CONGENITAL DIAPHRAGMATIC HERNIA

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Introduction

- Congenital diaphragmatic hernia (CDH) is a common malformation characterized by a defect in the posterolateral diaphragm, the foramen of Bochdalek, through which the abdominal viscera migrate into the chest during fetal life.

Epidemiology

- Incidence- 1 in 2000 to 5000 per live birth.
- One third of neonates with CDH are stillborn.
- So, the exact prevalence of the disease is underestimated.

- When still births are counted with live birth, the incidence is more common in females.
- Infants with isolated CDH are typically premature, macrosomic male.

- One third are associated with a major congenital anomaly.
- Approximately 80% are left sided.
- Bilateral defects are rare and are associated with other major anomalies.
- Although the exact etiology remains unknown, mothers that are thin or underweight may have an increased risk of bearing an infant with CDH.
- CDH may be due to the exposure of genetically predisposed or susceptible individuals to environmental factors.
- Exposure to a number of pharmacologic agents and environmental hazards has been implicated in its development.
- These include insecticides and drugs, such as phenmetrazine, thalidomide, quinine, cadmium, lead, and nitrofen.
- Retinoid-regulated target genes may be responsible for CDH development. (Vitamin A deficiency is seen in patients of CDH)

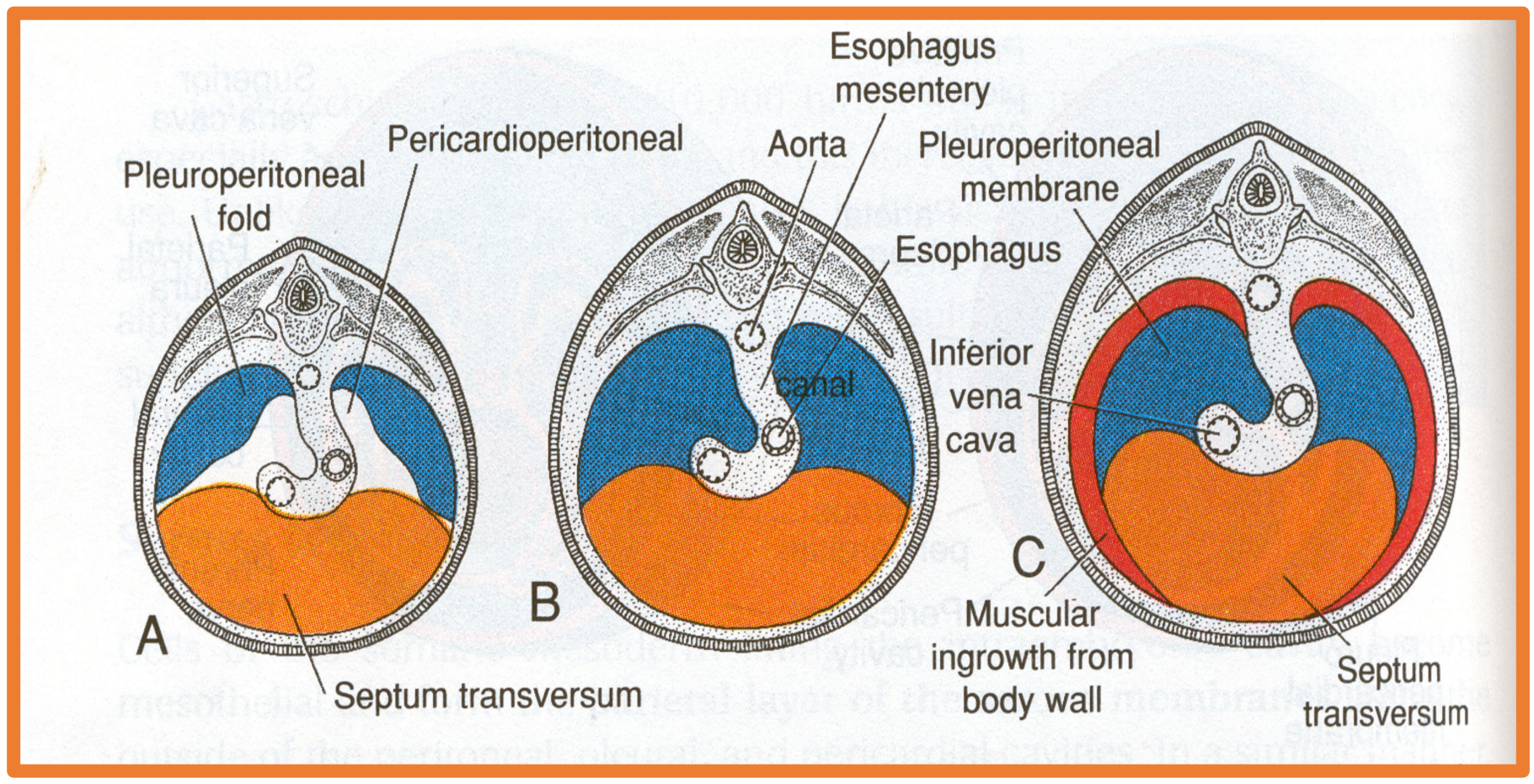
Associated anomalies

- Approximately 50% of CDH are isolated defects.
- Others are associated with anomalies of the cardiovascular (27.5%), urogenital (17.7%), musculoskeletal (15.7%), and central nervous (9.8%) systems (CNS).
- Anomalies as a consequence of diaphragmatic defect:
  - lung hypoplasia, intestinal malrotation, some cardiac malformations, and patent ductus arteriosus (PDA) are considered to be consequences of the diaphragmatic defect.
- Non-CDH-related defects are estimated to occur in 40–60% of cases and can involve the cardiovascular, CNS, gastrointestinal, and genitourinary systems.

