Congenital developmental defects of the respiratory system

Congenital developmental defects of the airways are often life-threatening for newborns and require urgent surgical treatment, or they can remain unrecognized for a long time and cause repeated and chronic bronchopulmonary diseases.

Choan stenosis and atresia is only rarely a separate congenital developmental defect (CDD), it usually occurs together with other CDDs (gothic palate, septal deviation, congenital heart defects, spinal defects or intellectual disorders). It can be bony or membranous. It is the most common congenital anomaly in the nasal area, less severe unilateral atresia manifests itself by restriction of breathing, difficulty in drinking and purulent secretion from the nose.

Clinical picture of bilateral atresia: immediately after birth, the child suffocates (the newborn breathes only through the nose), cyanosis. In milder forms, the difficulties worsen when drinking, there is a risk of aspiration of food (it can even kill a newborn). Diagnosis: impossibility of inserting the probe through the nasal cavity into the pharynx (max. 5 cm), x-ray with contrast in the lateral projection.

Surgical treatment. First aid: keeping the mouth open (using an oropharyngeal tube or endotracheal intubation).

Congenital defects of the larynx

Atresia (or stenosis) of the larynx

A rare anomaly causing upper airway obstruction in the fetus. Distal from the atresia/stenosis, the airways are widened, the lungs are enlarged - echogenic on percussion, the diaphragm is flat to inverted, fetal ascites or hydrops has developed. It can be recognized by ultrasonography.

Subglottic stenosis can be congenital, but more often it is iatrogenic (e.g. after a long intubation), rarely it can be caused by a tumor or hemangioma. A definitive diagnosis requires an endoscopic examination.

Rete laryngis, diaphragm of the larynx

This rare anomaly results from incomplete recanalization of the larynx during the 10th week. At the level of the vocal cords, a membranous network (lat. rete) is formed, which partially closes the airways.

The diaphragm of the larynx is located at or below the level of the vocal cords, most often in the anterior commissure. It is the cause of congenital stridor in about 5%. The difficulty depends on the strength and size of the membrane. In contrast to malacia, dysphonia and a barking cough often appear.

Laryngomalacia (stridor laryngis congenitus)

Congenital laryngeal stridor (stridor laryngis congenitus), or laryngomalacia, is caused by delayed development of the cartilages of the larynx (mainly the epiglottis), which leads to their abnormal softness. The epiglottis, together with the aryepiglottic eyelashes, is sucked into the laryngeal entrance during inspiration, causing upper airway obstruction.

Clinically, it manifests immediately after birth or in the first weeks with inspiratory stridor, which improves in the prone position. It can be constant or intermittent. As a rule, stridor is worsened by crying, restlessness or infection. Congenital stridor is evidenced by the fact that it lessens or disappears when the child's mandible is pushed forward.

Specific therapy is not necessary, for children an elevated position is advantageous (especially during feeding). It usually resolves spontaneously within a year.

Congenital defects of the trachea

Congenital stenosis of the trachea

Congenital stenosis of the trachea can be diffuse, funnel-shaped (in the region of the bifurcation) or segmental. Clinical picture: stridor, shortness of breath, repeated respiratory infections. pathways, may cause obstructive emphysema. Diagnosis: x-ray, tracheoscopy.

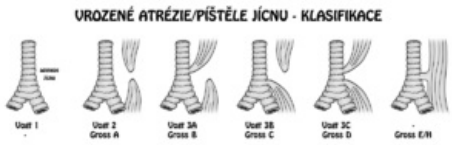
Therapy: sometimes a tracheostomy is necessary. Difficulties may improve with age. Reconstructive procedures, intraluminal stent.

Tracheomalacia

Tracheomalacia is increased pliability of cartilage or its complete absence and replacement by a membrane, due to which the wall collapses during exhalation. It can be manifested only when exhaling forcefully or when coughing. Diagnosis is endoscopic, treatment is usually not necessary.

Tracheoesophageal fistula

Tracheoesophageal fistula is associated with esophageal atresia in up to 90% of cases. Clinical picture: soon after birth, fogging of the mouth and nose with foamy liquid, shortness of breath, cyanosis (caused by aspiration of saliva or food). Diagnosis: insertion of a probe into the stomach (sometimes it is necessary to fill the esophagus with a contrast probe under X-ray control), aspiration pneumonia may be detected on a lung image. Therapy: surgery, intensive pre- and post-operative care. Fistula without esophageal atresia may be clinically silent at first and manifest in older children because there is often a membrane or mucous membrane in the mouth or the mouth is small and closes with compression when swallowing. Clinical picture: cough and cyanosis, especially when drinking, distension of the stomach and intestines with air, aspiration pneumonia.



