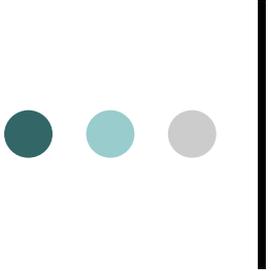


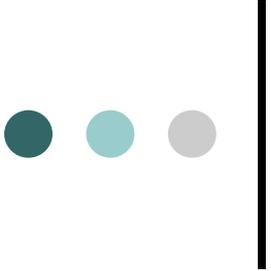
# Fetal Brain Anomalies

## Part II

指導 洪正修主任 楊明智主任  
陳志堯醫師

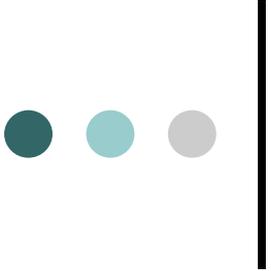
2007. 1. 19

- 
- Dandy-Walker continuum
  - Holoprosencephaly
  - Chiari II malformation



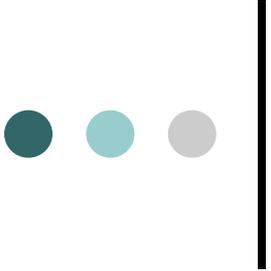
# Dandy-Walker Malformation

- Dandy-Walker continuum
- Dandy-Walker complex
- Dandy-Walker spectrum



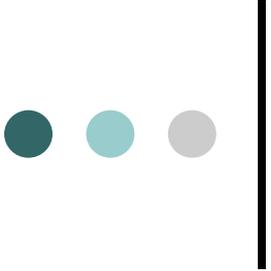
# History

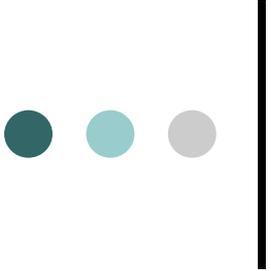
- In 1914, Blackfan and Dandy described some alterations of the posterior fossa as (a) **cystic dilatation of the fourth ventricle**, (b) **hypoplasia of the cerebellar vermis**, (c) **separation of the cerebellar hemispheres**, (d) **dilatation of the mesencephalic aqueduct** and (e) **absence of the mediate and lateral apertures** of the fourth ventricle.
- **These abnormalities were referred to as Dandy-Walker malformation. In the 1970 and 80 different definitions were introduced for similar abnormalities of posterior fossa and then the classic definition of the Dandy-Walker malformation.**

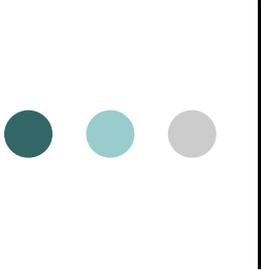


# Classification

- *Dandy-Walker malformation: increase of the posterior fossa, complete or partially agenesis of the cerebellar vermis, and a tentorium elevation.*

- 
- *Variant of Dandy-Walker: hypoplasia of the cerebellar vermis in different degrees with or without increase of the posterior fossa.*

