Fetal Brain Advanced: Midline Anomalies are EASY

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Important Sonographic Clue to the Diagnosis Midline Anomalies

- Absence of the normal cavum septi pellucidi (CSP) during the routine anatomical survey using axial scan.
- May be indicative of a HPE, AGCC or SOD

Normal CSP

Absent CSP

HPE

AGCC

SOD

The Normal CSP in Axial Plane

A fluid filled structure between the leaves of the septum pellucidum

Do NOT Confuse the Fornix for the CSP

Absence of the CSP in the Axial Plane

- The next step is to obtain the Coronal plane
- Most useful in the evaluation of the CSP
**Evaluation of the CSP: Coronal Plane**

- If the fetus is in a cephalic presentation, TVS is the easiest way to get this plane.
- Scan using the anterior fontanelle.

**Absence of the CSP in the Axial Plane**

- In addition to the coronal plane, a median plane of the brain is indicated.
- Since, abnormalities of corpus callosum are among the most common midline anomalies and have absent CSP.

**Best planes to Image Corpus Callosum**

- Transabdominally: Transfontal view
- Transvaginally: Median plane

**Corpus callosum & Cavum Septi Pellucidi**

- The rostrum (beak), genu (knee), corpus (body) and the splenium (tail).
- Cavum septi pellucidi & vergae.

**Corpus Callosum & Cavum Septi Pellucidi**

- CC develops in an anterior to posterior fashion. Exception: Rostrum & anterior genu which develop last.

- Best time to image is after 20 weeks.
Corpus Callosum & Pericallosal Arteries

• Before 18 weeks, using only gray scale, the corpus callosum may not be evident.
• However, using color Doppler the pericallosal artery is a proof of its presence.

Pericallosal Arteries

Corpus callosum Cavum Septi Pellucidi

• The presence of normal pericallosal arteries predicts normal development of the corpus callosum.

Corpus Callosum & Cavum Septi Pellucidi

• The development of corpus callosum is closely associated with that of the CSP.
  • There cannot be a CSP without a covering corpus callosum.
  • However, a corpus callosum can be present in the absence of the CSP such as in septal agenesis as the result of SOD.

Midline developmental anomalies or Disorders of Prosencephalic Development

• Prosencephalic development, is the major event following neurulation.
  • Its development peaks between 2nd - 3rd month of the pregnancy.
  • Three major events:
    • Formation, cleavage and midline prosencephalic development.
    • Failure of this developmental sequence results in a spectrum of pathologies affecting the forebrain as well as the face.
Disorders of Midline Development: AGCC, ASP, SOD

- Basic malformation: is complete or partial absence of the main commissural fiber tracts that connects the cerebral hemispheres.
- Depending on the region affected it will result in a specific abnormality

<table>
<thead>
<tr>
<th>REGION AFFECTED</th>
<th>DISORDER</th>
</tr>
</thead>
<tbody>
<tr>
<td>Commissural plate</td>
<td>Agenesis of corpus callosum</td>
</tr>
<tr>
<td>Commissural and commissural plates</td>
<td>Agenesis of corpus callosum and/or septo-optic dysplasia</td>
</tr>
<tr>
<td>Commissural, chiasmatic, and hypothalamic plates</td>
<td>Septo-opto-hypothalamic dysplasia</td>
</tr>
</tbody>
</table>

Agenesis of the corpus callosum

- Most common of the midline anomalies
- Spectrum of abnormality:
  - Complete or partial agenesis
  - Dysgenesis (abnormal shape)
  - Hyperplasia/hypoplasia (increased/decreased thickness)

Agenesis of the Corpus callosum: Axial plane - Indirect Sonographic Findings

- Non-visualization of the CSP
- Upwardly displaced 3rd ventricle

Agenesis of the Corpus callosum: Axial plane - Direct Sonographic Findings

- Widened interhemispheric space
- Parallel and 'tear-drop' shaped ventricles (colpocephaly)

Agenesis of the Corpus callosum: Coronal Plane - Indirect Sonographic Findings

- Lateral ventricles are parallel, slit-like and crescent shape
- 3rd ventricle elevated, dilated, and abnormally shaped

Agenesis of the Corpus callosum: Median Plane - Direct Sonographic Findings

- Complete or partial absence
- Color Doppler absence of the normal pericallosal artery
• No CSP
• 'Tear-drop' shape & parallel lateral ventricles
• Colpocephaly

• Widely separated, vertically oriented lateral ventricles ('viking’s helmet’ sign)
• Interhemispheric fissure meets 3rd ventricle

• Radial gyri & sulci on the median surface ('sunburst sign').
• Seen only when sulci & gyri develop

• Absent cingulate gyrus (CG), radial array of sulci in a ‘Sunburst’ appearance

• Absent corpus callosum & CSP

• AGCC may be associated with midline cysts & lipoma
  - Type 1 cyst: communicates with the ventricular system
  - Type 2 cyst: does not communicate with the ventricular system and are associated with cortical malformations

• Rare, may interfere with the growth & development of the corpus callosum between 11-20th week.
• The degree of the anomaly is in relation to the size & location of the lipoma.
AGCC With Interhemispheric Cyst and Posterior Fossa Abnormality

