Congenital Diaphragmatic Hernia (CDH)

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

Congenital Diaphragmatic Hernia (CDH) is a condition where a baby is born with a hole in the diaphragm. The diaphragm is the muscle that separates the chest from the belly. Normally, the heart and lungs are in the chest and the intestines and other organs are in the belly. When there is a hole in the diaphragm, organs from the abdomen can go through the hole into the chest cavity. As the baby grows in the womb, their lungs are compressed by these abdominal organs. The lungs are smaller because they lack the space to grow. In addition, the blood vessels of the lung are thicker, making it sometimes difficult for oxygen in the blood to get to the lungs.

The cause of this defect is not fully known. CDH occurs in about 1.9 in every 10,000 births. The size of the hole can vary. Larger holes are associated with more problems because the lungs are usually smaller. Sometimes the lungs are so small that the babies are not able to breathe adequately after they are born, and they don't survive. Most diaphragmatic hernias occur on the left side (80%) and will often contain stomach, spleen, small and large intestine. Those on the right side will often contain portions of the liver.

Signs and Symptoms - "What symptoms will my child have?"

Diaphragmatic hernias are mostly discovered during routine prenatal ultrasounds of the mother. An MRI (magnetic resonance imaging radiology test that uses magnetic waves to create images of the inside of the body) of the developing fetus can sometimes be done to predict how big the lungs are and how badly they are being compressed by the intestines and other organs. If a fetus is diagnosed with CDH, his or her birth is planned at a hospital with the capability of caring for babies with this diagnosis. The baby may have severe breathing difficulties, low oxygen levels in the blood and problems with blood pressure. These symptoms can be so serious that they can be life-threatening.

Some babies with CDH that do not have symptoms right after birth may later develop problems tolerating their diet or may begin to have difficulty breathing. Some may be discovered after an X-ray is taken for respiratory symptoms such as coughing or wheezing.

Diagnosis - "What tests are done to find out what my child has?"

- A thorough **physical examination** by an experienced health care provider is needed.
- **Chest X-ray** usually shows intestinal loops in the chest which confirms the diagnosis of CDH.

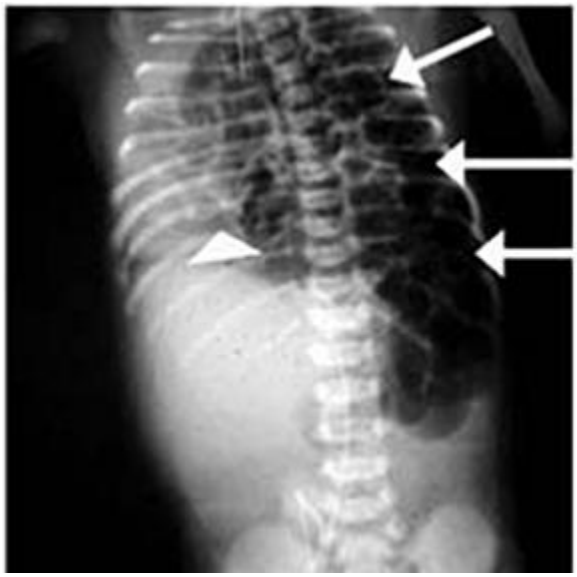

Figure 1: Chest Xray showing "bubbles" in the left chest indicating intestine pushing into the chest (small arrows) and heart being pushed over to the right (single large arrow).

- **Blood test** to check routine labs and to look for oxygen levels in the blood which show how well the lungs are working.
- **Echocardiogram ("ECHO") or ultrasound of the heart** will be performed to look for any congenital heart problems and to determine if the heart is being affected by the increased pressure in the lungs from the abnormal growth of lungs. Some babies also have problems with their heart, which may occur in up to 15% of babies with a CDH. This study may also indicate how bad the blood pressure is within the lungs (pulmonary hypertension).
- An **ultrasound of the head** is usually obtained to make sure that there are no abnormalities or bleeding into/around the brain.

Conditions that mimic this condition:

- *Diaphragm Eventration:* An abnormal elevation of one side of the diaphragm. This may be congenital (baby is born with it) causing the diaphragm muscle to be very thin, or it may occur after an injury to the nerve that controls the diaphragm, which then weakens the muscle.
- *Foramen of Morgagni Diaphragmatic Hernia:* This is a defect in the center and front part of the diaphragm. These types of hernias account for less than 2% of diaphragmatic hernias and typically present in older children and adults.
- *Hiatal Hernia:* When the center opening in the diaphragm enlarges, the stomach can then go through the hole into the middle part of the chest. These types of hernias may be associated with symptoms of reflux such as vomiting and heartburn.

Treatment - "What will be done to make my child better?"

It is important to know that in babies with CDH, returning the intestines into the belly and repairing the hole in the diaphragm will not cure the breathing problems. The breathing problems are a result of (1) smaller lung size and (2) pulmonary hypertension.

Pulmonary hypertension is increased pressure in the arteries of the lungs. Under normal circumstances, blood goes through the vessels in the lungs. Blood picks up oxygen and gets rid of carbon dioxide in the lungs. However, if the pressure is high in these vessels, blood is shunted away from the lungs. Exchange of oxygen and carbon dioxide does not happen effectively between the blood and the lungs. Pulmonary hypertension is a life-threatening problem.

In most cases, the blood pressure within the lungs goes down within several days after delivery. The main issue after birth is to support breathing (bringing oxygen into the body and getting rid of carbon dioxide) without causing permanent and severe damage to the lungs.

Medical therapy: The baby is admitted to the neonatal intensive care unit and a breathing tube is placed to help with breathing. A machine (ventilator) is used to regulate breathing, give oxygen, and get rid of carbon dioxide. There are several types of machines that deliver increasing and different types of support. The baby's breathing needs will determine the machine used.

