Congenital Diaphragmatic Hernia (CDH)

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- It's a discontinuity in the diaphragm resulting in herniation of the abdominal content into the chest .

- In both Europe and the United States, the prevalence of CDH is ¬ 2.3–2.4 per 10,000 live births

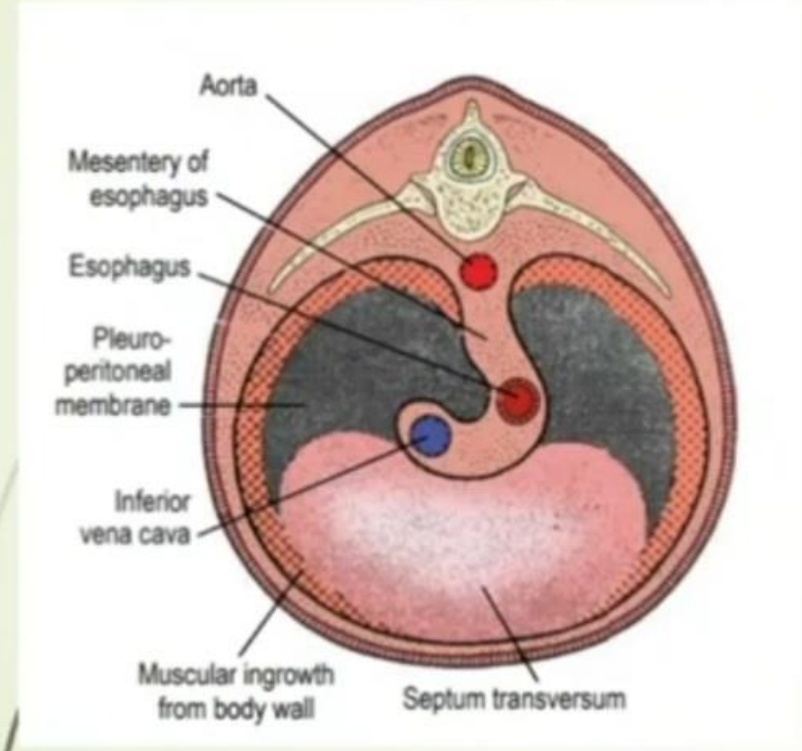
- Male>female

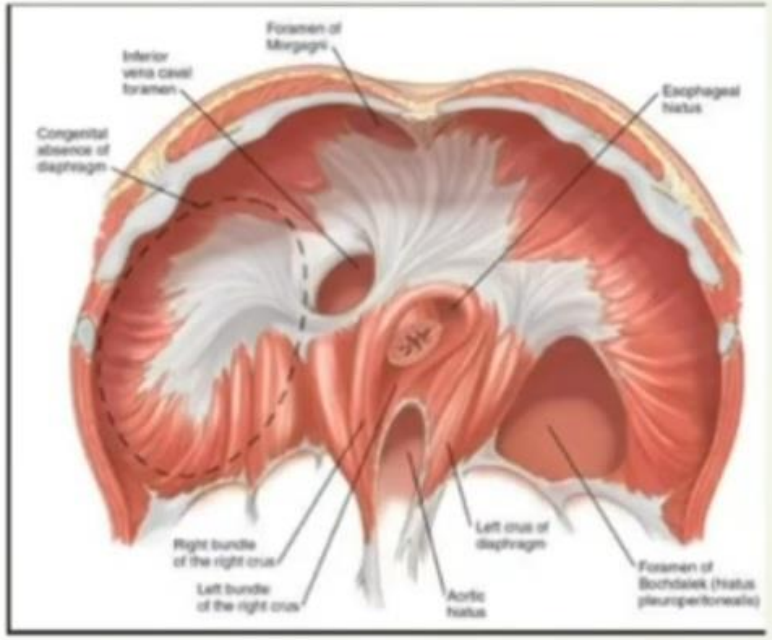
Associated Anomalies

- 60% of CDH cases are isolated , better survival
- **Genetic and chromosomal abnormalities** (e.g Trisomy 21,13,18) cause found in 1/3 of the cases
- **Associated anomalies** : cardiovascular (27.5%)
  urogenital (17.7%)
  musculoskeletal (15.7%)
  central nervous system (9.8%)
- **Syndromes** : Fryns and Donnai–Barrow syndromes, Simpson–Golabi–Behmel ,Beckwith–Wiedemann ,CHARGE , Pentalogy of Cantrell

- Isolated type has higher survival rate (up to 70-85%) compared to 20% in non-isolated type
- Major congenital heart disease is a significant contributor to morbidity and mortality in newborns with CDH.

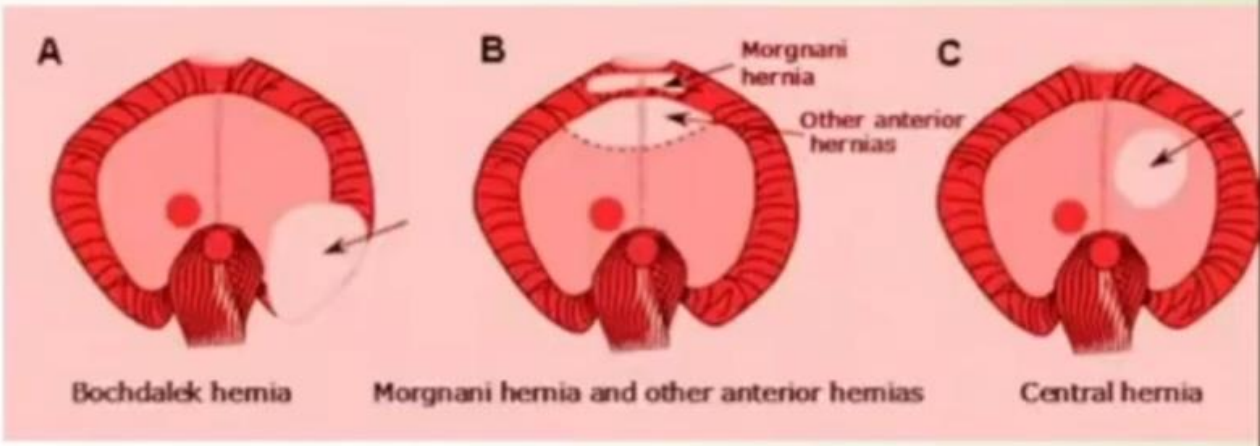
Embryology

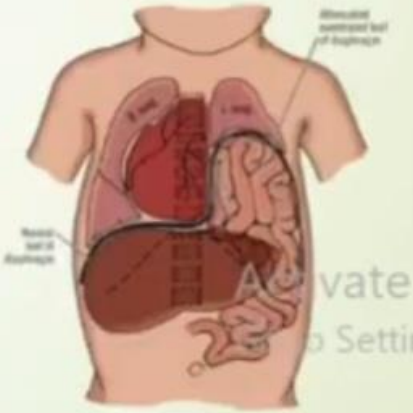




Types :

