



CONGENITAL LUNG LESIONS*

*This information is intended to supplement your consultation with members of the Fetal Treatment Program regarding your unborn child with a suspected lung lesion. The same information is also available on our web site: www.fetal-treatment.org. Underlined words are explained in a glossary at the end.

WHAT ARE CONGENITAL LUNG LESIONS?

There are three broad categories of congenital lung lesions: Cystic adenomatoid malformations (also called CCAM), bronchopulmonary sequestrations (or sequestrations, for short) and bronchogenic cysts. All three types represent the abnormal development of lung tissue, which may occur during normal fetal development. Why these lesions form is not really known; how they differ from each other may have to do with the timing and location of their development.

In the embryo, the lungs develop as an outpouching of what will become the esophagus: a “bud” develops, which elongates to become the trachea, then divides to become the left and right main stem bronchi. Further division of each bronchus will, like the branches of a growing tree, form the “tracheobronchial” tree of airways. At the same time, the lung parenchyma (the actual lung tissue responsible for exchange of oxygen and gases into and out of the bloodstream) develops around each of those airway branches.

A bronchogenic cyst is nothing more than an airway branch that buds off, loses all connections with the rest of the tracheobronchial tree, and does not connect with actual tissue. As a result, a cyst forms, which is lined by the same cells one finds in the trachea or the bronchi.

A sequestration represents a portion of lung (with a bronchus and some lung tissue) that has completely separated from the rest of the lung. If this separation occurs relatively early in embryonic life (before the 6th week of pregnancy), this “mini-lung” is usually fully developed and separate from the rest of the organ. It also contains its own blood supply: an artery that is directly connected to the aorta, for example. However, because it is separate from the rest of the lung, no air goes in or out of this “extralobar” sequestration.

If the separation from the rest of the lung occurs later (between the 8th and the 12th week of pregnancy), it usually remains within the normal lung (but may still have separate blood vessels). Even though it is not connected to the rest of the tracheobronchial tree by a normal bronchus, there may be microscopic communications with the rest of the lung: in this type, the “intralobar” sequestration, some air (and bacteria) may get trapped in the lesion after birth, which may cause recurrent infections.

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The congenital cystic adenomatoid malformation (CCAM) is less easily explained, but consists of some or all types of lung and bronchial cells and structures, arranged in a disorderly fashion. It is not a tumor, however: the cells themselves are not malignant and do not grow, invade other organs or spread to other parts of the body; rather, it is a “clumsily put together” part of lung, that doesn’t function properly. Because a CCAM tends to develop within the normal lung, microscopic communications with that lung also place the child at risk for infections, from trapped air and bacteria.

While the above classification is helpful, it is not always clear-cut, and many “hybrid” forms exist: lesions that have characteristics of more than one type. In addition, in up to 50% of sequestrations, part of the otherwise normal lung tissue is replaced by a CCAM.

Congenital lobar emphysema is one more form of congenital lung lesions, but it is much less common than the ones mentioned above. In congenital lobar emphysema, or CLE, one of the bronchi of a lung may be partially and/or intermittently blocked, causing accumulation of fluid and distension of the lung tissue connected to that bronchus. While the cause of this condition is different, its course and treatment is remarkably similar to that of the lesions described above.

HOW COMMON IS IT?

Congenital lung lesions are seen in approximately in 1 of every 3,000 live births. They are most often isolated findings, not associated with chromosomal or genetic disorders. CCAMs and sequestrations are sometimes seen in fetuses with a congenital diaphragmatic hernia, and can be an incidental finding (i.e., discovered by chance) in otherwise healthy children or children with unrelated anomalies.

WHAT CAN HAPPEN BEFORE BIRTH?

