Congenital conditions of the Head and Neck

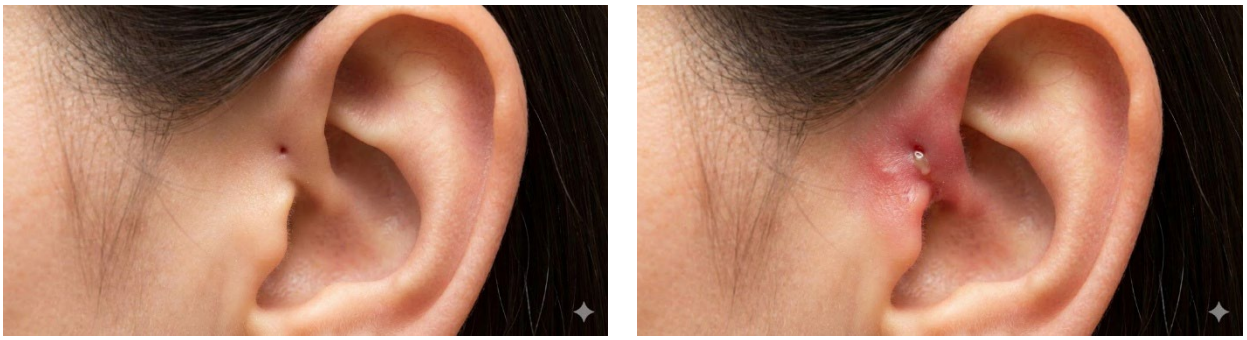
It is impossible to discuss all the congenital conditions of the Head and Neck. The most important will be covered here.

Congenital conditions

Origin	Conditions
Pinna	Pre-auricular pits, sinus and cysts
Branchial apparatus	Branchial clefts and pouch defects
Skin	Dermoid and Epidermoid cysts
Thyroid	Thyroglossal duct cyst
Lymphatic	Macro Micro Mixed
Vascular	Haemangioma Capillary Venous AV fistulas
Larynx	Laryngomalacia Vocal cord palsy Web Subglottic stenosis
Mouth	Ankyloglossia Torus

Pre-auricular

Pre-auricular cysts, also known as pre-auricular pits or sinuses, are benign congenital malformations that occur in the soft tissues in front of the ear. They are characterized by small openings that may lead to a sinus tract under the skin, which can sometimes become infected or form cysts. Most individuals with preauricular cysts are asymptomatic and may not realize they have them until an examination reveals the condition or an infection occurs. Once infected, the treatment of choice is antibiotics and / or aspiration. Try to avoid an incision and drainage, as this will results in a higher recurrence rate after formal surgical removal. Uninfected ones are surgically removed if the patient desires so.



The picture on the left shows a pre-auricular pit / sinus, that is infected on the right. Often, there can be a palpable / infected cyst as well.

Branchial apparatus conditions

Epidemiology

- 19% of paediatric cervical masses
- 2-3% are bilateral
- Tendency to cluster in families
- Manifest mostly in early adulthood
- Peak incidence 3rd decade
- No ethnic or gender predilection