- Posterio-lateral Hernia
  * 90%
  * left sided in 80% , right 19% , bilateral 1%*
- Anterior Hernia (Morgagni)
  *2%
- Central Hernia
  * rare
- Diaphragmatic Eventration



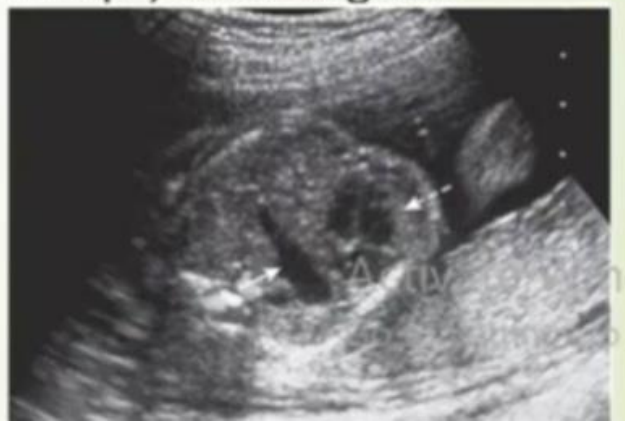


Why its important :

- High/long-term morbidity and mortality rate
- Antenatally : fetal hydrops and stillbirth
- Postnatally : chronic lung disease from CDH-associated pulmonary vascular hypertension (CDH-PH), and lung hypoplasia. Heart failure due to Persistent Pulmonary hypertension and Right-to-left shunting
- Neurocognitive delay, gastroesophageal reflux disease (GERD), musculoskeletal deformities, and operative complications including bowel obstruction or hernia recurrence

DIAGNOSIS:

- ¬50–70% of infants are identified antenatally
- The diagnosis of CDH is most often first between the 18th and 22nd weeks of pregnancy on ultrasound (US) screening exams
- Fetal US features include polyhydramnios, intrathoracic fluid-filled bowel loops, an echogenic chest mass, mediastinal shift, and/or an intrathoracic stomach/liver



- Two distinct US measurements have been utilized to risk stratify infants with CDH:

  (1) observed-to-expected (O/E) LHR (lung-Head ratio) , sever when ratio <25%

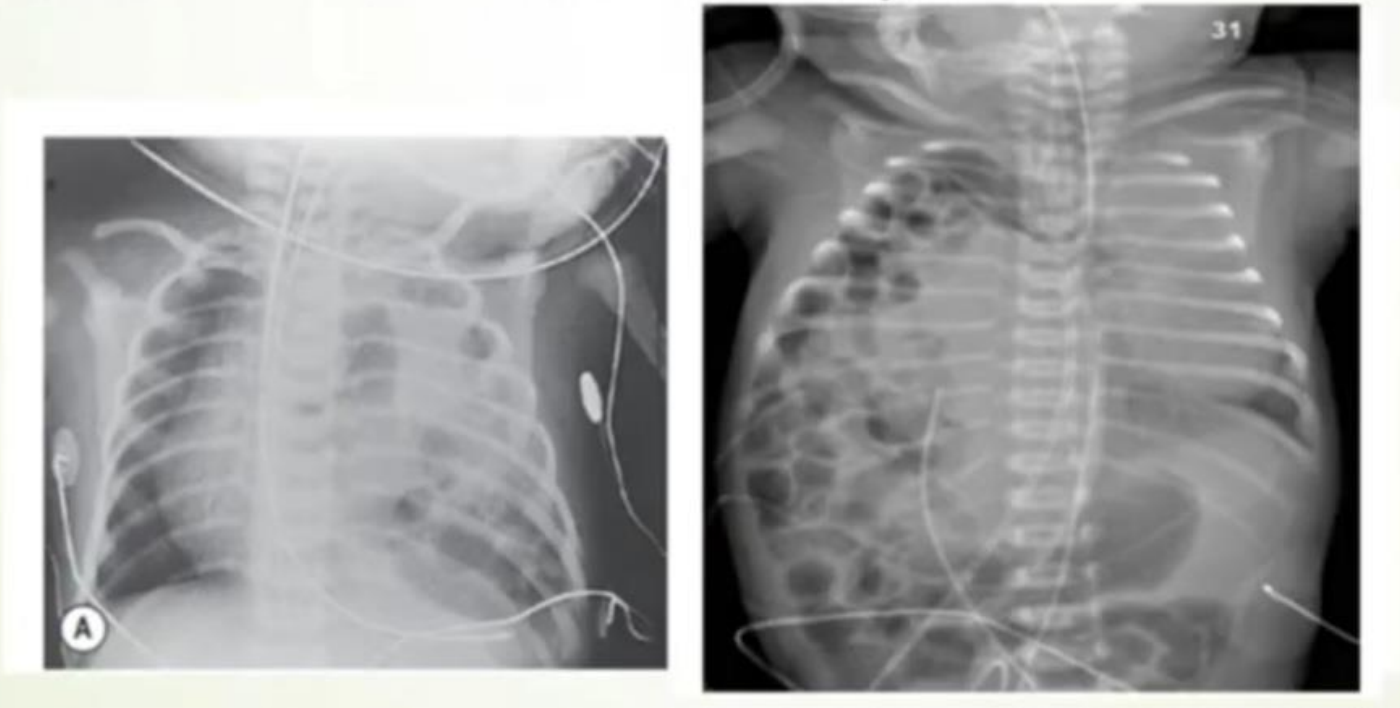
  (2) liver herniation into the hemithorax.

- Fetal MRI is an excellent modality to confirm and volumetric measurements of the fetal lung (total fetal lung volume [TFLV]).

- Survival rates with O/E TFLV 35%, survival ranged from 75–89%

Diagnosis

- Typically present with respiratory distress
- physical examination : Respiratory distress signs ,scaphoid abdomen ,displaced cardiac impulse, bowel sounds may be auscultated within the thoracic cavity with a decrease in breath sounds bilaterally.



Management :

- **PRENATAL CARE**:

Family counselling:

Chromosomal screening via amniocentesis for karyotyping and chromosome microarray analysis

Look for associated cardiac anomaly /fetal echocardiogram

observe lung development , hydrops fetalis

Delivery planning (site and timing)

Antenatal Intervention in severe cases

Randomized Trial of Fetal Surgery for Severe Left Diaphragmatic Hernia

Deprest JA et al. DOI: 10.1056/NEJMoa2027030

CLINICAL PROBLEM

Observational studies have shown that fetoscopic endoluminal tracheal occlusion (FETO) is associated with increased survival among infants with severe pulmonary hypoplasia due to isolated congenital diaphragmatic hernia on the left side, but data from randomized trials are lacking.

CLINICAL TRIAL

Design: An open-label, randomized, controlled trial was conducted to compare FETO with expectant care among women carrying singleton fetuses with isolated severe congenital diaphragmatic hernia on the left side.

Intervention: 95 women underwent randomization; 47 were assigned to undergo FETO at 27 to 29 weeks of gestation, and 48 were assigned to expectant care. The primary outcome was infant survival to discharge from the neonatal intensive care unit.

