GOTTESFELD-HOHLER MEMORIAL FOUNDATION
ULTRASOUND EDUCATION AND RESEARCH IN OB/GYN

28th Annual OB/GYN Ultrasound Update for Clinical Practice

Fetal Brain Advanced: Midline Anomalies are *EASY*

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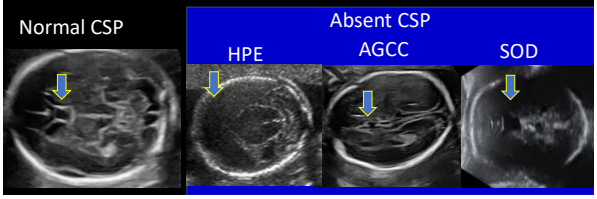
1

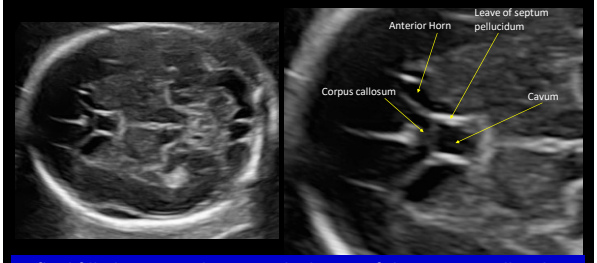
Important Sonographic Clue to the Diagnosis Midline Anomalies

- *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



2

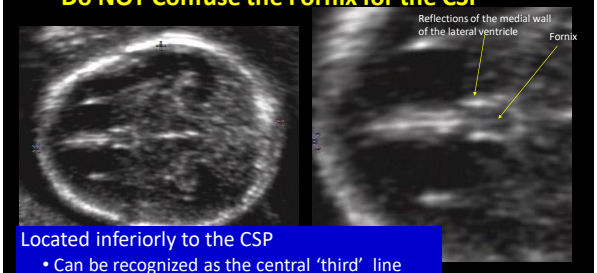
The Normal CSP in Axial Plane



A fluid filled structure between the leaves of the septum pellucidum

3

Do NOT Confuse the Fornix for the CSP



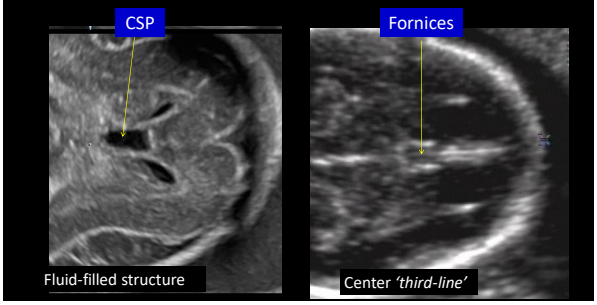
Located inferiorly to the CSP

- Can be recognized as the central 'third' line

CALLEN PW, ET AL . Columns of the fornix, not to be mistaken for the cavum septi pellucidi on prenatal sonography. J Ultrasound Med 2008;27:25-31.

4

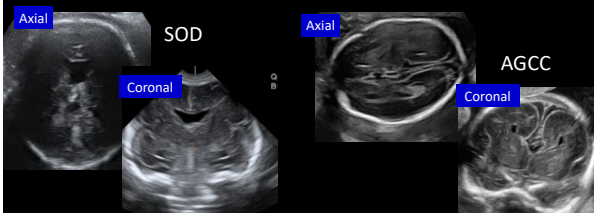
CSP vs. Fornices: Axial Plane



5

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*



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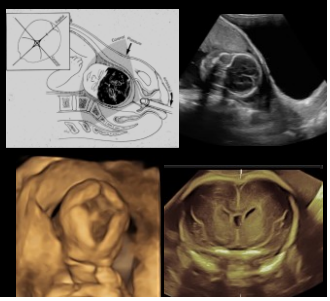
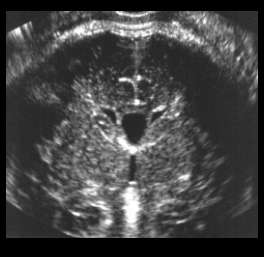
Evaluation of the CSP: Coronal Plane

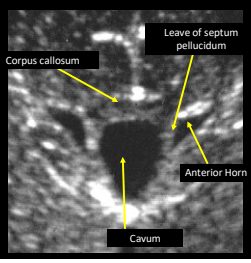
- If the fetus is in a cephalic presentation TVS is the easiest way to get this plane
- Scan using the anterior fontanelle



7

Evaluation of the CSP: Coronal Plane

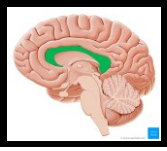


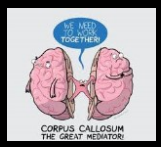


8

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.





