

Anticipating and Managing the Child and Family with Complex Congenital Anomalies



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Anticipating Complex Anomalies

- Why are we talking about this?
 - » Congenital anomalies are seldom first diagnosed in 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 misinformation.
 - » We are sometimes a huge source of mis-information.
 - » “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 hypertension



Congenital Diaphragmatic Hernia

Anticipating Complex Anomalies

Congenital Diaphragmatic Hernia

How is the diagnosis made?

- Polyhydramnios
- Fetal ultrasound
- Magnetic resonance imaging



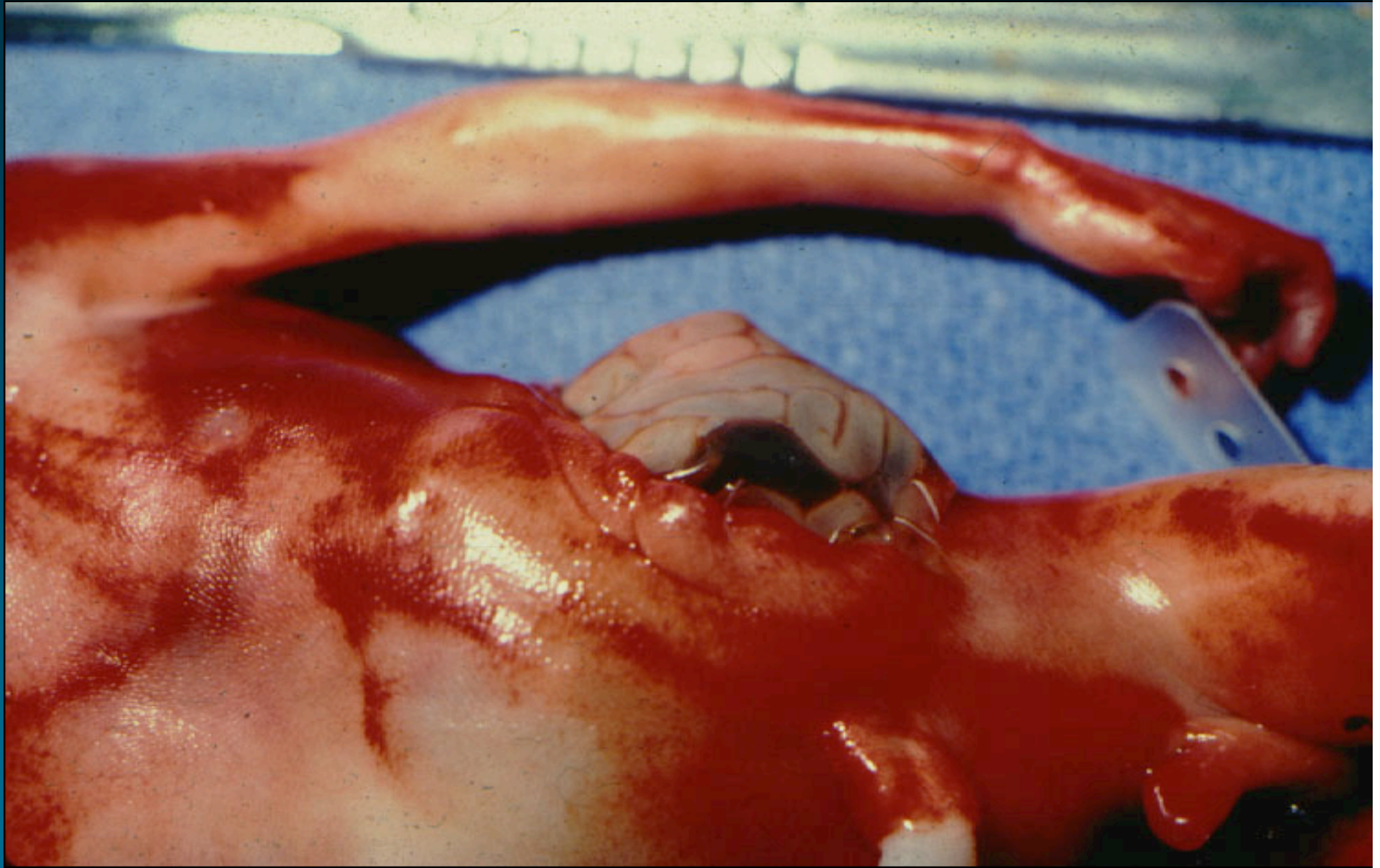
Congenital Diaphragmatic Hernia - Ultrasound

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 wall/axial 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
- **Omphalocele**
 - » Herniation of viscera into umbilicus, covered by umbilical membrane
 - » Often associated with other anomalies (cardiac, pulmonary, trisomy 13)