RESULTS

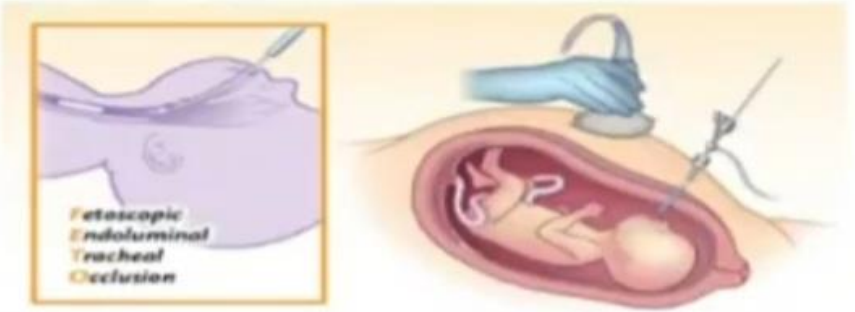
Efficacy: As compared with expectant care, FETO improved survival to discharge, and the benefit was sustained to 6 months of age. The trial was stopped early for efficacy after the third interim analysis.

Safety: FETO increased the risks of preterm, prelabor rupture of membranes and preterm birth.

LIMITATIONS AND REMAINING QUESTIONS

- Data on longer-term outcomes are needed, including neurodevelopmental outcomes.
- These results should not be generalized to centers without extensive experience in fetoscopy and FETO.

Links: Full article | NEJM Quick Take | Editorial



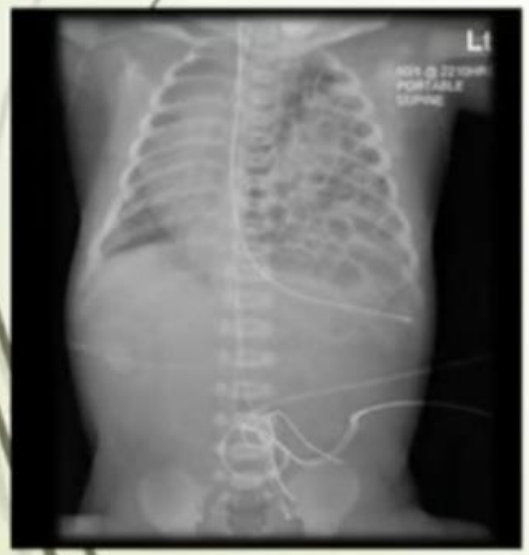
CONCLUSIONS

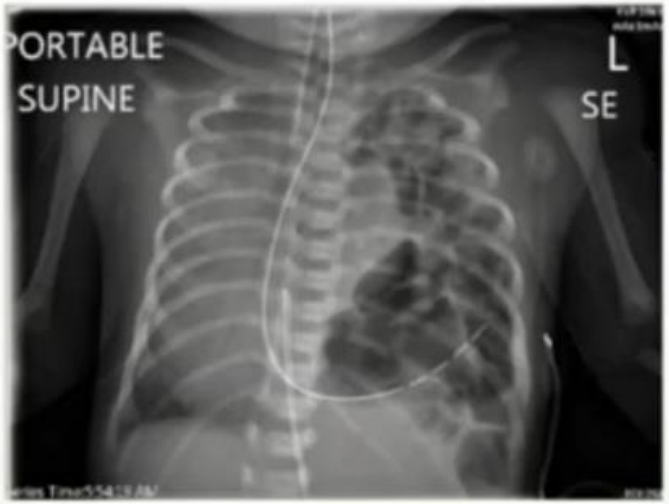
The use of FETO in fetuses with isolated severe congenital diaphragmatic hernia on the left side resulted in increased survival to hospital discharge but an increased risk of preterm, prelabor rupture of membranes and preterm birth.

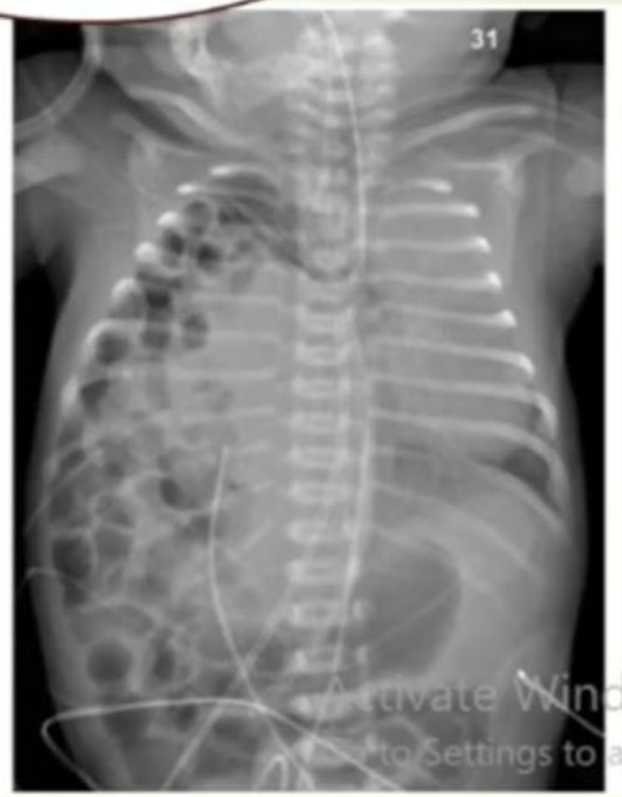
Goal is to resuscitate and stabilize baby especially those in cardiopulmonary distress.

- Immediate intubation post-delivery
- Mechanical ventilation /gentle hypercapnic ventilation
- NGT
- IV access and IVF
- Correction of acid-base status /avoid hypoxia and acidosis
- HFOV
- ECMO if no improvement
- Inotropic agents
- Manage pulmonary hypertension /Reverse right to left shunting with Nitric Oxide (iNO) , Sildenafil (PDE-5 inhibitor)
- No evidence to support surfactant and steroid