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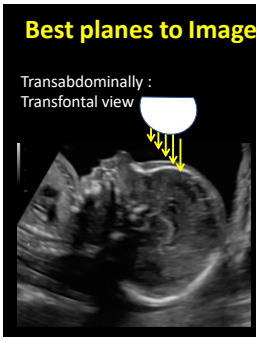
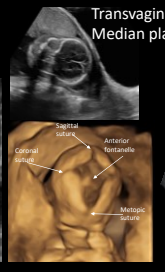
Best planes to Image Corpus Callosum

Transabdominally :
Transfontal view

Transvaginally:
Median plane





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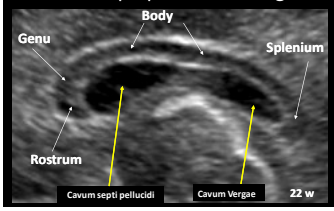
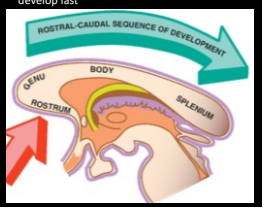
Corpus callosum & Cavum Septi Pellucidi

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



CC develops in a anterior to posterior fashion
Exception: Rostrum & anterior genu which develop last



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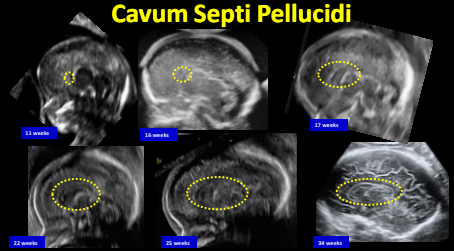
Corpus Callosum & Cavum Septi Pellucidi

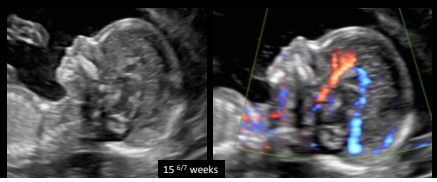


Best time to image is after 20 weeks

12

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



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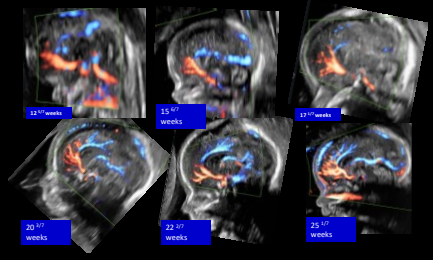
Pericallosal Arteries



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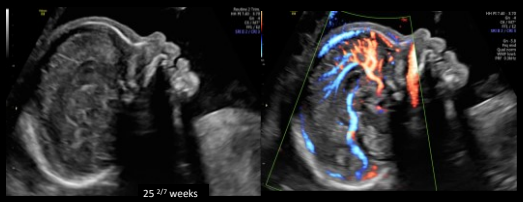
Corpus callosum Cavum Septi Pellucidi

- The presence of normal pericallosal arteries predicts normal development of the corpus callosum



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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

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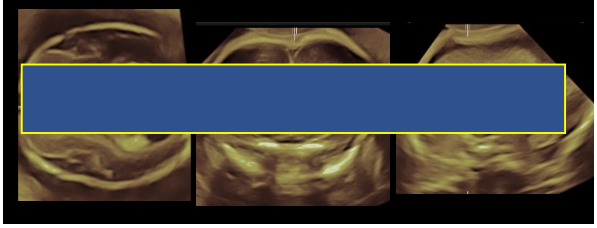
Midline developmental anomalies or Disorders of Prosencephalic Development



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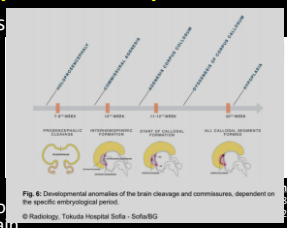
Midline developmental anomalies or Disorders of Prosencephalic Development

- 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 o pathologies affecting the forebrain as well as the face.