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

Anticipating Complex Anomalies

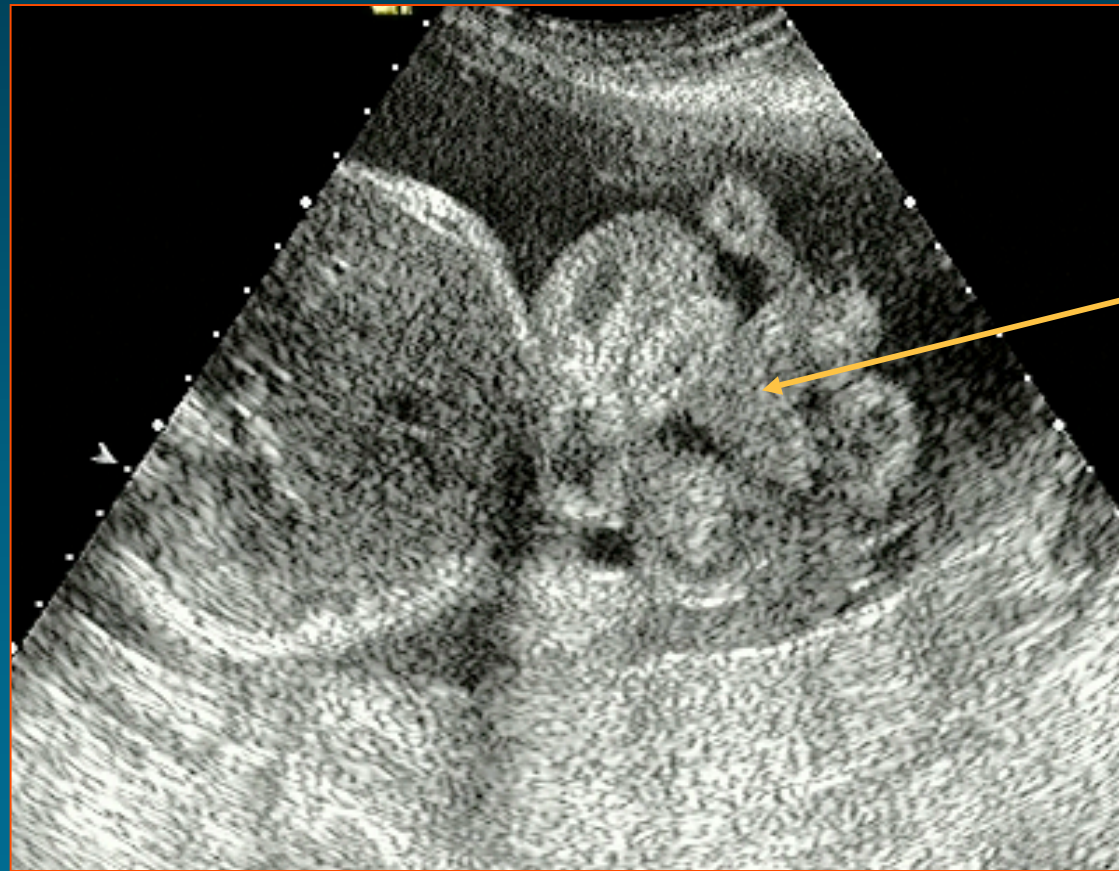
Abdominal Wall Defects

How is the diagnosis made?

- Ultrasound
- Chromosome analysis

Anticipating Complex Anomalies

Gastroschisis



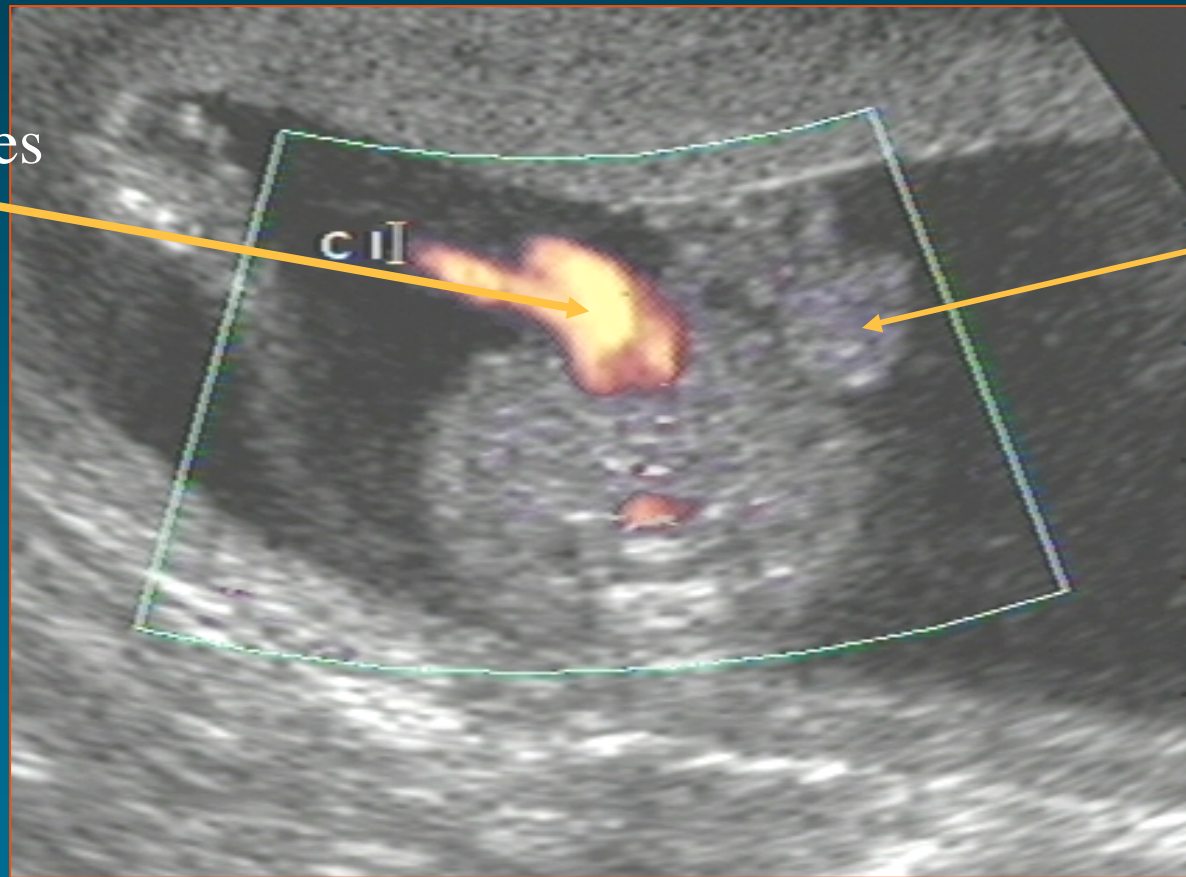
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Anticipating Complex Anomalies