Aim :
preductal SaO2 >80-85%
PH >7.2
PaCO2 50-70mmHg
PIP **< 25 cmH2O**



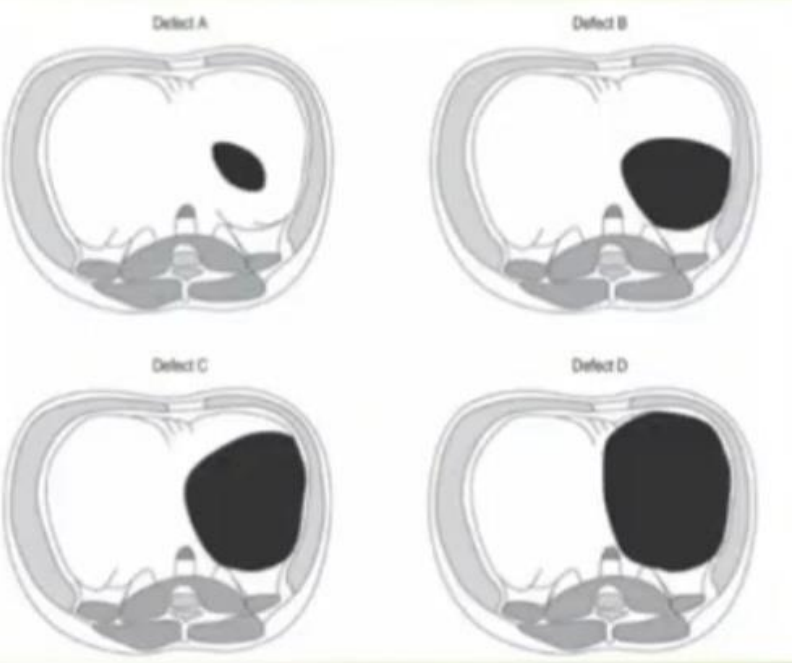




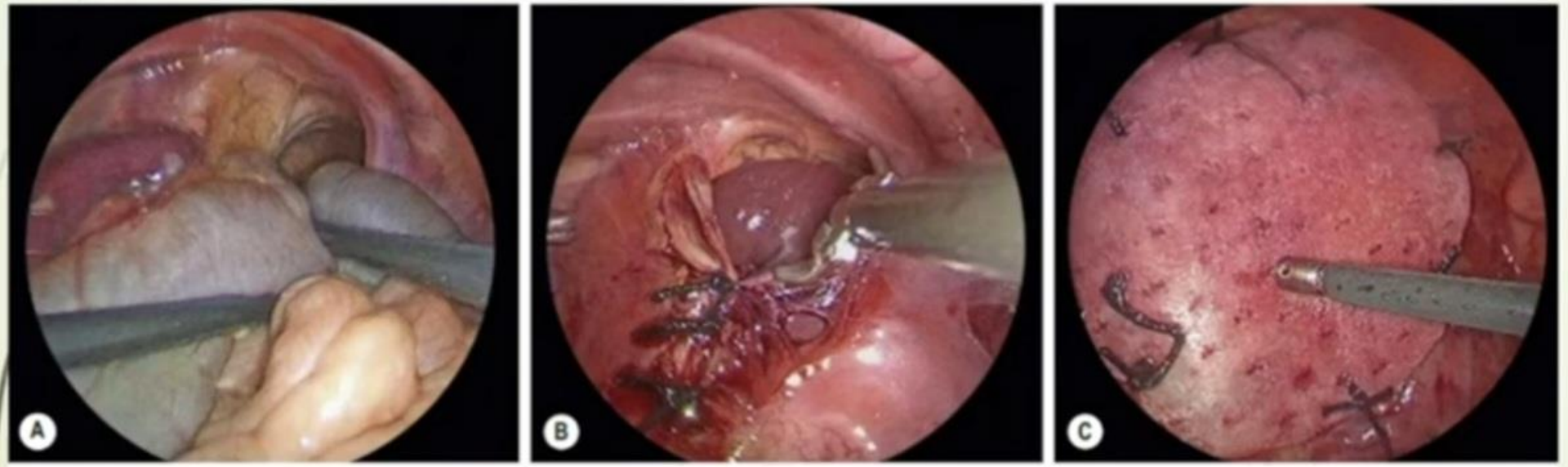
- Surgery : its not an urgent surgery , should be done when stable

  Open vs MIS (either abdominal or thoracic approach)

- The vast majority of open neonatal repairs for CDH are through a subcostal incision (>90%)



- MIS : laparoscopic /thoracoscopic



Management :

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Outcome

- Depends on CDH Defect size, associated anomalies, concomitant major congenital heart disease, and prematurity(impose the greatest influence on patient survival in CDH).
- Survival inversely correlates with the prematurity . Those born <28weeks have 32% chance to survive compared to 73% for those born at 37 weeks
- Congenital heart defects : (survival rate 36% (major defect) to 67% (minor) and 73% (No defect)

