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 misinformation.
- “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?

What is it/are they?

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

Anticipating Complex Anomalies

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

What should parents know?

• What is it/are they?
• How is the diagnosis made?
• What should parents know?
• What can be done about it?
• What are the outcomes?
How is the diagnosis made?
- Polyhydramnios
- Fetal ultrasound
- Magnetic resonance imaging

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
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
- Omphalocoele
  - 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
Gastroschisis

What are the potential long term concerns?
- Cardiac
- Pulmonary
- Foregut
- Chest wall axial skeleton
- Neuromotor

Anticipating Complex Anomalies
Gastroschisis

How is the diagnosis made?
- Ultrasound
- Chromosome analysis
Anticipating Complex Anomalies

Abdominal Wall Defects

What do we do about it?

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

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)

Abdominal Wall Defects

What are the long term problems?

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

Congenital Tumors

What are they?

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

How is the diagnosis made?

- CAT scan (baby only)
- MRI
- Ultrasound
- Serum markers
Anticipating Complex Anomalies
Sacrocoxygeal Teratoma

What do we do about it?

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

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 be resolve before birth or be associated with genetic syndrome.

**Cystic hygroma**

What are the long term problems?

- Chemotherapy morbidity
- Operative morbidity
- Follow-up

**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

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

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

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

What are the ultrasound observations?

- Dilated bowel
- Echogenic bowel
- Intra-abdominal calcifications

Anticipating Complex Anomalies
Broncho-Pulmonary (Lung Bud) Anomalies

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Intestinal Abnormalities

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# Anticipating Complex Anomalies

## Duodenal Atresia

### What do the ultrasound findings mean?
- Often, nothing… but sometimes they do
- Atresias, duodenal and other
- Hirschsprung’s disease
- Cystic fibrosis
- Ano-rectal malformations

## 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

### 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.

## Summary

- 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

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**Prenatal Tracheal Ligation**
Anticipating Complex Anomalies
Gastroschisis

lung bud/broncho-pulmonary malformation