Development of diaphragm

- The fully developed diaphragm is derived from four distinct components:
  1. the anterior central tendon forms from the septum transversum
  2. the dorsolateral portions form from the pleuroperitoneal membranes
  3. the dorsal crura evolve from the esophageal mesentery
  4. the muscular portion of the diaphragm develops from the thoracic intercostal muscle groups.
- The pleuroperitoneal folds grow ventrally and fuse with the septum transversum and dorsal mesentery of the esophagus during gestational week 6.
- Complete closure of the canal takes place during week 8 of gestation.
- Anatomically, the right side closes before the left.
- Neuro-muscularization of the diaphragm is the last in the development and matures the diaphragm.

Development of Diaphragm



Pathology of CDH

- Failure of closure of pleuro-peritoneal canal
- Most common area is a postero-lateral defect (Bochdalek)
- Left side more common
- Herniated contents
  - Left- left lobe of liver, spleen and bowel
  - Right- Liver and other viscera.

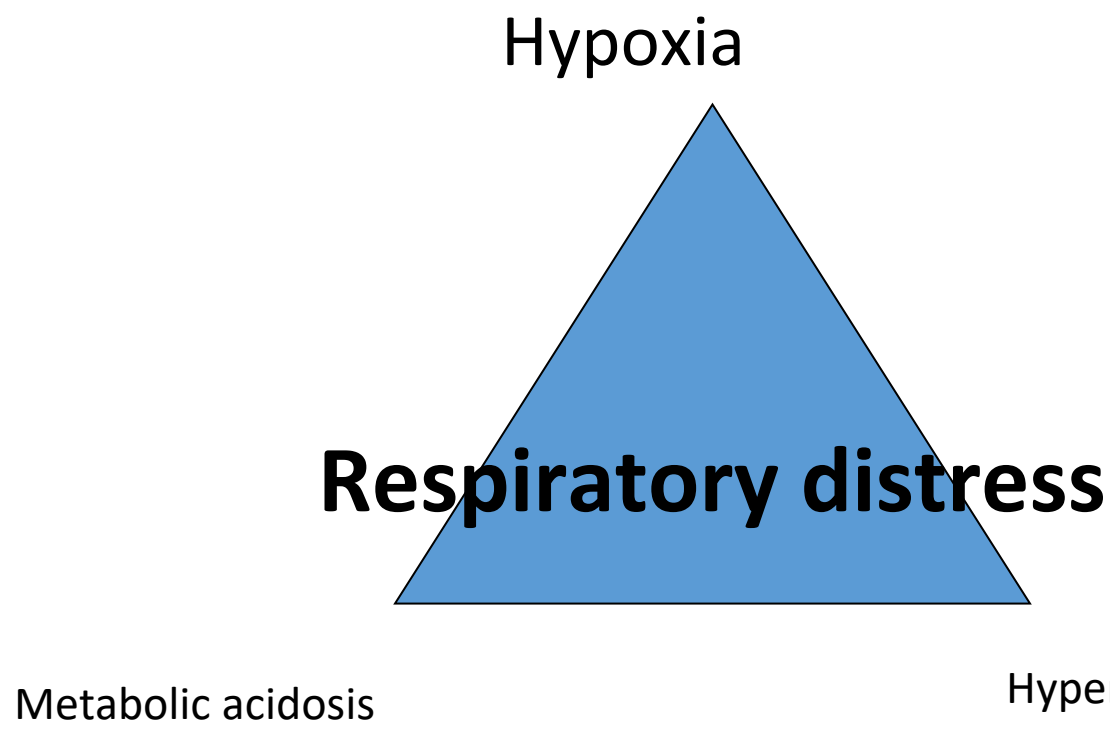
Lung development in CDH

- Both the lungs are affected-Lung hypoplasia.
- Ipsilateral > Contralateral
- No. of bronchial branches – greatly reduced
- Alveolar development severely affected
- Increased muscle mass in the conducting airways

