# Anticipating and Managing the Child and Family with Complex Congenital Anomalies

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# Anticipating Complex Anomalies

- Why are we talking about this?
  - » Congential anomalies are seldom first diagnosed i the labor room.
  - » Antenatal imaging techniques are increasingly accurate.
  - » Antenatal diagnosis is made increasingly early.
  - » The internet is a huge source of out-of-context mis information.
  - » We are sometimes a huge source of mis-informatic
  - » "Doctor" comes from the Latin verb, *to teach"*
  - » An educated consumer is a happy consumer.

# Anticipating Complex Anomalies "Center for Pre-Natal Pediatrics"

## Who is on the "team"?

- Obstetrician
- Perinatologist
- Radiologist
- Geneticist
- Prenatal counselor
- Neonatologist
- Pediatrician
- Many others
- Pediatric surgeon

# Anticipating Complex Anomalies

What are we going to talk about?

- Congenital diaphragmatic hernia
- Abdominal wall defects
- Congenital tumors
- Lung bud malformations
- Intestinal abnormalities

# Anticipating Complex Anomalies

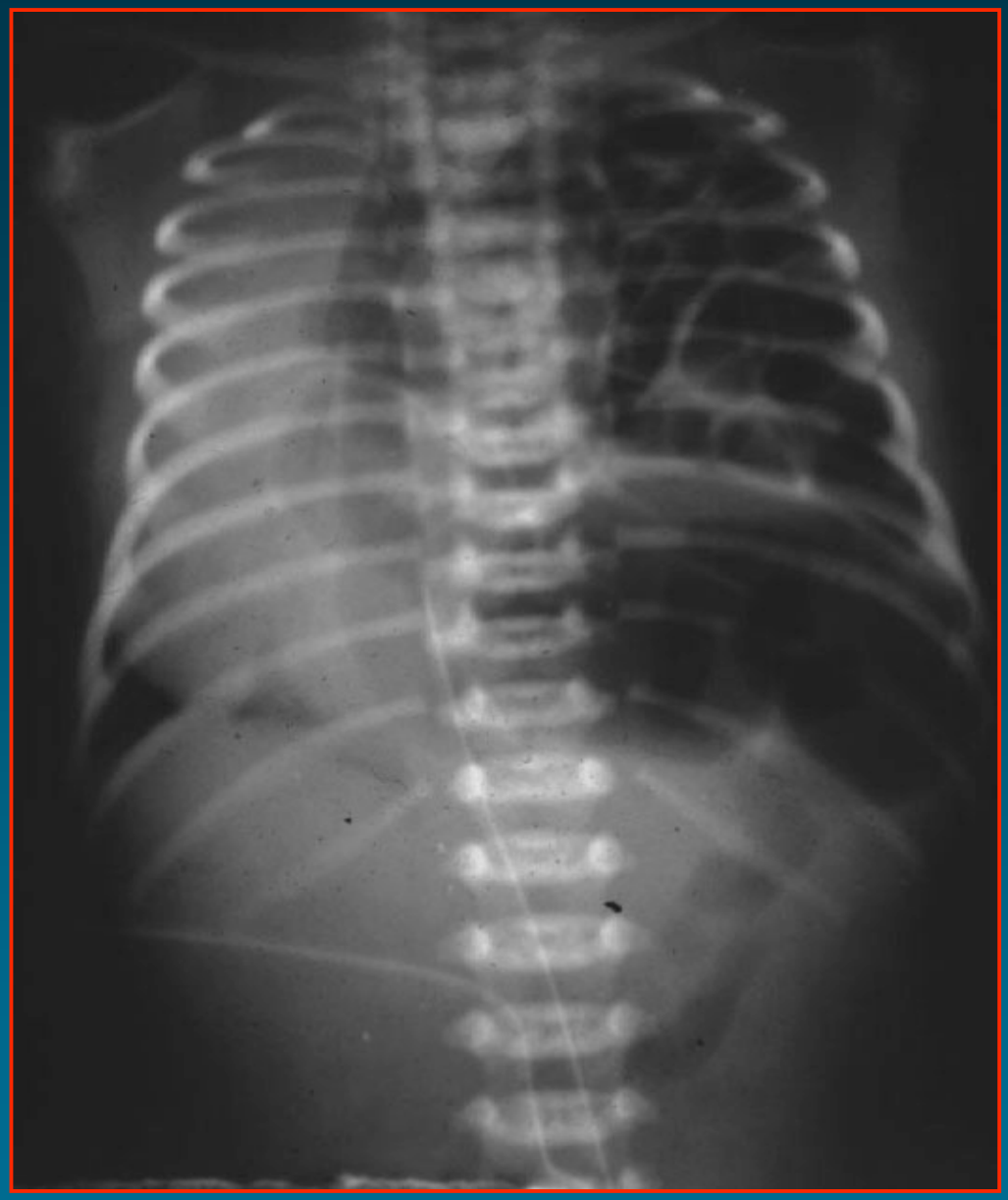
## What are we going to talk about?

- What is it/are they?
- How is the diagnosis made?
- What should parents know?
- What can be done about it?
- What are the outcomes?

# Anticipating Complex Anomalies
## *Congenital Diaphragmatic Hernia*

### What is it?

- Disturbed lung development early in gestation resulting from abnormal development of the diaphragm and causing a complex mix of pulmonary hypoplasia and pulmonary hypertensio



Congenital Diaphragmatic Hernia

# Anticipating Complex Anomalies
## *Congenital Diaphragmatic Hernia*

How is the diagnosis made?