Classification of congenital esophageal atresia

Occurs approximately 1:3000. It arises as a result of abnormal separation of the trachea from the esophagus by the tracheoesophageal septum (can also cause esophageal atresia). In most cases (90%), the upper part of the esophagus ends blindly, the lower part is connected to the trachea by a fistula. Isolated esophageal atresia and tr.e. fistula without esophageal atresia occurs less frequently (4%). Other defects occur rarely (less than 1%). The occurrence of these abnormalities is often associated with many other congenital defects, e.g. heart defects. Polyhydramnios is sometimes a complication, as fluids cannot pass freely through the digestive tract. Another possible complication is the passage of stomach contents into the trachea and lungs with subsequent inflammation.

An interesting feature is the VACTERL syndrome (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula, Esophageal atresia, Renal anomalies, Limb defects). It is a group of birth defects that occur together much more often than would be the case if they occurred individually. However, the cause is unknown.

Developmental defects of the lungs and bronchial tree

Major developmental abnormalities of the lungs and bronchial tree are relatively rare. Exceptionally, for example, a blind-ending trachea with the absence of lungs or agenesis of one lung occurs.https://www.wikiskripta.eu/w/Soubor:Congenital_Pulmonary_Airway_Malformation.webm

However, abnormal branching of the bronchial tree occurs significantly more often. This sometimes causes supernumerary lobes to form. However, these developmental variations have little functional significance; the only complication can be during bronchoscopy.

The occurrence of ectopic lung lobes that depart from the trachea or esophagus is interesting. Developmentally, they probably arise from supernumerary laryngotracheal protrusions of the foregut, which are formed independently of the main respiratory system.

Bronchomalacia[edit | edit source]

Bronchomalacia is a congenital hypoplasia or absence of cartilage rings. Clinical picture: may manifest as lobar emphysema in the newborn (but may be latent and manifest with frequent lung infections).

Special form: Williams-Campbell syndrome – congenital absence of cartilage from the segmental bronchi onwards (trachea and bronchi are always OK).

Symptoms: chronic cough, shortness of breath, wheezing in inspiration and expiration, recurrent bronchopneumonia with purulent exudation. Diagnosis: x-ray – hyperinflation of the lungs and increased honeycomb pattern diffusely bilaterally, bronchocinetography – demonstrates generalized saccular bronchiectasis with a noticeable change in the lumen depending on breathing. Therapy: administration of ATB, corticoids, mucolytics and permanent therapeutic physical education (this will alleviate the course).

Lung agenesis

Lung agenesis is the complete absence of bronchi, lung tissue and vascular supply. Bilateral agenesis is rare (it occurs in anencephaly), more often it is unilateral. Aplasia is more common - a rudimentary bronchus is formed. Clinical picture: unilateral can be well tolerated if it is not associated with other malformations, the other lung is usually hypertrophic, herniation on the affected side. Diagnosis: on the X-ray there is shadowing of the hemithorax with a significant shift of the heart and mediastinum to the affected side and elevation of the diaphragm, asymmetry of the chest may become apparent only in the further development of the child, the diagnosis can be confirmed by bronchoscopy, possibly angiography. Therapy: prevention and consistent treatment of all respiratory infections.

Lung hypoplasia

Hypoplasia of the lungs is associated with hypoplasia of the bronchial tree (reduction in the number of branches) and the pulmonary canal. It occurs most often in children with diaphragmatic herniation.

Pulmonary sequestration

Pulmonary sequestration is a mass of lung tissue that has no connection to the tracheobronchial tree and is supplied by an anomalous artery arising directly from the thoracic or abdominal aorta. There are two forms: extralobar sequestration - the sequestration has its own visceral pleura, often associated with other VVV - and intralobar sequestration - it does not have its own visceral pleura. It is most often located in the posterobasal

segment of the lower lobe. The sequestered lung is usually degenerated, fibrotic, inflammatory, or cystic. Clinical manifestation - in the first years of life it can manifest as left-right shunt, in older age as recurrent pneumonia. Diagnosis: x-ray – sharply demarcated shadowing reminiscent of an infiltrate or atelectasis or the character of a cyst or cysts. Therapy: surgical with a very good prognosis.