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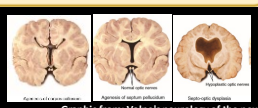
Disorders of Midline Development: AGCC, ASP, SOD

- 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 2.2 Disorders of Midline Prosencephalic Development

REGION AFFECTED	DISORDER
Commissural plate	Agenesis of corpus callosum and/or septum pellucidum
Commissural and chiasmatic plates	Septo-optic dysplasia
Commissural, chiasmatic, and hypothalamic plates	Septo-optic–hypothalamic dysplasia



Graphic from: Volpe's neurology of the newborn
Joseph J. Volpe editor.; Joseph J Volpe,2018, Chapter 2

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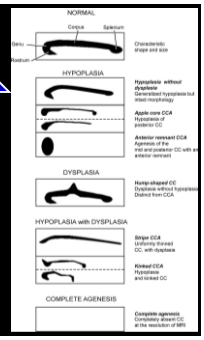
Agenesis of the corpus callosum

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



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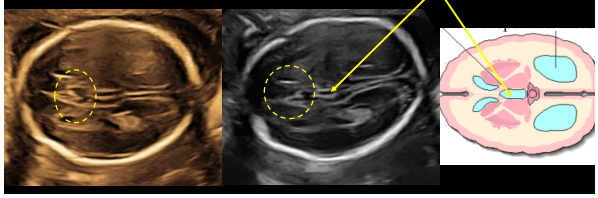
Agenesis of the Corpus callosum: Axial plane- Indirect Sonographic Findings

Non-visualization of the CSP

Upwardly displaced 3rd ventricle



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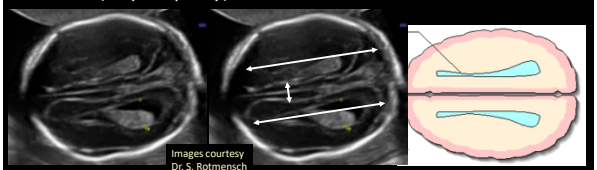
Agenesis of the Corpus callosum: Axial plane- Indirect Sonographic Findings

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



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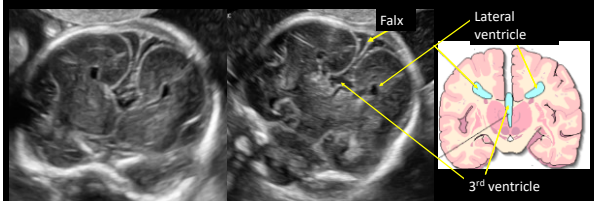
Agenesis of the Corpus callosum: Coronal Plane - Indirect Sonographic Findings

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



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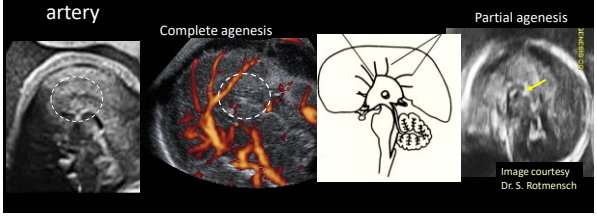
Agenesis of the Corpus callosum: Median Plane- Direct Sonographic Findings

- Complete or partial absence
- Color Doppler absence of the normal pericallosal artery



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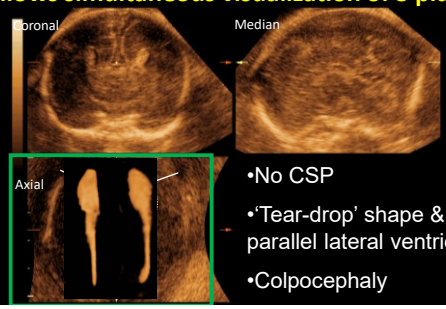
3D allows simultaneous visualization of 3 planes



- No CSP
- 'Tear-drop' shape & parallel lateral ventricles
- Colpocephaly

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3D allows simultaneous visualization of 3 planes

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

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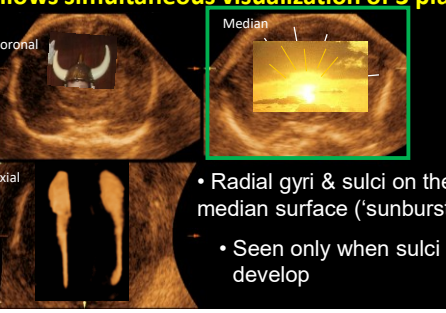
3D allows simultaneous visualization of 3 planes