Gastroschisis

umbilical arteries



gastroschisis

Anticipating Complex Anomalies

Abdominal Wall Defects

What do we do about it?

- Early delivery for gastroschisis if gut is very thickened(maybe), term for omphaloceol
- 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?

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

Anticipating Complex Anomalies

Congenital Tumors

What are they?

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

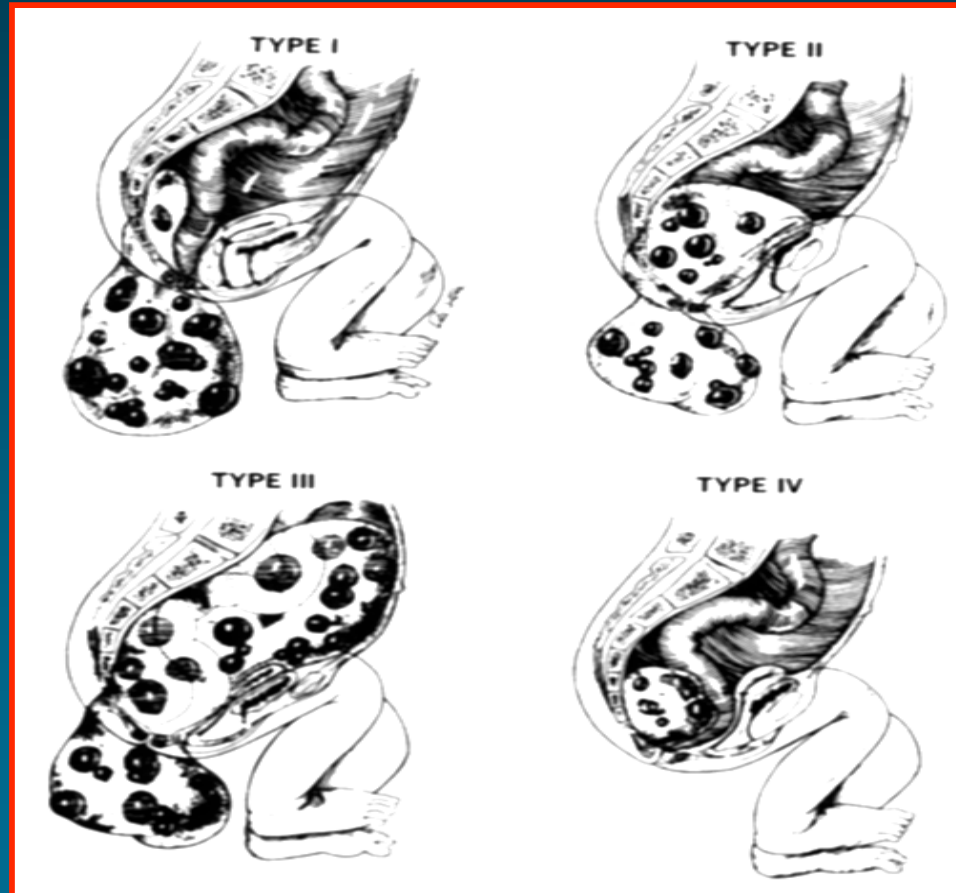
Anticipating Complex Anomalies

Congenital Tumors

How is the diagnosis made?

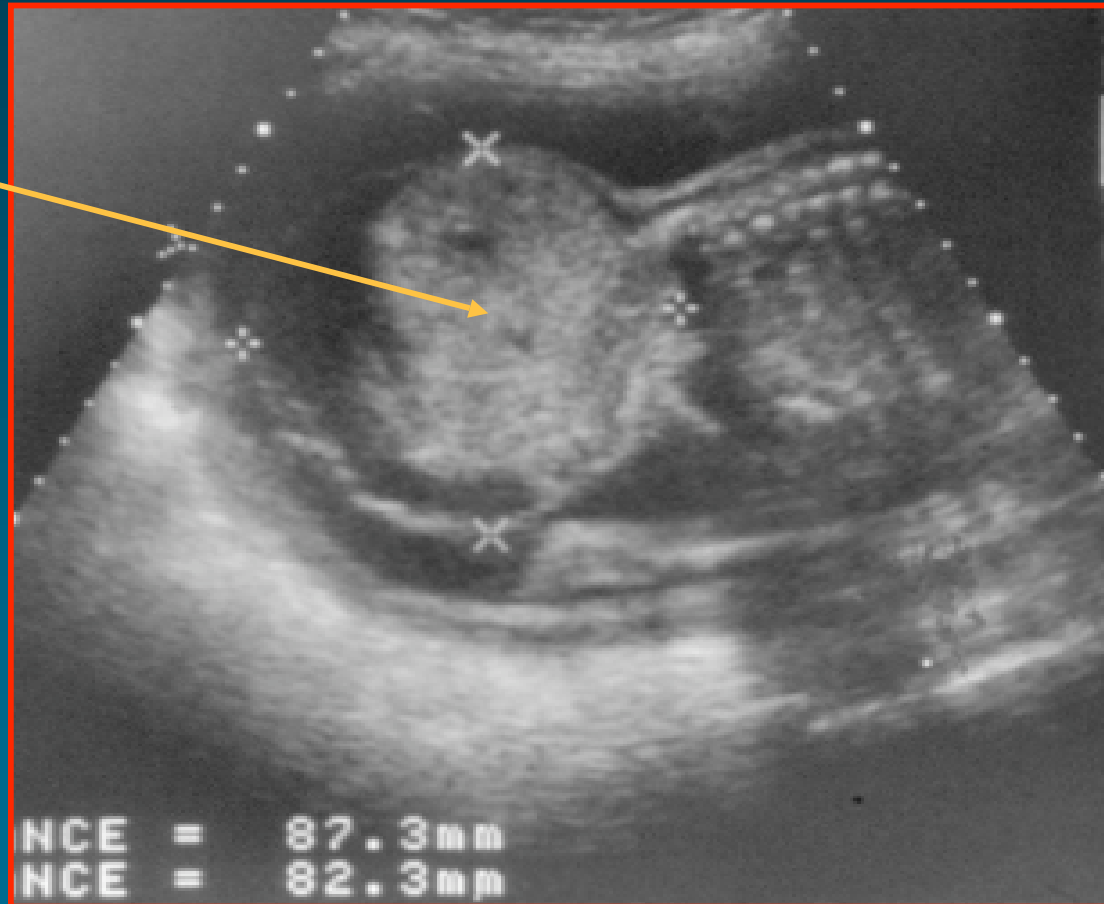
- CAT scan (baby only)
- MRI
- Ultrasound
- Serum markers

Anticipating Complex Anomalies sacrocoxygeal teratoma



Anticipating Complex Anomalies sacrocoxygeal teratoma

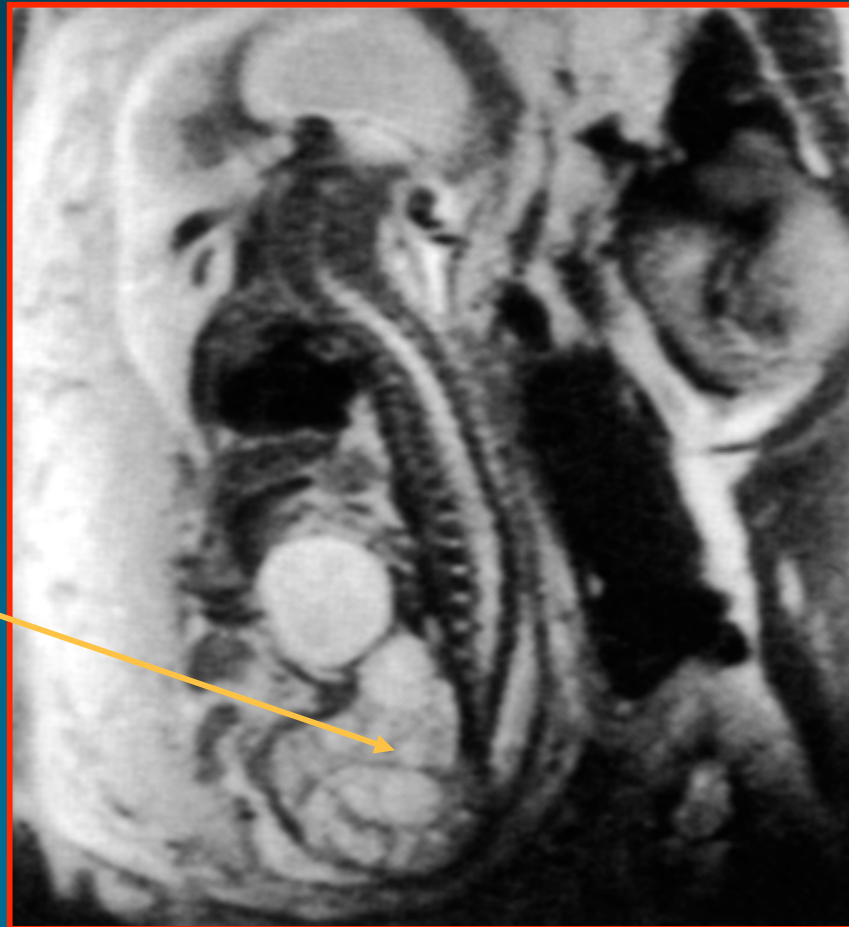
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Anticipating Complex Anomalies

