Respiratory system – Embryological Anomalies Summary

Anomalies of the nasal cavity

-Cleft Lip – Failure of fusion between the maxillary and the medial nasal prominences
Can be unilateral or bilateral, complete, or incomplete

-Cleft palate – Failed formation of the secondary palate or failure of fusion of:
1) The palatine shelves
2) The palatine shelves with the nasal septum and/or primary palate
Can be unilateral or bilateral, complete, or incomplete

-Cleft soft palate – Failure of fusion of the 2 folds that grow from the palatine shelves (8^{th} week)

-Cleft uvula – Failure of fusion of the 2 parts that'll form the uvula (11^{th} week)

Anomalies of the Trachea

-**Tracheoesophageal fistula** is the most common anomaly in the lower respiratory tract. It's associated with a defect in partitioning of the esophagus and trachea by the tracheoesophageal septum, this anomaly is **more predominant in males**, can present in many varietes:

1) Proximal esophageal atresia with TEF **(Most common variety –** 90% of cases**)**

2) Double Atresia (4% of cases)

3) H-Type, TEF with no esophageal atresia (4% of cases)

4) Atresia and double TEF (1% of cases)

5) Distal esophageal atresia and proximal TEF (1% of cases)

Infants with common type TEF and esophageal atresia **cough and choke** (excessive saliva in the mouth), **milk regurgitates** when they try to swallow, they have **polyhydramnios**, also fluids and gastric contents may enter trachea causing **pneumonia**.

Patients of the previous anomaly also are associated with other birth defects **(VACTERL)** (**V**ertebral anomalies, **A**nal atresia, **C**ardiac abnormalities (ASD,VSD, present in 33%), **T**EF, **E**sophageal atresia, **R**enal anomalies**, L**imb defects)

-Tracheal atresia and stenosis uncommon, associated with one of the varieties of TEF (as this anomaly results from unequal partitioning of the foregut into esophagus and trachea).

-Incomplete tracheal atresia partial obstruction to airflow, due to "web tissue" formation.

Anomalies of the Larynx

-Laryngeal Atresia aka (**CHAOS** – **C**ongenital **H**igh **A**irway **O**bstruction **S**yndrome), rare, may cause obstruction to the upper fetal airway, causes **lung enlargement distal** to the stenosis (distally: the airways become dilated, the lungs are hyperplastic).

Accompanied with a diaphragm that is flattened or inverted, fetal hydrops (accumulation of fluid in two or more compartments) and/or ascites.

GOLD STANDARD for its diagnosis is **Prenatal Ultrasonography,** which shows *"echoes".*

Anomalies of the lungs and bronchial tree

Most of these are rare

-Tracheal atresia with lung agenesis / agenesis of one lung

Agenesis causes absence, so if this leads to insufficient oxygen supply, death occurs.

-Abnormal division of the bronchial tree (Excessive development)

1) Supernumerary lobes (so the lung might end up having 3 lobes on the right, and 4 on the left!

2) Ectopic lung lobes, they might arise from the trachea or even the esophagus, caused by additional respiratory buds of the foregut

-Neonatal Respiratory distress syndrome aka **Hyaline membrane disease**
accounts for 20% of **deaths** among newborns
accounts for 30% of **diseases** among newborns

Causes: 1) Surfactant deficiency is a major cause of RDS
2) Intrauterine asphyxia by causing irreversible changes in type 2 pnuemocytes (making them incapable of producing surfactants)

Major stimulators of fetal surfactant production are **Corticosteroids ~** (Betamethasone), **Thyroxine,** so hypothyroidism maternally has to be treated, also we may give corticosteroids to the mother if early delivery is at risk!
In this syndrome, the lungs are underinflated and the alveoli contain hyaline membranes.
Luckily, treatment that includes supplementary oxygen and artificial surfactant help in the survival of more than 90% of neonates with RDS!

-Congenital cysts of the lung this is the most important clinically

The cysts are formed by dilation of terminal or larger bronchi, may be one or multiple, under radiography, honeycomb appearance of lungs is seen!

They're removed surgically (even before birth – intrauterine) because they might restrict the inflation of the lungs decreasing oxygen supply, also these cysts drain poorly thus causing chronic frequent infections!

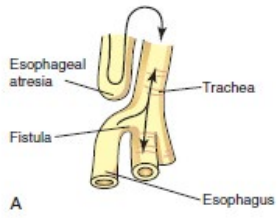
-Oligohydramnios and lung development

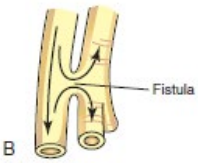
Amniotic fluid is very important for the maturation of the lungs! So when oligohydramnios (insufficient amounts of amniotic fluid) is severe and chronic, lung development is retarded, this might end up in severe pulmonary hypoplasia.