A tube is placed through the mouth into the stomach to drain air or fluid. A full stomach can compress the lung and lead to more problems breathing.

Medicines are usually given intravenously for sedation and pain. These medicines can calm the baby and decrease the pressures within the lung.

Catheters will be placed in veins (IV) or arteries to deliver medications and also to draw blood to check oxygen levels.

Surgery: Surgical options depend upon how well the baby is able to absorb oxygen (oxygenate) and breath off carbon dioxide (ventilate). If, despite breathing machine management and medications, the baby is not able to oxygenate or ventilate, the baby may need to have a specialized procedure called ECMO (see below).

- **ECMO** (Extracorporeal membrane oxygenation) is a type of heart-lung bypass. At the bedside, cannulas (large catheters) are placed into the blood vessels in the neck. Oxygen-poor blood is drained from the baby through the cannula in the neck and put through a machine where oxygen is infused into the blood and carbon dioxide is removed. This oxygen-rich blood is then returned back into the baby. Heparin, a medicine that thins the blood, is given to the baby to keep the ECMO circuit from clotting.
- **Preoperative preparation:** Echocardiogram (heart ultrasound) and head ultrasound are usually required prior to ECMO.
- **Benefits and risks:** ECMO is used when other medical treatment options fail. ECMO takes on the job of the heart and lungs while pulmonary hypertension resolves. The heart and lungs are able to rest. A baby can only stay on ECMO for a brief time, up to 21 days. If the baby's lung function does not get better at that time, then the prognosis is poor. Risks of ECMO are bleeding, dislodgement of the cannula, and clotting of the circuit. Placement of the cannulas can also be risky.
- If the lung function and pulmonary hypertension gets better, the baby is taken off ECMO and placed back on a regular ventilator. Repair can be done either on ECMO or after the baby is taken off.

Congenital diaphragmatic hernia repair: Timing of surgical repair is typically dependent on the baby's stability and degree of pulmonary hypertension. Sometimes, surgery may be performed while the baby is still on ECMO. The type of surgery will be up to the surgeon and can often be performed through an incision on the abdomen or through the chest. Minimal access approaches (laparoscopy or thoracoscopy) may also be used. For large defects in the diaphragm, a patch (synthetic piece of material) may have to be used to close the hole.

- **Postoperative care:** Following the surgery, the baby will need the same monitoring and support as before the surgery. Often, the baby will need to remain on the ventilator to support the lung function and may need medications to lower lung blood vessel pressure and other medications to raise overall blood pressure.
- **Risks/Benefits:** The primary risks of surgery include bleeding, infection, injury to the lung, heart or abdominal organs; respiratory failure and death. Survival rates vary from 60-90%. There is also a risk of recurrence of the hernia; recurrence is greater in those that had a patch placed at the time of surgery. Benefits of the surgery include the replacement of the abdominal organs in the abdomen, which allows the heart and lungs to resume their normal positions and also allows the lungs to fully expand and to grow normally.

- Sometimes, if there are a lot of organs that were originally in the chest, replacing them all back in the belly may make the belly too tight. A tight belly would decrease the blood flow to the organs, creating a dangerous situation. In these cases, the abdomen is left open with the organs contained in a silastic silo or closed with a large temporary patch. The belly is then closed in stages.
- **Informed consent:** A consent form is a legal document that states the tests, treatments or procedures that your child may need and the doctor or practitioner that will perform them. Before surgery, your doctor should tell you what the operation is, the goal of the surgery and other possible treatment options that are available. Your doctor should explain the risks and benefits of the surgery. You give your permission when you sign the consent form. You can have someone sign this form for you if you are not able to sign it. You have the right to understand your child's medical care in words you know. Before you sign the consent form, make sure all of your questions are answered. It is important to know that during surgery, there are things that can happen that your doctor may have not predicted before going in. He or she will explain these to you after the surgery.

Home Care - "What do I need to do once my child goes home?"

Diet: Your child will likely be on a typical infant formula or breast milk but may require nutritional supplementation to increase calories and protein. In some cases, if the lungs continue to be weak, feeding by mouth may be too much to coordinate with breathing. The baby may require feeding by tube through the nose or a gastrostomy tube (G tube).

Activity: Normal activity after the wounds have healed is allowed. Ask the surgeon when "tummy time" is allowed.

Wound care: Wounds are kept dry for the first three days after surgery, then the wounds may be washed but not soaked for a week. Most of the time, if the baby was born with CDH and repair was performed, he or she should be able to be bathed normally by the time he or she gets home.

Medicines: The baby may need to continue on some of the medicines for pulmonary hypertension if he/she continues to have difficulty with breathing.

What to call the doctor for: Call the doctor for fevers, worsening breathing, vomiting, pain or any redness/drainage from the incisions.

Follow-up care: You will need close follow-up with the pediatric surgeon as well as your pediatrician and possible pediatric pulmonologist.

Long Term Outcomes - "Are there future conditions to worry about?"

- Survival rates for all infants with CDH vary from 60-90%. The most common long-term problems are issues with breathing and follow-up with lung specialists may be needed.
- Some children may not have the same exercise tolerance as their peers due to the lung abnormalities.
- There is a higher incidence of developmental delay in children with CDH—motor and language problems may be evident. Hearing problems may also be present.
- A high incidence of gastroesophageal reflux (acid coming up from the stomach into the esophagus) is often seen in babies with CDH and some eventually need surgery to correct this problem.
- Hernia recurrence (the hole opens up and the hernia comes back) is seen in up to 50% in babies requiring a patch and in 10% of those that did not need a patch. Therefore, regular follow-up is needed.

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