Sacrocoxygeal Teratoma

teratoma



Anticipating Complex Anomalies

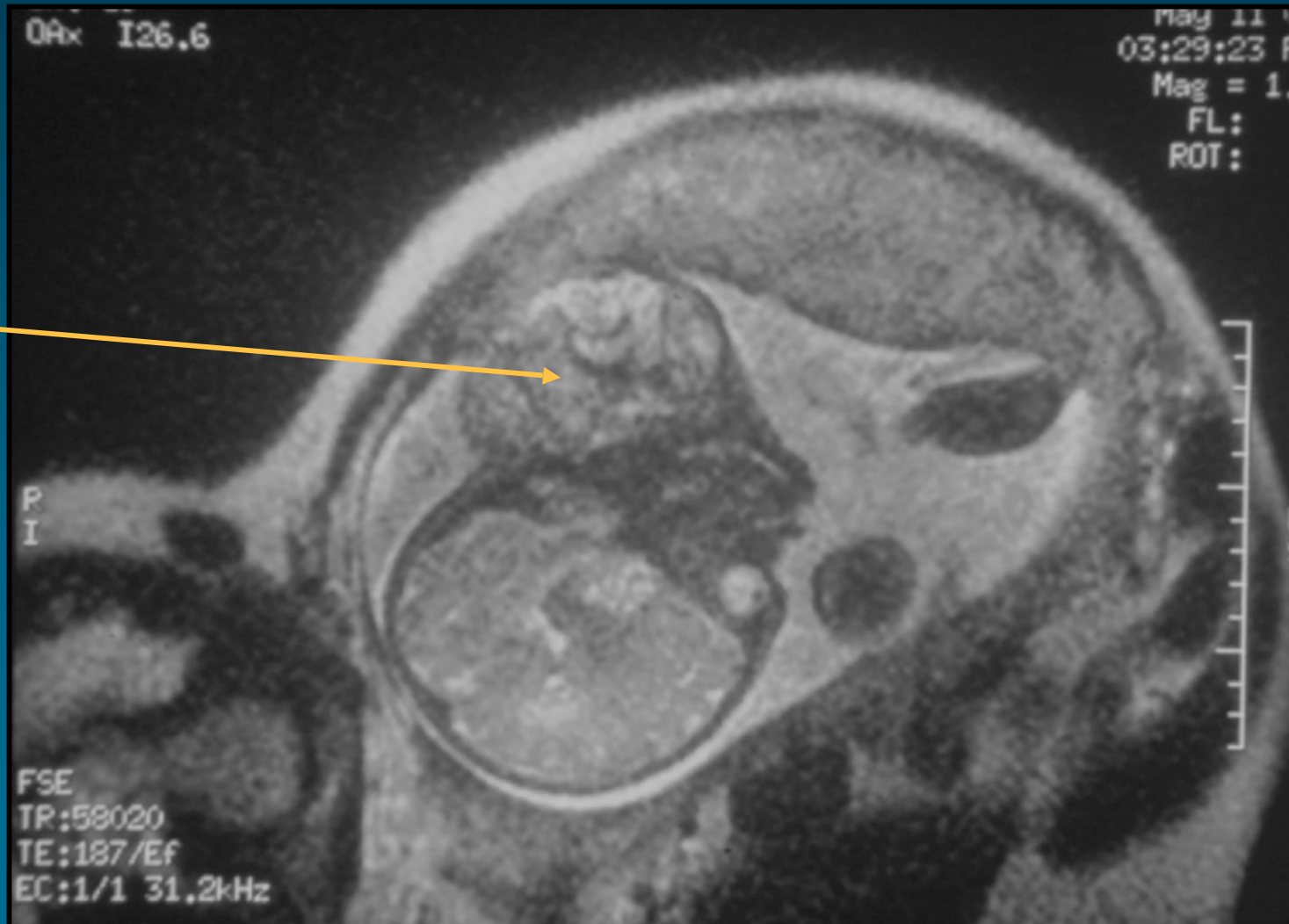
Congenital Tumors

What do we do about it?