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

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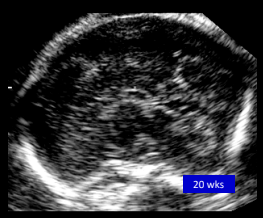
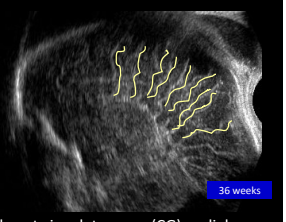
Direct Sonographic Signs: Median Plane 2nd vs. 3rd Trimester



•Absent corpus callosum & CSP



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

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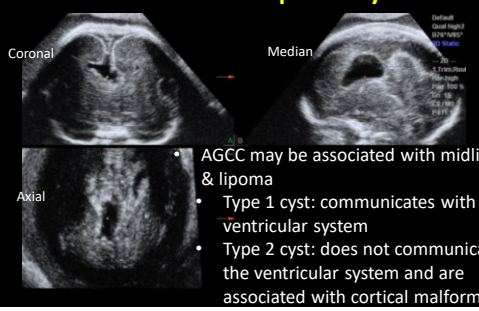
3D AGCC With Interhemispheric Cyst



- 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

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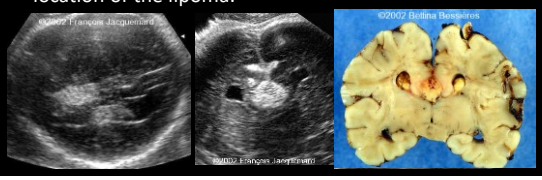
Pericallosal Lipoma

- Rare, may interfere with the growth a development of the corpus callosum between 11-20th week.
- The degree of the anomaly is in relation to the size and location of the lipoma.



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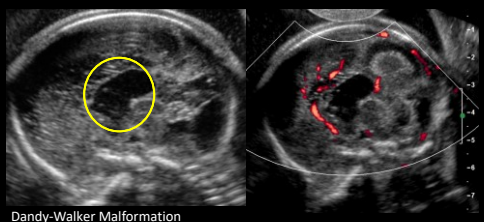
AGCC With Interhemispheric Cyst and Posterior Fossa Abnormality



Dandy-Walker Malformation

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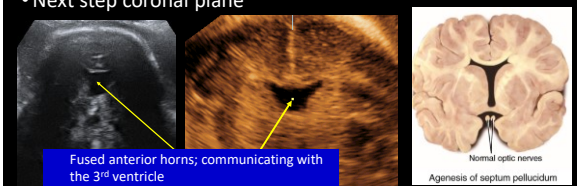
Agenesis Septi Pellucidi

- 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



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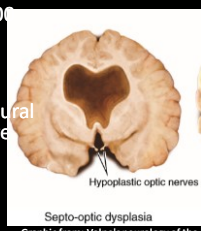
Septo-optic Dysplasia (aka De Morsier syndrome)

De Morsier, 1956

- Rare condition ~ 1.9 and 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

Graphic from: Volpe's neurology of the newborn
Joseph J. Volpe editor.; Joseph J Volpe,2018, Chapter 2

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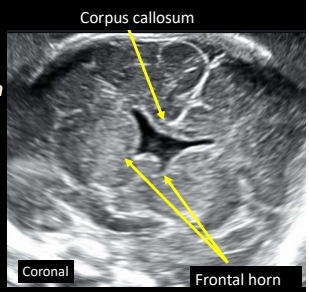
Septo-optic dysplasia: Sonographic Findings

- 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)*



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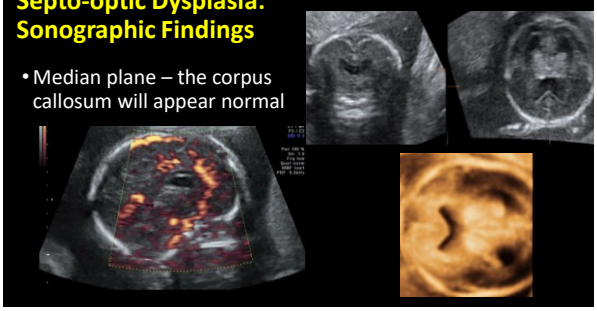
Septo-optic Dysplasia: Sonographic Findings

