It Happened…
What Now
How Can We Prepare
Prenatal Diagnosis Conference Case Studies

I have no Financial Disclosures

Objectives

» To support and highlight the importance of a collaborative multidisciplinary team approach when developing a plan of care for mother and baby
» To facilitate and lead perinatal diagnosis conference case reviews demonstrating the benefits of open communication, wholeness, and excellence
Prenatal Diagnosis Conference (PDC)

» Who are we

» What do we do

» Why

Who are we: Members of the Team

» Child Life Specialists
» Clinical Ethics
» Neonatology
» Palliative Care
» Pathology
» Radiology
» Sonographers
» Students

» Clinical Genetics
» Maternal Fetal Medicine
» Nursing
» Pediatric Cardiology, Pediatric Surgery, and other Pediatric Subspecialties (ENT, Neurosurgery, etc.)
What Do We Do

» Prenatally:
  ~ Show radiographic imaging
  ~ Review the clinical progression
» Disclose expectations for the plan of care
» Discuss controversial/complicating factors
» Postnatally: share the clinical course

Why

» Establish acceptable goals
» Optimize outcomes for mother and baby
» Enhance the patient experience
» Improve the satisfaction of the staff
» Foster learning

IT HAPPENED....

Birth Defects Occurring More Commonly: Abdominal Wall Defects
Unique Case
During 2005-2006, in the Inland Empire, Rates of Omphalocele Were Fairly Constant

In 2006, the Inland Empire had the Highest Reported Rate of Gastrochisis in the State
Abdominal Wall Defects

- Prenatal detection
  - Ultrasound
  - AFP

- Differential diagnosis
  - More common: gastroschisis and omphalocele
  - Less common: exstrophy (bladder, cloaca), amniotic band syndrome, limb body wall complex and/or Pentalogy of Cantrell

Physiologic Herniation

Normal Cord Insertion
Gastroschisis

- ~4-5/10,000
  - 2/3 simple (95% survival)
  - 1/3 complex (75% survival)
- Malformation (vascular and/or genetic) versus disruption (teratogens) versus deformation
- Para-umbilical defect—usually <4 cm and right sided
- All layers of abdominal wall
- Consequences
  - Poor growth, meconium passage, infection/inflammation, preterm birth, stillbirth
  - Bowel hypo-peristalsis/dilation, bowel atresia/malrotation, bowel necrosis and subsequent short gut syndrome

Defect to the right of the umbilical cord
Intra-amniotic bowel loops

Omphalocele
- Extrusion of abdominal contents into the base of cord
- Mass covered by peritoneum, amnion, and Wharton’s jelly (absent abdominal muscles, fascia, and skin)
- Associated anomalies (>2/3) and chromosomal anomalies (1/3) are common
- Preterm birth, poor growth, stillbirth, and lung hypoplasia (with a large omphalocele)

Omphalocele
- ~2/10,000 (30-70% survival)
- Extrusion of abdominal contents into the base of cord
- Mass covered by peritoneum, amnion, and Wharton’s jelly (absent abdominal muscles, fascia, and skin)
- Associated anomalies (>2/3) and chromosomal anomalies (1/3) are common
- Preterm birth, poor growth, stillbirth, and lung hypoplasia (with a large omphalocele)
CASE PRESENTATIONS
CASE 1: COMPLICATED GASTROSCISIS

CASE 2: ANTERIOR ABDOMINAL WALL DEFECTS

Fetal Anemia: Diagnosis & Treatment
Non-invasive Techniques

MCA Doppler
- Increased cardiac output → faster velocities
- In most cases, 1.5 MoM accurately predicts moderate to severe anemia with 98% precision
- Measurements can be initiated as early as 16-18 weeks

≥2 Constitutes Hydrops
- Ascites
- Pericardial effusion
- Pleural effusion
- Skin edema (>5 mm)
- Polyhydramnios (MVP >8 cm)
- Placentomegaly (>4 cm)
Intrauterine Transfusion (IUT)

- IUT is associated with a 1-2% rate of fetal loss
- Source of Red Blood Cells
  - Fresh adult red cells (CMV seronegative, leukocyte reduced, irradiated, O negative)
  - Fresh preferred to stored (enhanced 2,3-DPG)
- After initial transfusion should use MoM of 1.32 (mixing of adult and fetal blood cells changes the viscosity of blood and decreases the predictive value of MCA)

Outcome

- Survival ~84%
  - Hydropic = 70% versus Non-hydropic = 92%
  - Severe hydrops → 39% versus Mild hydrops → reversal in ~90%
- Need for top-up transfusion in neonatal life
- Long term neurologic effects
  - Hearing loss has been reported in association with high bilirubin levels (5-10x prevalence vs. the general population)
  - Normal neurologic outcome in 90% of survivors
Fetal Anemia: Alpha Thalassemia

Alpha-Thalassemia: deletions of the α-globulin gene on 16p

- Single deletion (α-α): clinically insignificant
- 2 deletions=trait/carriers: α-thalassemia minor
  - Clinically mild microcytic asymptomatic anemia
  - cis (α/-α):
    - More likely in Southeast Asian ancestry
    - Offspring have increased risk of HbH or Bart’s disease
  - trans (α/-α) deletions are more common in those of African descent

- 3 deletions (α/-α): HbH disease
  - Mild to moderate hemolytic anemia
- 4 deletions (α/-α): Hb Bart’s disease
  - α-thalassemia major
  - Associated with hydrops fetalis, IUFD, preeclampsia
CASE 3: ALPHA THALASSEMIA

Background

- Homozygous (4 gene deletion) alpha thalassemia is associated with hemoglobin (Hb) Bart hydrops fetalis
- Without intrauterine therapy, homozygous alpha thalassemia pregnancies generally result in fetal or neonatal demise
- Traditional techniques to detect fetal anemia, have proven unreliable in fetal homozygous alpha thalassemia cases
Case Discussion

» Consensus: offer IUT
   ~ if the couple provided informed consent
   ~ bone marrow transplant could be curative

» Transplant Medicine consultation

» The couple opted to pursue intervention