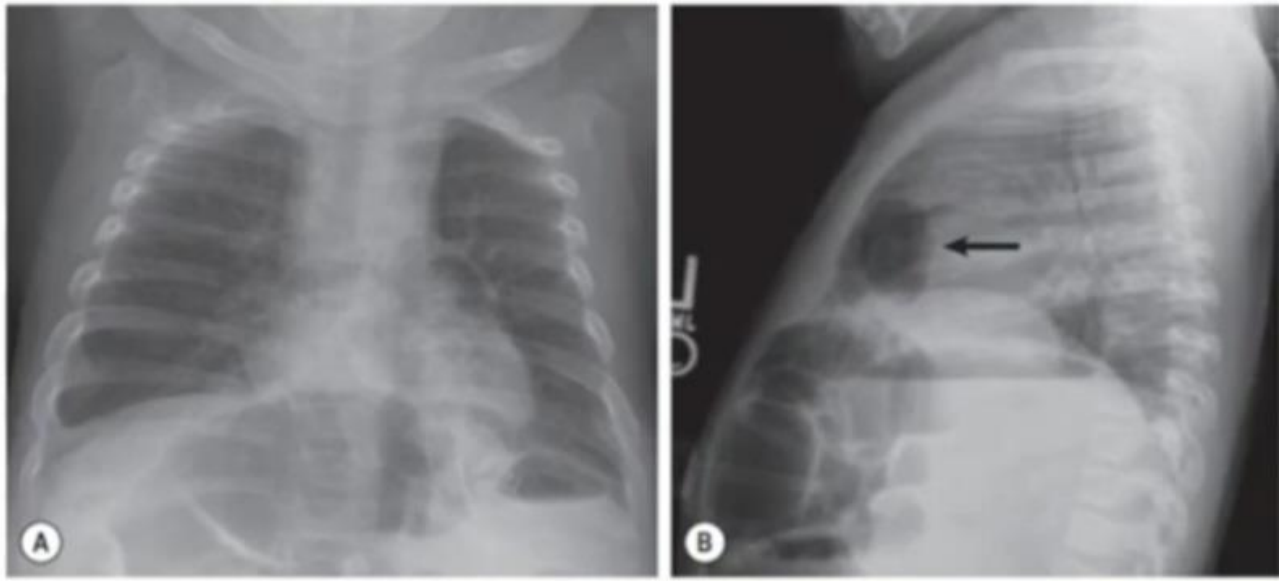
Follow up

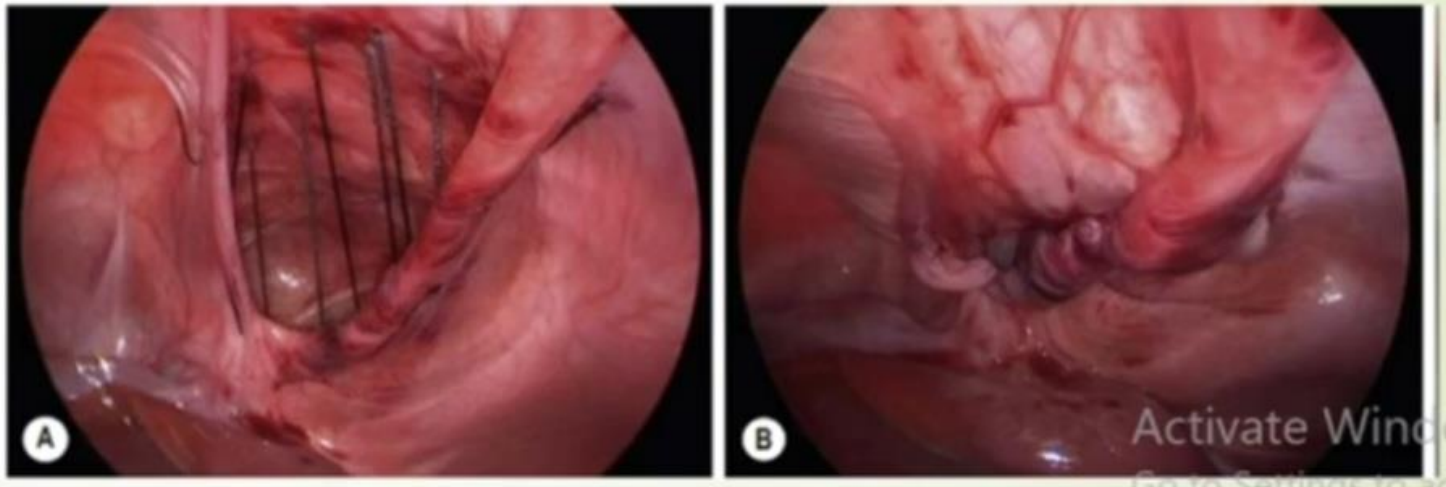
Table 24.1 Incidence of Morbidities in Long-Term Follow-Up of Patients With CDH

Morbidity	Estimated Frequency
Pulmonary	
Tracheostomy	4%
Decreased exercise tolerance	7–35%
Chronic recurrent pneumonia	7%
Neurodevelopmental	
Neuropathologic lesions	50%
Cognitive and motor dysfunction	20–73%
Emotional/behavioral problems	11–23%
Gastrointestinal	
Gastroesophageal reflux disease	50–100%
Long-term carcinoma risk	Unclear
Failure to thrive	50–60%
Musculoskeletal	
Pectus deformities	14–80%
Chest asymmetry	48%
Scoliosis	4–50%
Surgical complications	
Recurrence	6–24%
Small bowel obstruction	1–8% (early)
Impact on family Caregiver impact Financial/time burden	20–40%

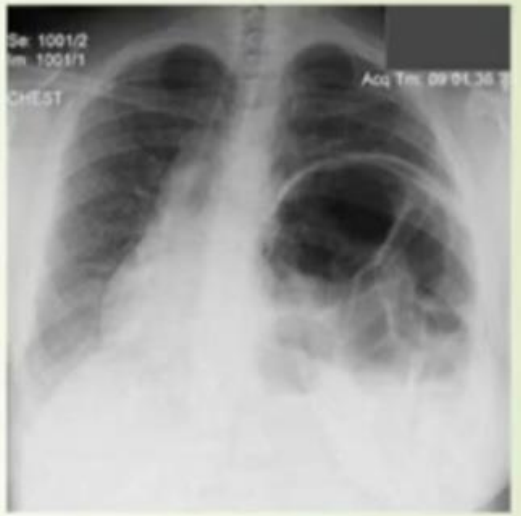
Morgagni

- Antero-medial defect
- <2% of all CDH
- Associated with trisomy 21 , congenital heart disease
- Usually asymptomatic
- Open or Laparoscopic repair





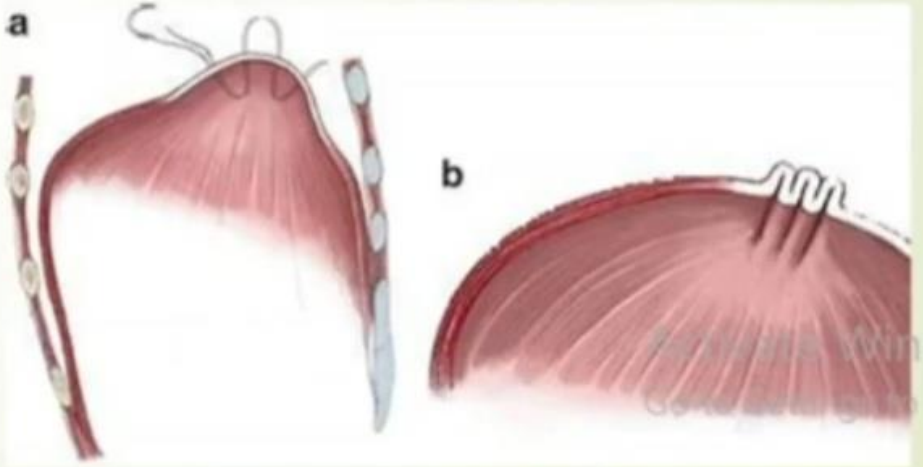
Eventration



- Abnormal elevation of diaphragm .
- My present acutely with resp distress in neonatal period, prolong intubation /difficult to extubate , or later in life with recurrent chest infection
- Diagnosis : elevated hemidiaphragm on Xray + Paradoxical movement in US
- Management :

mild and asymptomatic : conservative

severe : surgical repair (plication)



Esophageal Atresia /Tracheoesophageal Fistula (EA/TEF)

Embryology

- At 4th week of gestation the foregut starts to differentiate into
  - → Resp bud/ laryngotracheal diverticulum (ventrally) which will invaginate into the mesenchyme
  - → Esophageal bud (dorsally)
- Formation of lateral tracheoesophageal folds that fuse in the midline and create the tracheoesophageal septum.
- At 6–7 weeks of gestation, the separation between trachea and esophagus is completed.
- Incomplete fusion of the folds results in a defective tracheoesophageal septum and abnormal connection between the trachea and esophagus

Epidemiology

- 1 in 2500–3000 live births.
- There is a slight male preponderance of 1.26:1
- The risk for a second child with EA/TEF among parents of one affected child is 0.5–2%, increasing to 20% when more than one child is affected. The empirical risk of an affected child born to an affected person is 3–4%.

