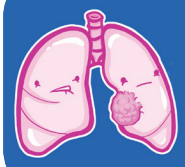

NOTES
LOWER RESPIRATORY TRACT CONGENITAL MALFORMATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Structural anomalies of lung during embryonic development

DIAGNOSIS

Prenatal ultrasound, radiography, CT scan, MRI

SIGNS & SYMPTOMS

- Asymptomatic/respiratory distress

TREATMENT

- Surgical resection, respiratory support

CONGENITAL PULMONARY AIRWAY MALFORMATION

osms.it/cpam

PATHOLOGY & CAUSES

- Congenital pulmonary airway malformation (CPAM)
- AKA congenital cystic adenomatoid disorder → part of/entire lung lobe replaced by non-functional cysts
- Result of abnormalities in branching morphogenesis of lung
- Rare in general; most common congenital lung malformation

TYPES

- *CPAM Type 0:* rare; multiple small cysts composed of cartilage, mucus cells; lethal anomaly
- *CPAM Type 1:* most common; large (> 2cm/0.78in) multiloculated cysts; good prognosis
- *CPAM Type 2:* small uniform cysts; associated with other congenital malformations (e.g. esophageal atresia with esophageal fistula); poor prognosis
- *CPAM Type 3:* large; not true cysts; communicates with surrounding parenchyma; "adenomatoid" type; more common in individuals who are biologically male
- *CPAM Type 4:* rare

CAUSES

- Possibly, *in utero* airway obstruction/atresia; definitive cause unknown

RISK FACTORS

- Occurs sporadically
- Not related to maternal factors
- No genetic predisposition except Type 4 (associated with familial pleuropulmonary blastoma syndrome)

COMPLICATIONS

- Pulmonary hypoplasia
- Mediastinal shift, putting pressure against heart
- Respiratory infections
- Rapid growth → venous outflow obstruction, cardiac failure, hydrops fetalis, death
- Longstanding CPAMs → cancer

SIGNS & SYMPTOMS

- *75% of individuals:* asymptomatic
- *25% of individuals:* cyanosis, pneumothorax, respiratory distress, tachypnea, intercostal retractions, grunting
- Hyperresonance on percussion, diminished vesicular murmur, asymmetrical thorax

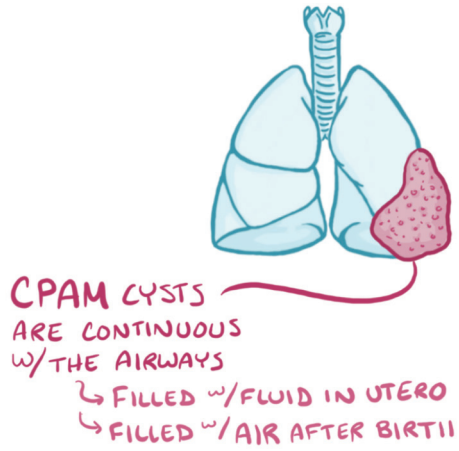

Figure 124.1 Illustration depicting continuous pulmonary airway malformation.

DIAGNOSIS

- Definitive diagnosis usually not possible without surgical resection, histopathological evaluation

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Echogenic mass appearing in the chest, displacement of heart, flat/everted diaphragm, absence of visible lung tissue

MRI/CT scan

- Delineate pathology

TREATMENT

SURGERY

- Minimally invasive surgical resection (thoracoscopy)
- *Large cysts: in utero* placement of Harrison thoracoamniotic shunt
- *Rare:* fetal surgery *in utero*; surgical delivery, *ex utero* intrapartum treatment (EXIT) procedure

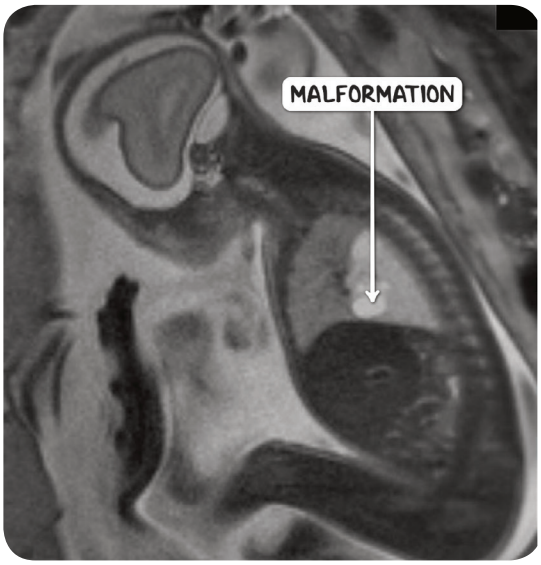

Figure 124.2 A fetal MRI demonstrating a congenital pulmonary airway malformation.

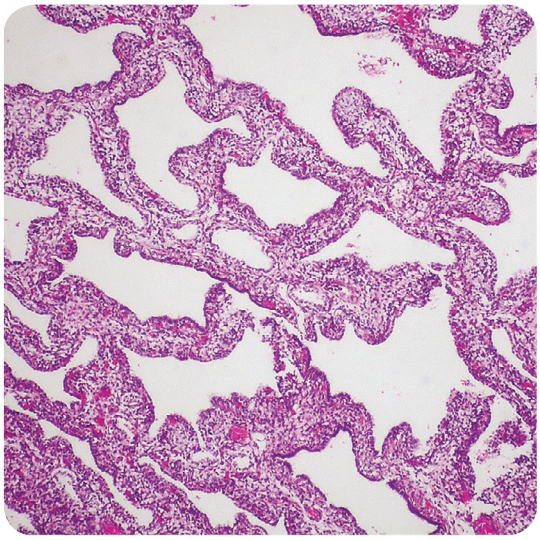

Figure 124.3 The histological appearance of a congenital cystic type II malformation. There are multiple small cystic spaces lined by immature respiratory tissue.

