

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

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Charles J. H. Stolar, M.D.  
Director, Division of Pediatric Surgery  
Columbia University,  
College of Physicians and Surgeons  
Cornell University  
Weill Cornell College of Medicine  
Children's Hospital of New York  
New York-Presbyterian Hospital



### 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

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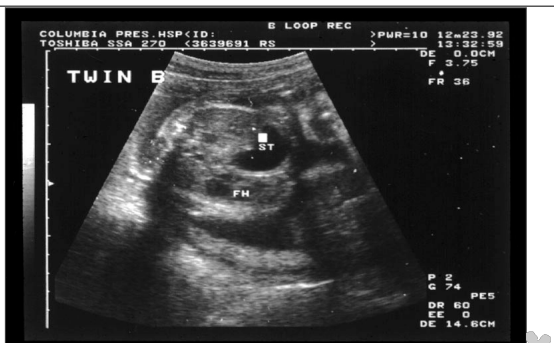


Congenital Diaphragmatic Hernia

### 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

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## 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?

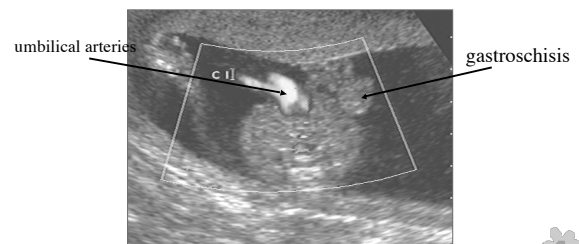
- Ultrasound
- Chromosome analysis

## Anticipating Complex Anomalies *Gastroschisis*



gastroschisis

## Anticipating Complex Anomalies *Gastroschisis*



umbilical arteries

c il

gastroschisis

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## Anticipating Complex Anomalies *Abdominal Wall Defects*

What do we do about it?

- Early delivery for gastroschisis if gut is very thickened(máybe), 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?

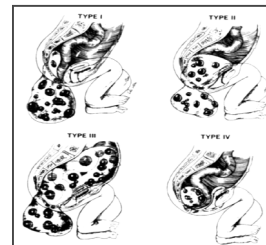
- Teratomas – sacrocoxygeal, etc.
- Mediastinal
- Neuroblastoma
- Cystic hygroma

## Anticipating Complex Anomalies *Congenital Tumors*

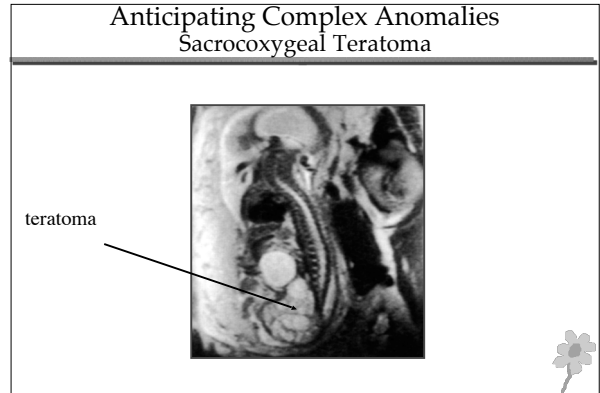
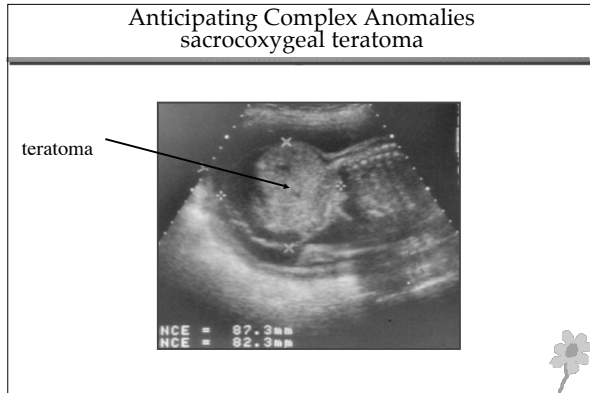
How is the diagnosis made?