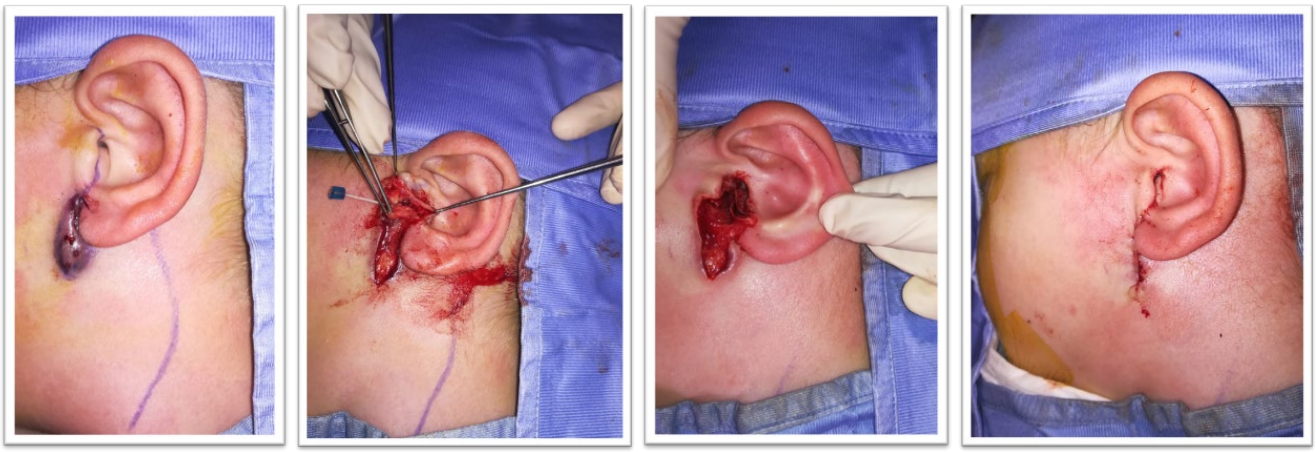
Presentation:

- Mostly asymptomatic lumps or sinuses
- Lateral neck cyst
- Infection or enlargement
- Lateral neck fistula / sinus
- Starts to drain

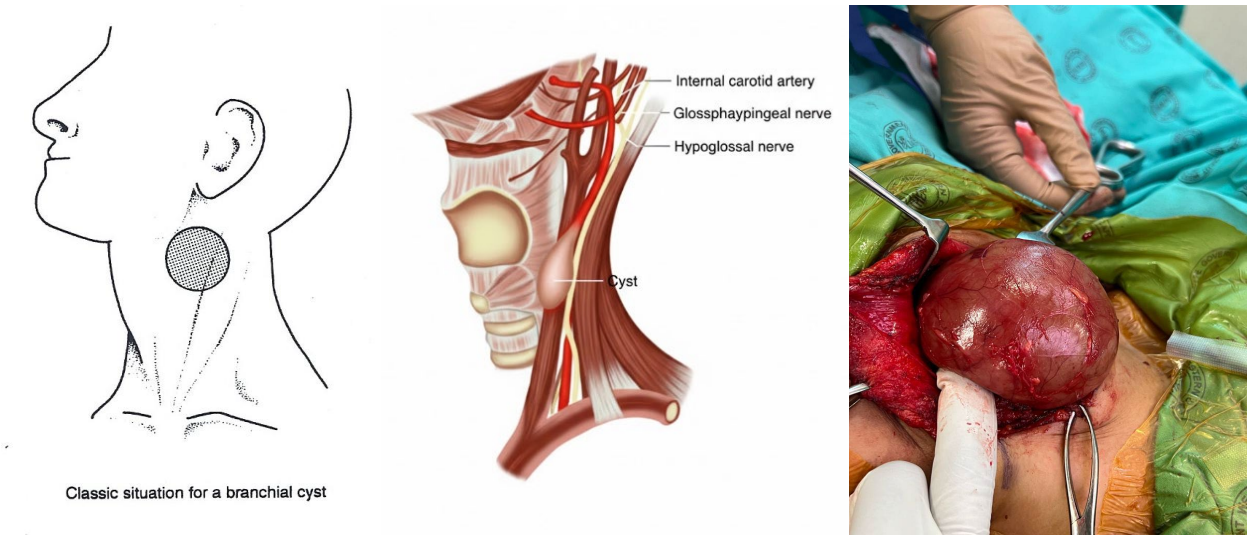
Types:

- First (5-8%)
  - External opening lies on a line between tragus and hyoid bone
    - Can also be post-auricular
    - Often inconspicuous
  - May be a second opening (fistula)
    - Anterior to tragus
    - In EAC at the osseocartilaginous junction
  - Classified into 2 types by Work (1972)
- Second (90-95%)
  - Commonest of the branchial cleft anomalies
  - Sinuses are more common
    - Open low in the neck
    - Anterior to the SCM
    - Leak mucoid or clear fluid
  - Fistulas are rare
    - Not always patent throughout length
    - Communicate with supratonsillar fossa
  - Cysts
    - Inferior to angle of mandible and anterior to SCM
  - Association with branchio-oto-renal syndrome
- Third (rare)
- Fourth (rare)