Pulmonary vasculature in CDH

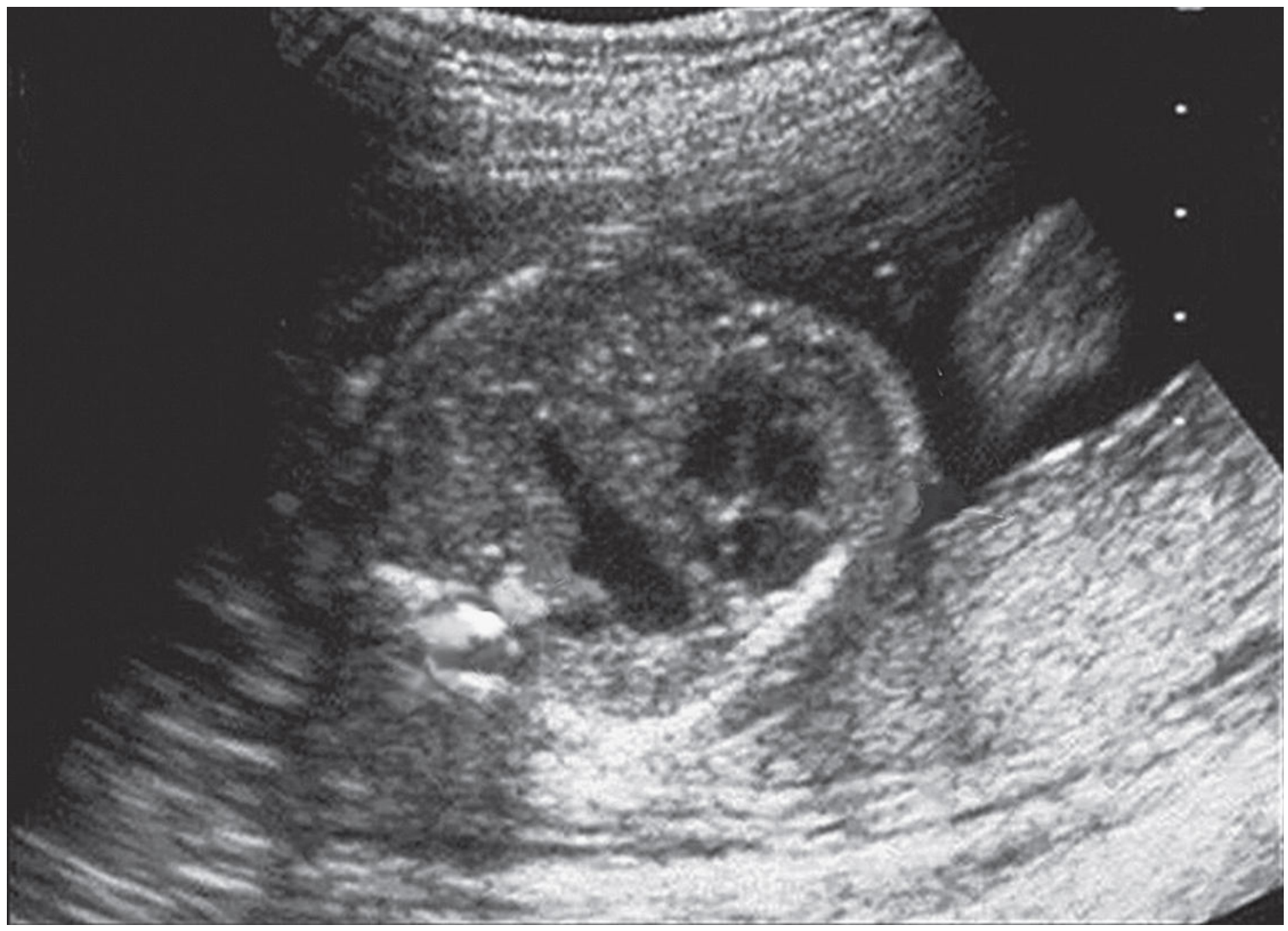
- Both the lungs are affected.
- Reduction in the total no. of branches
- Significant adventitial and medial wall thickening
- No significant changes in pulmonary venous structure
- Increased susceptibility to PPH
  - hypoxia, acidosis, hypothermia, stress

Problems: in CDH



Diagnosis: Prenatal Diagnosis

- Prenatal USG:
  - Mean gestational age at discovery is 24weeks.
  - Presence of polyhydramnios (80% cases of CDH)- due to kinking of the gastro-esophageal junction by translocation of the stomach into the thorax with resultant foregut obstruction.
  - Presence of stomach in the fetal thorax at the same cross-sectional level of heart.
  - Three-dimensional estimation of the fetal lung volume: important prognostic indicator.
  - Lung-to-head ratio has been the most widely used prognostic indicator.
- Fetal MRI



Fetal ultrasound image at the level of the four chamber heart (dotted arrow). Gastric bubble (solid arrow) at the level of the four-chamber heart suggests CDH.