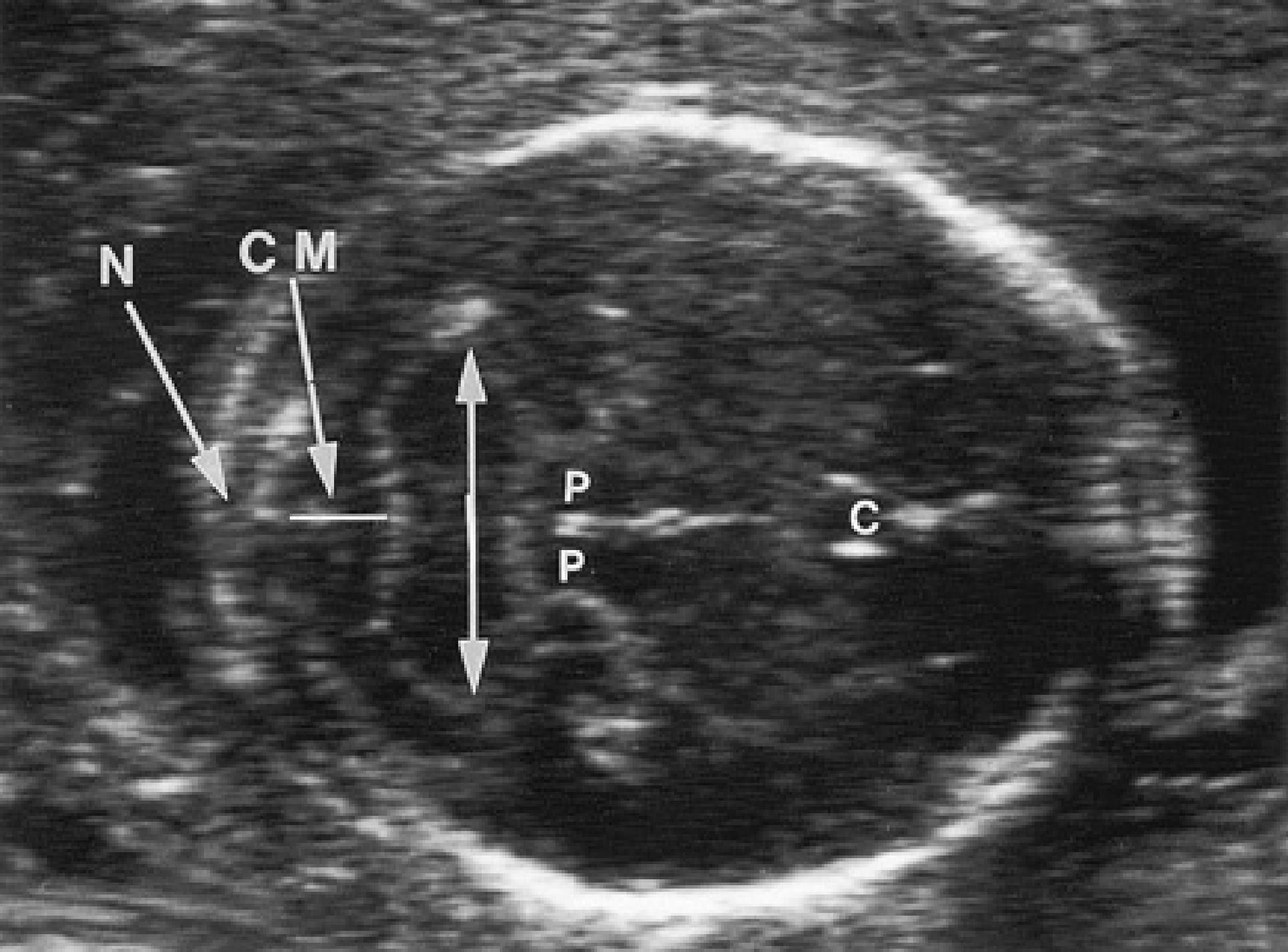
- 
- *Mega-cisterna magna: increase of the cisterna magna with integrity of the cerebellar vermis and fourth ventricle.*

