Common congenital malformations of the nose

Congenital bony stenosis/atresia

Congenital nasal bony stenosis usually presents in the neonatal period with stertor, respiratory distress, failure to thrive, difficulty feeding, or as an airway emergency at birth.

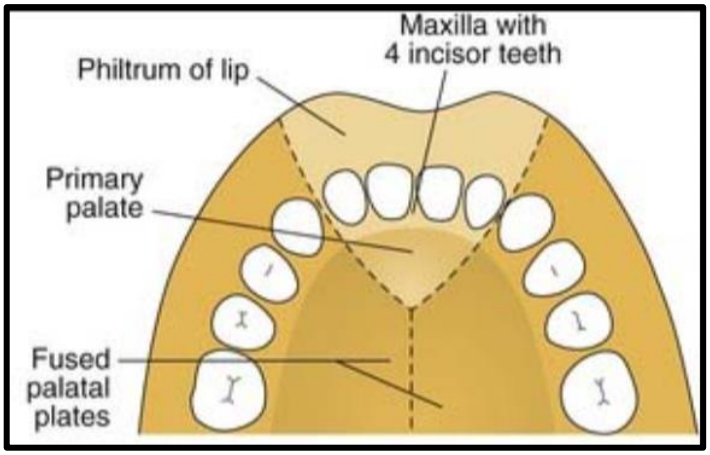

Fig 4. Anatomy of the midline palatal suture

Initial diagnostic assessment relies on passing a size 8 French catheter through the nasal passages. An inability to pass the catheter in the proximal first 1cm, confirms congenital nasal pyriform aperture stenosis. An inability to pass the catheter >3cm through the nasal passages indicates choanal atresia. Both pyriform aperture stenosis and choanal stenosis/atresia may be accompanied by nasal cavity stenosis.

The palate develops from the primary palate and 2 lateral palatal plates. Current thinking is that congenital pyriform aperture stenosis (CNPAS) is the result of premature ossification of the midline palatal suture causing palatal synostosis. This contributes to midface hypoplasia, which is associated with the malformation.

A CT scan confirms the diagnosis. Pyriform aperture length <11mm is the criteria for diagnosis in a term infant. Pyriform aperture stenosis may be associated with a solitary median maxillary central (mega) incisor. If this is present, an MRI of the brain.

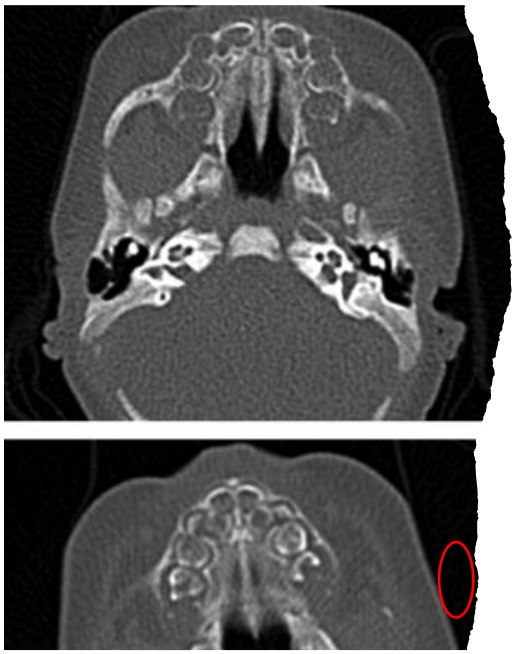

Fig 5. Anatomy of the midline palatal suture

CNPAS may occur in isolation or with other craniofacial or intracranial malformations (60-70% of cases). The most reported malformation is holoprocencephaly. Holoprocencephaly is a spectrum of midline developmental defects. Solitary median maxillary central incisor syndrome (SMMCI) is the most common and least severe (60% cases) form, and is identified at birth as single central maxillary alveolus & absent upper labial frenulum.