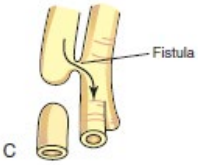
-Lung hypoplasia – *in infants with congenital diaphragmatic hernia* ***(CDH)***

Lungs are compressed by the abnormally positioned abdominal viscera so it can't develop normally, characterized by decreased lung volume.
Most of infants with this birth defect die of pulmonary insufficiency (despite optimal postnatal care, because their lungs are too hypoplastic to support extrauterine life)

Done by: Ahmad AlHurani ***Good luck!***







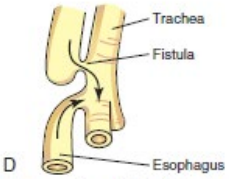
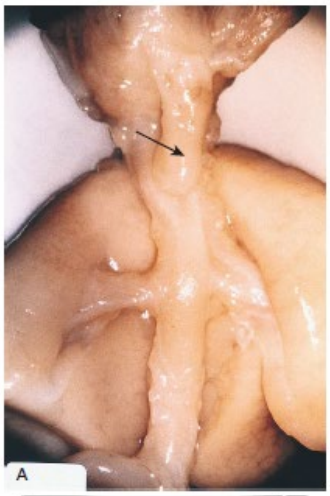


Figure 11–5 The four main varieties of tracheoesophageal fistula (TEF) are shown in order of frequency. Possible directions of the flow of the contents are indicated by *arrows*. **A,** Esophageal atresia is associated with TEF in more than 85% of cases. **B,** Fistula between the trachea and esophagus; this type of birth defect accounts for approximately 4% of cases. **C,** Atresia of the proximal esophagus ending in a tracheoesophageal fistula with the distal esophagus having a blind pouch. Air cannot enter the distal esophagus and stomach. **D,** Atresia of the proximal segment of the esophagus with fistulas between the trachea and both the proximal and distal segments of the esophagus. Air can enter the distal esophagus and stomach. All neonates born with TEF have esophageal dysmotility disorders, and most have reflux (regurgitation of contents of the stomach).



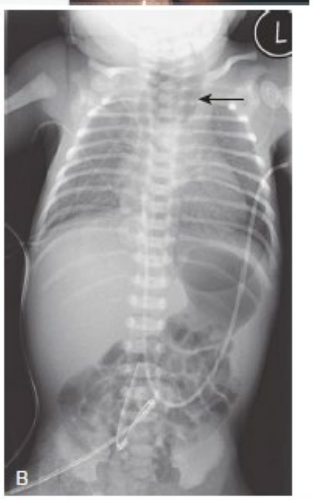
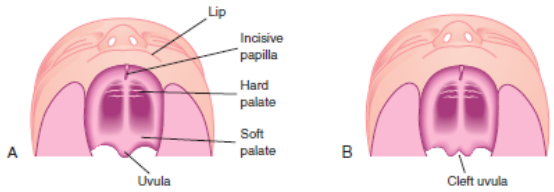
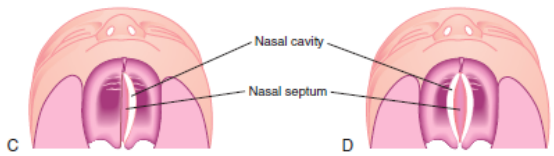
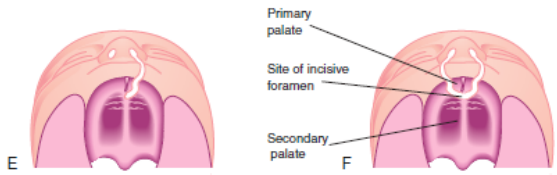


Figure 11–6 **A,** Tracheoesophageal fistula in a 17-week fetus. The upper esophageal segment ends blindly *(arrow)*. *(**A,** From Kalousek DK, Fitch N, Paradice BA: Pathology of the Human Embryo and Previable Fetus. New York, Springer Verlag, 1990.)* **B,** Radiograph of an infant with esophageal atresia. Air in the distal gastrointestinal tract indicates the presence of a tracheoesophageal fistula (*arrow*, blind proximal esophageal sac).







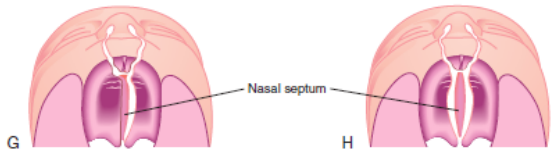


Figure 10–33 Various types of cleft lip and cleft palate. **A,** Normal lip and palate. **B,** Cleft uvula. **C,** Unilateral cleft of the posterior (secondary) palate. **D,** Bilateral cleft of the posterior palate. **E,** Complete unilateral cleft of the lip and alveolar process of the maxilla with a unilateral cleft of the anterior (primary) palate. **F,** Complete bilateral cleft of the lip and the alveolar processes of the maxillae with bilateral cleft of the anterior palate. **G,** Complete bilateral cleft of the lip and the alveolar processes of the maxillae with bilateral cleft of the anterior palate and unilateral cleft of the posterior palate. **H,** Complete bilateral cleft of the lip and the alveolar processes of the maxillae with complete bilateral cleft of the anterior and posterior palate.