- Polyhydramnios
- Fetal ultrasound
- Magnetic resonance imaging




Congenital Diaphragmatic Hernia - Ultrasound

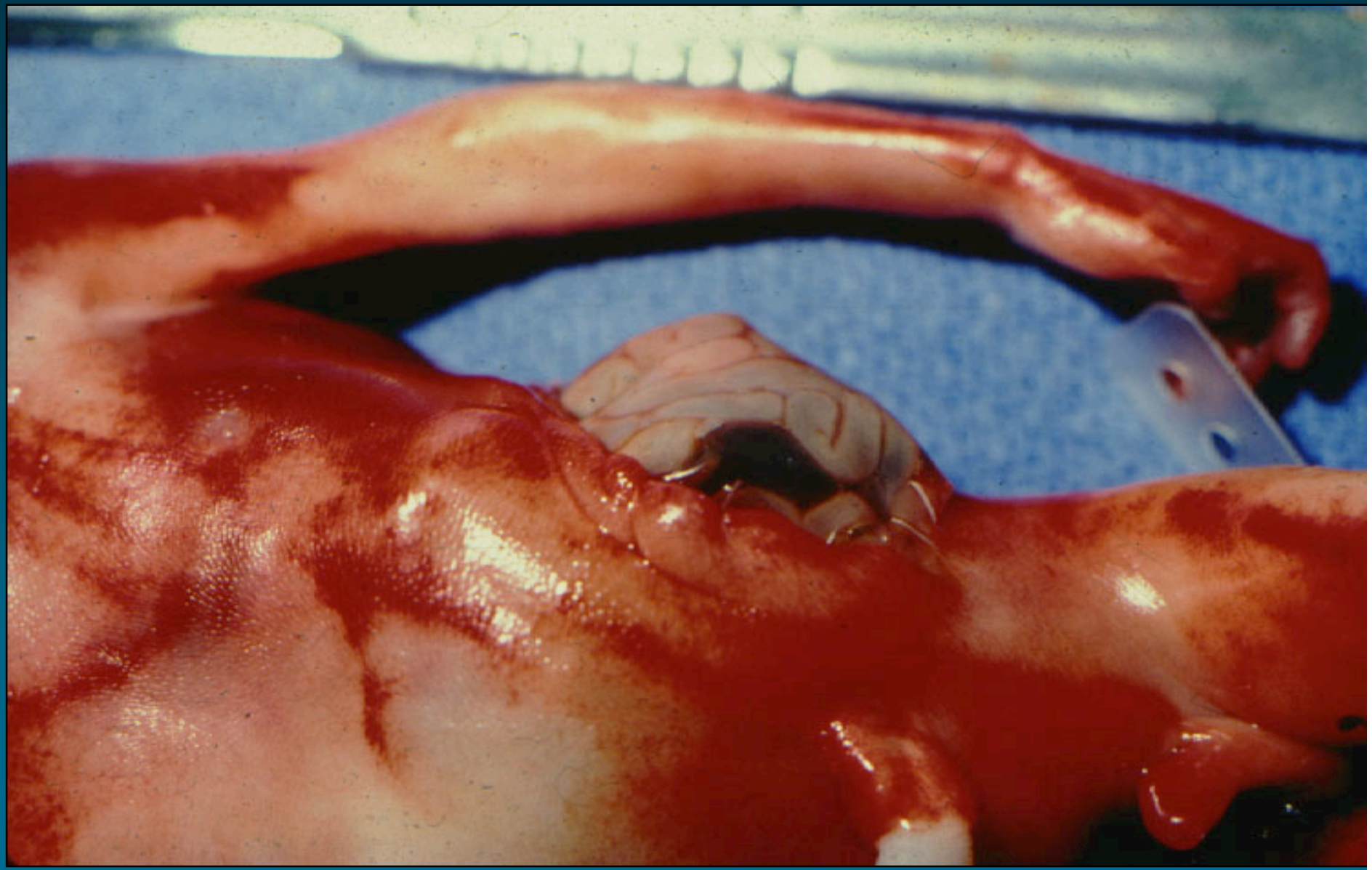


# Congenital Diaphragmatic Hernia - MRI

# Anticipating Complex Anomalies
## *Congenital Diaphragmatic Hernia*

What are we to do about it?

- Fetal intervention is contraindicated
- Elective vaginal delivery at term
- Pre-operative resuscitation until pulmonary hypertension resolves
- Permissive hypercapnea/spontaneous respiration
- Elective surgery
- Extracorporeal life support only if enough lung



Congenital Diaphragmatic Hernia - Fetal Repair

# Anticipating Complex Anomalies
## *Congenital Diaphragmatic Hernia*

What should we tell parents?

- Embryology as we understand it
- Pathophysiology as we understand it
- Possible treatment algorithms
- Possible outcomes
- Misinformation correction
- Cautious optimism, *not* gloom and doom

# Anticipating Complex Anomalies
## *Congenital Diaphragmatic Hernia*

What are the potential long term concerns?

- Cardiac
- Pulmonary
- Foregut
- Chest wallaxial skeleton
- Neuromotor

# Anticipating Complex Anomalies
## *Abdominal Wall Defects*

## What are they?

- Gastroschisis
  - » Rupture of right vitelline vein with herniation of uncovered viscera right of umbilicus
  - » Usually isolated anomaly, occasional atresia, apple peel deformity
  - » Serositis from amniotic fluid exposure; foreshortened gut

- Omphalocoel
  - » Herniation of viscera into umbilicus, covered by umbilical membra
  - » Often associated with other anomalies(cardiac, pulmonary, trisomy

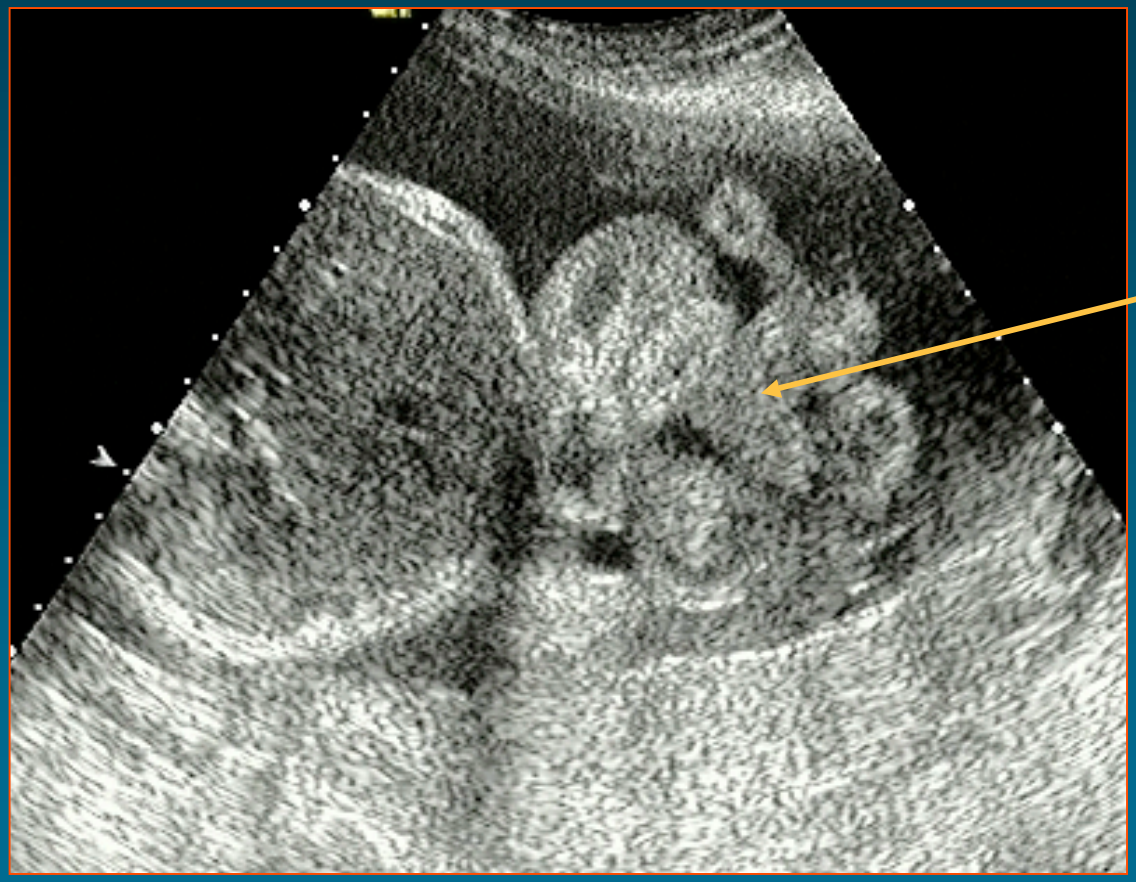
Both defects feature non-fixed midgut and result in loss of abdominal do

# Anticipating Complex Anomalies
## *Abdominal Wall Defects*

How is the diagnosis made?