Clinical presentation

- Newborns with CDH typically present with respiratory distress.
- Immediate respiratory distress with associated low Apgar scores to an initial stable period and a delay in respiratory distress for 24 to 48 hours.
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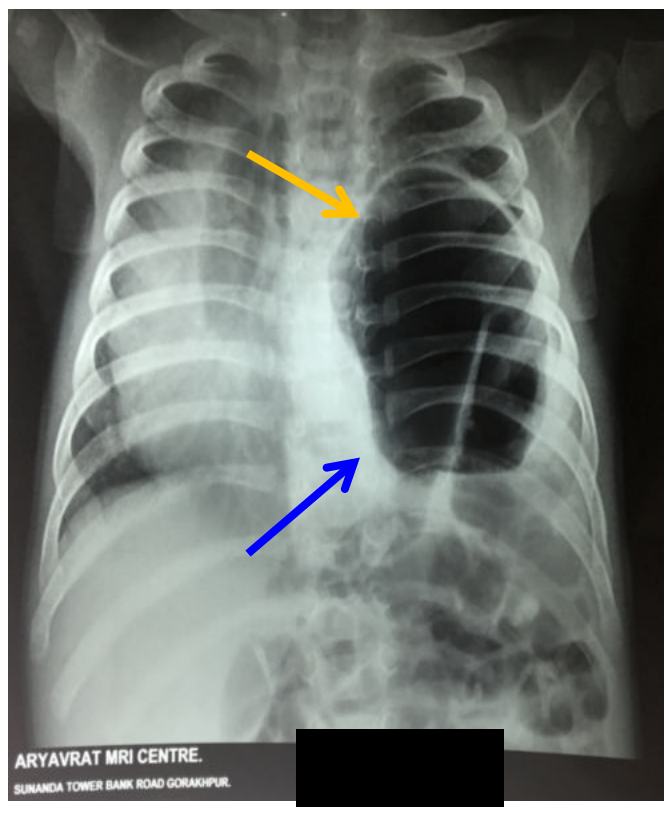
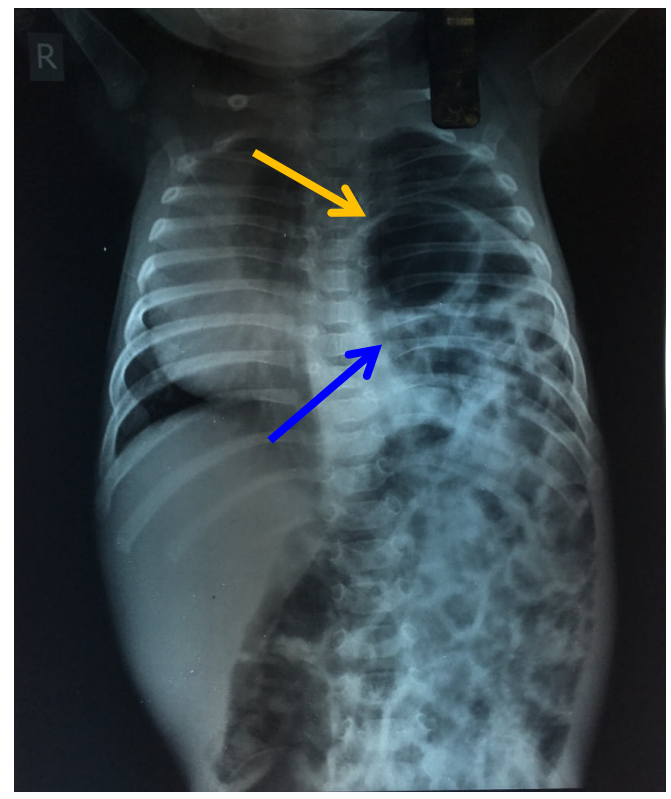
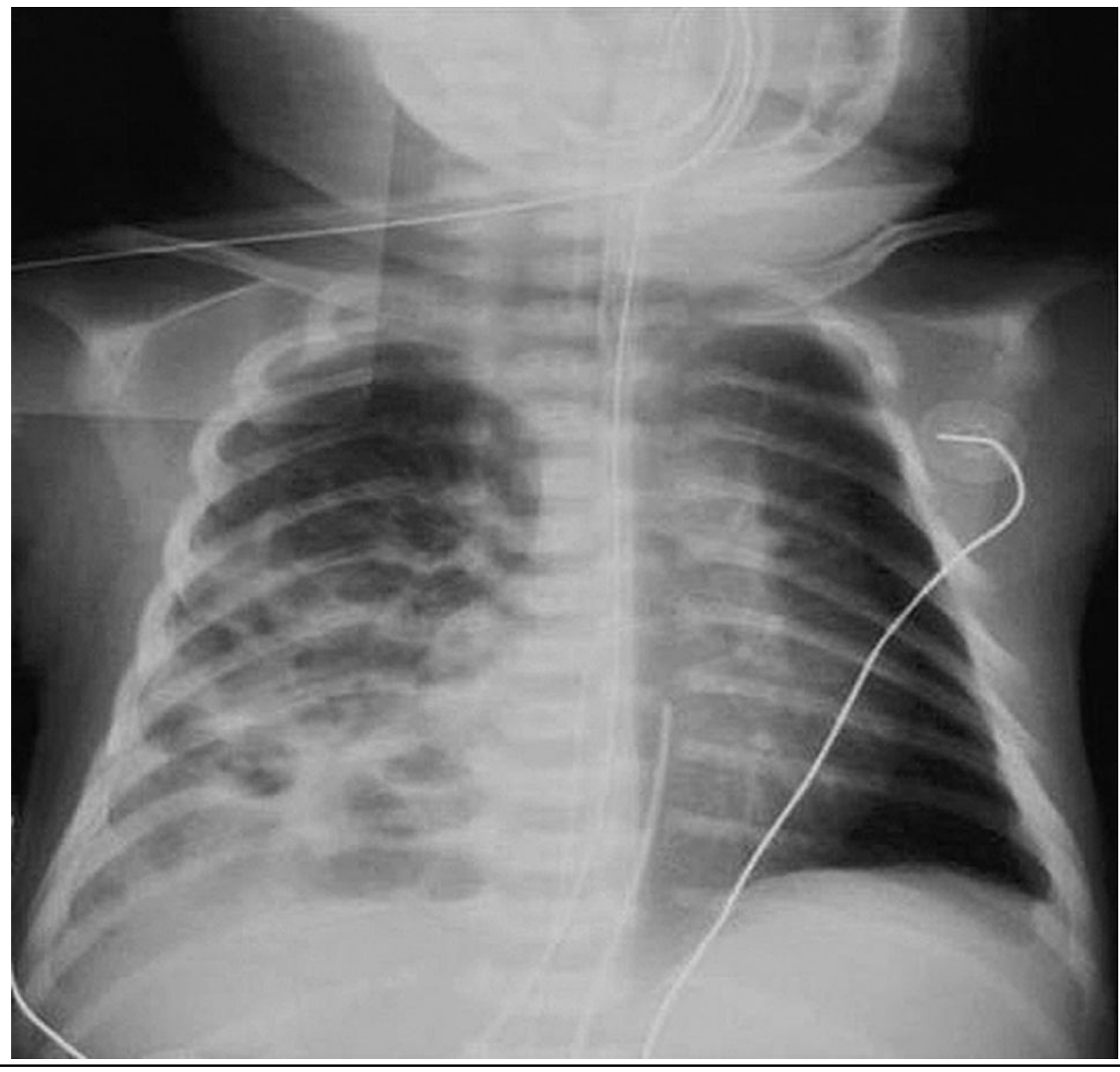
- Initial signs associated with respiratory distress include tachypnea, chest wall retractions, grunting, cyanosis, and pallor.