Most congenital lung lesions do not become visible until early in the second trimester. They often grow, sometimes fast, causing the lung on the same side to be compressed. In addition, the heart may be pushed to the other side, and even the lung on the other side may be compressed. If the lesion becomes very large, it may start to affect the well-being of the fetus: extreme compression of the heart and the large blood vessels of the chest may impair the heart’s function, and this could lead to hydrops (heart failure) and death. If compression of organs in the chest is severe and continues for a long time during pregnancy, the baby’s lungs may not function well at birth (pulmonary hypoplasia).

In most cases, however, growth of these lesions is limited, and they tend to become smaller toward the end of the second trimester. In approximately 75% of the cases, the lesion regresses either partially or completely by the time the baby is born; not uncommonly, the lesion is not visible by prenatal ultrasound anymore.

Of course, premature birth can affect the outcome, particularly if it occurs as the lesion is still large and compressing the lungs. However, the presence of a congenital lung lesion does not in itself increase the risk of prematurity.

It is important to realize that the accuracy of the diagnosis, although very good, is not 100% perfect. The condition most resembling a congenital lung lesion is congenital diaphragmatic hernia. In this condition, a hole in the diaphragm allows intestines and other abdominal organs to move into the chest cavity, thereby compressing the lungs. The effect of a diaphragmatic hernia on the lungs of the fetus is similar to that of a congenital lung lesion. However, a diaphragmatic hernia does not get better during pregnancy, and many of these infants are born with hypoplastic

lungs. With current imaging techniques (ultrasound and MRI), the diagnosis of a diaphragmatic hernia can almost always be differentiated from a lung lesion (bronchogenic cyst, CCAM or sequestration).

WHAT CAN BE DONE BEFORE BIRTH?

In most cases, the lesion will eventually regress without causing any permanent damage. If this can be predicted to occur, there is of course no reason to intervene before birth. It will be important, though, to follow the lesion closely, usually with weekly ultrasounds, until it is clear that the lesion is getting smaller. In addition, it may be helpful to obtain a fetal MRI (magnetic resonance imaging): this test may give more information than the ultrasound about the exact appearance of the lesion, possible feeding blood vessels, and the condition of the surrounding normal lung. It can help differentiate cystic lung lesions from congenital lobar emphysema or diaphragmatic hernia.

If the lesion seems to grow too much, and early signs of heart failure are seen, something may have to be done. If this occurs after 25-26 weeks of pregnancy, it may be safer to think about early delivery, rather than to continue the pregnancy or even to intervene directly on the fetus. This is only recommended if the life of the fetus is believed to be at risk, since such extreme prematurity carries an important risk of complications.

If signs of heart failure occur earlier in pregnancy (before 23-24 weeks), early delivery is not an option, since infants of that gestational age cannot survive outside the womb. Intervention on the fetus may be possible, although this is obviously an invasive and risky procedure. If the lesion consists mainly of one or two large cysts, it may be possible to remove the fluid inside those cysts, thereby collapsing the lesion and allowing heart and lungs to function better. This procedure, called thoracentesis, is usually performed under local anesthesia and constant ultrasound guidance. A long, thin needle is introduced directly into the womb, and into the fetus's chest. Fluid is aspirated from the lesion, and the needle is withdrawn, or a "double pigtail" catheter is left behind. This will then continue to drain fluid from the cyst into the amniotic cavity.

In rare cases, the lesion continues to grow and threatens the well-being of the fetus, but no single large cyst can be identified. In these cases, when the lesion is mostly solid, the only option may be to remove the lesion surgically. This type of fetal surgery is the most invasive and riskiest form of intervention on the fetus, and can only be performed in specialized centers. Although this can be life-saving, not all babies can be saved, and the procedure carries some risks to the mother as well.

If the lesion does regress, as is seen in the majority of cases, no prenatal intervention is needed. It may be important to plan for the delivery, however:

1. Mode and timing of delivery

While Cesarean section can sometimes be indicated for certain conditions of the fetus, there is no need for it in the case of a congenital lung lesion. Of course, a Cesarean section may still be performed for obstetrical reasons.

