Pharyngeal Clefts

The 5-week embryo is characterized by the presence of four pharyngeal clefts, of which only one contributes to the definitive structure of the embryo.

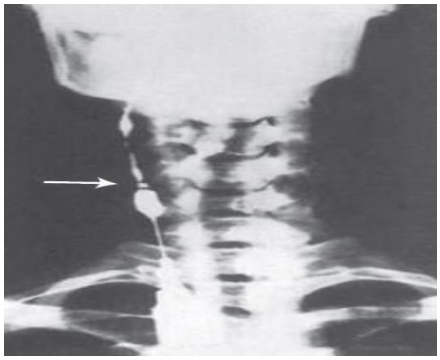
- The first cleft penetrates gives rise to the **external auditory meatus.** The epithelial lining at the bottom of the meatus participates in formation of the **eardrum**.

- Active proliferation of mesenchymal tissue in the second arch causes it to overlap the third and fourth arches. Finally, it merges with the **epicardial ridge** in the lower part of the neck, and the second, third, and fourth clefts lose contact with the outside.

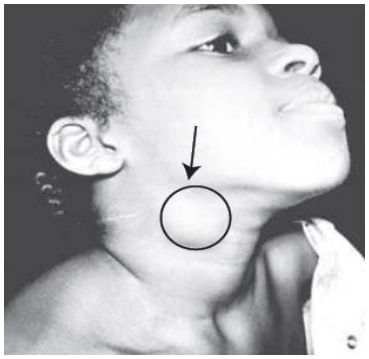
- The clefts form a cavity lined with ectodermal epithelium, the **cervical sinus,** but with further development this sinus disappears.

CLINICAL CONSIDERATIONS

A. **Pharyngeal fistula occurs** when **pharyngeal pouch 2** and **pharyngeal cleft 2** persist, thereby forming a patent opening from the internal tonsillar area to the external neck. It is generally found along the **anterior border of the sternocleidomastoid muscle**. The radiograph B after injection of a contrast medium shows the course of the fistula through the neck (*arrow*). The fistula may begin inside the throat near the tonsils, travel through the neck, and open to the outside near the anterior border of the sternocleidomastoid muscle.

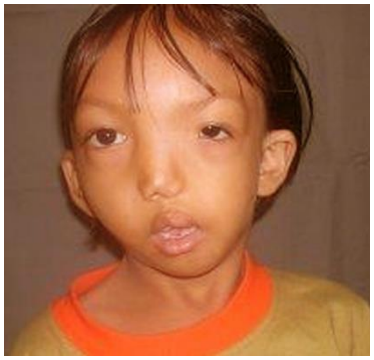


B. Pharyngeal cyst occurs when parts of the **pharyngeal grooves 2, 3, and 4** that are normally obliterated persist, thereby forming a cyst. It is generally found near the **angle of the mandible**. The photograph shows a fluid-filled cyst (*dotted circle*) near the angle of the mandible (*arrow*).



B. **DiGeorge syndrome DS (22q11 syndrome)** is caused by a microdeletion of a region in chromosome 22q11. This results in the failure of **pharyngeal pouches 3 and 4** to differentiate into the thymus and parathyroid glands.

DS is usually accompanied by facial anomalies resembling first arch syndrome (micrognathia, low-set ears) due to abnormal neural crest cell migration, cardiovascular anomalies due to abnormal neural crest cell migration during formation of the aorticopulmonary septum, immunodeficiency due to the absence of the thymus gland, and hypocalcemia due to the absence of parathyroid glands.



d. **Ectopic Thymic and Parathyroid Tissue** Because glandular tissue derived from the pouches undergoes migration, it is not unusual for accessory glands or remnants of tissue to persist along the pathway. This is true particularly for thymic tissue, which may remain in the neck, and for the parathyroid glands.

E.Oculoauriculovertebral spectrum (Goldenhar syndrome)

It includes a number of craniofacial abnormalities that usually involve the maxillary, temporal, and zygomatíc bones, which are small and flat. Ear (anotia, microtia), eye (tumors and dermoids in the eye ball], and vertebral (fused and hemivertebrae, spína bifida) are commonly observed in these patient.

Asymmetry is present in 65% of the cases. Other malformatíons, which occur in 50% of cases, include cardiac abnormalities, such as tetralogy of Fallot and ventricular septal defects.