Association

- Environmental factors t: use of methimazole in early pregnancy
  - Prolonged use of contraceptive pills
  - Progesterone and estrogen exposure
  - Maternal diabetes
  - Thalidomide exposure
  - Fetal alcohol syndrome
  - Maternal phenylketonuria
- Chromosomal anomalies are found in 6–10% of the patients(trisomy 18,21)
- Genetic : MYCN haploinsufficiency in Feingold syndrome, CHD7 in CHARGE syndrome, and SOX2 in the anophthalmia–esophageal–genital (AEG) and other syndromes e.g Opitz G/BB syndrome, oculo-auriculo-vertebral syndrome, Bartsocas–Papas syndrome, Fryns syndrome

Associated anomalies

- Cardiac (13–34%)

  Vertebral (6–21%)

  Limb (5–19%)

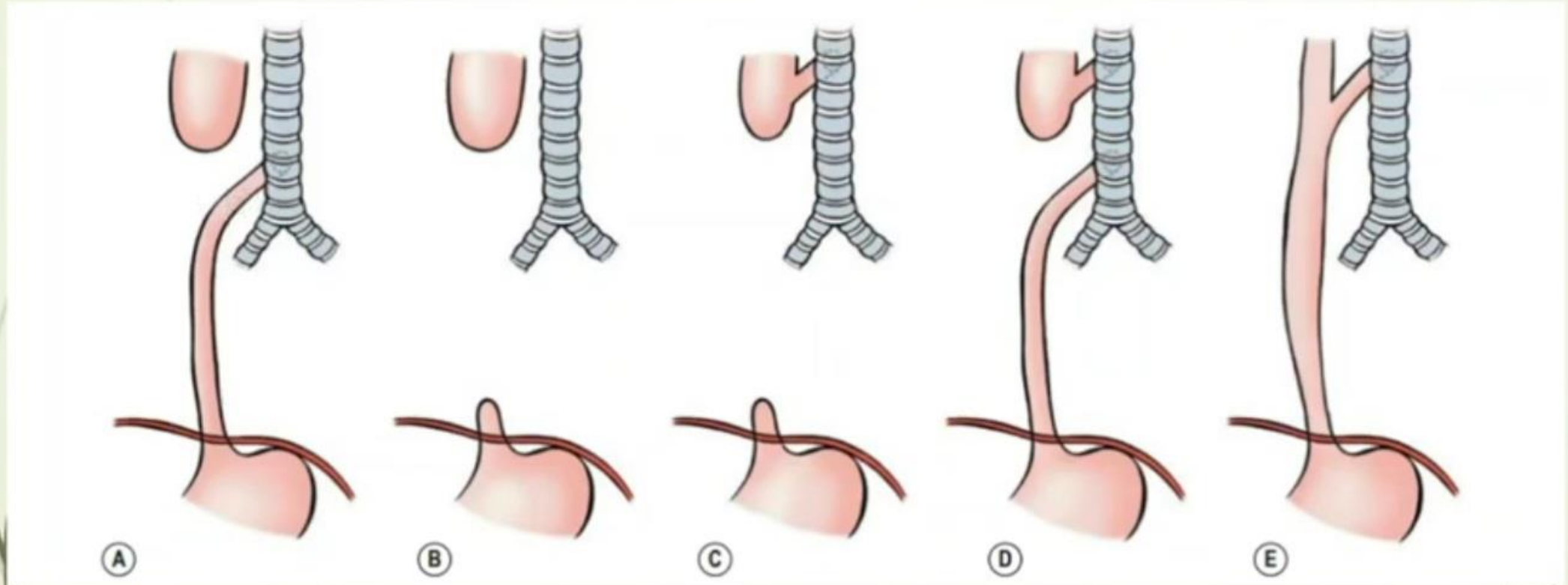
  Anorectal (10–16%)

  Renal (5–14%)

- Non-random associations have been documented : **VACTERL** association (Vertebral, Anorectal, Cardiac, Tracheo-Esophageal, Renal, and Limb abnormalities), and the **CHARGE** association (Coloboma, Heart defects, Atresia of the choanae, developmental Retardation, Genital hypoplasia, and Ear defect)

Types

- Gross Classification:



85% 7% 2% 1% 4%

Diagnosis

- Antenatally :

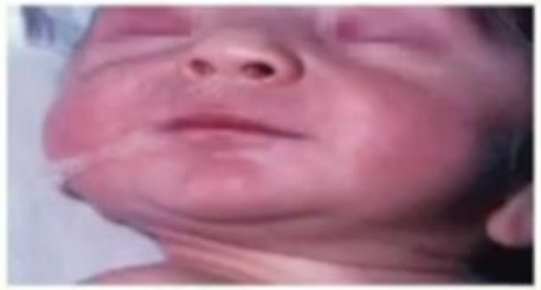
Fetal US : Polyhydraminos , small/absent stomach bubble , pouch sign , associated cardiac/renal anomalies

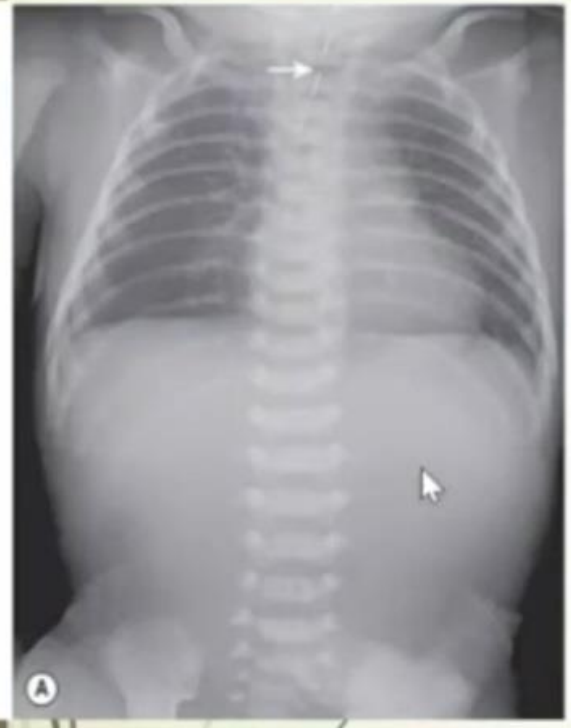
- Postnatally :

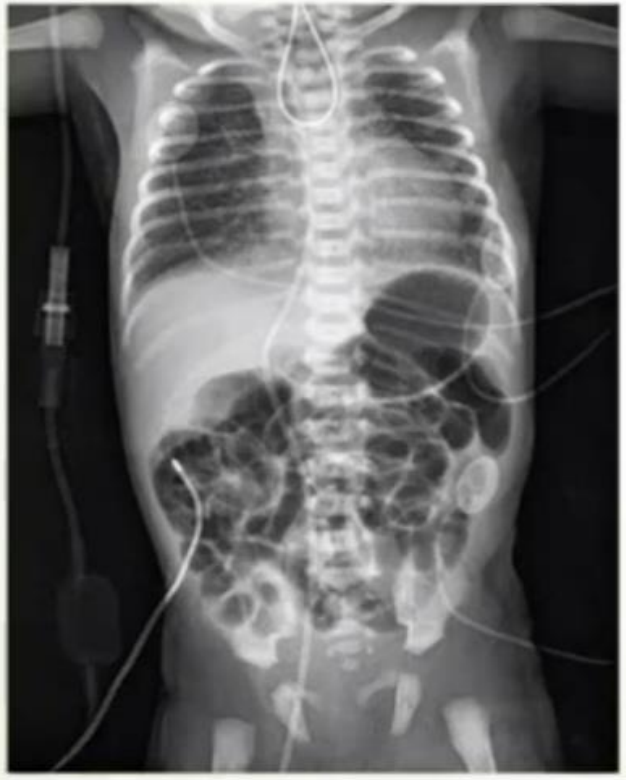
Drooling of saliva , respiratory distress