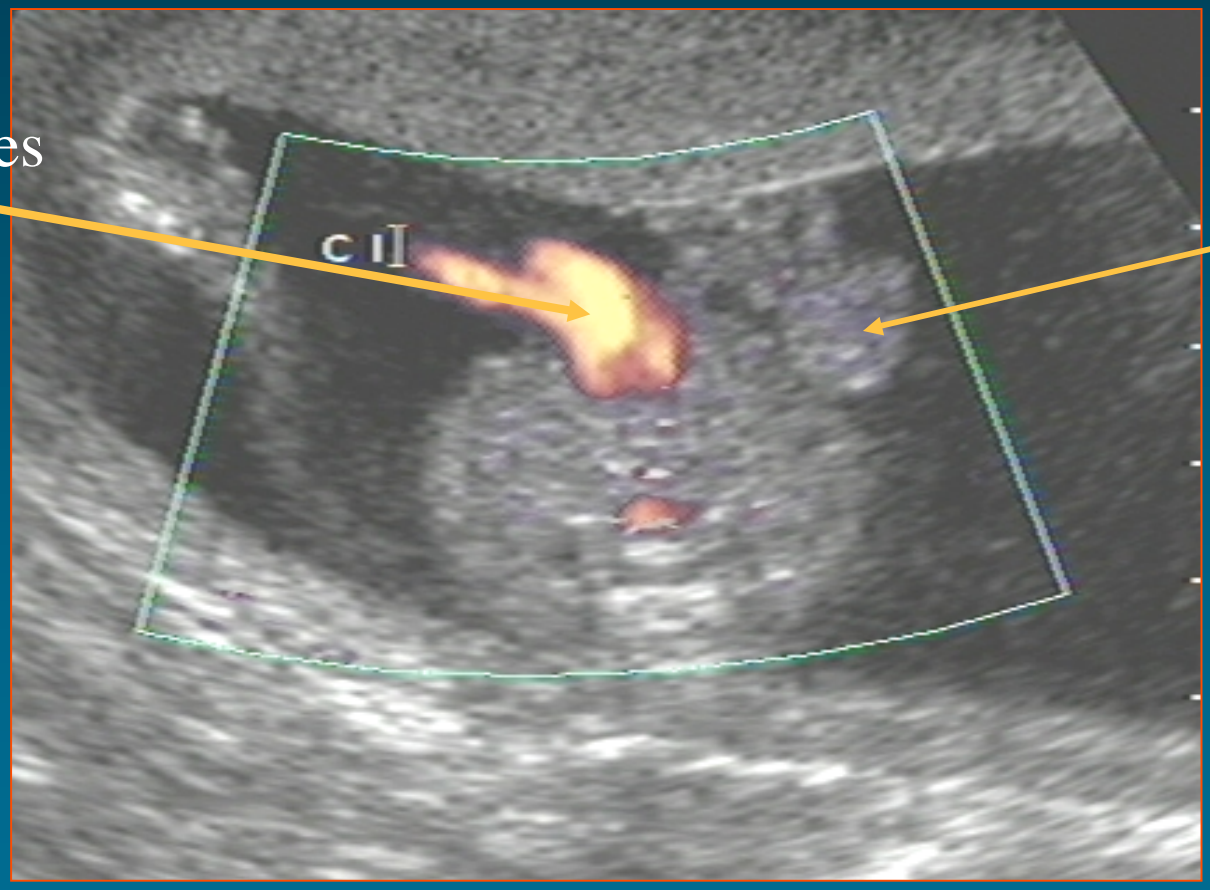
- Ultrasound
- Chromosome analysis

# Anticipating Complex Anomalies
## Gastroschisis



# Anticipating Complex Anomalies
## Gastroschisis

umbilical arteries



gastrosc

# Anticipating Complex Anomalies
## *Abdominal Wall Defects*

What do we do about it?

- Early delivery for gastroschisis if gut is very thickened(maybe), term for omphalocoel
- Caesarian section *may* be indicated
- Primary reduction and closure if possible
- Staged closure if large
- Respiratory and nutritional support
- No belly button

# Anticipating Complex Anomalies
## *Abdominal Wall Defects*

What should parents know?

- Most (not all) of these defects can be fixed
- Sometimes there is not enough midgut to support life
- Sometimes associated problems are limiting (pulmonary / cardiac)

# Anticipating Complex Anomalies
## *Abdominal Wall Defects*

What are the long term problems?

- Gastrointestinałnutritional
- Intestinal obstruction
- Cardiac/pulmonary
- Psychiatric – no belly button!

# Anticipating Complex Anomalies
## *Congenital Tumors*

What are they?

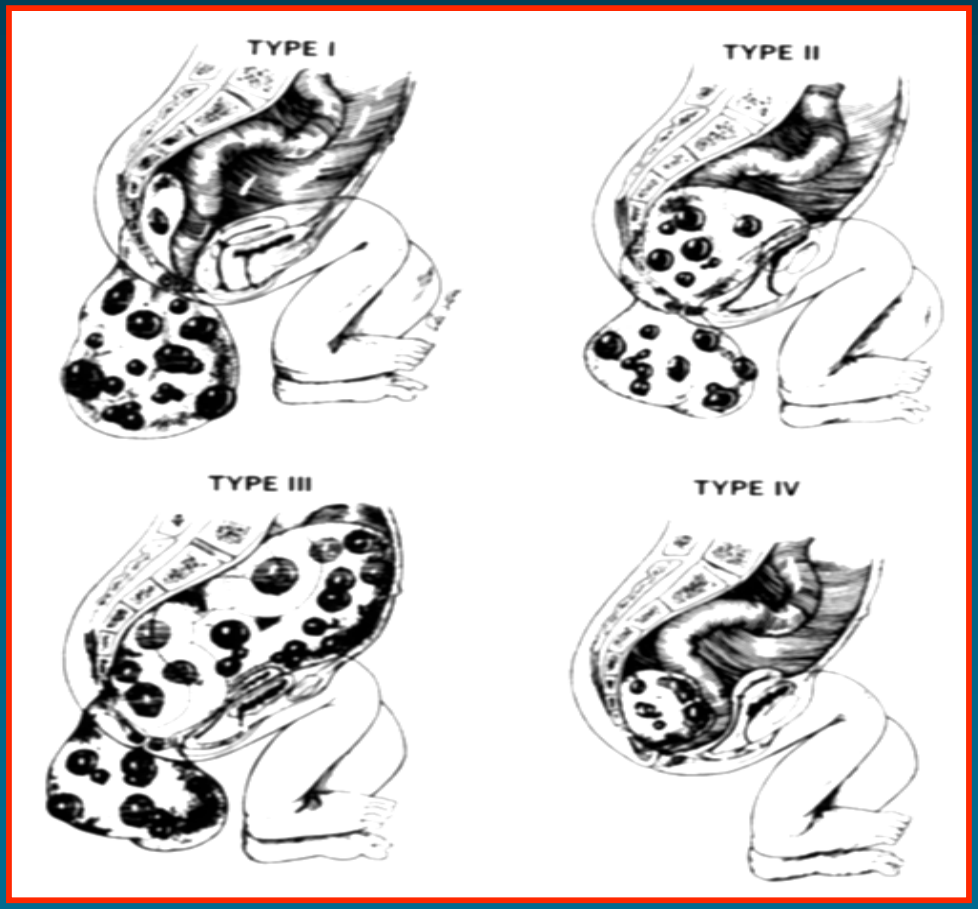
- Teratomas – sacrocoxcygeal, etc.
- Mediastinal
- Neuroblastoma
- Cystic hygroma