- On physical examination:
  - scaphoid abdomen and an increased chest diameter.
  - The point of maximal cardiac impulse is often displaced, suggesting mediastinal shift.
  - Bowel sounds may be auscultated within the chest cavity with a decrease in breath sounds bilaterally.
  - Chest excursion may be reduced, suggesting a lower tidal volume.

- The diagnosis of CDH is typically confirmed by a chest radiograph demonstrating intestinal loops within the thorax.
- The abdominal cavity may have minimal to no gas.







Right-sided CDH.

- Occasionally, CDH may be completely asymptomatic and is only discovered incidentally.

- Older patients who present later in life have a much better prognosis due to milder or absent associated complications, such as pulmonary hypoplasia and hypertension.

Prenatal Care

- Referral to tertiary care centres where respiratory distress of neonates can be managed.

- Prenatal corticosteroids:
  - To enhance the lung development in the premature infants
  - Role in CDH is not determined.

Pre-operative care

- Resuscitation:
  - Cardio-respiratory system stabilisation
  - Endotracheal intubation
  - Nasogastric tube insertion
  - Ventilation by mask and Ambu bag is contraindicated to avoid distention of the stomach and intestines that may be in the thoracic cavity.
  - Arterial and venous access through umbilicus
  - Infant to be properly sedated
  - Ventilation at low pressures and high rate

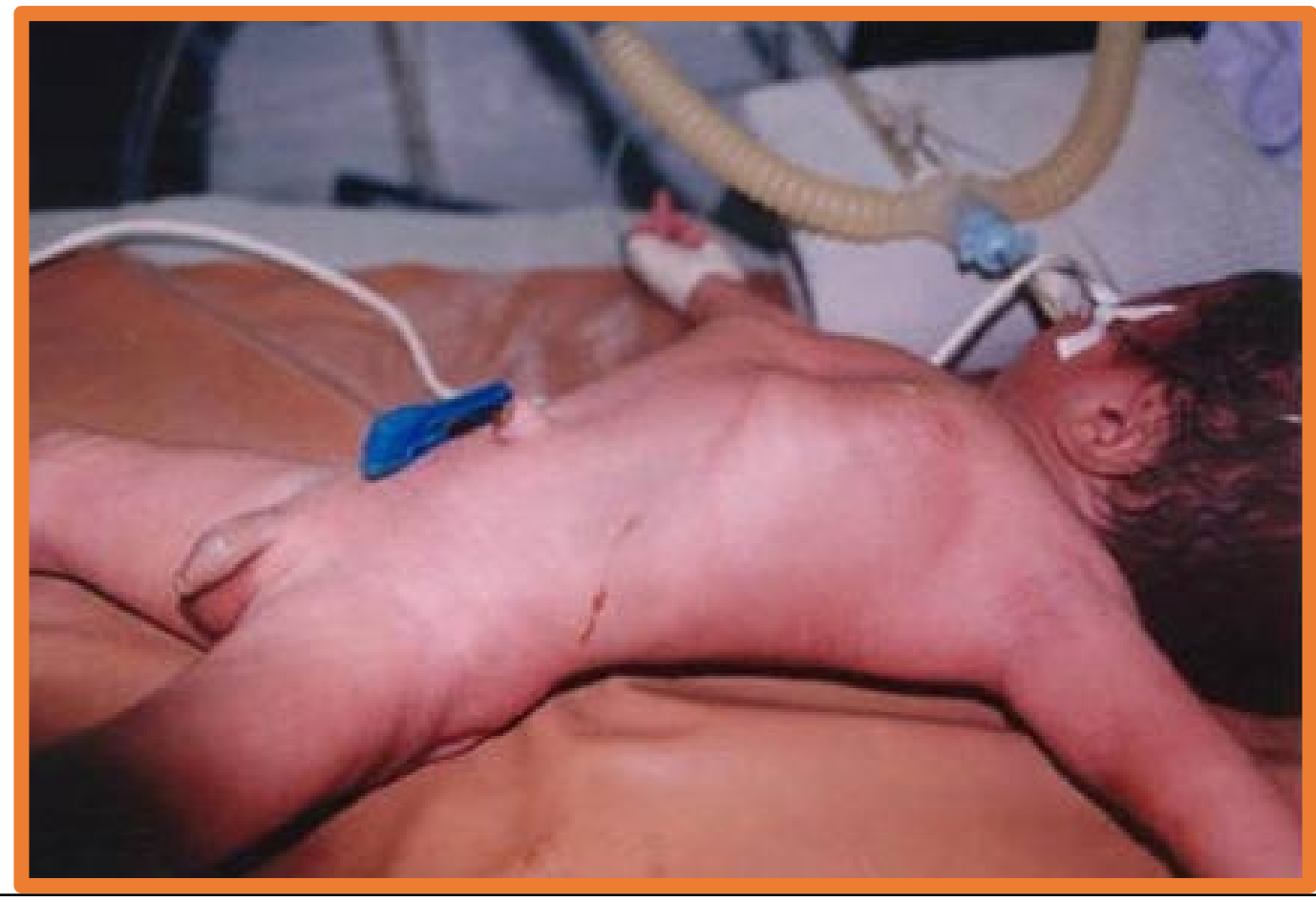
- Pharmacology:
  - Drugs to reduce pulmonary hypertension:
    - calcium channel blockers, prostacyclin derivatives, endothelin receptor antagonists, and phosphodiesterase-5 inhibitors such as sildenafil