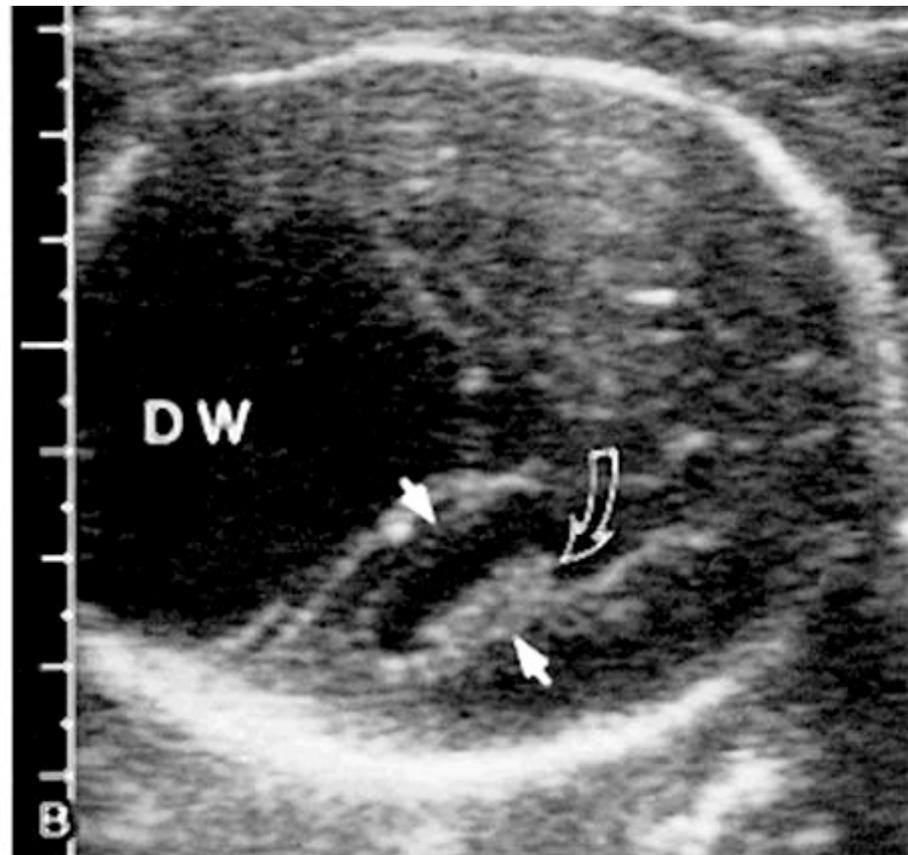
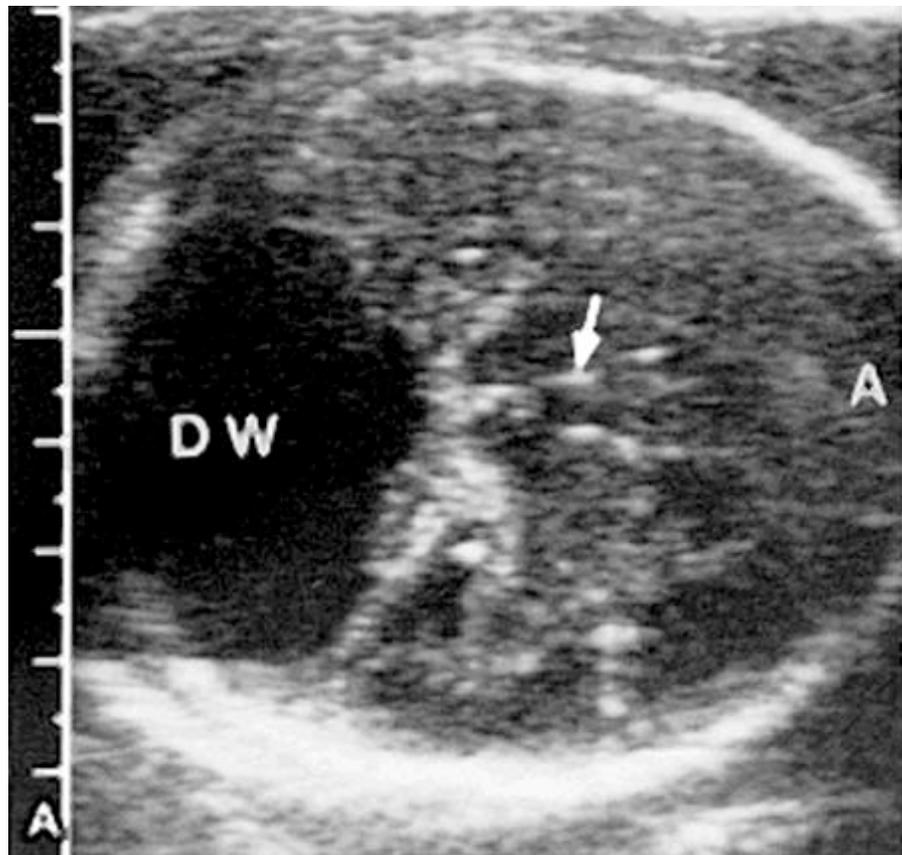


