Spina Bifida

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Introduction

Spinal Dysraphisms include:

• **Open spinal dysraphism**
  – Myelomeningocele

• **Closed spinal dysraphism**
  – With a subcutaneous mass
    • Meningocele
  – Without a subcutaneous mass
    • Posterior spina bifida
Spina Bifida

A: Normal
B: Spina Bifida Occulta
C: Meningocele
D: Myelomeningocele
Epidemiology

- Incidence of spina bifida:
  - 3.4-4.1/10,000 live births.\(^1\)
- Hispanics have higher risk, African Americans have lower risk.\(^2\)
- Highest rates are found in Ireland, Great Britain, Pakistan, India, and Egypt.

1 MMWR weekly, 5/04
2 Frey & Hauser, Epilepsia 2003
Maternal factors

• Age: small risk.
• Obesity: BMI > 29 doubles risk.
• Socioeconomic status: Higher rates in low SES populations.
• Smoking: protective.
• Diabetes mellitus: increased risk.
• Alcohol and recreational drugs: no change in risk

Frey & Hauser, Epilepsia, 2003
Environmental Factors

Exposures associated with development of NTDs

- Valproic acid
- Carbamazepine
- Clomiphene citrate
- Hyperthermia
- Folic acid antagonists
- Folic acid deficiency
# Epidemiology

## Estimated Incidence of Neural Tube Defects Based Upon Risk Factors for a United States Population

<table>
<thead>
<tr>
<th>Population</th>
<th>Incidence/1000 live births</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Mother as reference</strong></td>
<td></td>
</tr>
<tr>
<td>General incidence</td>
<td>1.4 to 1.6</td>
</tr>
<tr>
<td>Women with diabetes mellitus</td>
<td>20</td>
</tr>
<tr>
<td>Women on valproic acid in the first trimester</td>
<td>10 to 20</td>
</tr>
<tr>
<td><strong>Fetus as reference</strong></td>
<td></td>
</tr>
<tr>
<td>One affected sibling</td>
<td>15 to 30</td>
</tr>
<tr>
<td>Two affected siblings</td>
<td>57</td>
</tr>
<tr>
<td>Parent with neural tube defect</td>
<td>11</td>
</tr>
<tr>
<td>Half sibling with neural tube defect</td>
<td>8</td>
</tr>
<tr>
<td>First cousin affected (child of mother's sister)</td>
<td>10</td>
</tr>
<tr>
<td>Other affected first cousins</td>
<td>3</td>
</tr>
</tbody>
</table>

† Adapted from data in Main, DM, Mennuti, MT. Obstet Gynecol 1986; 67:1.
Folate

- Unknown mechanism of action.
- Mixed results on primary protective effect.
- Recurrent NTDs reduced by 75%.\(^1\)
- 26% decline in NTDs post folate fortification in the US.\(^2\)

1 Frey & Hauser, Epilepsia 2003
2 MMWR 5/04
Normal Embryology

Formation of neural plate

- Notochord-induced formation of neural plate.
- Neural plate cells proliferate and come together midline.
- Somites and mesoderm form laterally.

Kaufman, BA, Ped Clin N Am, 2004
Normal Embryology

Neural tube formation

- Midline fusion of neural plate.
- Ectoderm undergoes disjunction, fusing to form future skin.

Kaufman, BA, Ped Clin N Am, 2004
Normal Embryology

Neural tube formation

• Fusion of neural folds begins in cervical region and proceeds in both cephalad and caudal directions.
• Normal delay in fusion at both ends so that neuropores form open communication between the lumen of the neural tube and amniotic cavity.
• Closure of cranial neuropore occurs 25d post-conception, caudal end 2d later.
• NTDs from failure of this closure occur between 3rd and 4th wk post-conception (5th and 6th wk GA).
Prenatal Diagnosis: Serum markers

Alpha fetoprotein (AFP) is synthesized by the fetal yolk sack, GI, and liver.