# Anticipating Complex Anomalies
## *Congenital Tumors*

How is the diagnosis made?

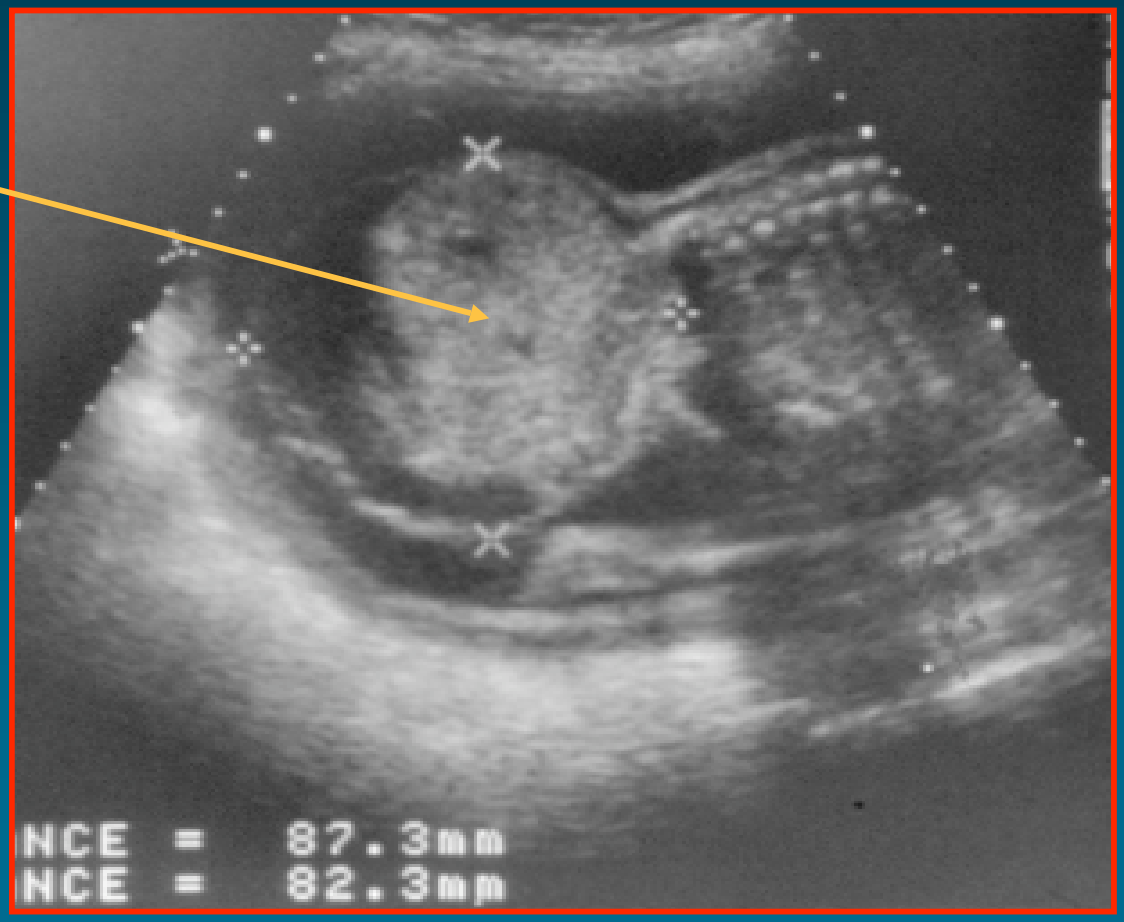
- CAT scan (baby only)
- MRI
- Ultrasound
- Serum markers

# Anticipating Complex Anomalies
## sacrocoxygeal teratoma



# Anticipating Complex Anomalies
# sacrocoxygeal teratoma

teratoma



# Anticipating Complex Anomalies
## Sacrocoxygeal Teratoma

teratoma



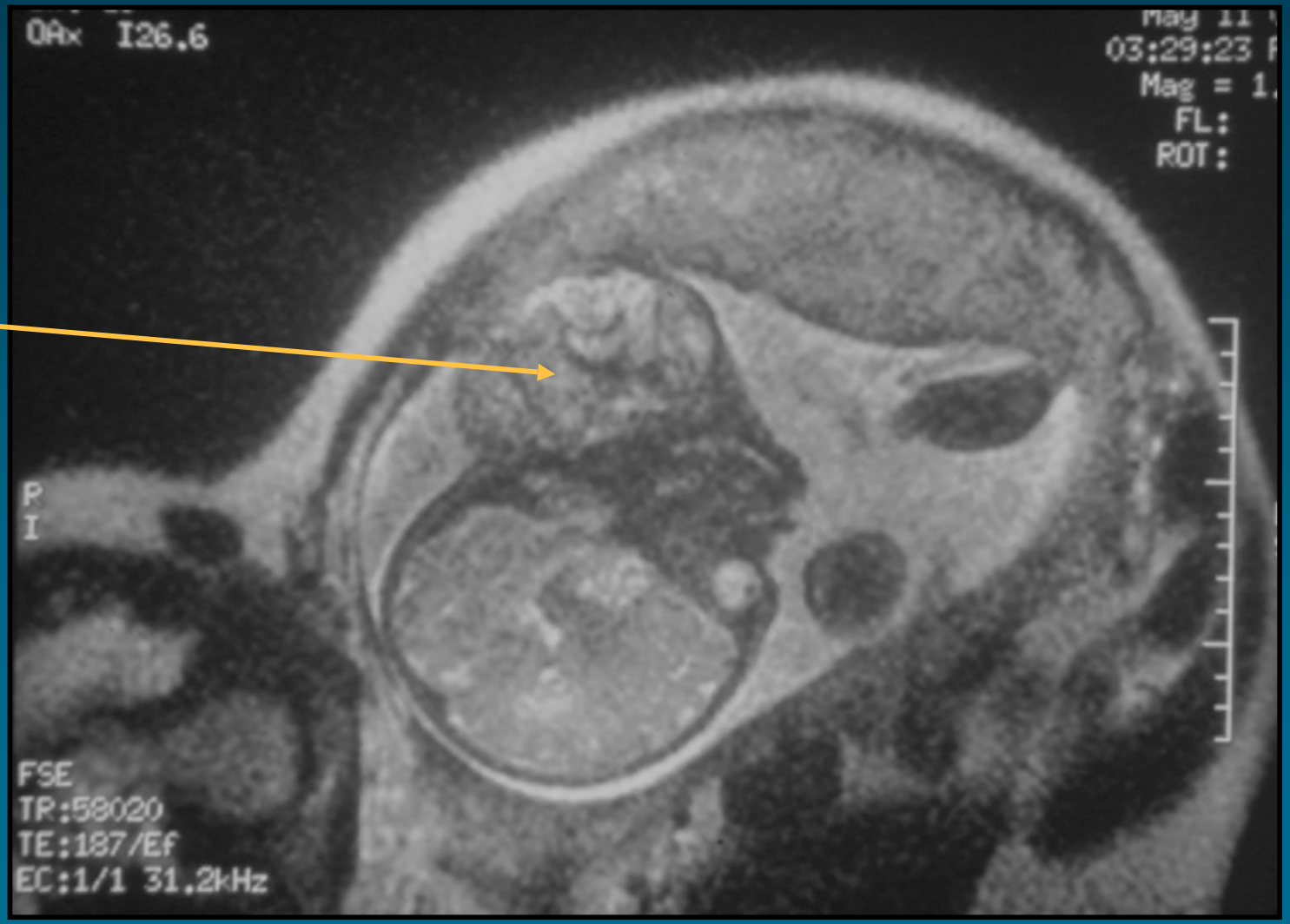
# Anticipating Complex Anomalies
## *Congenital Tumors*

What do we do about it?

- Diagnosis specific treatment at birth
- ***E***t***X***ra-uterine ***I***n***T***rapartum (EXIT)
- Anecdotal fetal intervention

# Anticipating Complex Anomalies
## *congenital tumors*

cervical teraroma

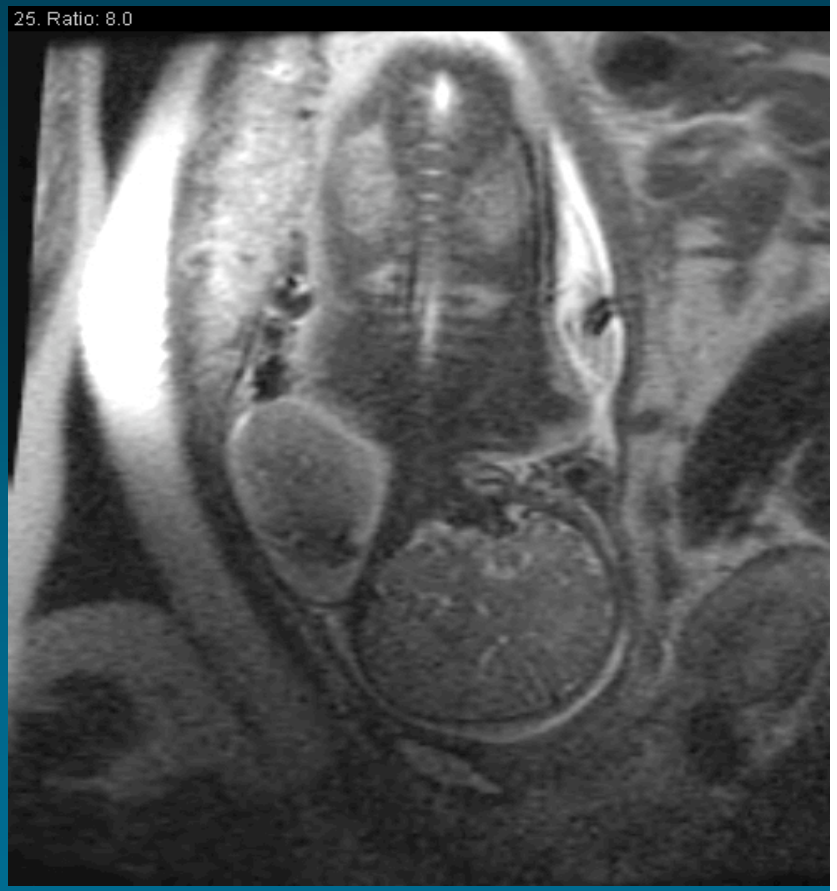


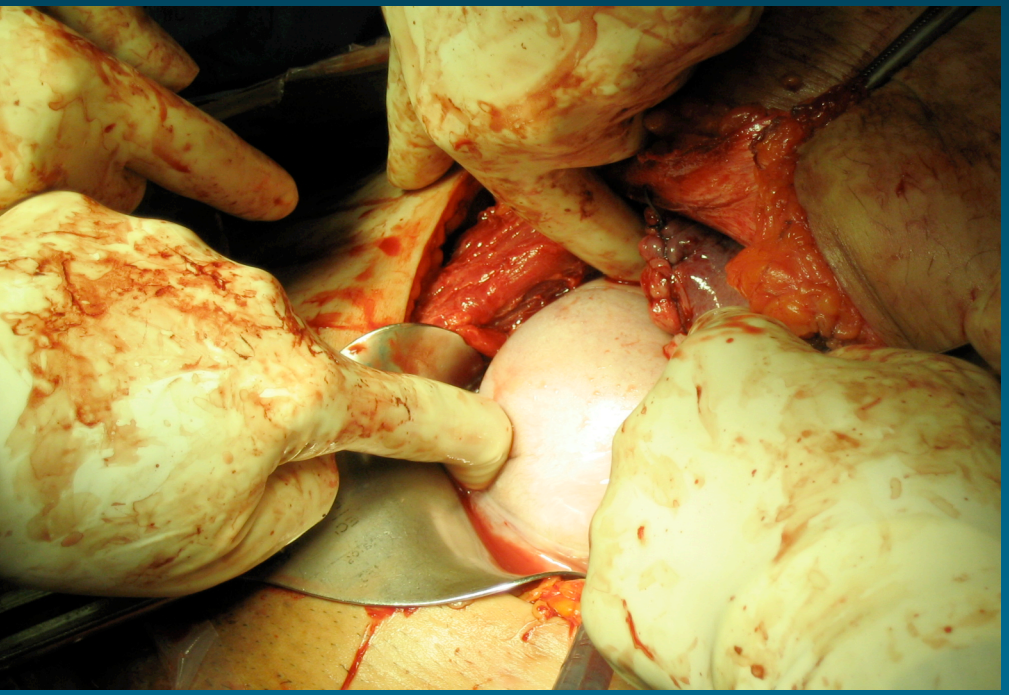
# Anticipating Complex Anomalies
## *congenital tumors*

