The common congenital lung malformations have traditionally been divided into separate pathological entities such as pulmonary airway malformations, cystic adenomatoid malformations, congenital lobar emphysema, intra or extra pulmonary sequestrations, bronchogenic and foregut cysts. Some may produce respiratory compromise immediately after birth or even in utero. The management of such life threatening congenital lesions is non-controversial and rests on a surgical approach.

The challenge that now faces physicians is that the increasing use of routine antenatal ultrasound evaluations in developed countries has increased the prenatal detection of fetal lung anomalies. Experience has shown not only that many of those lesions seem to involute over time during pregnancy but also that many infants are totally asymptomatic at birth. Furthermore, the traditional classification of congenital lung malformations may be difficult to apply antenatally. In fact, hybrid lesions combining features of congenital cystic adenomatoid malformations (CAM) and pulmonary sequestrations are described increasingly frequently and the usual pathological divisions between the respiratory malformations may in fact represent the extremes of a spectrum of embryologic anomalies. This brought some authors to suggest the descriptive term Congenital Thoracic Malformations (CTM) to supersede several previous pathological entities such as CAM, sequestration and a variety of congenital thoracic cysts. To complicate management further, we do not have much data on the natural history of antenatally detected chest malformations, as most studies are retrospectively based. Because of these factors, the management of lung malformations especially in infants with few or no symptoms has become controversial. The following thoughts aim at exposing the controversies and will hopefully reinforce the idea that we urgently need to systemize and structure prospective observations of the CTM in order to manage them based on proper scientific grounds. The management of other malformations such as pulmonary hypoplasia or agenesis and for which no specific therapy exists will not be discussed.
Many congenital lower respiratory tract malformations have a cystic component. Bronchogenic and foregut cysts are single congenital malformations that are most often located in the mediastinal region. Central lesions can produce compression of the major airways with respiratory distress, cough and wheezing. Smaller cysts can be asymptomatic and can be discovered incidentally on chest X-Ray as a round mediastinal mass with or without overinflation or atelectasis of varying portion of the ipsilateral lung. Foregut or enteric cysts can present as a posterior mediastinum mass and may contain other tissues such as neural or gastric mucosa that may even ulcerate and bleed. The most common complication is infection with exacerbation of respiratory symptoms due to the distension and inflammation of the cyst.

Congenital lobar emphysema is a condition characterized by overinflation of one or more lung lobes. It tends to localize in the upper lobes and is often associated with other cardiac malformations. Emphysema is a misnomer since there is no “destruction” of lung parenchyma. Congenital lobar overinflation would somewhat be a better term and it can be produced by an obstruction of the supplying bronchi (abnormal cartilage, valvelike mucosal folds, extrinsic compression…) or by an increase in the number of expanded alveoli (polyaveolar lobe). The impact of this malformation is mainly mechanical due to its effect on surrounding structures. Symptoms most often occur at birth with large lesions but it can be discovered at any age.

CAM is a malformation of the terminal lung structures with varying combination of cysts and solid tissue. It may affect varying areas of the lung from a unique segment to the entire lung. Large lesions can be present in utero and compress the surrounding chest structures producing early respiratory compromise. Others are much smaller and can present from infancy to adult life with spontaneous pneumothorax or persistent “infiltrates” on chest X-Ray. However, most are diagnosed within the first 2 years of life. CAM have been classified by Stocker largely based on the size of the cysts they tend to contain. In type I CAM, macroscopic cysts (1 or more) predominate. In type II, microscopic or small numerous cysts are present. Type III CAM is a solid mass of airless
tissue. Traditionally, CAM have been aggressively approached with surgical excision due to repetitive descriptions of malignant transformation occurring within the malformation. Rhabdomyosarcomas, pulmonary blastomas and bronchiolar carcinomas have been reported to develop in both children and young adults.

A pulmonary sequestration is a mass of bronchopulmonary tissue that may lie within a pleural lining surrounding adjacent normal lung parenchyma (intrapulmonary sequestration) or be surrounded by its own pleural covering (extrapulmonary sequestration). By definition, sequestrations have abnormal or absent communication with the airways. Many are supplied by anomalous arteries arising directly from the aorta but they usually drain normally to the pulmonary venous return and left atrium. Most are the size of a lung segment or less and they tend to be found in the lower lobes. Pulmonary sequestrations can be found incidentally on a chest X-Ray but symptoms tend to occur more frequently with increasing age. Due to large volume of blood flow, some sequestration may behave as arteriovenous fistula early in life and produce symptoms of cardiac failure. Most will however manifest as recurrent localized pneumonitis with fever, purulent sputum or even hemoptysis. Some may contain cystic elements and the majority of extralobar sequestrations are associated with some CAM tissue. This has led some authors to speculate that the malignancies sometimes linked to sequestrations are in fact related to a CAM component.