Dandy-Walker Malformation

Agenesis Septi Pellucidi

- Rare ~ 2-3/100,00 can be partial or complete; isolated or associated with other brain anomalies e.g SOD, HPE
- Axial scan non-visualization of the normal CSP
- Next step coronal plane

Fused anterior horns; communicating with the 3rd ventricle

Septo-optic Dysplasia (aka De Morsier syndrome)

- Rare condition ~ 1.9 to 2.5 per 100,000 births (EUROCAT)
- Is part of the continuum of midline developmental disorders.
- Defective development of the commissural and chiasmatic plates, often also involve the hypothalamic plate
- Triad of abnormality
  - Absence of the septi pellucidi/Abnormal corpus callosum
  - Optic nerve underdevelopment
  - Pituitary hypoplasia (diagnosed postnatally)

Septo-optic dysplasia: Sonographic Findings

- Axial plane- Non-visualization of the cavum septi pellucidi
- Coronal plane – needed for the diagnosis
  - Corpus callosum is seen
  - Absence of the septi pellucidi
  - Fused and down pointing frontal horns (key to the diagnosis)

Septo-optic Dysplasia: Sonographic Findings

- Median plane – the corpus callosum will appear normal

TVS 3D of SOD simultaneous visualization of 3 planes
Isolated Agenesis Septi Pellucidi vs. SOD: Tough Diagnosis

- Both have absent cavum septi pellucidi (CSP) and fused anterior horns.

Isolated ASP vs. SOD: Evaluating the Optic Nerves & Chiasma - Tough Diagnosis

- To diagnose SOD is important to evaluate the optic nerves and optic chiasm.
- 3D ultrasound can help image the optic nerves.

Holoprosencephaly

- Basic Malformation: Failure of horizontal, transverse, and sagittal cleavage of the prosencephalon.
- The original classification by De Myer described 3 histological types:
  - Alobar
  - Semilobar
  - Lobar
- Fourth type: Middle interhemispheric variant
- Other more subtle types: septopreoptic variant and interhypothalamic adhesion

Alobar HPE: Ultrasound Findings

- Most amenable to US diagnosis
- Single ventricle
- Absent midline structures
- Corpus callosum, CSP, Falx
- Fusion of thalamus & basal ganglia
- Posterior fossa ± normal
- Hydrocephaly is present in most cases; usually with a dorsal cyst.
- 92% have dorsal cysts
The Face Predicts the Brain: Alobar HPE

- Spectrum of facial anomalies
- The most severe facial abnormalities are associated with the most severe brain abnormalities.
  - The face predicts the brain
- However, severe brain abnormality is not always associated with severe facial defects.
- ~10% of alobar HPE is NOT associated with "significant" facial abnormalities
Alobar HPE: Ultrasound Findings
- Non-visualization of the ‘butterfly sign’
- Detection rates for HPE is 100%

First Trimester

Semilobar Holoprosencephaly
- US findings similar to Alobar
- Failure of separation of the anterior hemisphere
  - Absent: falx, CSP, anterior portion of the corpus callosum
- The posterior portion of the interhemispheric fissure is present.
  - Well developed posterior horns
- Microcephaly is common
- 28% dorsal cyst of the 3rd ventricle due to fusion of the thalamus and impaired flow of CSF

Semilobar HPE: Ultrasound Findings
- Single ventricle, absent midline structures
- Face: Hypotelorism, midline CLP
- Microcephaly
- Posterior horns well developed

Lobar HPE: Ultrasound Findings
- Findings are subtle
- Axial scan absent CSP
- Fusion of the FH of the LV; wide communication with the 3rd ventricle.
- Corpus callosum: Normal or hypoplastic
- Falx is present; IHF is fully formed; thalami are not fused

First Trimester

Alobar HPE: Facial Ultrasound Findings
- Abnormal profile
- Proboscis
- Single orbit

Semilobar HPE: Ultrasound Findings
- Coronal plane
- Absence of CSP
- Fused frontal horns
- Fornices are fused form a thick fascicle (bright echogenic dot)

Lobar HPE: Ultrasound Findings
- Coronal plane
- Absence of CSP
- Fused frontal horns
- Fornices are fused form a thick fascicle (bright echogenic dot)
Lobar HPE: Color Doppler Findings

• Sagittal plane the anterior cerebral artery (ACA) displaced anteriorly to lie directly underneath the frontal bone 'snake under the skull sign'
• Cerebral hemispheres nearly fully separated
• Microcephaly is common; 9% dorsal cyst of the 3rd ventricle

Lobar HPE vs. Isolated ASP vs. SOD: Tough diagnosis

• All three have absent cavum septi pellucidi (CSP) and fused anterior horns

To summarize...

• The CSP
  • Is an important landmark in the antenatal sonographic evaluation of the brain
  • Is part of the of the 2nd trimester anatomy scan
  • It’s presence is a marker for normal development of the forebrain
  • Non-visualization in the axial plane is associated with midline brain abnormalities
  • Next step is the coronal plane
  • Corpus callosum is best seen in the sagittal plane

Beware of Secondary disruption of the CSP resulting from hydrocephaly may mimic absent CSP