As mentioned before, prematurity may increase the risk of complications for the newborn baby. Since the lesions usually regress toward the end of the second trimester or the beginning of the third trimester, pre-term delivery is usually *not* indicated, unless there are signs that the fetus is in trouble.

2. Place of delivery

Because of the risks of lung failure in the newborn infant, and the possibility of early hydrops in the fetus, it is recommended that the baby be born in a hospital that has immediate access to a tertiary neonatal intensive care unit. In some cases, the lesion is still sufficiently large at birth (particularly if the baby is born prematurely) that immediate surgical intervention is necessary. For that reason, presence of pediatric surgical specialists is also advisable.

WHAT WILL HAPPEN AT BIRTH?

If everything goes as planned, you will deliver at a tertiary care center with direct access to a neonatal intensive care unit. The neonatologists will be present at delivery, so that they can immediately assess your baby and start treatment, if necessary. At the same time, the pediatric surgeons will be alerted. In many cases, however, you will be able to see (and hold) your baby after delivery.

Your baby will be "stabilized" in the intensive care unit. An intravenous line will be placed in an arm or a leg, so that fluids can be given. If your baby shows signs of distress, it is possible that he will be intubated, so that we can help him breathe better.

If it is clear that there are no other major problems, your baby will undergo imaging tests to look for the lesion. If he is stable and breathing well, a chest X-ray and/or an ultrasound will be obtained, usually within one or two days of birth. Even if the lesion had "disappeared" by prenatal ultrasound, it can usually be found by ultrasound or chest X-ray after birth, since imaging the baby directly can show more details than when the fetus is still in the womb.

Babies who breathe well and show no other signs of distress don't need immediate intervention. Typically, your baby will be allowed to go home, and plans will be made to be seen by a pediatric surgeon. Even if there are no symptoms, surgical intervention may be recommended later in infancy (typically, between 6 and 18 months), to avoid long-term complications of the lesion. In some cases, this can be done using minimally invasive techniques (thoracoscopy); in others, the operation will be performed using a thoracotomy.

If the infant has breathing difficulties and it is felt that this is due to the lesion, more urgent intervention may be necessary. This can be done immediately after birth, or later in the newborn period. The results of the operation and the outcome for your baby depend primarily on the severity of the condition and the degree of prematurity. Surgical removal of part of a lung is an invasive procedure, but one that can be performed safely in even the smallest of patients. Even if a portion of normal lung has to be removed or if the lesion has prevented full development of the normal lung, full recovery is likely: normally, lungs continue to grow until a child is several years old.

COMPLICATIONS AND LONG-TERM OUTCOME

The overall outcome of congenital lung lesions (CCAM, bronchogenic cyst, sequestration) is generally excellent, if the lesion has substantially shrunk by the time of birth. As mentioned before, CCAM and intra-lobar sequestrations are at a significant risk of recurrent infections (pneumonia). The infected cysts may look like lung abscesses – the knowledge of an underlying cystic lesion greatly facilitates the diagnosis. Pneumonia and other lung infections are treated with antibiotics, but recurrence of infections can be avoided by surgically removing the lesion.

In a small number of patients, the lung lesion (usually a CCAM) may harbor a malignant tumor later in life. This is another reason to recommend surgical removal of the lesion in early childhood, even if your child has never shown any symptoms.

GLOSSARY

Amniotic cavity: The space within the uterus in which the fetus resides, and bound by the amniotic membrane

Aorta: the main artery in the body; it comes directly off the left ventricle of the heart, and carries oxygen-rich blood to the entire body

Bronchogenic cyst: A lesion, found in the fetus, that develops when a portion of bronchus buds off from the rest of the lung and airways. Because the lining of the airways normally secretes some fluid, a bronchogenic cyst is filled with the same type of fluid, and appears as a fluid-filled mass

Bronchopulmonary: related to the lung and airways. **Bronchopulmonary sequestration:** see Sequestration