First branchial cleft operation type I

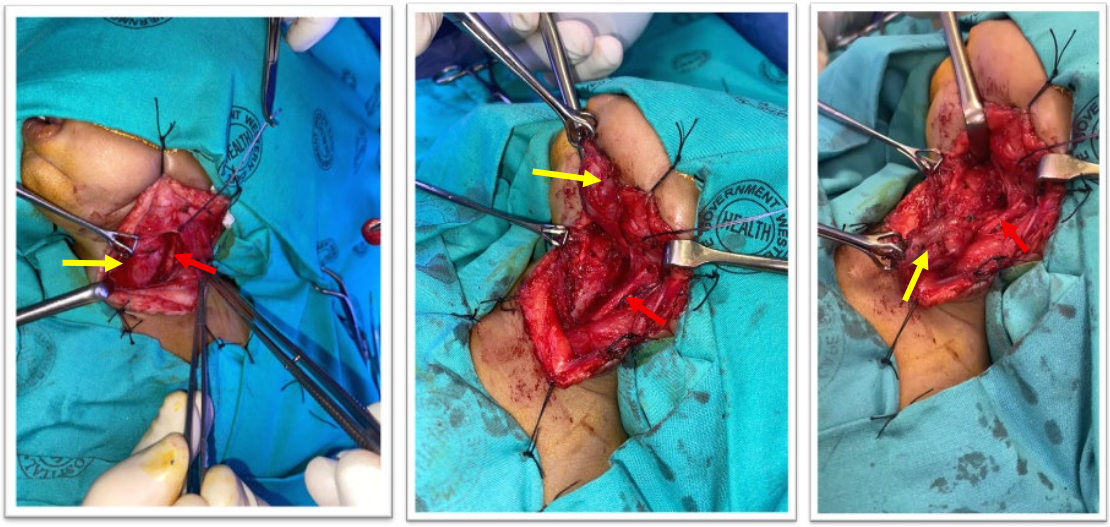


Second branchial cleft classic position and operation



Three pictures showing the classical position on the left side (level IIa), its embryological pathway and tract with a possible opening at the supratonsillar fossa, and an intra-operative picture of a very large second branchial cleft cyst.

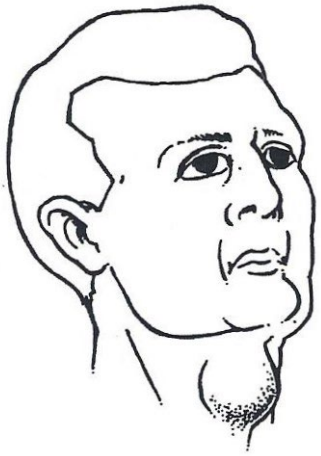
Fourth branchial cleft operation



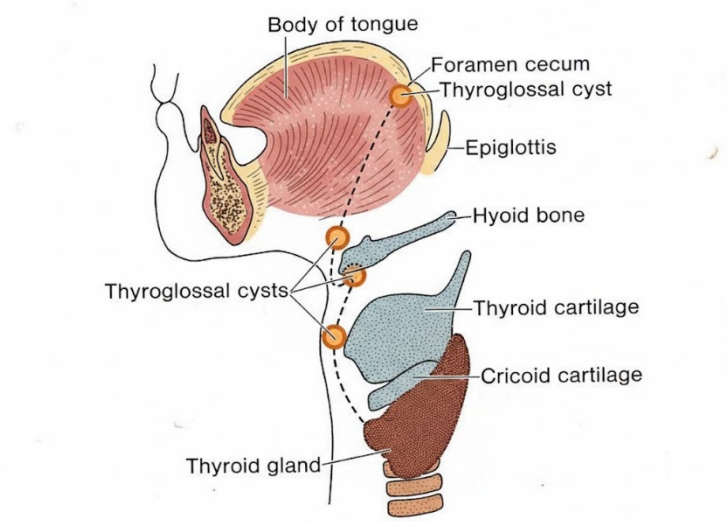
A fourth branchial cleft cyst. The yellow arrow points to the cyst and the red to the carotid artery.

Thyroglossal duct cysts

It is said to compose one third of all congenital neck masses. Typically, in the midline position but can be off to the side in some cases. Rarely occurs in children and typically seen in young adults as the cyst accumulate enough mucous. It can rapidly enlarge when cysts become secondarily infected or when there is haemorrhage. There is a very small chance of developing into a thyroid cancer. It can occur anywhere along the line of development of the thyroid between the base of the tongue and the thyroid itself (see picture below). Occasionally there is an associated fistula, due to the cyst having burst. Surgical excision is the preferred treatment and entails removal of the cyst and its tract, with the body of the hyoid bone (Sistrunk operation). However, before surgery it is important to investigate if the thyroid gland is in its normal position.



Thyroglossal cyst This is the usual site. In addition to moving with swallowing, a thyroglossal cyst also moves upwards when the tongue is protruded.



Embryological tract of the descending thyroid gland, with typical areas where a thyroglossal duct cyst can occur. The most common area is over the hyoid bone.

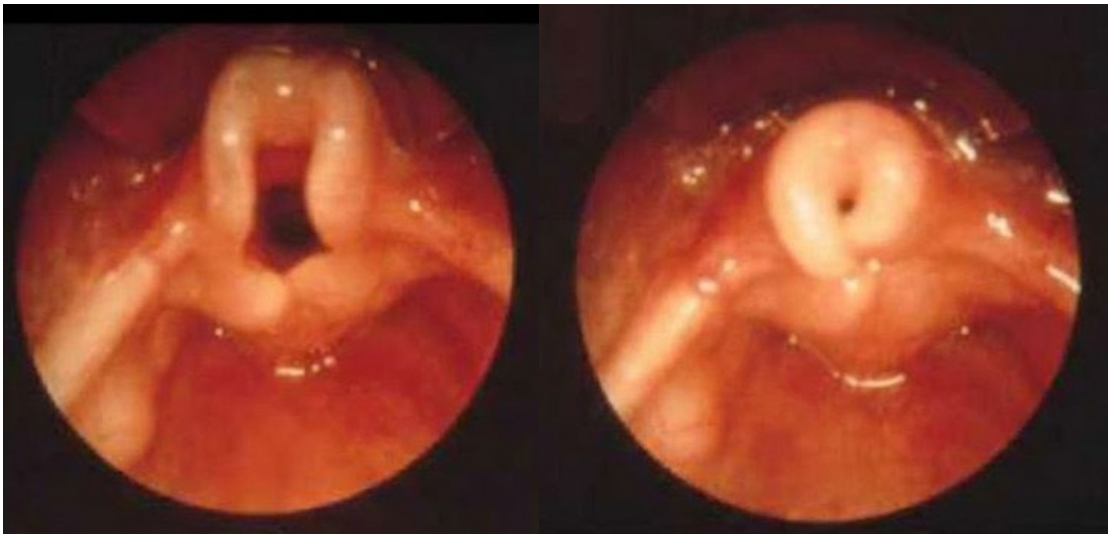
Laryngomalacia

Laryngomalacia is the most common reason for stridor in children. It causes 60% of all laryngeal disease in children. Typically, the children develop an intermittent, low tone, inspiratory stridor within 2 weeks after birth (the stridor is typically not present at birth). In general, it worsens up and to 9-12 months and 75% will clear up

spontaneously at 18 months. Rarely it can last up to 5 years. Signs of a complicated laryngomalacia (and therefore referral to an ENT) include:

- Feeding problems
    - As long as the babies does not need to "come up for air" while breast or bottle feeding, interventions are usually not needed.
- Failure to thrive
- Apnoea
- Cyanosis
- Pectus excavatum

Various theories try to explain the underlying pathology such as a neuro-muscular problems, floppy cartilage, and hypotonia. The most common finding is that of an omega shaped epiglottis, but various others have been described such as short aryepiglottic folds, posterior collapse, mucosa prolapsing into larynx from the arytenoids, and a deep narrow glottic inlet (see pictures below). Mild laryngomalacia will improve with crying, as opposed to severe forms which will worsen with crying. The babies also improve when turned to the prone position. Reflux plays an aggravating role, and babies are routinely put onto PPI. Only 20% will require intervention and it is usually the group with complicated features mentioned above. Therefore, the majority is managed by explaining the disease process to the parents, given advice regarding PPI and positing of the child, and follow up.



Two pictures demonstrating the typical omega shaped epiglottis on the left and near complete closure on inspiration on the right (causing the stridor).

Vocal cord palsy

It is the second most common congenital condition in the larynx after laryngomalacia. It is most commonly due to intracranial, mediastinal, or heart pathologies. This includes Arnold Chiari malformations and various heart defects. The clinical picture and management depend on whether it is unilateral or bilateral.

	Unilateral	**Bilateral**
Presentation	Fairly asymptomatic except for weak cry and aspiration	Can have catastrophic stridor
Spontaneous recovery	70% of non-iatrogenic palsies clears up in 6 months	50% of non-iatrogenic palsies clears up in 24-36 months
Intervention	Rarely interventions	Frequently a tracheostomy is placed for the child to grow to such a age (1-2 years) that a definitive procedure is planned

Congenital subglottic stenosis

Congenital subglottic stenosis is the most common reason for kids under the age of 1 year to require a tracheostomy. It comprises 20% of all congenital laryngeal conditions and frequently occurs with other problems such as Down syndrome and laryngeal clefts. It is due to a narrow cricoid (< 3.5 mm lumen in term babies) and presents with bi-phasic stridor. Sometimes kiddies present with prolonged croup-like symptoms. Any child with upper airway symptoms should be referred to an ENT specialist. Management entails waiting until the child grows and then performing an enlarging procedure either endo-luminal or external. In most cases cartilage grafts are used to achieve this.

Congenital haemangiomas

Is said to be the most common neonatal tumour and occurs in 4-10% of children below the age of one year. It is more common in females and 50% of them occurs in the head and neck area. The most common area affected in the head and neck is the subglottic area. Haemangiomas in trigeminal distribution should raise the suspicion of a subglottic haemangioma. Currently, congenital haemangiomas are divided into rapidly involuting, non-involuting, and partially involuting. Despite these different types, classical teaching involved that they enlarge up until the age of one year. Thereafter, 50% would disappear at the age of five years, 70% at seven years, and 90% and 9 years respectively. Management is complex, and these patients should be referred to ENT paediatric units.

Ankyloglossia

Ankyloglossia is commonly known as “tongue tie”. It is due to failure of tongue to separate from the mouth floor and occurs in varying degrees. The most common presentation is a short frenulum that can impair sucking and later on speech development. There is some debate whether it can cause teeth alignment problems. The prevalence is estimated at 4-11% but the cause is still unknown although genetics may play a role as it tends to occur in families. Sometimes it occurs in conjunction with upper lip tie as well. In general, it will be more severe in the first-time breast-feeding mother.