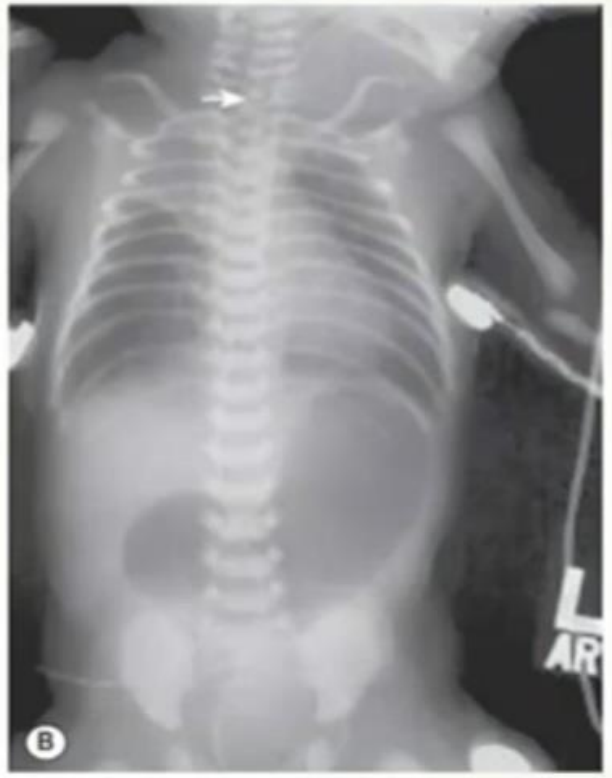
Confirm with insertion of NG/OG and CXR +/- upper contrast study

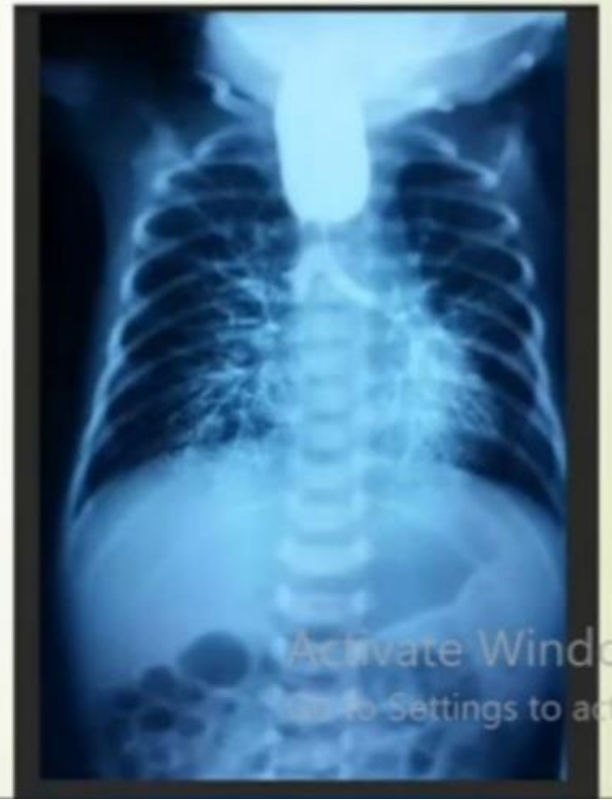
(coiling of tube at upper pouch /T2-T4 level)