- Diagnosis specific treatment at birth
- *Extra-uterine InTrapartum* (EXIT)
- Anecdotal fetal intervention

Anticipating Complex Anomalies *congenital tumors*

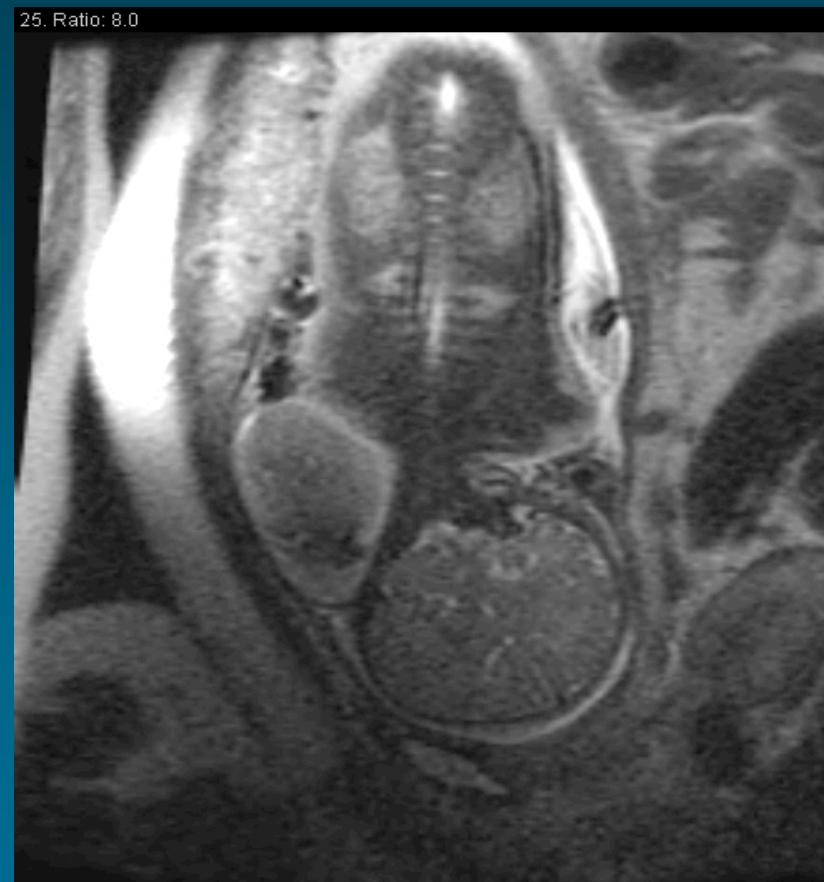
cervical
teraroma

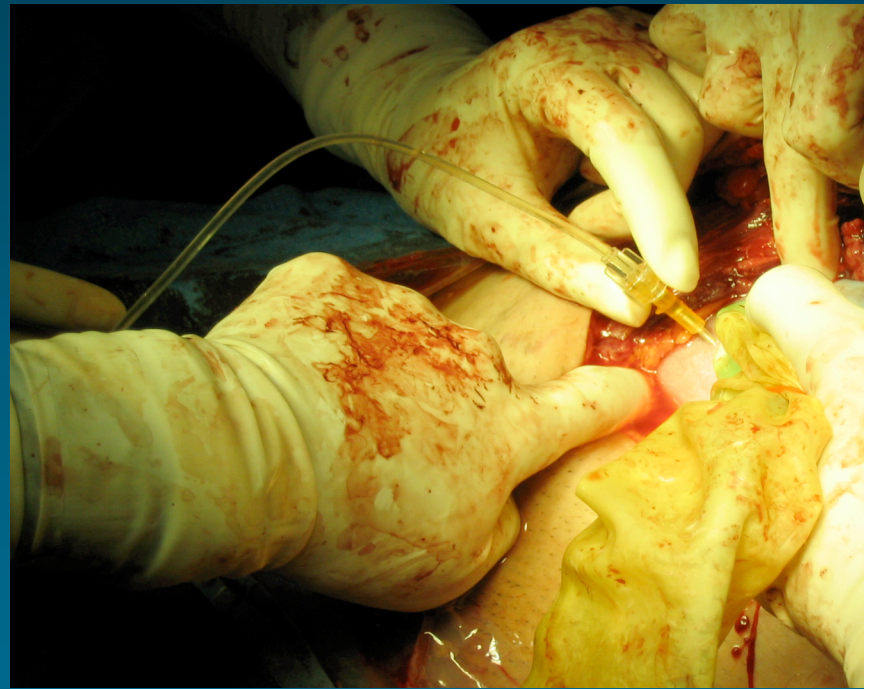


Anticipating Complex Anomalies *congenital tumors*



Cervical Teratoma







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 resolve before birth or be 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