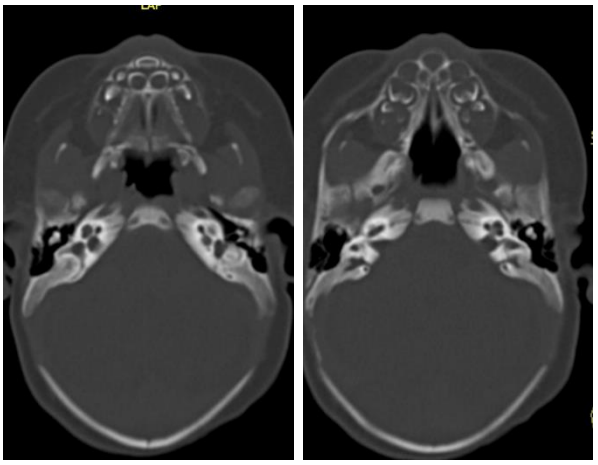

Fig 7. Solitary median maxillary central incisor

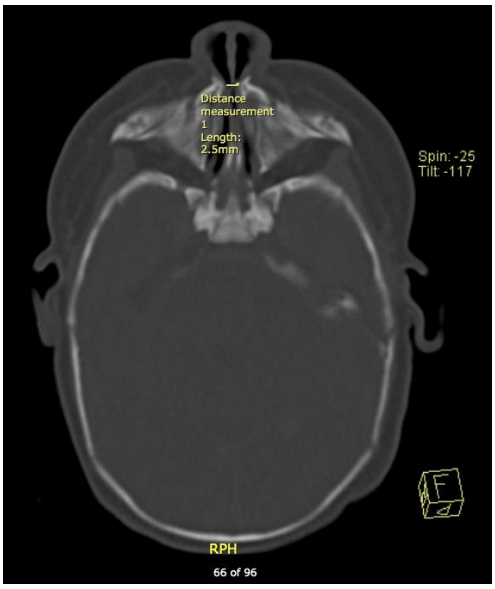

Fig 6. Pyriform aperture measurement

Severe forms of holoprosencephaly may be fatal and include:

- Microcephaly
- Hypoplasia of the corpus callosum or olfactory bulbs
- Cleft palate
- Pituitary dysfunction
- Oesophageal and duodenal atresia
- Neurological impairment

Choanal atresia is the consequence of failure of the buconasal membrane to rupture at 5-6 weeks during foetal development. It results in thickening of the medial pterygoid plates and lateral thickening of the vomer.

It may be unilateral or bilateral. It may be an isolated malformation or it may be associated with various syndromes.

Bilateral choanal atresia is most commonly associated with the following syndromes:

- CHARGE (up to 80% of children with CHARGE syndrome have choanal atresia)
- Treacher-Collins
- Crouzon

- Velocardiofacial (22q11 deletion)
- Craniosynostosis

Please watch the videos about embryonic facial development before reading about the following malformations.

Pierre Robin Sequence

Pierre Robin sequence (PRS) is characterized by the clinical triad of micrognathia (mandibular hypoplasia), glossoptosis (downward-backward displacement of the tongue), and upper airway obstruction.

Clinically these infants present with, mandibular hypoplasia that displaces the base of the tongue backward-and-downward which may obstruct the airway. It is commonly associated with a U-shaped cleft palate.

These findings may be isolated or part of a syndrome. The sequence was first described in 1891, but Pierre Robin published the first case of an infant with these characteristics in 1923.

Non-syndromic (isolated) PRS has been associated with mutations on chromosomes 2, 4, 11, or 17. Some evidence suggests SOX9 or KCNJ2 mutations (on chromosome 17) may affect the development of facial structures and cartilage development, leading to PRS.

Syndromic PRS has been recently reported to account for 60% of PRS. There have been 34 syndromes associated with syndromic PRS, the most common being Stickler syndrome.

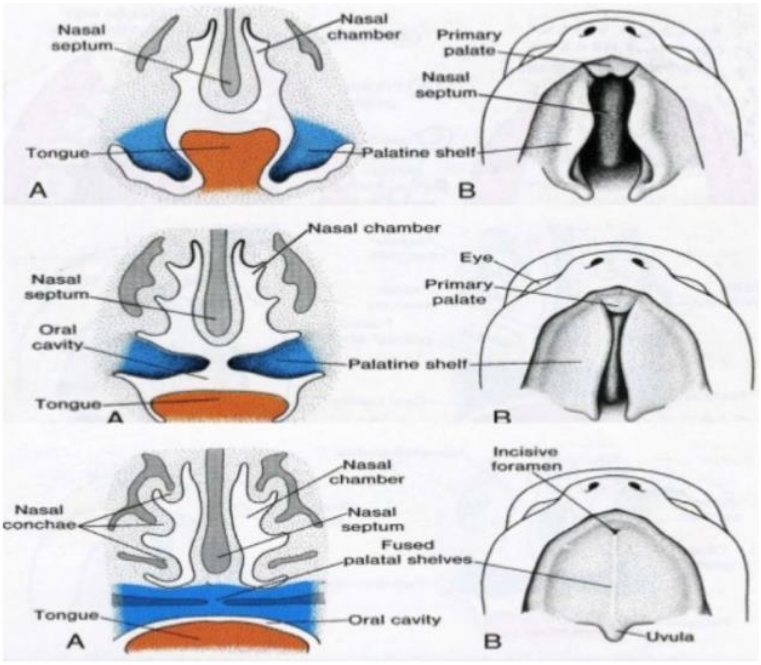

Fig 8. Normal fusion of the palatal shelves in the 8th embryological week of development

Other commonly associated syndromes:

- Velocardiofacial syndrome is a 22q11 deletion syndrome
- Treacher Collins syndrome

The branchial arches and some of these syndromes will be discussed in more detail in Chapter 7.

