Ultrasound and MRI: Complimentary Tools in Evaluating Fetal Anomalies

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I have no relevant financial relationships to disclose
Objectives

• Describe the advantages of using additional imaging modalities in the diagnosis of complex congenital anomalies

• Describe the management options of complex congenital anomalies
Magnetic Resonance Imaging
HASTE

- Half Fourier Acquisition Turbo Spin Echo
  - Single shot fast spin echo (GE)
- "Workhorse"
  - Anatomy
  - pathology
True FISP

- Fast imaging with steady-state precession
- FIESTA (GE)
- Bright blood sequence

- Useful:
  - Cardiac/mediastinal abnormalities
  - CDH
  - Polyhydramnios
  - Cine sequences
• **Limited contrast**

• **Useful:**
  - Meconium
  - Liver in CDH
Echoplanar Imaging

- EPI
- Very fast
- Sensitive to image artifacts & distortion

Useful:
- Osseous structures
- Calcifications
- Blood products
- Vessels
Ultrasound
Ultrasound
Ultrasound
**Ultrasound**

- Around since late 50s/early 60s
- Techniques improved in 70s
- Safe
- $ (expensive)
- Portable
- Experienced sonographers
- Experienced interpretation
- Gestational age is a factor
- Can adjust for fetal movement

**MRI**

- MRI in pregnancy since early 80s
- Fetal diagnostics improved in 90s
- Safe
  - Second and third trimester
- $$$ (very expensive)
- Stationary
- Experienced technicians
- Experienced interpretation
- Gestational age is a factor
- Fetal movement is a factor
Why Does This Matter?

- No perfect prenatal diagnostic imaging modality

- Optimal imaging improves our diagnostic abilities
  - More accurate diagnosis →
  - Improved counseling
  - Improved prognostication
  - Delineation of available management options
Available management options

- Routine care
- Change in delivery venue
- Mode of delivery
- Fetal therapy
- Postnatal surgery
- Palliative care
- Termination
Fetal Surgery - Prerequisites

- Accurate Prenatal Diagnosis
- No Associated Anomalies
- Defined Natural History
- Correctable lesion leading to fetal death or organ destruction
- Technical Feasibility
Clinical Examples
What is This?
A. Banana Sign
B. Strawberry Sign
C. Mango Sign
A Randomized Trial of Prenatal versus Postnatal Repair of Myelomeningocele

N. Scott Adzick, M.D., Elizabeth A. Thom, Ph.D., Catherine Y. Spong, M.D., John W. Brock III, M.D., Pamela K. Burrows, M.S., Mark P. Johnson, M.D., Lori J. Howell, R.N., M.S., Jody A. Farrell, R.N., M.S.N., Mary E. Dabrowiak, R.N., M.S.N., Leslie N. Sutton, M.D., Nalin Gupta, M.D., Ph.D., Noel B. Tulipan, M.D., Mary E. D'Alton, M.D., and Diana L. Farmer, M.D., for the MOMS Investigators*
Fetal MMC Repair
Who is a Candidate for fMMC Repair?

• Singleton
• Lesion Level T1-S1
• Hindbrain Herniation
• 19.0-25.9 weeks
• Normal karyotype
• 18 years of age and up
• No significant medical concerns
Who is NOT a Candidate for fMMC Repair?