- Median plane – the corpus callosum will appear normal



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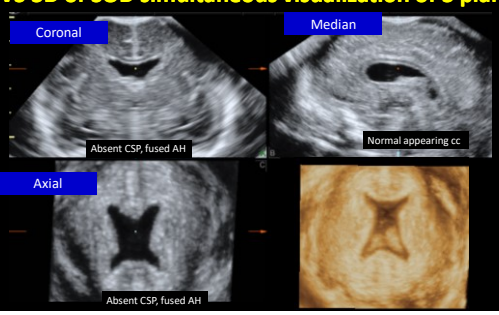
TVS 3D of SOD simultaneous visualization of 3 planes



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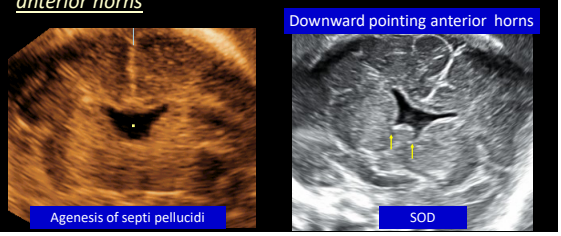
Isolated Agenesis Septi Pellucidi vs. SOD: Tough Diagnosis

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



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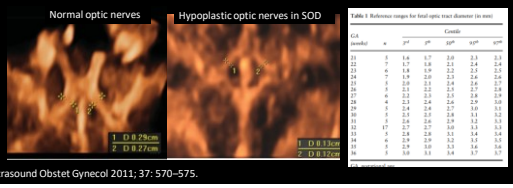
Isolated ASP vs. SOD: Evaluating the Optic Nerves & Chiasma- Tough Diagnosis

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



Ultrasound Obstet Gynecol 2011; 37: 570–575.

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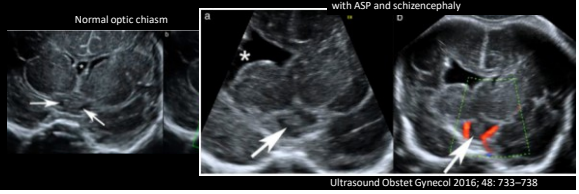
Isolated ASP vs. SOD: Evaluating the Optic Nerves & Chiasma- Tough Diagnosis

- The optic chiasm can be imaged and evaluated by ultrasound. However, a normal measurement does not exclude SOD



Ultrasound Obstet Gynecol 2016; 48: 733–738

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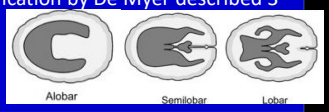
Holoprosencephaly

- *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

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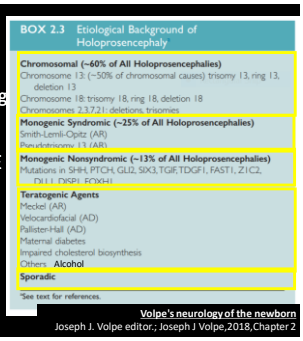
Holoprosencephaly

- Spectrum of disorders
  - Without clear distinction among the subtypes
- Alobar: Most common & severe accounts for 2/3 of HPE
- Prevalence:
  - 1:10,000 liveborn
  - 1:250 abortus
- 80% have craniofacial abnormalities
- Etiology



BOX 2.3 Etiological Background of Holoprosencephaly

Chromosomal (~60% of All Holoprosencephalies)
Chromosome 13: (~50% of chromosomal causes) trisomy 13, ring 13, deletion 13
Chromosome 18: trisomy 18, ring 18, deletion 18
Chromosomes 2,3,7,21: deletions, trisomies

Monogenic Syndromic (~25% of All Holoprosencephalies)
Smith-Lemli-Opitz (AR)
Pseudotrisomy 13 (AR)

Monogenic Nonsyndromic (~13% of All Holoprosencephalies)
Mutations in SHH, PTCH, GLI2, SIX3, TGIF, TDGF1, FAST1, ZIC2, DLL, DSRL, FOXH1

Teratogenic Agents
Meckel (AR)
Velocardiofacial (AD)
Pallister-Hall (AD)
Maternal diabetes
Impaired cholesterol biosynthesis
Others: Alcohol

Sporadic

*See text for references.