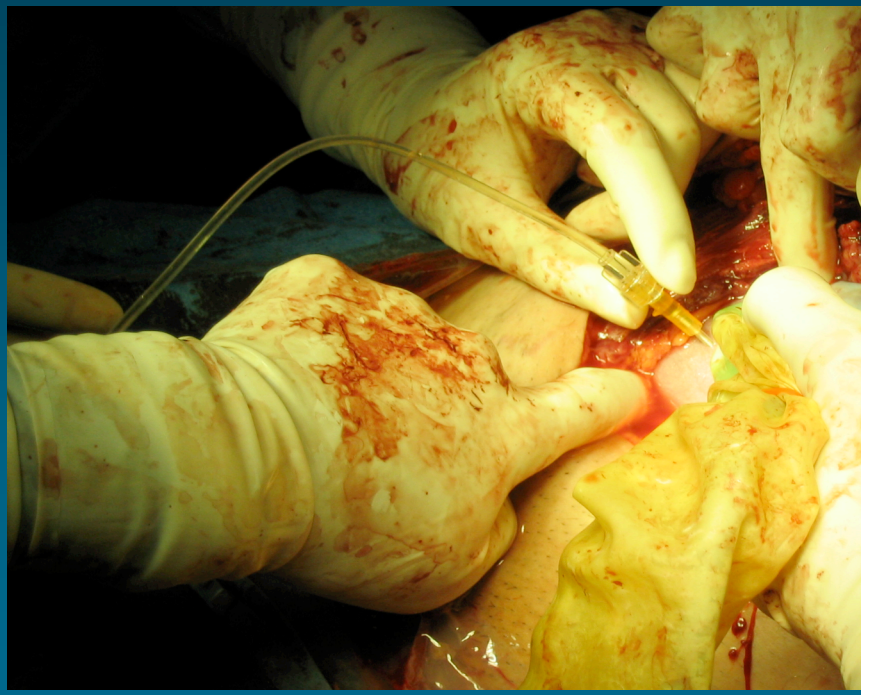


Cervical Teratoma


NUNEZ, PEGGI SG
NY PRESBYTERIAN HOSP
4632267
C5-2 OB/Gen
09 Feb 04
4:42:39 pm
TIb0.5 MI 1.4
Fr #142 20.0cm
HEAD
7.45cm
8.04cm


25. Ratio: 8.0









# Anticipating Complex Anomalies
## *Congenital Tumors*

### What should parents know?

- Early diagnosis is an opportunity for education and treatment at birth
- Most congenital tumors, while frightening, are curable
- Cystic hygroma may be resolve before birth or b associated with genetic syndrome

# Anticipating Complex Anomalies
## *Congenital Tumors*

What are the long term problems?

- Chemotherapy morbidity
- Operative morbidity
- Follow-up

# Anticipating Complex Anomalies
## *Broncho-Pulmonary(Lung Bud) Malformations*

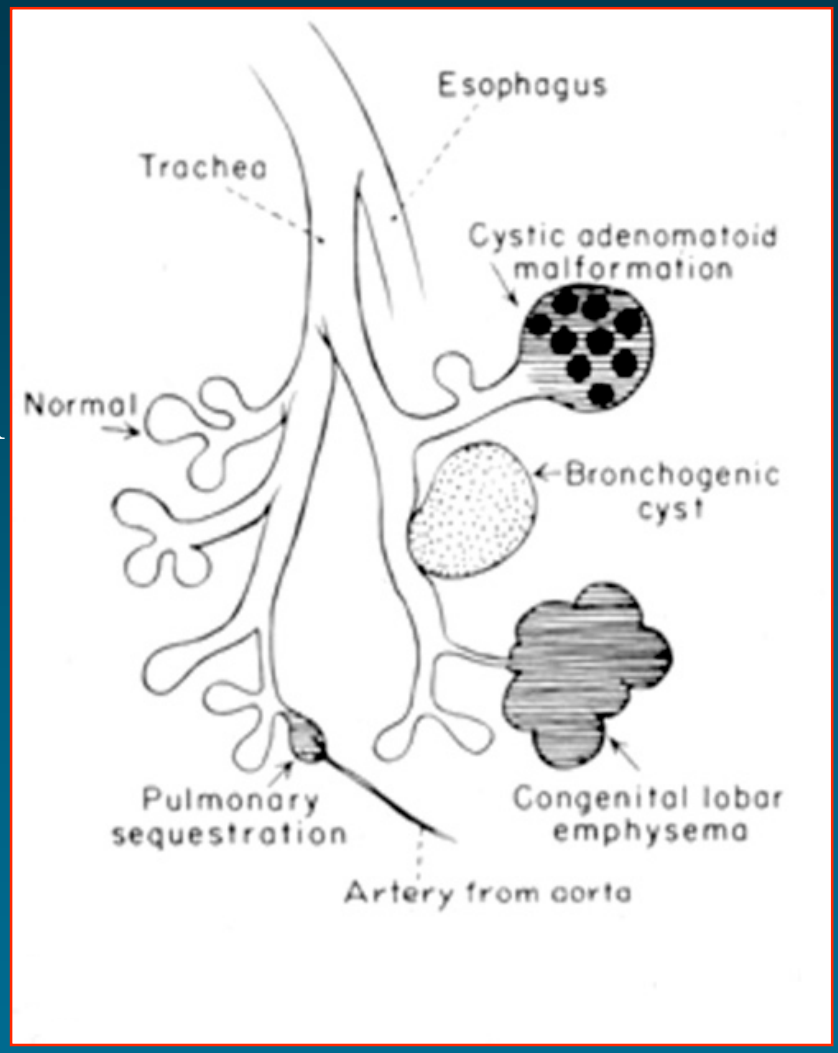
### What are they?

This is a collection of structural anomalies that arise from disordered embryogenesis of the foregut/lung bud/circulation and feature disorder "cross-talk" between foregut endoderm and mesoderm.

