Neural tube defects

A gamut of CNS anomalies that result from a failure of the normal process of neurulation. Frequently the lesions, which may be found anywhere in the cranium or spine, are open allowing cerebrospinal fluid (CSF) to pour into the amniotic cavity. Such lesions will cause elevation of maternal serum and amniotic fluid levels of alpha fetoprotein (AFP). Routine prenatal testing that reveals elevated MS-AFP levels is an indication for a Level II sonogram. When the sonogram fails to identify the cause for the elevated AFP, amniocentesis is recommended. Overall incidence varies but in the United States it is 1.5 - 2.0 per 1,000 births with lower incidence in Asians and Blacks.

<table>
<thead>
<tr>
<th>Terminology</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rachischisis</td>
<td>Completely open spine with herniation and/or destruction of the spinal cord.</td>
</tr>
<tr>
<td>Spina bifida occulta</td>
<td>Failure of closure of the vertebral column without any associated external abnormalities. Rarely associated with neurological complications.</td>
</tr>
<tr>
<td>Spina bifida apperta</td>
<td>Failure of closure of the vertebral column with apparent external abnormalities and neurological complications.</td>
</tr>
<tr>
<td>Meningocele</td>
<td>Herniation of the meninges through a schisis defect.</td>
</tr>
<tr>
<td>Meningomyelocele</td>
<td>Herniation of the meninges and neural tissue through a schisis defect.</td>
</tr>
</tbody>
</table>

Spina Bifida

General term for lack of closure of vertebral column. Prognosis depends on severity of lesion and is poorest in infants who have total paralysis below the lesion, kyphosis, hydrocephalus, and associated congenital defects.

Pathology:
- Presence of a meningomyelocele, a protruding sac which may contain meninges and spinal cord.
- May occur anywhere along the spinal column but lumbar region is most common.
- Variable in size and content.
**Spina Bifida**

**SONOGRAPHIC FINDINGS:**

**TRANSVERSE**
- Splaying of posterior ossification centers into a "U" or "V" shape.
- When sac is intact, a cystic structure may be seen extending from the back. Appearances include: a small, simple cystic structure, a cyst with *septations and/or solid matter.*

**SAGITTAL**
- Splaying of parallel lateral ossification centers.
- Associated with hydrocephalus and Arnold-Chiari II malformation

**INTRACRANIAL**
- **LEMON SIGN:** overlapping of the frontal bones creating a lemon shaped fetal head
- **BANANA SIGN:** effacement of the cisterna magna due to downward displacement of the cerebellum
- **ARNOLD-CHIARI** Type II MALFORMATION: displacement of the lower cerebellum into the upper cervical column with dislocation of the medulla oblongata and 4th ventricle
**Anencephaly**

Most common congenital anomaly of the CNS. Occurs 1:1,000 births. 4% chance of recurrence in subsequent pregnancy. More common in females than in males.

**Pathology**
- Absence of cranial vault and cerebral hemispheres (functioning brain).
- Portions of midbrain and brain stem may be present.
- Other features include bulging eyes (macrophthalmia), (thick tongue) macroglossia and a very short neck.

**Embryology**
- Occurs at 2-3 weeks when anterior neuropore fails to fuse.
- Open defect is covered by angiomatous stroma rather than skin or bone.
- Another theory suggests that an excess of cerebrospinal fluid causes disruption of the normally formed cerebral hemispheres.

**Sonographic Findings:**
- Major portions of cranium and intracranial structures are absent.
- Orbits and face are usually present.
- Fetal head should be identifiable by 12 weeks, definitively by 15 weeks at latest.
- Associated polyhydramnios (40-50%).
**ENCEPHALOCELE**
Occurs 1:2,500 births. Occipital encephaloceles are most frequent in Western world.

**PATHOLOGY**
- Bony defect in calvarium with herniation of brain and/or meninges outside of cranium.
- Classified as occipital, frontal, and/or parietal.
- Hydrocephalus is present in 80% of occipital lesions; Spina bifida in 7-15%

**EMBRYOLOGY** results from failure of surface ectoderm to separate from neuroectoderm leading to defect in cranium.

**SONOGRAPHIC FINDINGS:**
- Mass extending from calvarium. May be totally cystic, cystic with septations (meningocele), or may contain brain (encephalomeningo-myelocele).
- Cranial defect is occasionally seen.
- Cranial cavity appears small if a significant portion of brain is herniated.
- Associated with hydrocephalus, polyhydramnios.
INIENCEPHALY
A complex neural tube developmental abnormality characterized by:
- Exaggerated spinal lordosis
- Cervical rachischisis
- Occipital encephalocele

Other fetal anomalies are associated with iniencephaly in 84% of cases and include:
- Anencephaly
- Cephalocele
- Hydrocephaly
- Cyclopia
- Absence Of Mandible
- Cleft Lip And Palate
- Cardiovascular Anomalies
- Diaphragmatic Hernia
- Single Umbilical Artery
- Omphalocele
- Gastrochisis
- Situs Inversus
- Polycystic Kidneys
- Arthrogryposis
- Clubfoot

SONOGRAPHIC FINDINGS:
- Exaggerated, hyperextension of the fetal head
- Demonstration of cervical spina bifida
- Restriction of normal fetal head movements

Demonstration of external anomalies in a stillborn
Sagittal sonographic correlation