Volpe's neurology of the newborn
Joseph J. Volpe editor.; Joseph J Volpe,2018,Chapter 2

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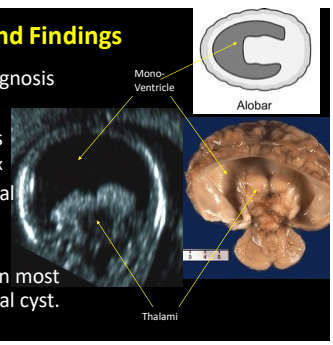
Alobar HPE: Ultrasound Findings

- 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



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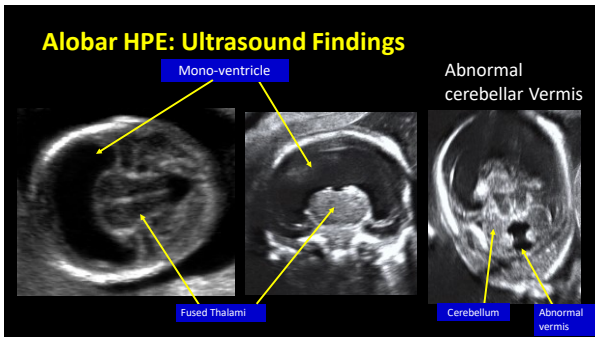
Alobar HPE: Ultrasound Findings



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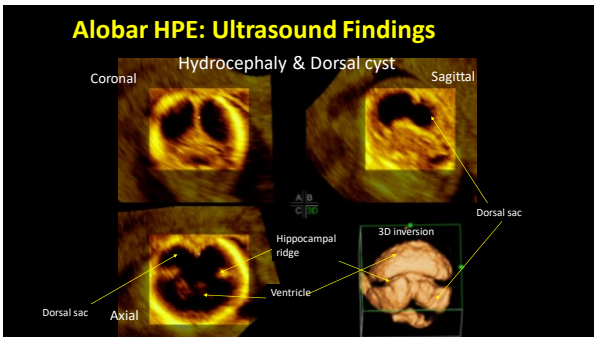
Alobar HPE: Ultrasound Findings



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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

Abnormal Face

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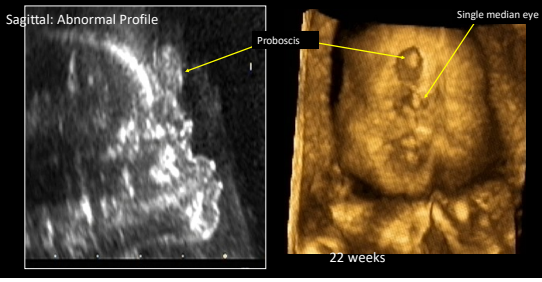
Alobar HPE: Facial Ultrasound Findings



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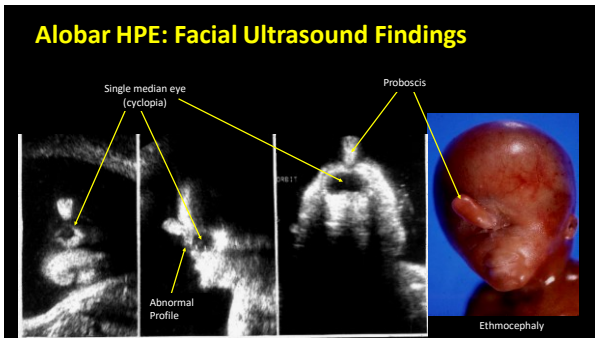
Alobar HPE: Facial Ultrasound Findings



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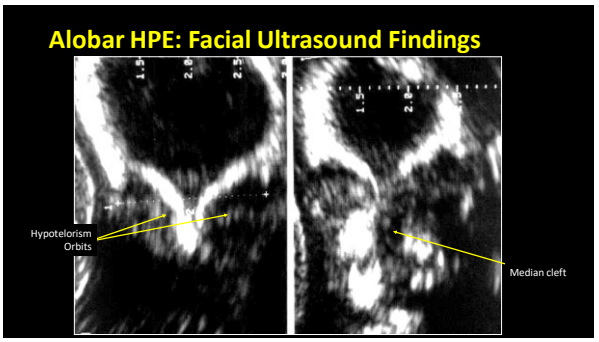
Alobar HPE: Facial Ultrasound Findings