Important points to ask and examine are the following:

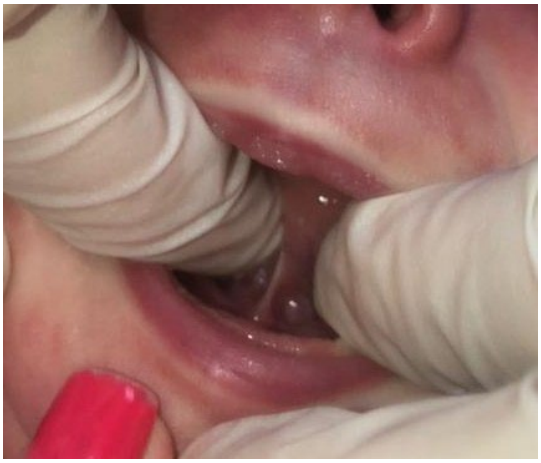
- Baby symptoms
    - Not sufficient weight gain
    - Aerophagia
    - Numerous feeding times required
    - Stop-start feeds
- Maternal symptoms and signs
    - Breast pain
    - Nipple problems
    - Breast does not empty fully with feeding
    - Mastitis

Various classifications exist, but the two most often used are:

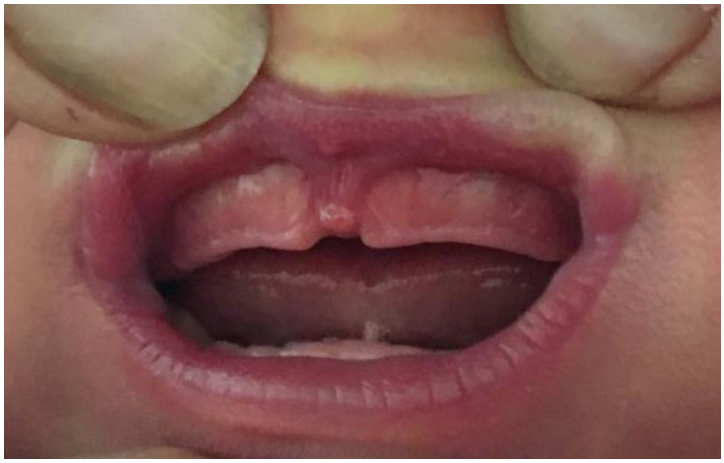
- Coryllos criteria
    - I – Frenulum attached to tip of tongue
    - II – Frenulum 2-4 mm behind tip of tongue
    - III – Frenulum attached to mid tongue
    - IV – Frenulum attached to base of tongue, but thick and inelastic
- Kotlow upper lip
    - I – No significant attachment
    - II – Attachment mostly into the gingival tissue

- III – Attachment in front of the anterior papilla
- IV – Attachment into the papilla or extending into the hard palate

The management of the babies remains controversial, mainly because of lack of high-level evidence. However, it is estimated that the USA will save $13 billion if 90% of mothers complied with exclusive breastfeeding for 6 months. In reality, 80% of mothers who initiate breastfeeding but will stop if tongue tie is not corrected. Maternal nipple pain is a sensitive indicator for tongue / lip problems. Furthermore, isolated posterior tongue-tie problems have been neglected / missed in the past as these babies often present with aerophagia and subsequent abdominal cramps and vomiting. The authors advise that patients should be referred to an ENT with a keen interest in the subject.



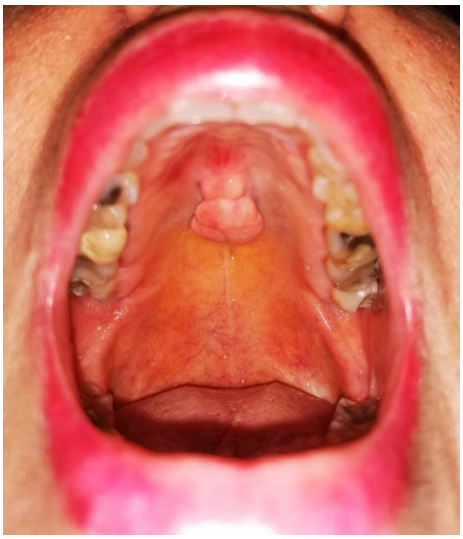
Picture showing a tongue tie (left).



Picture showing a lip and tongue tie (right).

Torus

Torus mandibularis and palatine are benign bony overgrowth. They typically occur on the inner table of the mandible and on the hard palate. The mucosa over them may be injured causing an ulcer. Surgical removal is indicated if they are symptomatic.



Picture showing a torus palatine.