Congenital Posterior Fossa Malformations

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Agenda

• Our Patient: History, Findings, and Diagnosis
• Facts about Dandy-Walker Complex
• Dandy Walker Malformation
• Dandy Walker Variant
• Mega Cisterna Magna
• Posterior Fossa Cyst
• Potential Causes
• Associated Anomalies
• Prognosis
• Our Patient: Outcome
• Summary
Our Patient: Clinical History

- Female G4P1 presenting for full fetal survey at 21 wks
  - h/o gestational diabetes
  - no family h/o genetic disorders
Our Patient: Full Fetal Survey at 21 Wks

Axial U/S

- Dilated 4th ventricle
- Posterior fossa cyst communicating with 4th ventricle
Our Patient: Summary of Findings

• Dilated Fourth Ventricle
• Absence of cerebellar vermis
• Enlargement of posterior fossa
• Findings consistent with a posterior fossa malformation
Our Patient: Differential Diagnosis

• Congenital CNS Anomalies
  – Chiari Malformation
  – Encephalocele
  – Holoprosencephaly
  – Septo-optic dysplasia
  – Dandy-Walker Malformation
Menu of Tests

• Fetal Ultrasound
• Fetal MRI

• Which is better?
MRI vs. U/S

- Study at Brigham and Women’s Hospital compared diagnosis of fetal anomalies with MRI vs. U/S
- Compared diagnosis of 27 fetuses with anomalies
- 57% of the cases involved CNS anomalies
- In 62% of those cases, both modalities correctly diagnosed the anomaly
- MRI provided additional information (such as agenesis of corpus callosum) in half of those cases
- Concluded that MRI may be value as an adjunct to U/S in evaluating CNS anomalies

Our Patient: Diagnosis

Findings on ultrasound were consistent with Dandy-Walker Malformation
Now that we have a diagnosis, let’s find out more about Dandy-Walker Complex
Dandy-Walker Complex

• Malformation of posterior fossa
  – Pathogenesis unknown but thought to be due to arrest of development of hindbrain around 7-10 week gestation

• Spectrum of disease that includes:
  – Dandy-Walker malformation
  – Dandy-Walker variant
  – Mega Cistern Magna
  – Posterior Fossa Arachnoid Cyst

• Occurs in 1:30,000 births

• Seen in 4-12% of all babies with hydrocephalus
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Dandy-Walker Malformation

- Triad of Malformations
  - Cystic dilation of fourth ventricle
  - Complete or partial agenesis of the cerebellar vermis
  - Enlarged posterior fossa with displacement of the tentorium and torcular and lateral sinus

- Diagnosed after 18 weeks – closure of cerebellar vermis should happen by that time
We now know how to recognize Dandy-Walker Malformations. Let’s look at some classic examples on Ultrasound and MRI
Companion Patient 2: Dandy-Walker Malformation on Ultrasound

Posterior fossa cyst communicating with dilated 4th ventricle

Courtesy of Dr. Debenedectis
Companion Patient 3: Dandy Walker Malformation on Ultrasound

Axial

Sagittal

Posterior fossa cyst communicating with dilated 4th ventricle

Courtesy of Dr. Debenedectis
Companion Patient 4: Dandy Walker Malformation On U/S and MRI

**Axial**

**T2/C-/Axial**

*Posterior fossa cyst communicating with dilated 4th ventricle*

Kolble N et al. Prenatal Diagnosis 2000; 20:318-327
Companion Patient 5: Dandy-Walker Malformation on MRI

- Dilated 3rd ventricle
- Posterior fossa cyst communicating with dilated 4th ventricle
- Agenesis of corpus callosum

Poe L B et al. Radiographics 1989; 9:801-826
Companion Patient 6: Dandy-Walker malformation on MRI

T2/C-/Sagittal

- Normal corpus callosum
- Posterior fossa cyst displacing cerebellum and tentorium

Levine D et al. Radiology 1999;211:609-617
Companion Patient 7: Dandy-Walker on MRI

- Posterior fossa cyst communicating with dilated 4th ventricle
- Ventriculomegaly

T2/C-/Coronal

T2/C-/Axial

Courtesy of Dr. Debenedectis
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Dandy-Walker Variant

– Cerebellar dysgenesis without enlargement of the posterior fossa
– Variable hypoplasia of the cerebellar vermis
– Better prognosis
Companion Patient 8: Dandy-Walker Variant on U/S

Axial

Cleft of cerebellar vermis

Ecker J L et al. Prenatal Diagnosis 2000;20:328-332
Companion Patient 9: Dandy-Walker variant on Ultrasound

Axial

Cleft of cerebellar vermis

Companion Patient 10: Dandy-Walker variant on MRI

T2/C-/Axial

Cleft of cerebellar vermis

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Mega Cisterna Magna

- Enlarged posterior fossa caused by enlarged cisterna magna
- Cisterna magna drains fourth ventricle
- Cerebellum and fourth ventricle are normal
- Treated with cisternotomy
Companion Patient 11: Mega Cisterna Magna on MRI

T1/C-/Sagittal

Mega Cisterna Magna visualized as a retrocerebellar fluid collection

Hamid HA. Egypt Journal of Human Genetics 2007;8:115-120.
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Posterior Fossa Arachnoid Cyst

- Retrocerebellar arachnoid cyst that displaces cerebellum anteriorly
- Causes mass effect on cerebellum and fourth ventricle
- Looks similar to Mega Cisterna Magna on MRI
  - Differentiate the two with ventriculogram (Mega Cisterna Magna communicates with subarachnoid space)
- Treated with marsupialization of cyst
Companion Patient 9: Posterior Fossa Arachnoid Cyst on MRI

T1/C-/Sagittal

Posterior fossa cyst causing compression of cerebellum and fourth ventricle

Hamid HA. Egypt Journal of Human Genetics 2007;8:115-120.
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Potential Causes

– Chromosomal Aneuploidy
– Sporadic Mutation
– Environment Exposure – Alcohol, Rubella
– Multifactorial etiology
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Associated Anomalies

• Intracranial
  – Ventriculomegaly (Seen in companion pt. 7)
  – Agenesis of Corpus Callosum (Seen in companion pt. 5)

• Extracranial
  – Cardiac Defects (most common)
  – Renal Abnormalities
  – Facial Abnormalities
  – Extremity abnormalities
Our Patient: Hydronephrosis on Ultrasound

Coronal

Right and Left kidney with dilated calyces
Companion Patient 10: Micrognathia associated with a Dandy-Walker variant on Ultrasound

Sagittal

Mernagh J R et al. Radiographics 1999;19:S229-S241
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Prognosis

• Variable and hard to predict
  – Some have mental and physical handicaps while others develop normally
  – ~25% mortality rate – mostly due to associated anomalies
  – Prognosis worse in children with chromosomal aneuploidy and associated anomalies

• Treatment aimed at control of hydrocephaly
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Our Patient: Outcome

- Amniocentesis was normal

- Patient decided not to terminate pregnancy and delivered baby at 35 wks

- Received further treatment at Children’s Hospital of Boston Neurology
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• Posterior Fossa Malformations exist in a spectrum of diseases collectively known as the Dandy-Walker complex
• Dandy-Walker Malformation is the most severe and involves a triad of malformations easily diagnosed with U/S and MRI
• Dandy-Walker complex is associated with many other congenital abnormalities and chromosomal aneuploidy which can be predictive of a worse prognosis
References

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