Bronchus: airway; tube-like structure whose function is to allow air to travel to and from the lung tissue

Congenital: condition that is present at or before birth. A congenital lung lesion is a lung lesion that is present in the fetus

Congenital diaphragmatic hernia (CDH): a hole in the diaphragm found in the fetus; it causes the intestines and other abdominal organs to move into the chest and compress the lungs, and often causes severe breathing problems at birth

Congenital lobar emphysema (CLE): a lung condition found in the fetus and the infant, whereby a portion of the lung is bloated and distended, because of a temporary and/or incomplete blockage of the corresponding airway. After birth, the lung tissue in that segment of the lung is distended and does not function properly

Cyst: a fluid-filled cavity

Cystic adenomatoid malformations (also called CCAM): a type of lung lesion that is composed of disorganized lung tissue. It can be mostly cystic (fluid-filled cavity), solid, or a combination of both

Diaphragm: the thin muscle layer that separates the chest from the abdominal cavity

“Double pigtail” catheter: a fine tube that can be left in a body cavity, such as the chest of the fetus. The pigtails refer to the curled ends of the catheter, which keep the catheter in place in that cavity

Esophagus: the food-pipe. In the embryo, the trachea (wind-pipe) and bronchi (the smaller airways) form from an outpouching of the upper portion of the esophagus

Extralobar: see Sequestration

Fetal: related to the fetus

Hybrid: that is composed of more than one type of tissue or structure. A hybrid lung lesion may be a sequestration with CCAM elements

Hydrops: heart failure in the fetus. It can manifest itself by enlargement of the heart, abnormal accumulation of fluid around the heart, around the lungs, in the abdominal cavity or under the skin

Intralobar: see Sequestration

Intubation: placement of a tube; refers specifically to the insertion of a plastic tube in someone's wind-pipe (trachea) and connecting it to a breathing machine (ventilator or respirator)

Lung abscess: a form of lung infection whereby the bacteria form a cavity surrounded by a thick wall through which blood vessels (and antibiotics) cannot penetrate. The abscess cavity is filled with pus, and treatment requires incision and drainage ("lancing")

Magnetic resonance imaging (MRI): A type of imaging test that does not use radiation, and provides very detailed images of the inside of the body. To obtain good images, the patient should lay still while the body is being scanned. Now that MRI machines take only a few seconds to do this, it is possible to obtain detailed images of the fetus

Malignant: cancerous

Minimally invasive surgery (thoracoscopy or laparoscopy): a newer form of surgical access to a body cavity: instead of making a wide incision and opening the cavity (chest or abdomen), small tubes are inserted and surgical procedures are performed inside the cavity using long, fine instruments and a miniaturized camera

Pulmonary hypoplasia: impaired growth and development of the lung. It can be caused by a variety of conditions, such as prolonged compression of the fetal lung by a large lesion in the chest or abdominal organs that have migrated through a hole in the diaphragm

Sequestration: A type of lung lesion found in the fetus. A sequestration develops when a portion of lung tissue (with its accompanying bronchus) becomes separated from the tracheobronchial tree. It contains normally organized lung tissue, but cannot function properly, since it is cut off from the normal airways. If the splitting off occurs early in the embryo, it will be completely separated from the normal lung (*extralobar* sequestration); if the splitting off occurs later, the sequestration will be found inside the normal lung (a so-called *intralobar* sequestration)

Trachea: the wind-pipe; the upper portion of the airways. It divides in two branches, the left and right main stem bronchus. Each main stem bronchus is the main airway to a lung

Tracheobronchial tree: another name for the airways. The trachea divides into two main stem bronchi, which in turn divide into smaller branches, resembling an (upside-down) tree

Thoracentesis: A technique whereby fluid is aspirated from the chest cavity (or cyst in the chest cavity) through a fine needle

Thoracotomy: an "open" operation of the chest, whereby a large incision is made to open the cavity