OB Ultrasound and the Pediatrician

Borderline Anomalies

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Disclosure

• No conflict of interest to disclose

Introduction

Commonly Diagnosed Anomalies

• Spina Bifida
• Hydrocephalus
• Hypoplastic Left Heart Syndrome
Borderline Anomalies

- New technology facilitated diagnosis
  - Technology advances faster than our understanding of fetal development
- Diagnosis tends to be imprecise
- Associated with normal development
- Associated with abnormal development
- Presence of additional U/S findings important
- Counseling regarding prognosis difficult (for isolated finding)
- Neonatal follow-up planning not standardized

Intracranial Anomalies: Posterior Fossa

Diagnosis?
Dandy-Walker Variant

Description

- Partial agenesis or hypoplasia of inferior vermis
- Cystic dilation of 4th ventricle
- Part of Dandy-Walker continuum

Dandy-Walker Variant

Associated anomalies

- Abnormal karyotype (30%)
- Ventriculomegaly
- Absent corpus callosum
- Cardiac defects
- Genito-urinary abnormalities
- Limb defects

Dandy-Walker Malformation Complex

Table 1. Reported Autopsy Pathology or Postmortem Imaging After Prenatal Diagnosis of Dandy-Walker Malformation

| Reference | GA (wk) | Agenesis | Partial | Discordant | Discordant | Cases (%)
|-----------|---------|----------|---------|------------|------------|----------
| Arute and Fang | 14-15 | 1 | 0 | 0 | 0 | 50.0%
| Sleigh et al | 15-16 | 2 | 0 | 0 | 0 | 50.0%
| Inoue et al | 16-17 | 3 | 0 | 0 | 0 | 50.0%
| Lossen et al | 17-18 | 4 | 0 | 0 | 0 | 50.0%
| De Ziegler et al | 18-19 | 5 | 0 | 0 | 0 | 50.0%
| Ong et al | 19-20 | 6 | 0 | 0 | 0 | 50.0%

- Ventricles
- Head
- Heart
- Limb
- Body

DWC

Natural history/Prognosis

- Extremely variable prognosis
- Range: normal development to severe handicap or death
- Depends largely on associated anomalies
- Utility of partial “variants” unclear
  - Only 41% concordance with autopsy findings

Fetal Cerebral Ventrices

- Head
- Limb
- Body
- Heart
- Ventricles
Mild Ventriculomegaly – Long Term Outcome

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Glioma &gt; 12 mm</th>
<th>Normal Males</th>
<th>Normal females</th>
<th>p*</th>
<th>p**</th>
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<tbody>
<tr>
<td>Brainstem</td>
<td>55.16 ± 8.14</td>
<td>56.02 ± 7.33</td>
<td>54.04 ± 7.15</td>
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Agenesis of the Corpus Callosum

- Failure of axons to cross midline and form corpus callosum
- May be complete or partial

Corpus Callosum – Normal Anatomy

Absence of the Corpus Callosum

Absence of the Corpus Callosum
### Absence of the Corpus Callosum

**Agenesis of the Corpus Callosum**

**Associated anomalies**

- Structural anomalies seen 60% of the time
- Chromosomal anomalies 10-20%
- Multiple syndromes described
- Numerous intracranial anomalies associated
- Cardiac, gastrointestinal, musculoskeletal, and renal defects common

### Borderline CNS Anomalies

**Antepartum Evaluation**

- Targeted ultrasound
- Fetal echocardiogram
- Amniocentesis
- Fetal MRI?
- Genetic consultation
- IgM titers for CMV, Toxoplasmosis, Parvovirus, Rubella (Ventriculomegaly, ACC)

### Agenesis of the Corpus Callosum

**Etiology**

- Incidence: 0.3-0.7% of general population
- Genetics
  - Most sporadic
  - Autosomal dominant, recessive, and x-linked described
- Teratogens
  - Alcohol, cocaine, valproate
  - CMV, Rubella

**Prognosis**

- Isolated
  - 75% normal or near normal at 3 years
  - Subtle cognitive defects may occur later