- Additional fetal anomalies
- Severe kyphosis
- Risk of preterm birth
  - Short cervix, history of preterm birth, bleeding in pregnancy
- Placental abruption/abnormal placentation
- BMI >35
- Contraindication to elective surgery
- Previous hysterotomy in active uterine segment
<table>
<thead>
<tr>
<th>Outcome</th>
<th>Prenatal Surgery (N=78)</th>
<th>Postnatal Surgery (N=80)</th>
<th>Relative Risk (95% CI)</th>
<th>P Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Primary outcome — no. (%)</td>
<td>53 (68)</td>
<td>78 (98)</td>
<td>0.70 (0.58–0.84)†</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Components of the primary outcome — no. (%)</td>
<td></td>
<td></td>
<td></td>
<td>&lt;0.001</td>
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<tr>
<td>Death before placement of shunt</td>
<td>2 (3)</td>
<td>0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Shunt criteria met</td>
<td>51 (65)</td>
<td>74 (92)</td>
<td></td>
<td></td>
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<tr>
<td>Shunt placed without meeting criteria</td>
<td>0</td>
<td>4 (5)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Placement of shunt — no. (%)</td>
<td>31 (40)</td>
<td>66 (82)</td>
<td>0.48 (0.36–0.64)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Any hindbrain hemiation — no./total no. (%)</td>
<td>45/70 (64)</td>
<td>66/69 (96)</td>
<td>0.67 (0.56–0.81)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Degree of hindbrain hemiation — no./total no. (%)</td>
<td></td>
<td></td>
<td></td>
<td>&lt;0.001‡</td>
</tr>
<tr>
<td>None</td>
<td>25/70 (36)</td>
<td>3/69 (4)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mild</td>
<td>28/70 (40)</td>
<td>20/69 (29)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Moderate</td>
<td>13/70 (19)</td>
<td>31/69 (45)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Severe</td>
<td>4/70 (6)</td>
<td>15/69 (22)</td>
<td></td>
<td></td>
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<tr>
<td>Any brainstem kinking — no./total no. (%)</td>
<td>14/70 (20)</td>
<td>33/69 (48)</td>
<td>0.42 (0.25–0.71)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Degree of brainstem kinking — no./total no. (%)</td>
<td></td>
<td></td>
<td></td>
<td>0.001‡</td>
</tr>
<tr>
<td>None</td>
<td>56/70 (80)</td>
<td>36/69 (52)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mild</td>
<td>4/70 (6)</td>
<td>8/69 (12)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Moderate</td>
<td>7/70 (10)</td>
<td>17/69 (25)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Severe</td>
<td>3/70 (4)</td>
<td>8/69 (12)</td>
<td></td>
<td></td>
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<tr>
<td>Abnormal location of fourth ventricle — no./total no. (%)</td>
<td>32/70 (46)</td>
<td>49/68 (72)</td>
<td>0.63 (0.47–0.85)</td>
<td>0.002</td>
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<tr>
<td>Location of fourth ventricle — no./total no. (%)</td>
<td></td>
<td></td>
<td></td>
<td>&lt;0.001‡</td>
</tr>
<tr>
<td>Normal</td>
<td>38/70 (54)</td>
<td>19/68 (28)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Low</td>
<td>28/70 (40)</td>
<td>29/68 (43)</td>
<td></td>
<td></td>
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<tr>
<td>At foramen magnum</td>
<td>1/70 (1)</td>
<td>8/68 (12)</td>
<td></td>
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<tr>
<td>Below foramen magnum</td>
<td>3/70 (4)</td>
<td>12/68 (18)</td>
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<tr>
<td>Syringomyelia — no./total no. (%)</td>
<td>27/69 (39)</td>
<td>39/67 (58)</td>
<td>0.67 (0.47–0.96)</td>
<td>0.03</td>
</tr>
<tr>
<td>Epidermoid cyst — no./total no. (%)</td>
<td>2/67 (3)</td>
<td>1/66 (2)</td>
<td>1.97 (0.18–21.20)</td>
<td>1.00</td>
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<tr>
<td>Surgery for tethered cord — no./total no. (%)</td>
<td>6/77 (8)</td>
<td>1/80 (1)</td>
<td>6.15 (0.76–50.00)</td>
<td>0.06</td>
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<tr>
<td>Chiari decompression surgery — no./total no. (%)</td>
<td>1/77 (1)</td>
<td>4/80 (5)</td>
<td>0.26 (0.03–2.24)</td>
<td>0.37</td>
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<tr>
<td>Shunt infection — no./total no. (%)</td>
<td>5/77 (6)</td>
<td>7/80 (9)</td>
<td>0.73 (0.24–2.21)</td>
<td>0.58</td>
</tr>
<tr>
<td>Outcome</td>
<td>Prenatal Surgery (N=78)</td>
<td>Postnatal Surgery (N=80)</td>
<td>Relative Risk (95% CI)</td>
<td>P Value</td>
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<tr>
<td>----------------------------------------------</td>
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<tr>
<td><strong>Maternal outcome</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chorioamniotic membrane separation — no. (%)</td>
<td>20 (26)</td>
<td>0</td>
<td>NA</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Pulmonary edema — no. (%)</td>
<td>5 (6)</td>
<td>0</td>
<td>NA</td>
<td>0.03</td>
</tr>
<tr>
<td>Modified biophysical profile &lt;8 — no. (%)†</td>
<td>13 (17)</td>
<td>6 (8)</td>
<td>2.22 (0.89–5.55)</td>
<td>0.08</td>
</tr>
<tr>
<td>Oligohydramnios — no. (%)</td>
<td>16 (21)</td>
<td>3 (4)</td>
<td>5.47 (1.66–18.04)</td>
<td>0.001</td>
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<tr>
<td>Placental abruption — no. (%)</td>
<td>5 (6)</td>
<td>0</td>
<td>NA</td>
<td>0.03</td>
</tr>
<tr>
<td>Gestational diabetes — no. (%)</td>
<td>4 (5)</td>
<td>5 (6)</td>
<td>0.82 (0.23–2.94)</td>
<td>1.00</td>
</tr>
<tr>
<td>Chorioamnionitis — no. (%)</td>
<td>2 (3)</td>
<td>0</td>
<td>NA</td>
<td>0.24</td>
</tr>
<tr>
<td>Preeclampsia or gestational hypertension — no. (%)</td>
<td>3 (4)</td>
<td>0</td>
<td>NA</td>
<td>0.12</td>
</tr>
<tr>
<td>Spontaneous membrane rupture — no. (%)</td>
<td>36 (46)</td>
<td>6 (8)</td>
<td>6.15 (2.75–13.78)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Spontaneous labor — no. (%)</td>
<td>30 (38)</td>
<td>11 (14)</td>
<td>2.80 (1.51–5.18)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Blood transfusion at delivery — no. (%)</td>
<td>7 (9)</td>
<td>1 (1)</td>
<td>7.18 (0.90–57.01)</td>
<td>0.03</td>
</tr>
<tr>
<td><strong>Status of hysterotomy site at delivery — no./total no. (%)</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Intact, well-healed</td>
<td>49/76 (64)</td>
<td></td>
<td></td>
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<tr>
<td>Very thin</td>
<td>19/76 (25)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Area of dehiscence</td>
<td>7/76 (9)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Complete dehiscence</td>
<td>1/76 (1)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Fetal or neonatal outcome</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bradycardia during fetal or neonatal repair — no. (%)</td>
<td>8 (10)</td>
<td>0</td>
<td>NA</td>
<td>0.003</td>
</tr>
<tr>
<td>Perinatal death — no. (%)</td>
<td>2 (3)</td>
<td>2 (2)</td>
<td>1.03 (0.14–7.10)</td>
<td>1.00</td>
</tr>
<tr>
<td>Gestational age at birth — wk</td>
<td>34.1±3.1</td>
<td>37.3±1.1</td>
<td></td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Gestational age at birth — no. (%)</td>
<td></td>
<td></td>
<td></td>
<td>&lt;0.001‡</td>
</tr>
<tr>
<td>&lt;30 wk</td>
<td>10 (13)</td>
<td>0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>30–34 wk</td>
<td>26 (33)</td>
<td>4 (5)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>35–36 wk</td>
<td>26 (33)</td>
<td>8 (10)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>≥37 wk</td>
<td>16 (21)</td>
<td>68 (85)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Fetal Risks Associated with OFS