With the increasing use of antenatal ultrasounds, thoracic malformations are becoming more and more frequently detected. Most will manifest as a mass of altered echogenicity with or without obvious cystic components. Unfortunately, the appearance of the lesion is often non-diagnostic and congenital pulmonary and non-pulmonary malformations can seldom be definitely identified on antenatal ultrasound alone. Sometimes, magnetic resonance imaging can be useful and provide additional information as to the nature of an antenatal chest mass. After detection, recommendations are that serial US should be used to follow up the lesion. These serial exams have allowed a better understanding of the natural antenatal evolution. For example, CAM typically reach a maximum size in relation to the overall chest volume at around 25 weeks of pregnancy. The prognosis is in
general rather good and 90 % of fetuses will do well during pregnancy as many lesions tend to regress over time and may even disappear totally. A large size lesion can however impede the development of the ipsilateral or even the contralateral lung with the risk of respiratory insufficiency and pulmonary hypertension in the newborn. The evolution of hydrops is also an ominous sign with an elevated risk of fetal or neonatal death.

Treatment of life threatening antenatal lung malformations by in utero or ex utero intrapartum surgery has been described (cyst aspiration, thoraco-amniotic shunting, percutaneous laser therapy…). Such procedures are usually reserved for fetuses with no other evidence of chromosomal or serious malformations and are associated with a significant risk of fetal death that precludes the dissemination of the techniques outside of specialized centers. After 32 weeks of gestation, early delivery can also be considered and urgent open surgery be performed. In general and in line with other chest malformations such as diaphragmatic hernias, delayed surgery following respiratory stabilization after birth is favoured.

Even if a lesion regresses over time and becomes undetectable after birth by US or chest X-ray, it is still recommended that CT-Scan be performed as some rather substantial lesions can be missed by other investigations. The lesion can then not only be delineated properly, but the investigation can include the use of contrast material to document abnormal vascular structures or communications with the esophagus. Such additional techniques can then be integrated with the other imaging processes and a more definite diagnosis can be expected.

While symptomatic lesions are obvious surgical candidates, the major challenge and the focus of the dominant controversy rest on the management of asymptomatic newborns with an antenatally diagnosed malformation. Should they be simply observed until complications arise or should they be electively operated on? Those who favour elective surgery argue that, over the long term, CTM can become infected which by itself can hamper surgical resection. CTM can also bleed, rupture to produce a pneumothorax, or induce sudden respiratory compromise. Furthermore, CAM can undergo malignant
transformation and the CAM portion of hybrid lesions can be missed by medical imaging. Finally, the follow-up of the lesion rests on repeated CT Scans, which by themselves carry their own long term risks of radiation exposure but also the short-term risk of repeated anesthesia as CT Scans in young children require sedation.

Once a surgical excision has been decided upon, the question then becomes: When is the best time to do it? Since the anesthesia and surgical risks decrease within the first few weeks and months of life, many authors recommend elective surgery after 3 months of age. Furthermore, the potential for alveolar growth decreases with age and the number of alveoli increases essentially for the first 2 years of life. It can thus be argued that a resection performed between 3 months and 2 years of life offers the best potential for compensatory lung growth and functional recovery, but the concept is still open to debate. Finally, post-op imaging should be performed to ensure complete resection of the lesion and improvement of its associated long-term risk.

In conclusion, a growing number of prenatally detected CTM will come to the attention of physicians. In the case of symptomatic neonatal malformations, surgery is still the standard of care. With asymptomatic lesions, many would argue that elective surgery within the first 2 years of life is still warranted in view of the risk of infection or malignant transformation. This classical approach however rests on an unquantified risk/benefit ratio over expectant management that precludes any firm recommendation for parents and physicians. Research is thus still needed to refine our approach to CTM by increasing our diagnostic identification of the lesions and by clarifying the natural history of asymptomatic congenital thoracic malformations.

Suggested readings:


