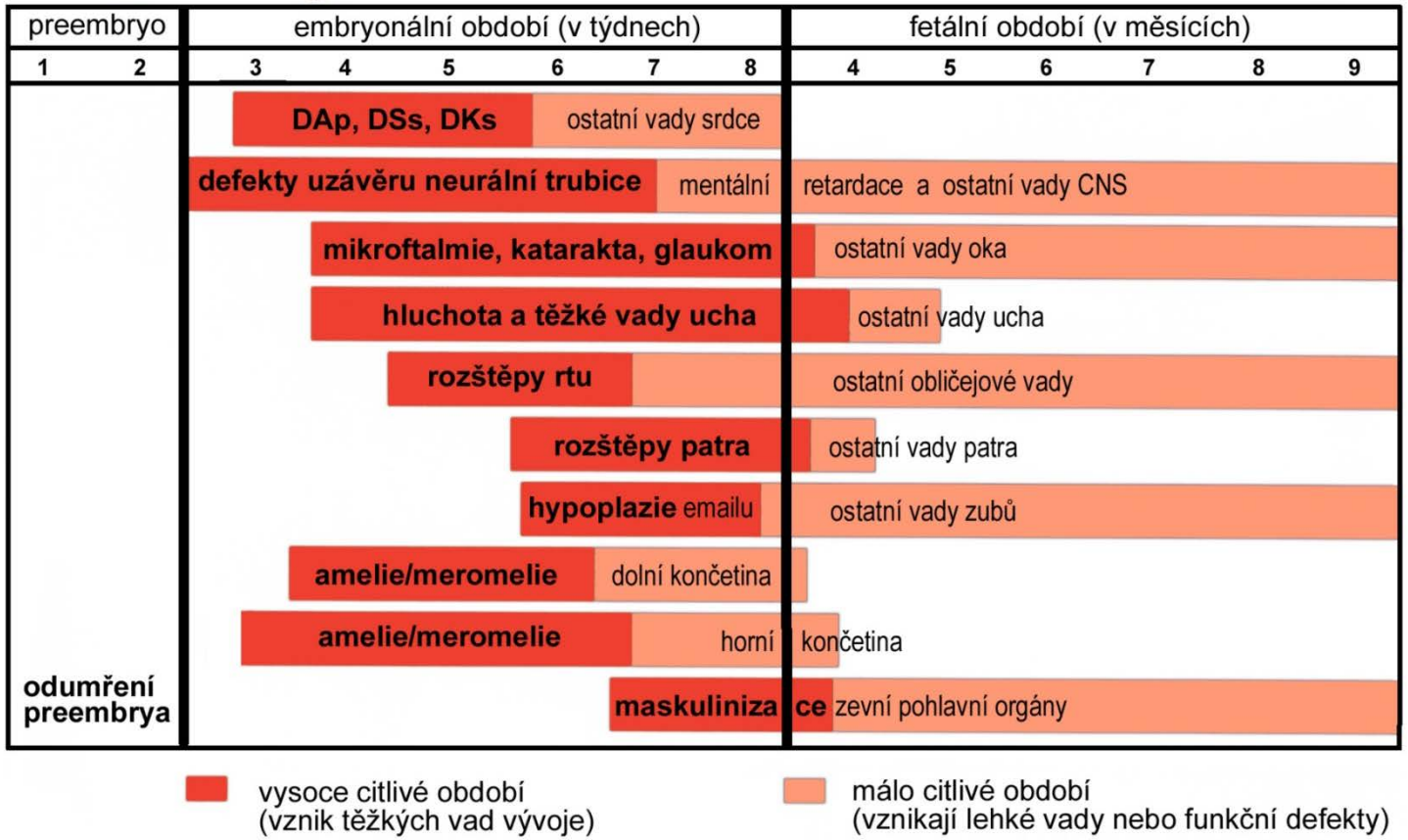
Associative occurrence of defects - non-random occurrence of two or more defects together for which the cause is not yet known

(VACTERL association - vertebral, anal, cardiac, tracheo-esophageal, renal, limbs anomalies)

Critical periods

Periods during which developing organs are more sensitive to teratogens and when a developmental defect is most likely to occur
Known for individual organs: e.g. in the brain and spinal cord - day 16 to 36, in the heart on days 19 to 38, for the eye on days 22 to 50, etc.





http://www.sci-news.com/othersciences/anthropology/newborn-babies-skills-pick-out-individual-words-06863.html