- Surgical injury
- Prematurity
- Drug effects
- Fetal/neonatal death
Maternal Risks Associated with OFS

- Bleeding
- Infection
  - Wound
  - Intraamniotic
- Preterm premature rupture of membranes
- Preterm labor/delivery
- Anesthesia complications
  - General
  - Regional
- Medication side effects
  - Tocolysis
- Prolonged hospitalization
- Pulmonary edema
- Impaired fertility
- Repeat cesarean delivery
- Uterine rupture
- Death
No Hindbrain Herniation

Hindbrain Herniation
Asymmetric Venticulomegaly

- Is there an etiology?
- If both normal size – likely normal variant
- If asymmetric ventriculomegaly:
  - Rapidly evolving is poor prognostic sign
  - Isolated, mild, stable – most likely normal outcome
  - Higher incidence of mental, psychomotor and behavior abnormalities

Senat Ultrasound Obstet Gynecol 1999;14:327-332
Sadan BJOG 2007;114:596-602
Achiron Obstet Gynecol 1997;89:233-7
Grade I-II IVH
Management: Serial scans and possible change of delivery venue and mode
Referral for possible midline cleft and abnormal profile
Hypothalamic Hamartoma
Pallister-Hall Syndrome

- Polydactyly or syndactyly
- Hypothalamic hamartoma
  - Seizures
  - Panhypopituitarism
- Bifid epiglottis
- Imperforate anus
- Renal abnormalities
- Mutation in GLI3 gene
Pallister-Hall Syndrome

