

## 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 mis-information.
  - » 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?

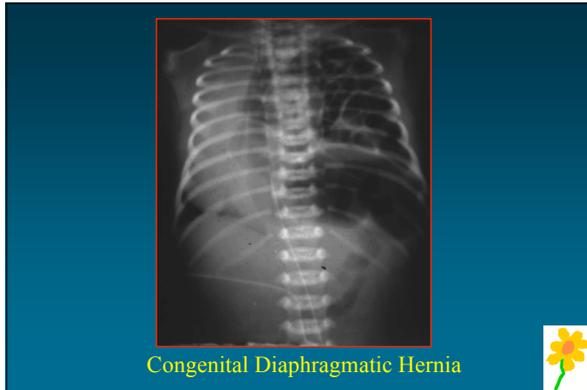


## Anticipating Complex Anomalies *Congenital Diaphragmatic Hernia*

### 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

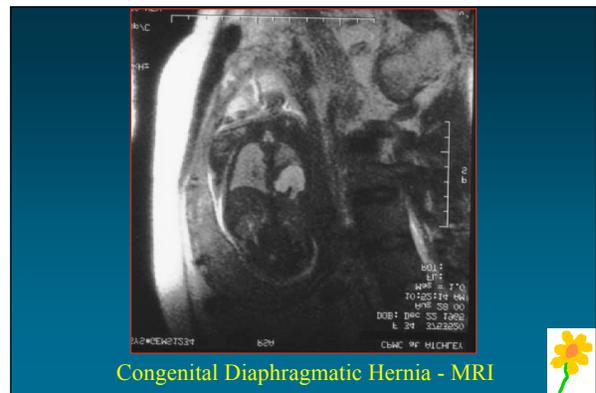
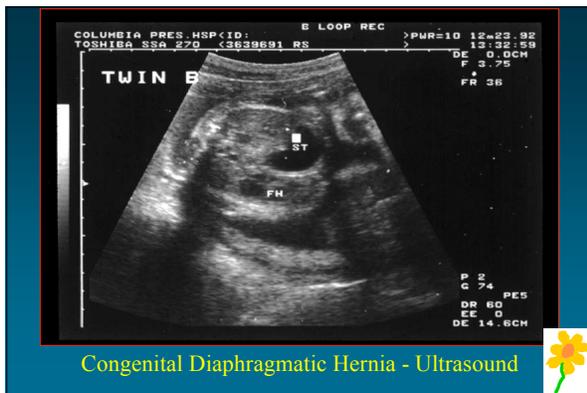




### Anticipating Complex Anomalies *Congenital Diaphragmatic Hernia*

How is the diagnosis made?

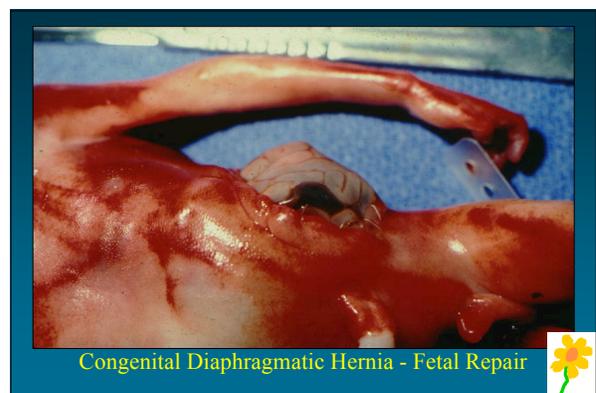
- Polyhydramnios
- Fetal ultrasound
- Magnetic resonance imaging



### 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



## 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 to 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 membranes
  - » Often associated with other anomalies (cardiac, pulmonary, trisomy 21)

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

## Anticipating Complex Anomalies Abdominal Wall Defects

How is the diagnosis made?

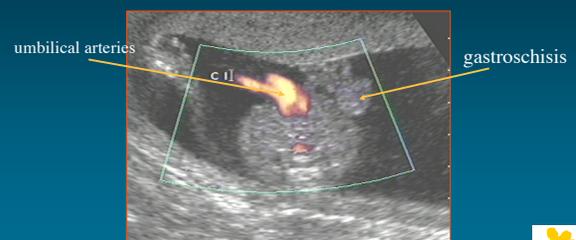
- Ultrasound
- Chromosome analysis

## Anticipating Complex Anomalies Gastroschisis



gastroschisis

## 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 omphalocele
- 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



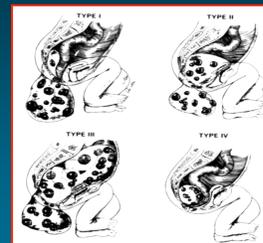
## Anticipating Complex Anomalies *Congenital Tumors*

How is the diagnosis made?