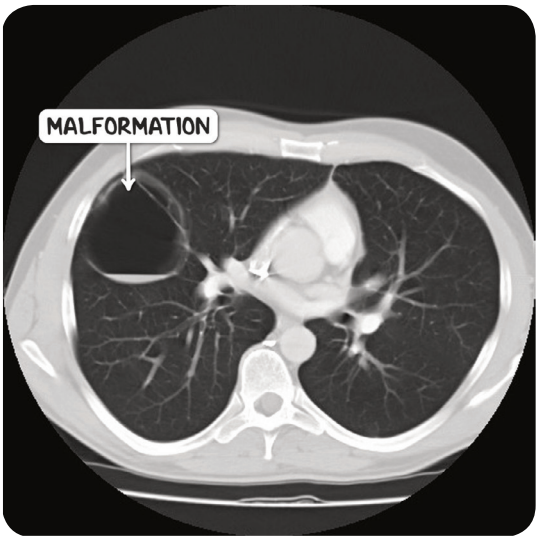

Figure 124.4 A CT scan of the chest demonstrating a pulmonary congenital cystic adenomatoid malformation, Type I, presenting as a single cyst of middle lobe in an adult.

PULMONARY HYPOPLASIA

osms.it/pulmonary_hypoplasia

PATHOLOGY & CAUSES

- Underdevelopment of lungs → low number/size of bronchopulmonary segments/alveoli
- Typically occurs prior to/after pseudoglandular stage (6–16 weeks gestation)

TYPES

- Primary
  - Idiopathic, not associated with maternal/fetal abnormalities; rare
- Secondary
  - Due to fetal abnormalities disrupting lung development
  - Associated with bilateral renal agenesis, congenital diaphragmatic hernia, congenital cystic adenomatoid malformation, fetal hydronephrosis, caudal regression syndrome, mediastinal tumor, dextrocardia, sacrococcygeal teratoma
  - Bilateral renal agenesis → oligohydramnios → decreased lung expansion, decreased mechanical stretching → decreased growth factors lung synthesis → pulmonary hypoplasia

RISK FACTORS

- *Decreased amniotic fluid:* severe oligohydramnios, mid-trimester rupture of membranes
- *Disruption of signaling pathways involved in growth:* sonic hedgehog (SHH) signaling pathway
- *Aberrant expression growth factors:* vascular endothelial growth factor (VEGF), epidermal growth factor (EGF), fibroblast growth factor (FGF)
- Early delivery

COMPLICATIONS

- Respiratory distress, chronic respiratory failure, bronchopulmonary dysplasia, pneumothorax, secondary scoliosis, impaired cardiac function
- Survival depends on degree of hypoplasia, cause of restricted growth

SIGNS & SYMPTOMS

- *Prenatal:* poor fetal movement, amniotic fluid leakage, oligohydramnios
- *Postnatal:* asymptomatic/severe respiratory distress, apnea, cyanosis
- Small, bell-shaped chest; heart displacement; decreased/absent breath sounds

DIAGNOSIS

DIAGNOSTIC IMAGING

3D ultrasound

- Total lung volume measurement

Doppler ultrasound/magnetic resonance angiography

- Shows lack of blood supply

CT scan/MRI

- Shows loss of lung volume

Radiography

LAB RESULTS

- Renal function (serum creatinine, blood urea, electrolyte levels)
  - Oligohydramnios

OTHER DIAGNOSTICS

- Lung weight, lung weight to body weight ratio, mean radial alveolar count (RAC), lung DNA

TREATMENT

OTHER INTERVENTIONS

- *Amnioinfusion:* instilling isotonic fluid into amniotic cavity
- *Amniopatch:* intra-amniotic injection of platelets, cryoprecipitate → seal amniotic fluid leak
- Treatment of underlying condition; respiratory support; in severe cases, fetal surgery

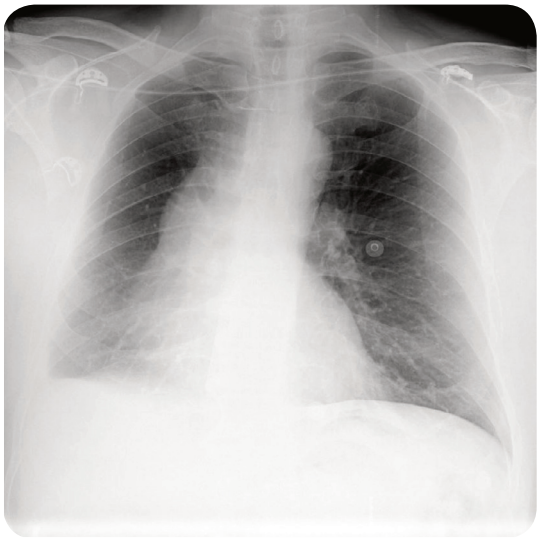

Figure 124.5 A chest X-ray demonstrating a volume defect of the right thoracic cavity caused by pulmonary hypoplasia.