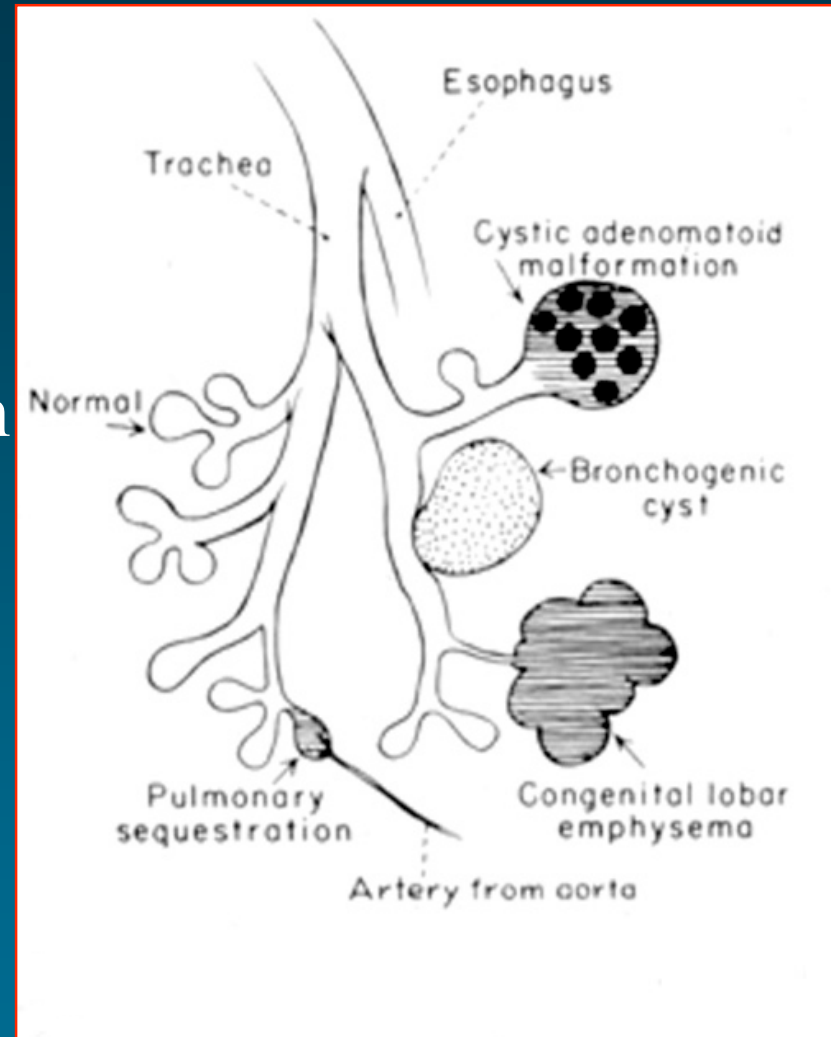
What are they?