- Congenital cystic adeomatoid malformation(CCAM)
- Bronchogenic cyst
- Pulmonary sequestration
- Congenital lobar emphysema

# Broncho-Pulmonary Foregut Malformations

- cystic adenomatoid malformation
- bronchogenic cyst
- lobar emphysema
- pulmonary sequestration

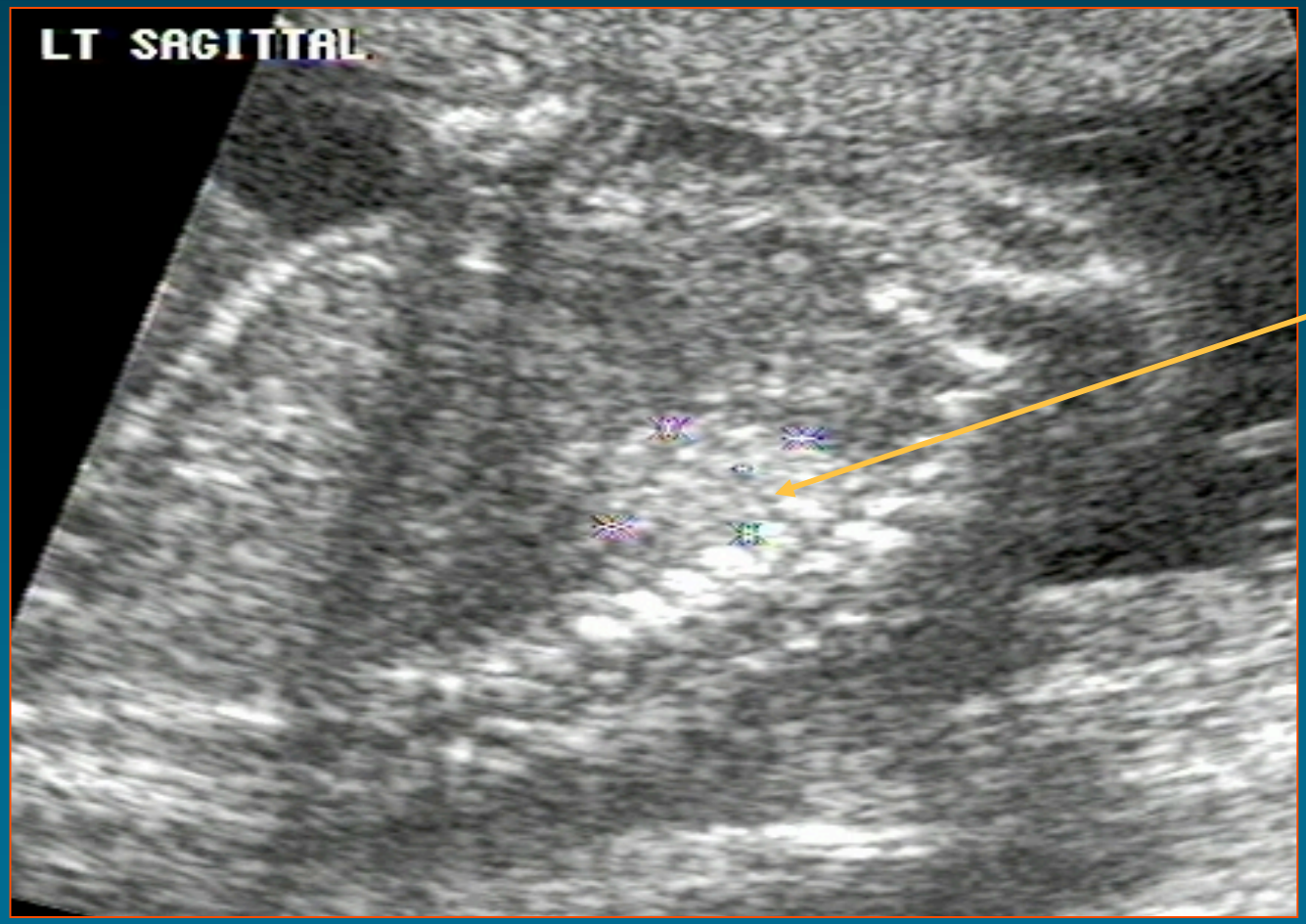


# Anticipating Complex Anomalies
## *Broncho-Pulmonary(Lung Bud) Malformations*

How is the diagnosis made?

- Ultrasound
- MRI

# Anticipating Complex Anomalies
## pulmonary sequestration



# Anticipating Complex Anomalies
## *Broncho-Pulmonary(Lung-Bud) Malformations*

What do we do about it?

- Follow throughout pregnancy
- Confirm diagnosis at birth
- Not all need operation
- Surgery is rarely an emergency
- Anatomic resection *vs* enucleation
- Fetal intervention with progressive hydrops

# Anticipating Complex Anomalies
## *Broncho-Pulmonary(Lung Bud) Anomalies*

What should parents know?

- Arise from errors in embryogenesis very early
- Usually isolated
- Almost all survive
- May or may not need surgery
- Surgery is indicated for lung growth, cardiac compromise, infection
- Malignancy risk is real but extremely rare…and not limited to the malformation

# Anticipating Complex Anomalies
## *Broncho-Pulmonary(Lung Bud) Malformations*

What are the long term problems?

- Follow-up for infectious/cardiac/malignancy risk treated non-operatively
- Pulmonary morbidity is related to acute surgical intervention or compromised lung growth

# Anticipating Complex Anomalies
# Intestinal Abnormalities

*What are the ultrasound observations?*

- *Dilated bowel*
- *Echogenic bowel*
- *Intra-abdominal calcifications*

# Anticipating Complex Anomalies
## Duodenal Atresia



# Anticipating Complex Anomalies
# Intestinal Abnormalities

*What do the ultrasound findings mean?*

- *Often, nothing, …but sometimes they do*
- *Atresias, duodenal and other*
- *Hirschsprung's disease*
- *Cystic fibrosis*
- *Ano-rectal malformations*

# Anticipating Complex Anomalies
# Intestinal Abnormalities

*What do we do about it?*

- *Chromosome analysis on parents and fetus*
- *monitor renal images*
- *detailed evaluation at birth*
  - *physical exam*
  - *radiology*
- *Surgery depends on diagnosis*