Maternal serum AFP in 2nd trim to assess for NTDs.
- 16-18wks GA optimal.
- Does not detect closed spina bifida.
- Increase is also a marker for other anomalies: aneuploidy, ventral wall defect, tumor
- Measured in Multiples of Means (MoMs). Values > 2.0 to 2.5 abnormal.
Prenatal Diagnosis: Ultrasound

1. Lemon sign: biconcave shape of the frontal calvarium.
2. Banana sign: posterior convexity of the cerebellum as it herniates through foramen magnum.
3. Splaying of vertebral ossification centers.
4. Cystic sac.
Spina Bifida Occulta

- Midline defect in vertebral bodies (usually L5 or S1) without protrusion of the spinal cord or meninges.
- May have tuft of hair over affected area.
- Most people are asymptomatic.
- Incidental finding or no clinical significance.

http://www.mercksource.com/ppdocs/us/common/dorlands/dorland/images/fig_s_0040.jpg
Spina Bifida Occulta: case

A 28-year-old male developed lower back pain at the age of 19 while lifting heavy boxes.

The pain got progressively worse through the years; described as shooting pain down leg. Associated with paresthesias.
Spina Bifida Occulta

Spina bifida occulta at the S1 segment associated with an elongated L5 spinous process (white arrow).

On CT, elongated spinous process shown to be impinging on spinal cord (image not shown).

www.chiroweb.com/archives/20/14/19.html
Meningocele

- Herniation of a CSF-filled sac lined by dura mater and arachnoid through a posterior spina bifida.
- Not assoc with hydrocephalus or neurologic defects.

http://www.geneva-link.ch/dupuisim/Images/MENINGOCELE.GIF
Meningocele

Sac overlying vertebral bodies (arrow)
Myelomeningocele

• Due to primary failure of neural tube to close or secondary reopening of closed neural tube.

• Herniation of spinal cord through defect.

• Assoc with Chiari II malformation.

http://www.geneva-link.ch/dupuisim/Images/MYELOMENINGOCELE.GIF
Myelomeningocele

Patient 1: large cystic sac in lumbar region
Myelomeningocele

Patient 2: cystic sac
Chiari II malformation

- Assoc. with myelomeningoceles.
- Caudal displacement of vermis, brainstem, and 4th ventricle.
- Lemon and banana sign on US.
  - Lemon = biconcave frontal calvarium.
  - Banana = cerebellar herniation through foramen magnum.

Gabbe: Obstetrics – Normal and Problem Pregnancies, 4th ed
Chiari II malformation
Chiari II malformation

Patient B: lemon-shaped calvarium
Other anomalies

• NTDs assoc with other abnormalities:
  – Hydrocephalus
  – Clubfeet
  – Congenital heart disease
  – Structural defects of airway, GI tract, ribs
  – Renal malformations
Other anomalies

**Hydrocephalus**

Due to blockage of 4\textsuperscript{th} ventricle by Chiari malformation or associated aqueductal stenosis.
Twenty-one postmenstrual weeks fetus showing hydrocephaly. All four images were obtained using transvaginal sonography. A, Posterior coronal section showing the very large posterior horns of the lateral ventricles. In the posterior fossa a relatively normal appearing cerebellum (between arrows) and the cisterna magna are seen. B, A more anterior section than A and also showing dilation of the lateral ventricles (LV). C, An axial section showing the large ventricles, the dangling choroid plexus, and the very thin cortical mantle. D, Oblique section showing the hydrocephaly. T = thalamus; CP = choroid plexus. Courtesy of Ana Monteagudo, MD.
Other anomalies

Multicystic dysplastic kidney
Our Patient

- MH is a 43 yo G3P0 at 17 and 3/7 wks GA
  - Cystic structure in L spine with splaying of pedicles and bilat club feet on U/S.
6/7 Ultrasound

Cystic structure in L spine (arrowheads): meningocele
Club feet
Our Patient

- Fetal MRI and amniocentesis on 6/14 at 18 & 3/7 wks GA

- Small bleb seen in the region of L-spine, but due to early GA and fetal motion, actual NTD itself not well visualized.
- Intracranial anatomy nl.
- No Chiari malformation.
- No ventriculomegaly.
- Club feet seen.
Our Patient

- Ultrasound on 7/19

Cyst over approx L4 measuring 0.9cm by 0.9cm.
Our Patient

• Ultrasound on 7/19

Transverse through meningocele (arrow)
Our Patient

- Awaiting results of amniocentesis and amniotic fluid AFP evaluation.
References

• Hochberg and Stone. Etiology, prenatal diagnosis, and prevention of neural tube defects. *Up-to-date*.
• Fishman and Villarreal. Myelomeningocele. *Up-to-date*
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