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

Broncho-Pulmonary Foregut Malformations

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



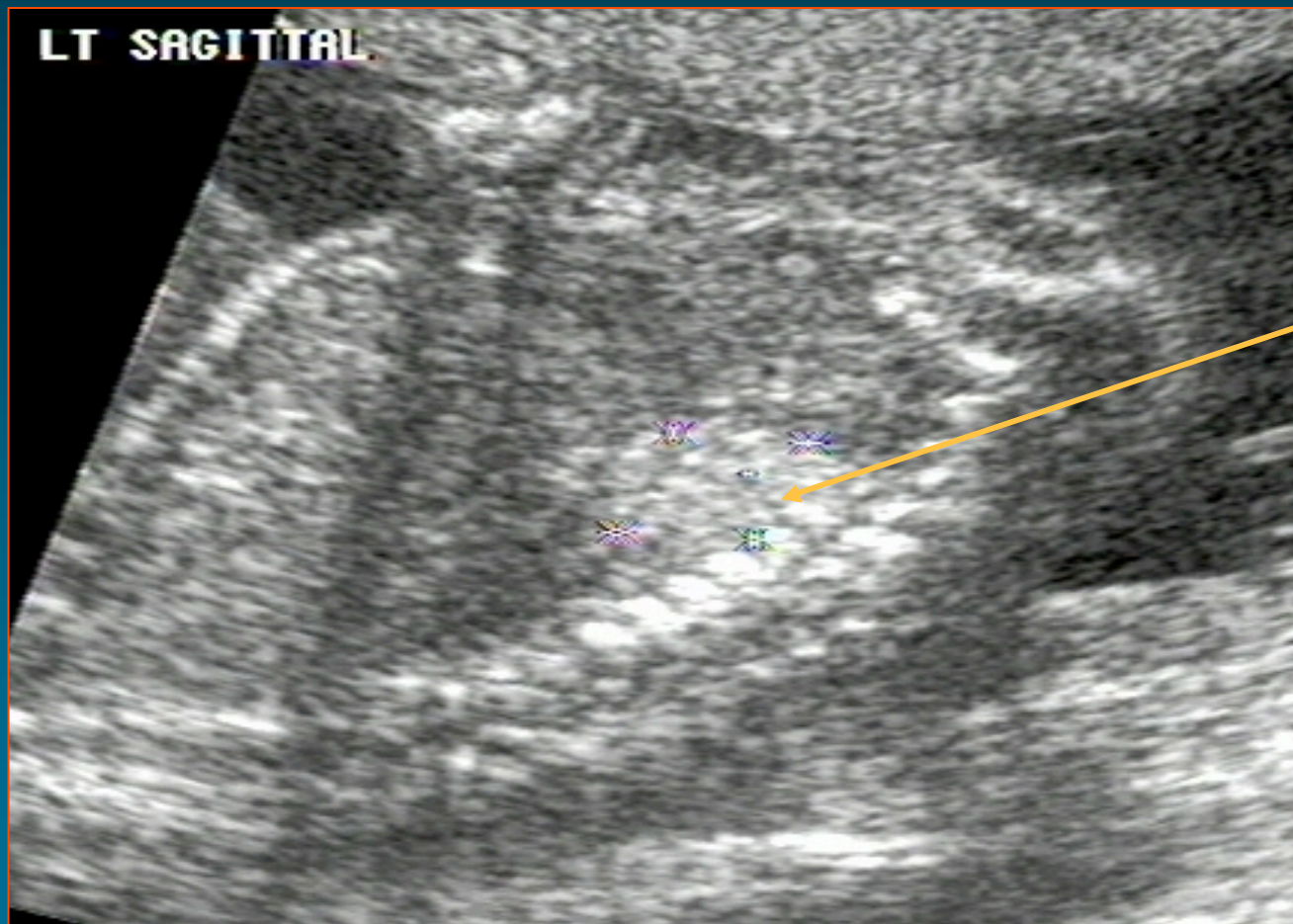
Anticipating Complex Anomalies

Broncho-Pulmonary(Lung Bud) Malformations

How is the diagnosis made?

- Ultrasound
- MRI

Anticipating Complex Anomalies pulmonary sequestration



lung
malform

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?

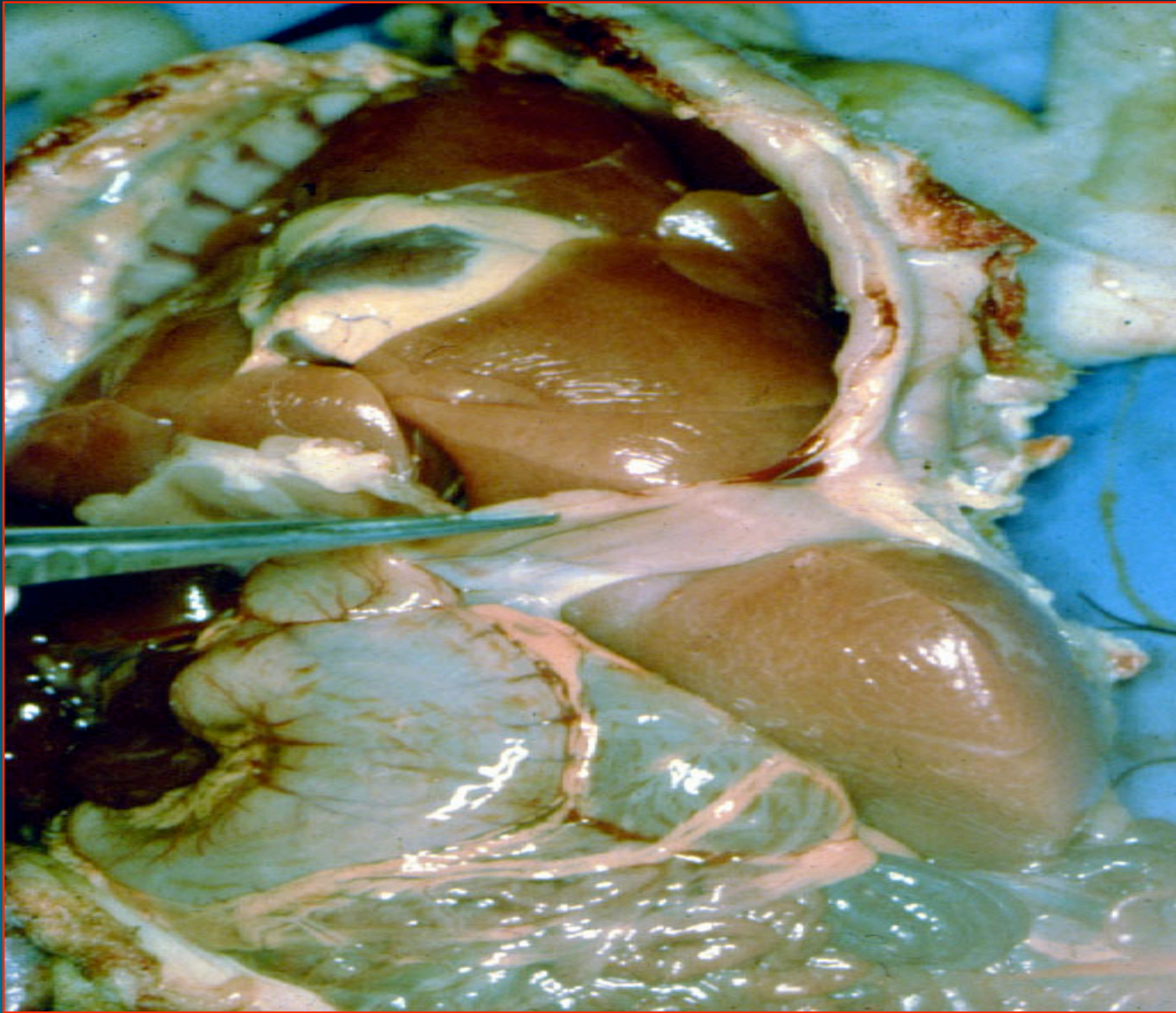
- *Almost all of these diagnoses can be fixed.*
- *The repair depends on the diagnosis.*
- *The exact diagnosis can't be made until birth.*
- *Some of the infants may have life-long nutritional concerns.*

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