PRS affects approximately 1 in 8,500 to 1 in 14,000 newborns a year.

Pierre Robin sequence (previously named Pierre Robin syndrome) is now correctly named a sequence because one initial malformation leads to a sequential chain of events causing the other anomalies. A syndrome, in contrast, is a set of anomalies that arise separately due to one underlying pathogenesis. In PRS, micrognathia is the first abnormality that leads to glossoptosis and, ultimately, airway obstruction and/or a cleft palate. The most widely accepted theory for its aetiology is the **Mechanical Theory.**

In the early first trimester, around the 7th week of gestation, the mandible typically grows ventrally and inferiorly. If mandibular growth is abnormal, the tongue is not able to follow the normal trajectory of growth and blocks the closure of the palatal cleft in the 11th week of gestation.

Midline nasal masses

Nasal dermoids, epidermoids, teratocysts, encephaloceles, meningocoeles, meningoencephalocoeles, and gliomas are rare congenital lesions that result from anomalous embryologic development. Differentiation between them and understanding of their pathology is needed to avoid unnecessary complications. In view of their potential intracranial connection, identification, appropriate diagnostic imaging modalities, and treatment are required.

These lesions are rare, occurring in an estimated 1 in 20,000 to 1 in 40,000 births.Understanding normal embryological development of the frontonasal region helps explain their origin. The fonticulus frontalis and the prenasal space are two transient spaces that allow extension of a dural diverticulum from the anterior cranial fossa to the skin. The fonticulus frontalis temporarily separates the frontal and nasal bones. The prenasal space is located between the nasal bones and developing nasal cartilage. With the progressive growth of the frontal and nasal bones, the dural diverticulum and prenasal space regress, and the cribriform plate forms from fusion of the fonticulus frontalis with the foramen cecum, a foramen that passes through the skull base just anterior to the crista galli. Developmental midline craniofacial masses arise due to failure of these processes to occur, causing incomplete regression and separation of the dura from the overlying skin.

link to article: Congenital frontonasal midline masses

Paranasal cysts

Nasolabial cysts, also known as nasoalveolar cysts or Klestadt cysts, are rare non-odontogenic, soft-tissue, developmental cysts occurring inferior to the nasal alar region. The cyst is derived from epithelial cells retained in the mesenchyme after fusion of the medial and lateral nasal processes and the maxillary prominence during foetal life or due to the persistence of epithelial remnants from the nasolacrimal duct extending between the lateral nasal process and the maxillary prominence.

The patient usually presents with a slowly enlarging asymptomatic swelling. Patients usually seek medical advice on secondary infection of the cyst or due to the resulting disfigurement.

A dentigerous cyst is an odontogenic cyst (also known as a follicular cyst) that forms around the crown of an unerupted tooth, such as a wisdom tooth. Dentigerous cysts result from a fluid accumulation surrounding and covering an unerupted tooth.

The so-called "globulomaxillary cyst", described as a fissural cyst, caused by entrapped epithelium between the nasal and maxillary process, is no longer considered for its own entity. Nevertheless, cystic lesions, which correspond to the previous position of globulomaxillary cysts, do still occur in daily practice. Further discussion is beyond the scope of this text.

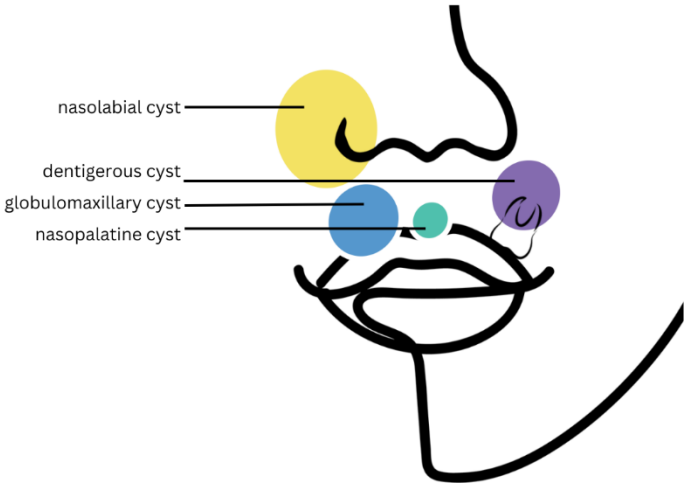

Fig 9. Paranasal cysts

The nasopalatine cyst is the most common epithelial and nonodontogenic cyst of the maxilla. The cyst originates from epithelial remnants from the nasopalatine duct.

Periapical or radicular cyst is the most common cyst of the jaws. It is considered an inflammatory rather than a developmental odontogenic cyst. This cyst is always associated with a nonvital tooth (no longer has any blood supply). Periapical cysts are not distinguishable radiographically from granulomas when small.