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

## Anticipating Complex Anomalies *sacrocoxygeal teratoma*



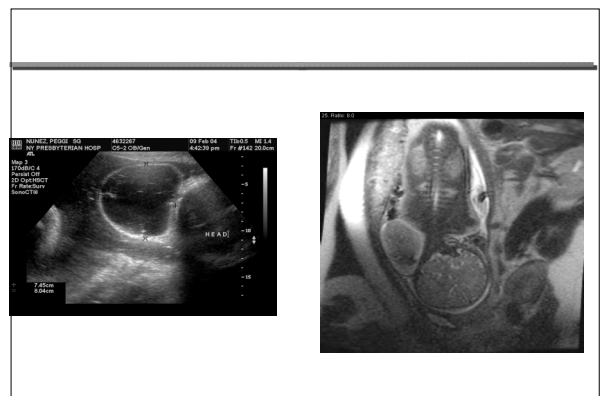
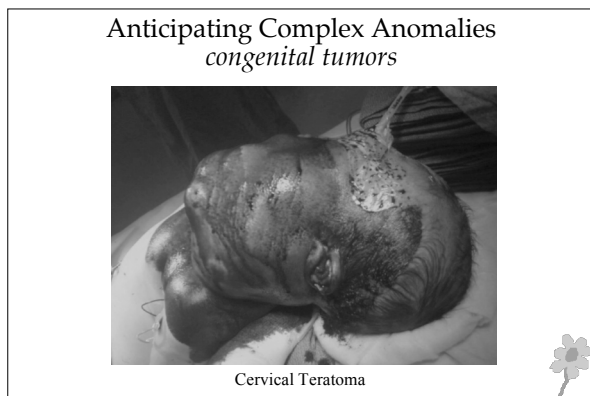
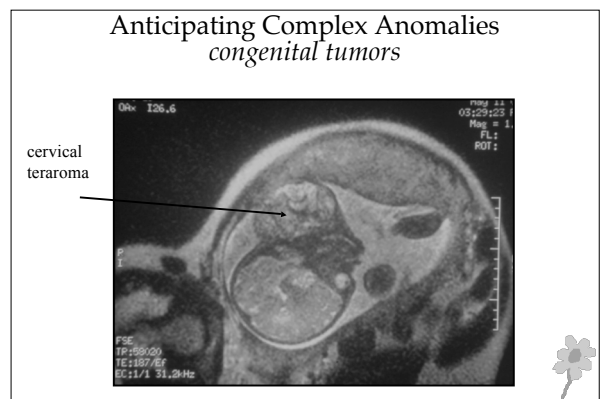
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Anticipating Complex Anomalies  
*Congenital Tumors*

What do we do about it?

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





### Anticipating Complex Anomalies *Congenital Tumors*

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

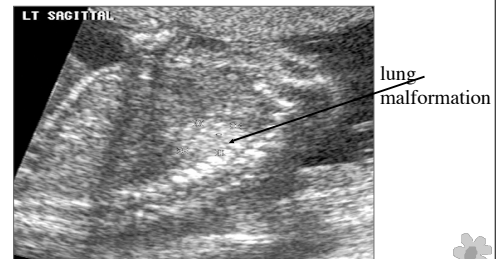
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### Anticipating Complex Anomalies *Broncho-Pulmonary(Lung Bud) Malformations*

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

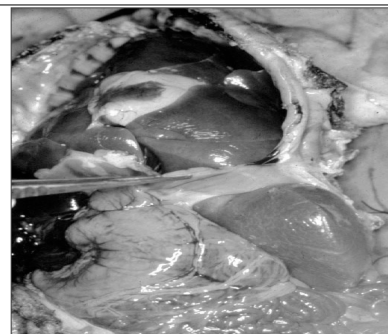
*What should parents know?*

- 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

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