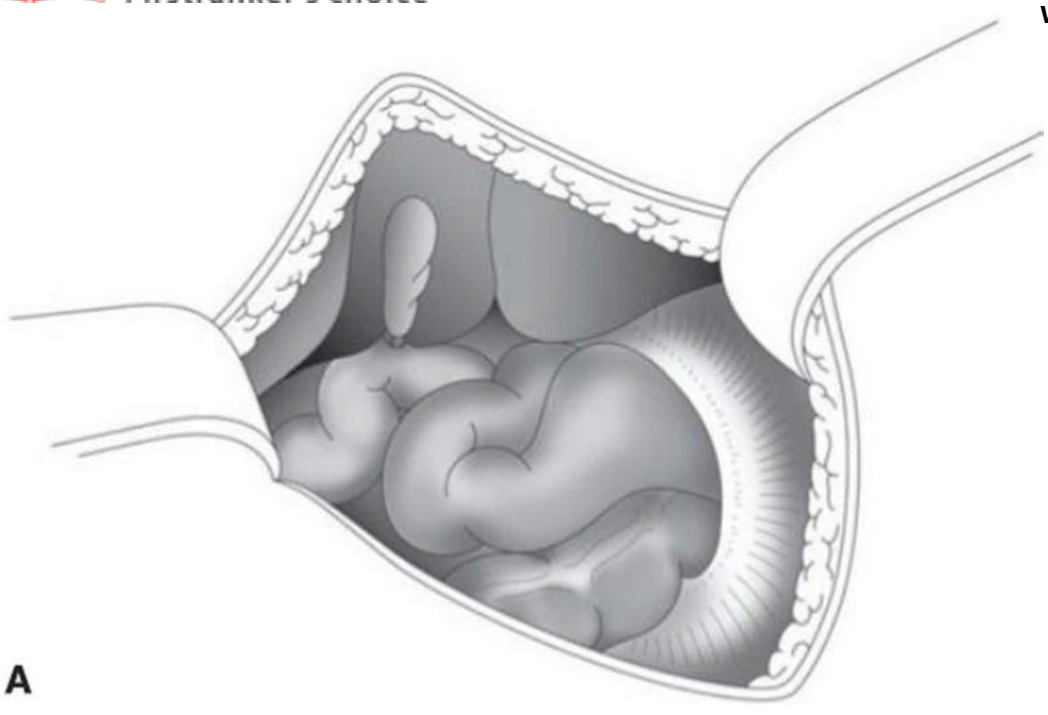
- Surfactants:
  - There is deficiency of surfactants in CDH.
  - May improve respiratory function: doubtful overall benefit

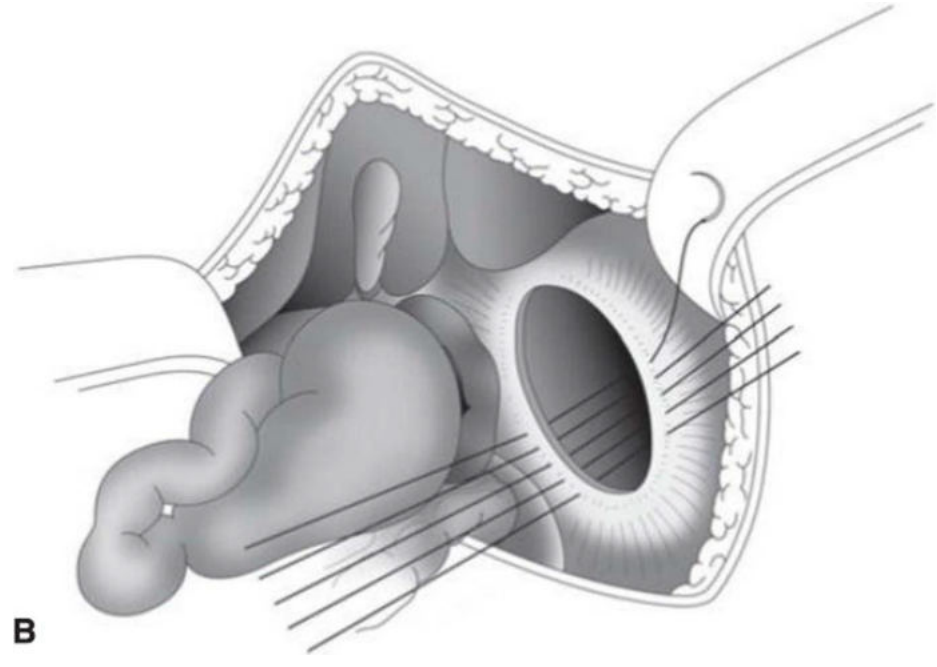
- Inhalational nitric oxide:
  - Potent vasodilator
  - May reduce pulmonary hypertension