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**Table 3**

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<th>CNS Anomalies</th>
<th>Cardiac/Gastrointestinal/Musculoskeletal/Renal Defects</th>
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<td>Autosomal dominant</td>
<td>Reversal of cerebral asymmetry, bicoronal plagiocephaly</td>
<td>Hypoplasia, micrognathia, hypospadias, vertebral defects</td>
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<td>Autosomal recessive</td>
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<td>Hypoplastic lungs, diaphragmatic hernia, congenital heart defects</td>
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<td>X-linked</td>
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Borderline CNS Anomalies (Isolated)
Neonatal Evaluation

- Fetal MRI
  - If abnormal,
    - Genetics
    - Neurosurgery/Neurology
    - Follow for developmental delays
    - Follow-up MRI

Choroid Plexus Cyst
Description

- One or more anechoic cysts in choroid plexus
  - First seen in 1st trimester
  - Disappear in third trimester

Choroid Plexus Cyst
Associated Abnormalities

- Seen in 1% of all pregnancies
- 50% of Trisomy 18 fetuses have CPC

Choroid Plexus Cyst
Management

- Always regress
- Targeted Ultrasound to r/o other abnormalities
- Isolated finding: excellent prognosis
- No pediatric follow-up indicated
Echogenic Cardiac Focus

Description

- Cardiac papillary muscle echogenicity
  - 78% left ventricle
  - 18% right ventricle
  - 4% Bilateral
- Occurs in 3-4% of all 2nd trimester fetuses
- Occurs in 9-12% Asian fetuses

Associated anomalies

- Increased risk of Trisomy 21 (LR 1.8)
  - Rarely clinically significant increase
- Look for other Trisomy 21 “markers”
- Increased risk of Trisomy 13
  - Never an isolated finding

Management

- Targeted ultrasound
- Isolated ECF almost always a normal finding
- No pediatric follow-up indicated
Mild Pelviectasis

Associated Anomalies

- Increased risk for Trisomy 21 (LR 1.6)
  - Not an important isolated marker
- Examine other Trisomy 21 “markers”
- Increased risk for Trisomy 13
  - Never an isolated finding
- Also weak association with turner’s Syndrome, XXX,Trisomy 8

Mild Fetal Pyelectasis

Issues with Prenatal Detection

- Precursor to renal pathology
- Allow prompt surgical correction to avoid irreversible damage to renal function
- Excessive follow-up
  - Increased medical expense
  - Overutilization of professional resources
  - Increase patient/family anxiety
- Uncertain postnatal follow-up

Mild Pelviectasis

Description

- Definitions
  - Pyelectasis
  - Transient or minimal hydronephrosis
- Ultrasound definition
  - >4cm AP diameter of renal pelvis 2nd trimester
  - >7cm 3rd trimester
- 3% of normal fetuses have MP
- 0.46% with MP have aneuploidy

Pyelectasis
Mild Pelviectasis

Prognosis

- Most cases are transient and idiopathic
- Postnatal workup if MP persists (RPD>7mm after 32 weeks)
- Causes
  - 48% transient
  - 15% physiologic (mild, nonprogressive)
  - 11% UPJ obstruction
  - 9% vesicoureteral reflux
  - 4% megaloureter
  - 2% multicystic dysplastic kidney disease
  - 2% ureterocele

Conclusions

- Prenatal detection important
- Excessive prenatal follow-up not indicated
- Postnatal follow-up indicated
  - 1st week
  - 1 month
    - 5% show pathology requiring surgery
  - Periodic ultrasound
  - Renogram
  - Antibiotics
  - Surgical intervention (few trials)

Summary

- Obstetrical Ultrasound has become an excellent tool for identifying structural anomalies in the fetus.
- Borderline anomalies are more difficult, but advancing technology allows increasing accuracy in detection.
- MRI has become an important adjunct to ultrasound. However, its precise role has yet to be determined.
- In utero detection allows appropriate postnatal follow-up and intervention.
- Evidence-based management is lacking.