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

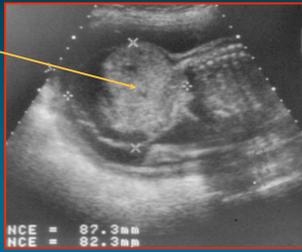


## Anticipating Complex Anomalies *sacrocoxygeal teratoma*



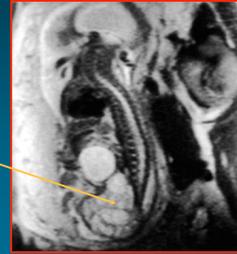
### Anticipating Complex Anomalies sacrocoxygeal teratoma

teratoma



### Anticipating Complex Anomalies Sacrocoxygeal Teratoma

teratoma



### Anticipating Complex Anomalies Congenital Tumors

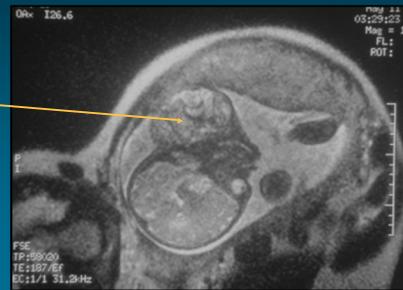
What do we do about it?

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



### Anticipating Complex Anomalies congenital tumors

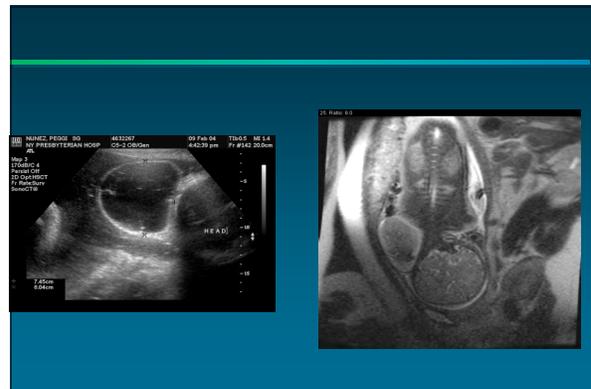
cervical  
teratoma

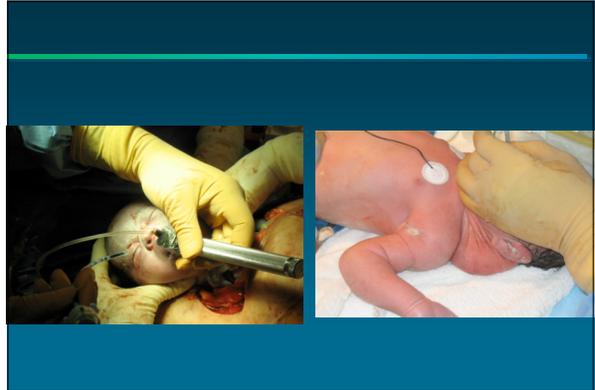


### Anticipating Complex Anomalies congenital tumors



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 disordered "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

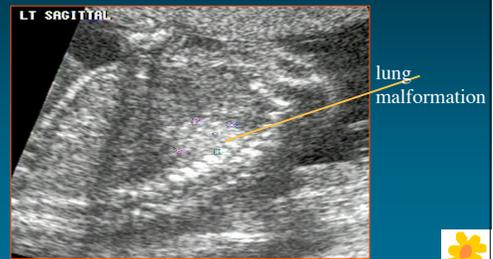


How is the diagnosis made?

- Ultrasound
- MRI



## Anticipating Complex Anomalies pulmonary sequestration



## 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 if 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

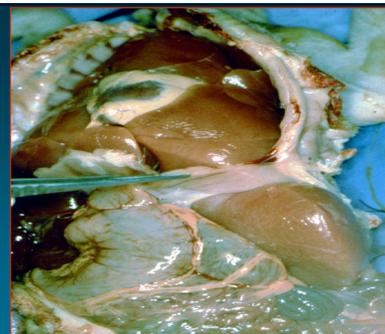
What should parents know?

- 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