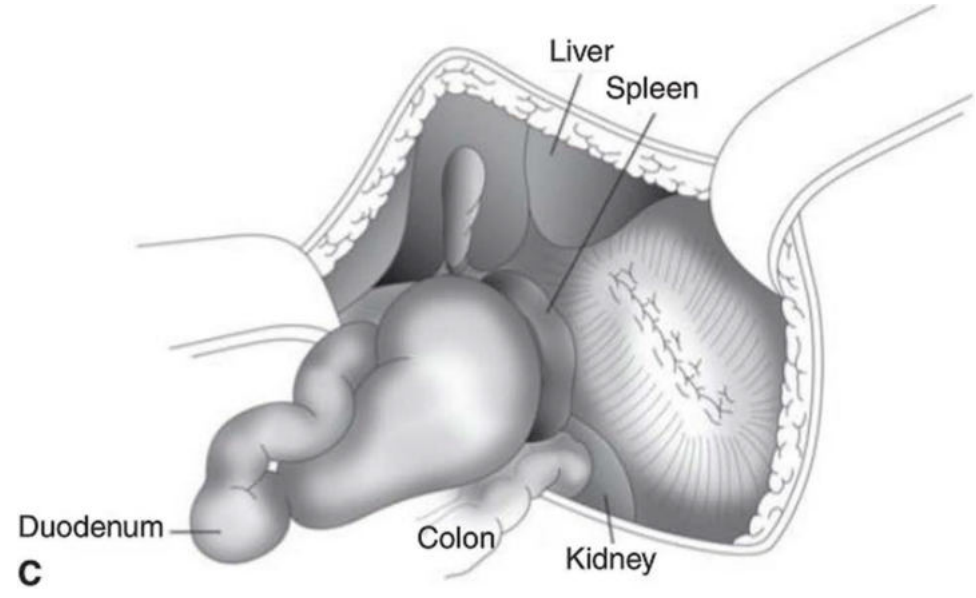
Surgery

- Timing of surgery:
  - CDH is a medical emergency not a surgical emergency.
  - Surgery once the baby is stabilised with minimal respiratory support and least pulmonary hypertension.
- Surgical steps:
  - Ipsilateral abdominal incision
  - Defect is exposed.
  - Reduction of the abdominal viscera
  - Closure of the defect : Primary or grafts
  - Wound closure