- Polydactyly or syndactyly
- Hypothalamic hamartoma
  - Seizures
  - Panhypopituitarism
- Bifid epiglottis
- Imperforate anus
- Renal abnormalities
- Mutation in GLI3 gene

Delivery venue changed in order to monitor for neonatal instability, airway concerns and hormone replacement if needed.
Abdominal Wall Defect
Limb-Body-Wall Complex

Severely Disorganized Spine
Large Defect
Short Umbilical Cord
Adhered to the Placenta
Lethal
Hydrops and Solid Abdominal Mass
Right Mesoblastic Nephroma
CHAOS:
Congenital High Airway Obstruction Syndrome
Prenatal diagnosis:
- Large echogenic lungs
- Flattened or inverted diaphragms
- Dilated airways distal to the obstruction
- Fetal ascites/hydrops

Etiology:
- Laryngeal atresia
- Laryngeal web
- Tracheal atresia
- Laryngeal cyst
CHAOS

- Previously thought to be lethal
- More recent series suggest that survival is possible despite ascites/hydrops
- Treatment is EXIT to secure airway
  - At CHOP: 12 fetuses with CHAOS
  - 6 terminated, IUFD, multiple congenital anomalies died
  - 6 survived to delivery
  - 5 of 6 survived neonatal period
  - 4 survived the first year

EXutero Intrapartum Treatment

- Surgical excision and airway established on placental bypass prior to delivery
- Potentially compromised ability to transition from intrauterine to extrauterine environment
  - Neck/mediastinal mass (lymphangioema, goiter, teratoma)
  - CHAOS
  - Large lung lesion (CCAM, bronchial atresia)
  - Severe micrognathia
- 1996-2011 - 75 cases
  - Overall survival 85%
EXIT Procedure

**Essential Components**

- Maintenance of uteroplacental bloodflow for prolonged periods after hysterotomy
- Complete uterine relaxation
- Maintenance of intrauterine volume
- Maternal homeostasis and hemostasis
- Control of the membranes
- Placental/cord avoidance
EXIT Procedure

**CHOP Team**

Maternal Anesthesiologist (1 ± fellow)
Fetal Anesthesiologist (1)
Scrub nurses (2)
Circulating nurses (2)
Airway cart nurse (1)
Level one operator
Sonologist (MFM)
Echocardiographer (Cardiology)
Surgeons - Operating Surgeon
  Airway surgeon
  Assistant surgeon
  2nd Assistant - (fetal position)
Neonatology Team
  Neonatology Attending
  Neonatology NPs
  Surgical APNs
  2nd OR team
  ECMO team
Maternal Team
  Uterine Closure
  Establish Uterine Tone
  Monitor for Hemorrhage
Postoperative Care
EXIT Procedure:
Specialized Equipment
EXIT at CHOP
62 EXIT procedures from 1996 - 2009

- Intrapartum Complications: 10% (6/60)
  - Atony and abruption
  - Atony and hemorrhage
  - Abruption and hemorrhage
  - Hemorrhage
  - Focal dehiscence of previous uterine scar

- Placental Abruption: 5% (3/60)
- Blood Transfusion: 8.3% (5/60)
- No uterine artery ligations, B-lynch sutures or hysterectomies
- No maternal deaths

Moldenhauer et al. AJOG 2009;201(6)Supp:S164-S165
## Uterine Incision

<table>
<thead>
<tr>
<th>Type</th>
<th>Number (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>LUS</td>
<td>36/60 (60)</td>
</tr>
<tr>
<td>Classical</td>
<td>4/60 (7)</td>
</tr>
<tr>
<td>Fundal</td>
<td>3/60 (5)</td>
</tr>
<tr>
<td>Posterior</td>
<td>6/60 (10)</td>
</tr>
<tr>
<td>LUS with Extension</td>
<td>11/60 (18)</td>
</tr>
</tbody>
</table>

EXIT at CHOP

Moldenhauer et al. AJOG 2009;201(6)Supp:S164-S165
Sacroccocygeal Teratoma
Sacrococcygeal Teratoma (SCT)

Teratoma - “Monstrous Tumor”
Fetal SCT - Pathophysiology

- AV “Steal”
- High Output Cardiac Failure (Hydrops)
Fetal SCT - Surgical Resection
22 weeks – no hydrops
Conclusions

• There is no perfect prenatal diagnostic imaging modality

• Ultrasound and MRI are complimentary in optimizing diagnosis
Thank You