# Anticipating Complex Anomalies
# Intestinal Abnormalities

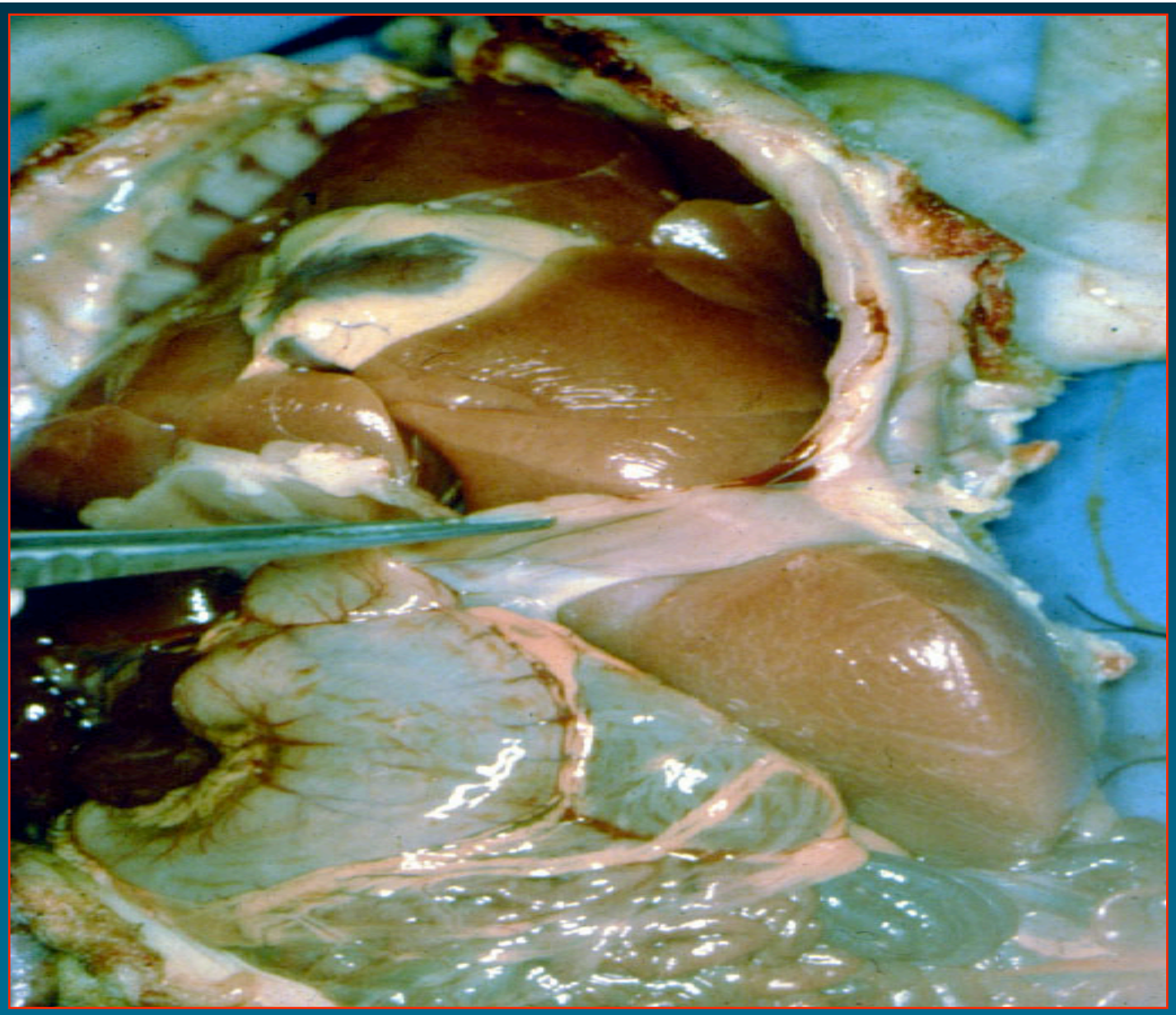
*What should parents know?*

- *Almost all of these diagnoses can be fixed.*
- *The repair depends on the diagnosis.*
- *The exact diagnosis can't be made until birth.*
- *Some of the infants may have life-long nutritional concerns.*

# Anticipating Complex Anomalies
## *Summary*

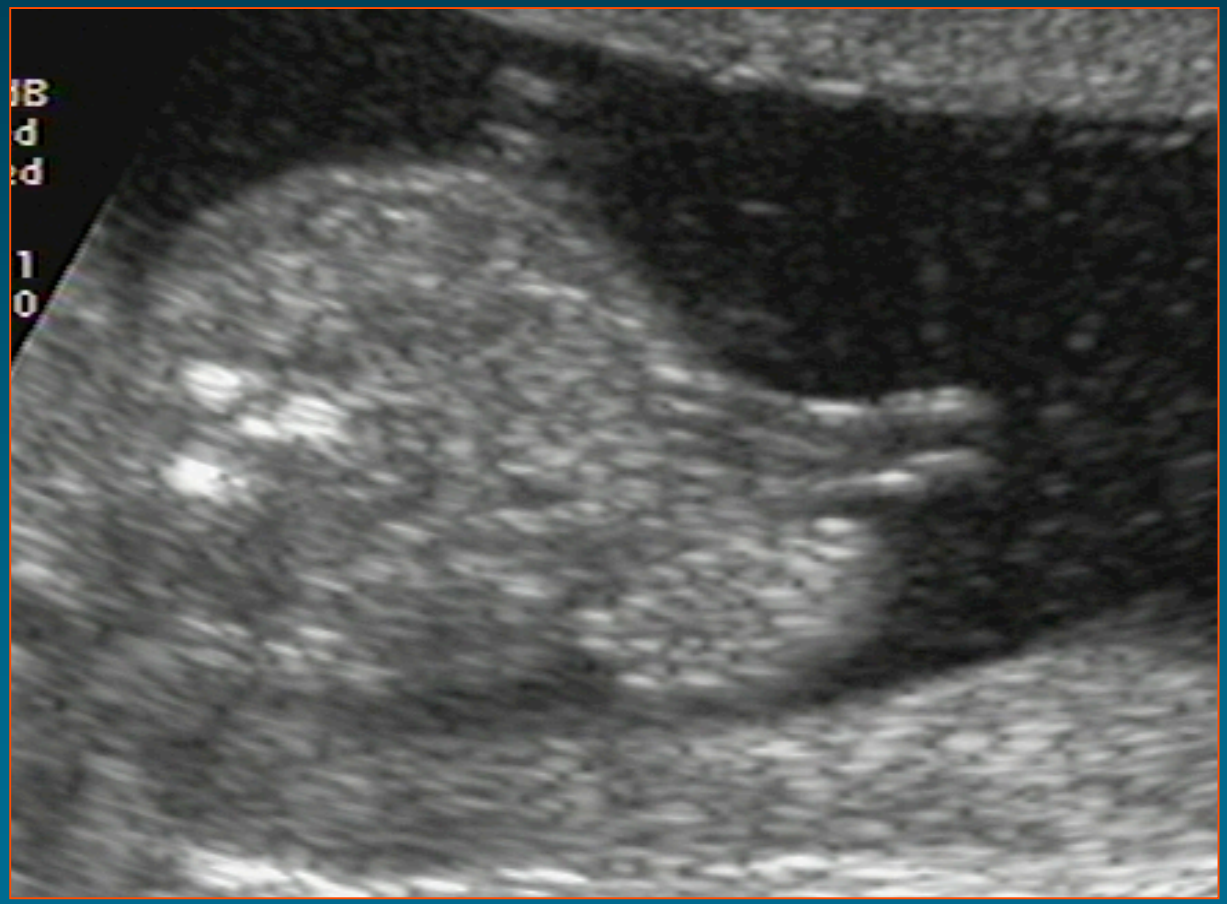
The important features of antenatal diagnosis are:

- Comprehensive multi-disciplinary consultation
- Parental education regarding what the problem is where it came from, what can be done, how the story will end
- Correction of misinformation
- Plan for rare fetal intervention
- Coordination of multidisciplinary care for parents and child
- Plan elective delivery and care for child in a full service facility…as needed



Prenatal Tracheal Ligation

# Anticipating Complex Anomalies
## Gastroschisis



# Anticipating Complex Anomalies
## lung bud / broncho-pulmonary malformation