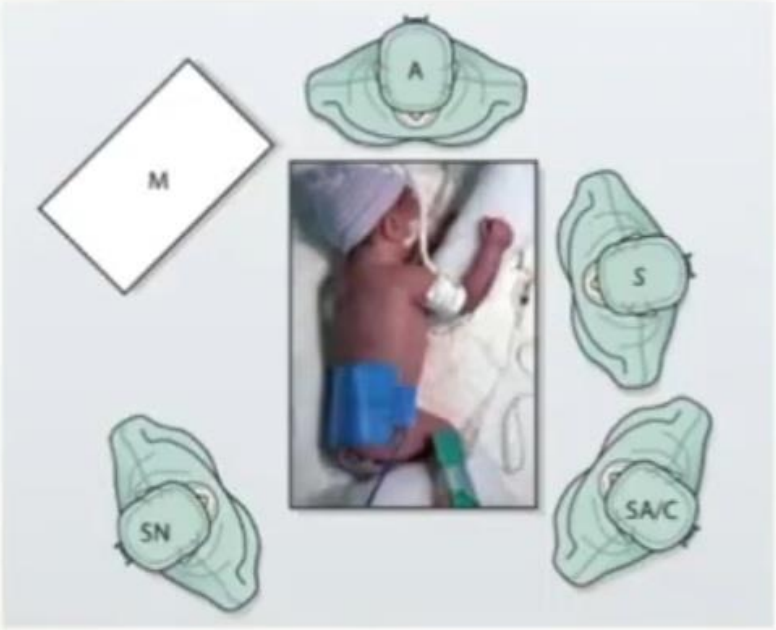


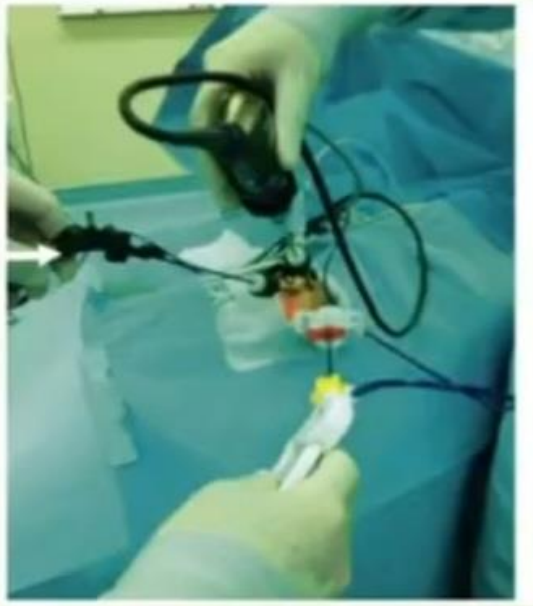
Management

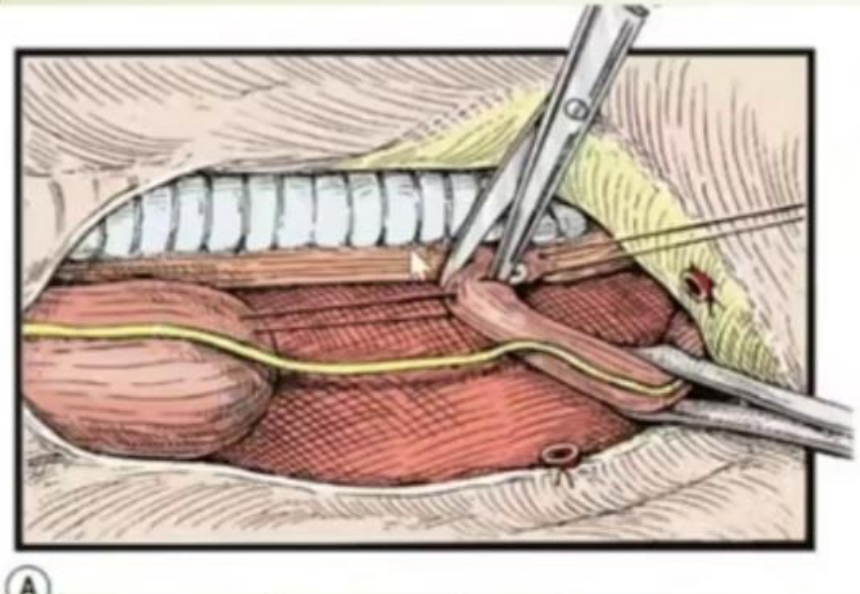
- **Its not an emergency procedure**
- Confirm the diagnose , resuscitate and rule out associated anomalies
- **Pre operative management :**
- NPO
- NG/OG on continuous suction
- IV access + IVF
- Upright position and on the side
- if in respiratory distress: endotracheal intubation (gentle ventilation)

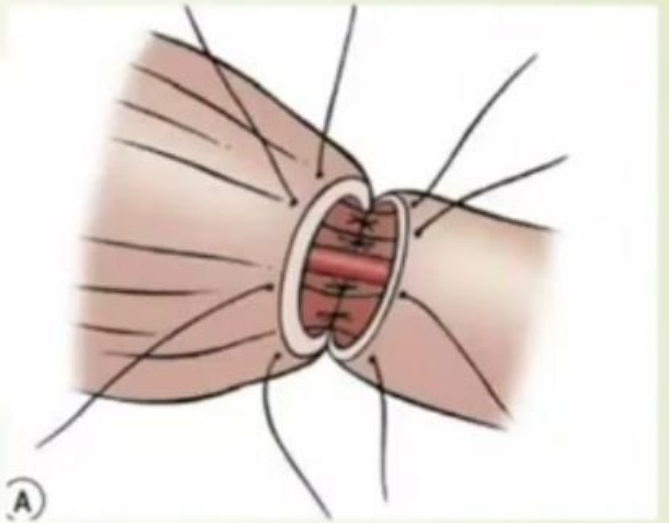
Surgery

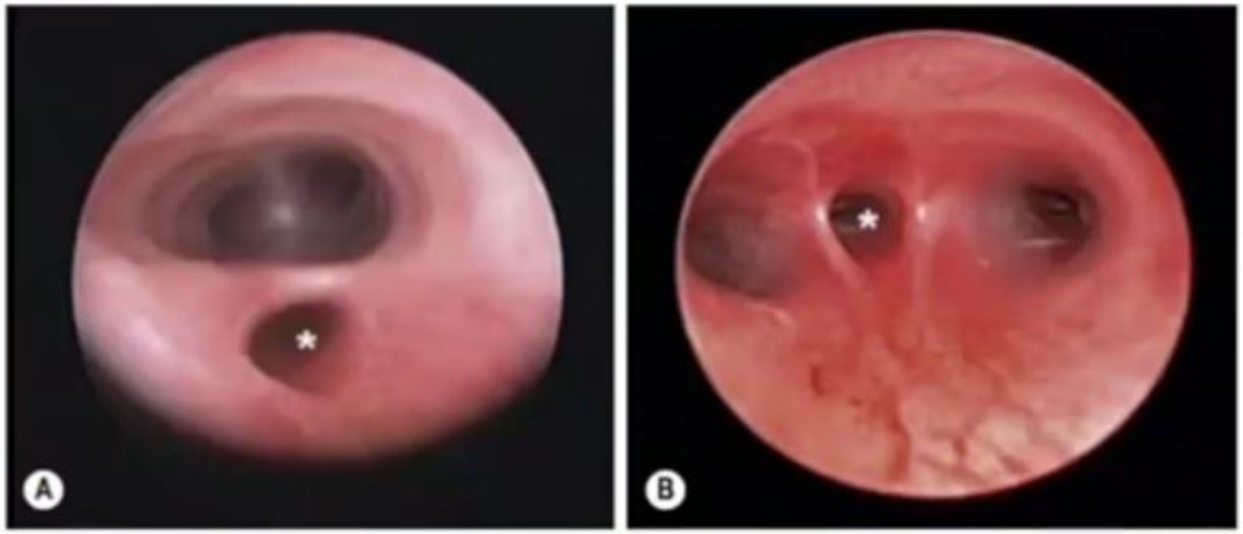
- Open (thoracotomy) Vs MIS (thoracoscopy) : fistula ligation and primary anastomosis +/- bronchoscopy

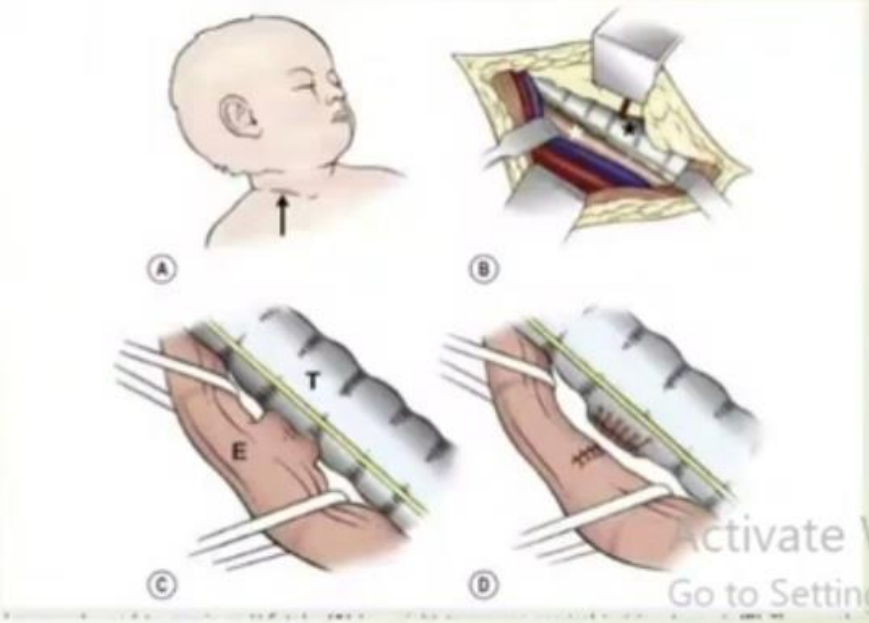












Complications

- Anastomotic Leaks (3.5-17%)
- Anastomotic Stricture (17-60%)
- Recurrent Tracheoesophageal Fistula (3-15%)
- Tracheomalacia
- Disordered esophageal peristalsis → GERD → Barret esophagus → Cancer
- Vocal Cord Dysfunction
- Respiratory Morbidity
- Thoracotomy-Related Morbidity