Congenital lung cysts

Congenital lung cysts are among the most common lung anomalies. They are solitary or multiple, usually located in one lung lobe. Histologically, we distinguish between bronchogenic cysts, alveolar cysts and a combination of both. Their size is different - from microscopic to huge across the entire hemithorax.

These arise from the dilation of terminal bronchioles, or larger bronchi. These cysts can either be small and multiple, which then cause the honeycomb appearance of the entire lung on an X-ray image, or there can be one or two large cysts. Cystic lung structures are usually poorly ventilated and are often the cause of chronic infections.

Clinical picture: a communicating (tension) cyst can mimic pneumothorax with deviation of the mediastinum. Congenital cysts must be distinguished from post-abscess pneumatoceles and lobar emphysema. The cyst has a typical border. When located on the left side, they can sometimes imitate a diaphragmatic hernia.

Therapy – extensive tension cysts present as an urgent condition requiring surgical treatment. Prognosis: secondary infection of the cyst with the formation of abscess, pyopneumothorax, bronchial fistula and general sepsis is common. The risk of cyst malignancy is also reported.

Cystic adenomatoid malformation of the lung

Cystic adenomatoid malformation of the lung is a special and probably the most common form of cystic lung. Excessive growth of terminal bronchioles and impaired alveolar differentiation. Cystic cavities communicate with large bronchi, they are lined with bronchial epithelium, there is no cartilage and mucous glands in their wall. Between the cysts is normal lung tissue. Diagnosis: the affected part of the lung is enlarged, there is shadowing on the x-ray (differently sized cystic elucidations in it). Often associated with other VVV, 50% of babies are born prematurely. Clinical picture: cyanosis and tachycardia soon after birth (with the onset of breathing, the cysts expand and the surrounding healthy tissue is suppressed). Therapy: surgical removal of the affected lobe (often affects only the segment, but resection of only the segment is not possible).

Congenital lobar emphysema

Congenital lobar emphysema is the opening of the lung lobe by the mechanism of valve closure of the bronchus. It most often affects one lobe (mainly the upper left). Causes: in the wall of the bronchus (anomalies of cartilage, ...), in the lumen of the bronchus (mucosal cilia), by compression from the outside (by a vessel, mediastinal cyst or tumor).

Clinical picture: progressive respiratory difficulties, tachypnea, dyspnea, cyanosis, cough, wheezing and stridor. Difficulties are greater the earlier after birth this occurs. The emphysema part compresses the surrounding parenchyma and radiates it from activity. Diagnosis: bulging of the chest in the affected area, weakened breathing and hypersonic percussion. X-ray: increased transparency of the affected lobe, filling almost the entire hemithorax, displacement of the mediastinum. Differential diagnosis: foreign body aspiration, congenital tension cyst, emphysematous bulla, postinfectious pneumatocele, diaphragmatic hernia.

Therapy: segment resection or lobectomy. Cases of spontaneous regression are also described. We can administer oxygen (not overpressure - it would worsen the expansion of the lobe), selective intubation of the main bronchus of a healthy lung is better.

Cardiovascular malformation causing pressure on the tracheobronchial tree[edit | edit source]

Double aortic arch (complete vascular ring) - compresses the trachea and esophagus - causes breathing and swallowing difficulties; basic examination: echocardiography, esophagogram,

incomplete vascular ring – it consists of the right-sided aortic arch with the left-sided lig. arteriosum.

Insufficient amount of surfactant[

Lack of surfactant (dysfunctional pneumocytes II, signaling pathway, etc.) causes respiratory distress syndrome (RDS). With a lack of surfactant, the surface tension on the alveolar fluid-air barrier rises, which causes a high risk of alveolar collapse during exhalation. The partially collapsed alveoli of an immature child contain a high-protein fluid with hyaline membranes and lamellar bodies apparently derived from the surfactant layer. RDS (formerly known as hyaline membrane syndrome) is responsible for approximately 20% of newborn deaths. Currently, however, there are procedures for applying artificial surfactant and inducing its production using glucocorticoids in premature babies. This enabled the survival of some babies from the middle of the sixth month of pregnancy.

Oligohydramnios and lung development

The presence of fluid in the lungs is an important stimulus for their development. If a severe form of oligohydramnios (lack of amniotic fluid) is present in a chronic form, lung development is delayed and severe pulmonary hypoplasia occurs.