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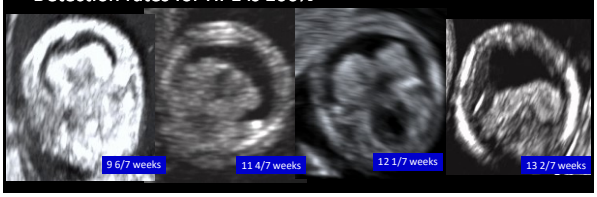
First Trimester

Alobar HPE: Ultrasound Findings

- Non-visualization of the 'butterfly sign'
- Detection rates for HPE is 100%



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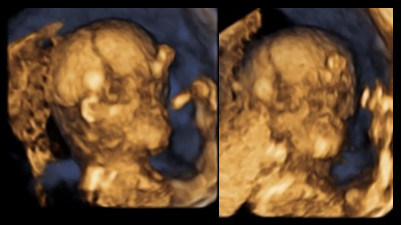
First Trimester

Alobar HPE: Facial Ultrasound Findings

- Abnormal profile
- Proboscis
- Single orbit



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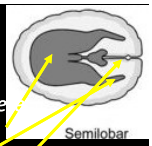
Semilobar Holoprosencephaly

- US findings similar to Alobar
- Failure of separation of the *anterior hemisphe*
  - 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



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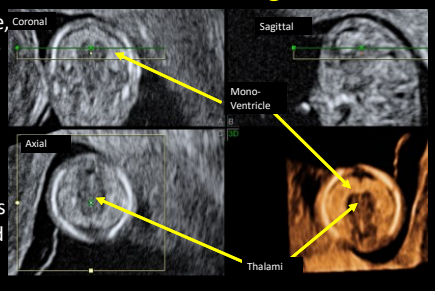
Semilobar HPE: Ultrasound Findings

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



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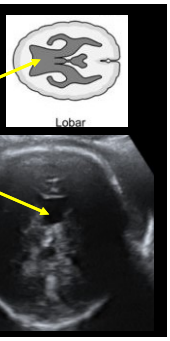
Lobar HPE: Ultrasound Findings

- 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



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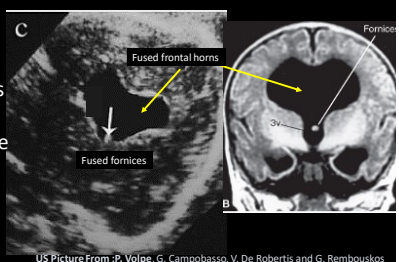
Lobar HPE: Ultrasound Findings

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



US Picture From :P. Volpe, G. Campobasso, V. De Robertis and G. Rembouskos Disorders of prosencephalic development *Prenat Diagn 2009; 29: 340–354.*

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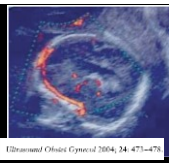
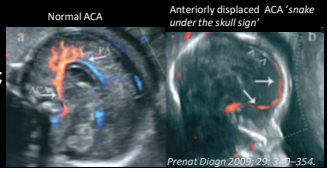
Lobar HPE: Color Doppler Findings

- 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





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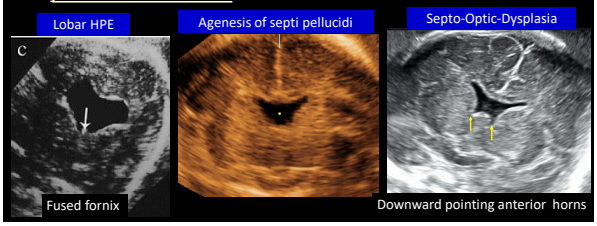
Lobar HPE vs. Isolated ASP vs. SOD: Tough diagnosis

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



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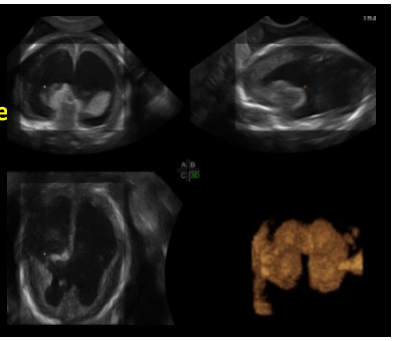
Beware of *Secondary* disruption of the CSP resulting from hydrocephaly may mimic absent CSP



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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

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