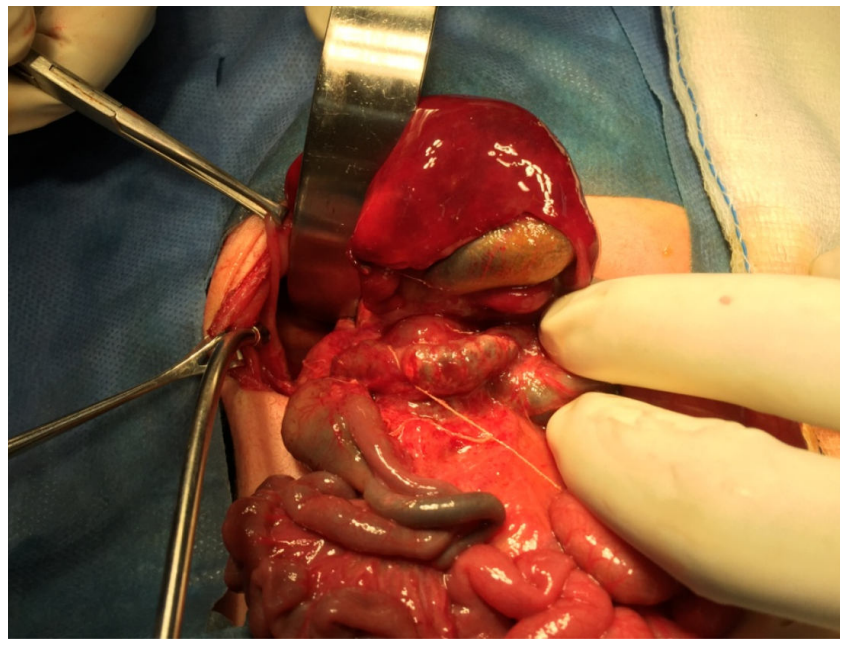


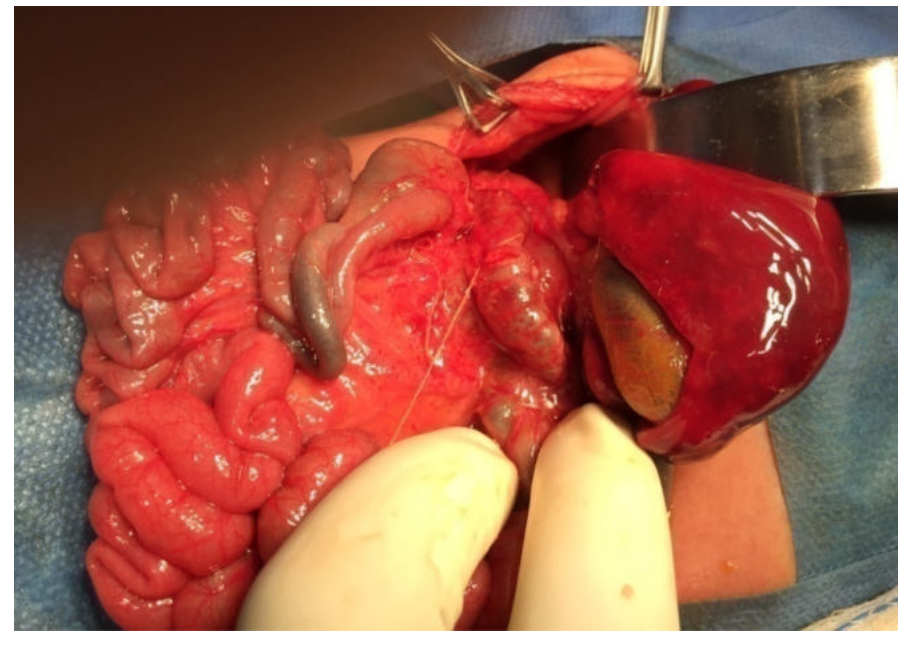






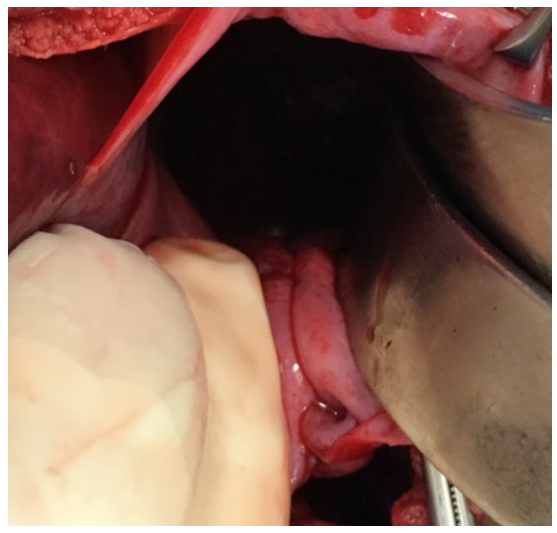
Case -3: (CDH)

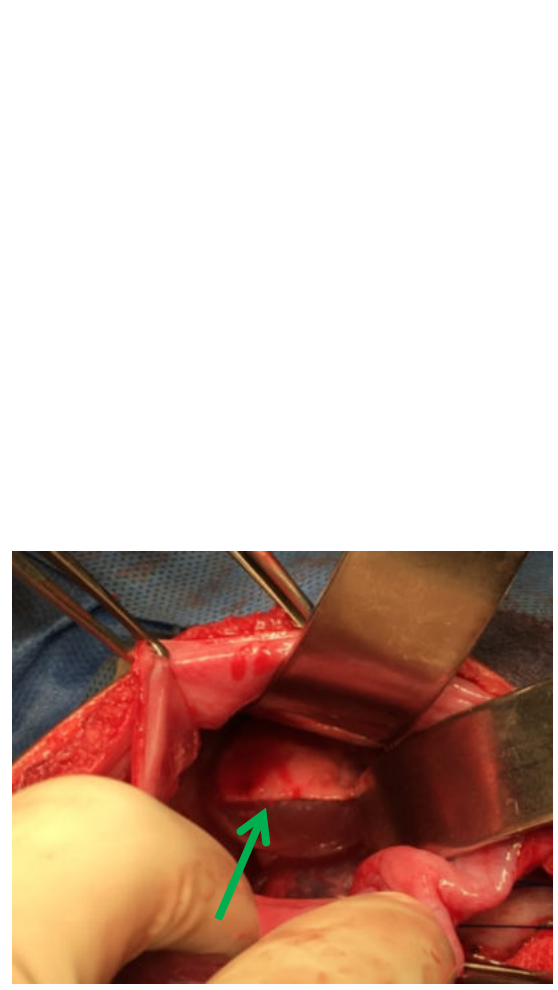


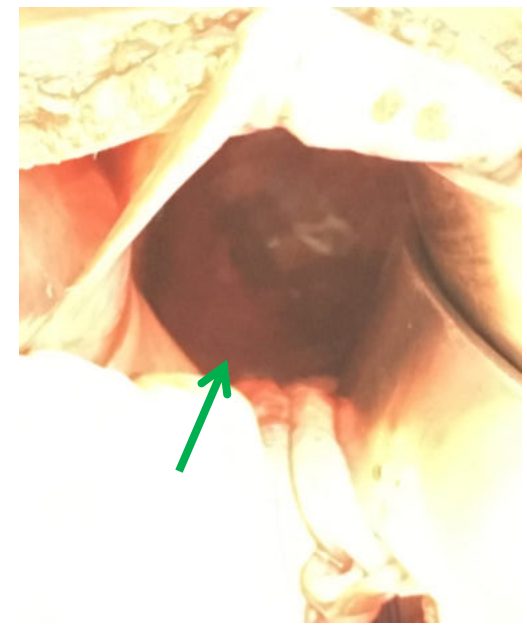


Intra-operative pictures showing the diaphragmatic defect and margins of diaphragm

Case -3: CDH







- Other surgical approaches
- Thoracic approach

- Minimal access surgery
  - Thoracoscopic approach

  - Laparoscopic approach

- Post surgery, the baby is kept on mechanical ventilatory supports with abdominal decompression with the help of nasogastric tube, rectal washes, and urinary catheterisation.

- Gradually, supports are weaned.

Outcome

- Survival rates at advanced centres are 60-90%.

- CDH survivors are at significant risk for chronic neurologic, developmental, gastrointestinal, nutritional, pulmonary, musculoskeletal, and other disorders.

- Late deaths have been reported in approximately 10% of initial survivors, mainly because of the consequences of persistent pulmonary hypertension.

- Respiratory risk:
  - Pneumonias
  - Reactive airway disease
  - Cor pulmonale
- Gastrointestinal:
  - Gastroesophageal reflux
  - Nutritional and growth related problems
  - Malrotation
- Musculoskeletal:
  - Chest wall deformities and scoliosis
- Neurodevelopmental abnormalities.
  - Problems in motor and cognitive skills.