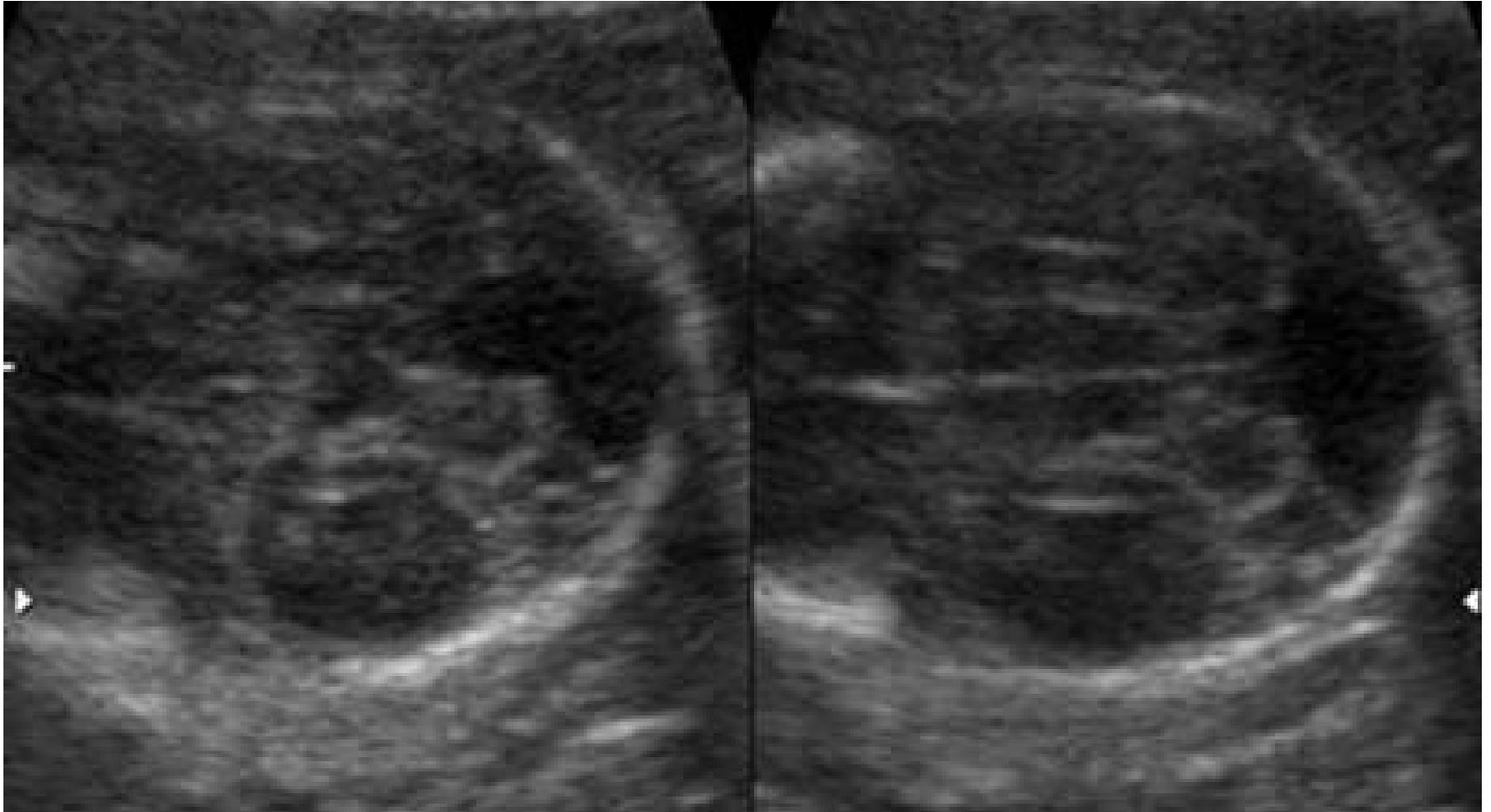
# Image Findings

- Large posterior fossa with big CSF cyst.
- 4<sup>th</sup> ventricle appears “open” and contiguous with PF cyst.
- Vermis severely hypoplastic or absent.

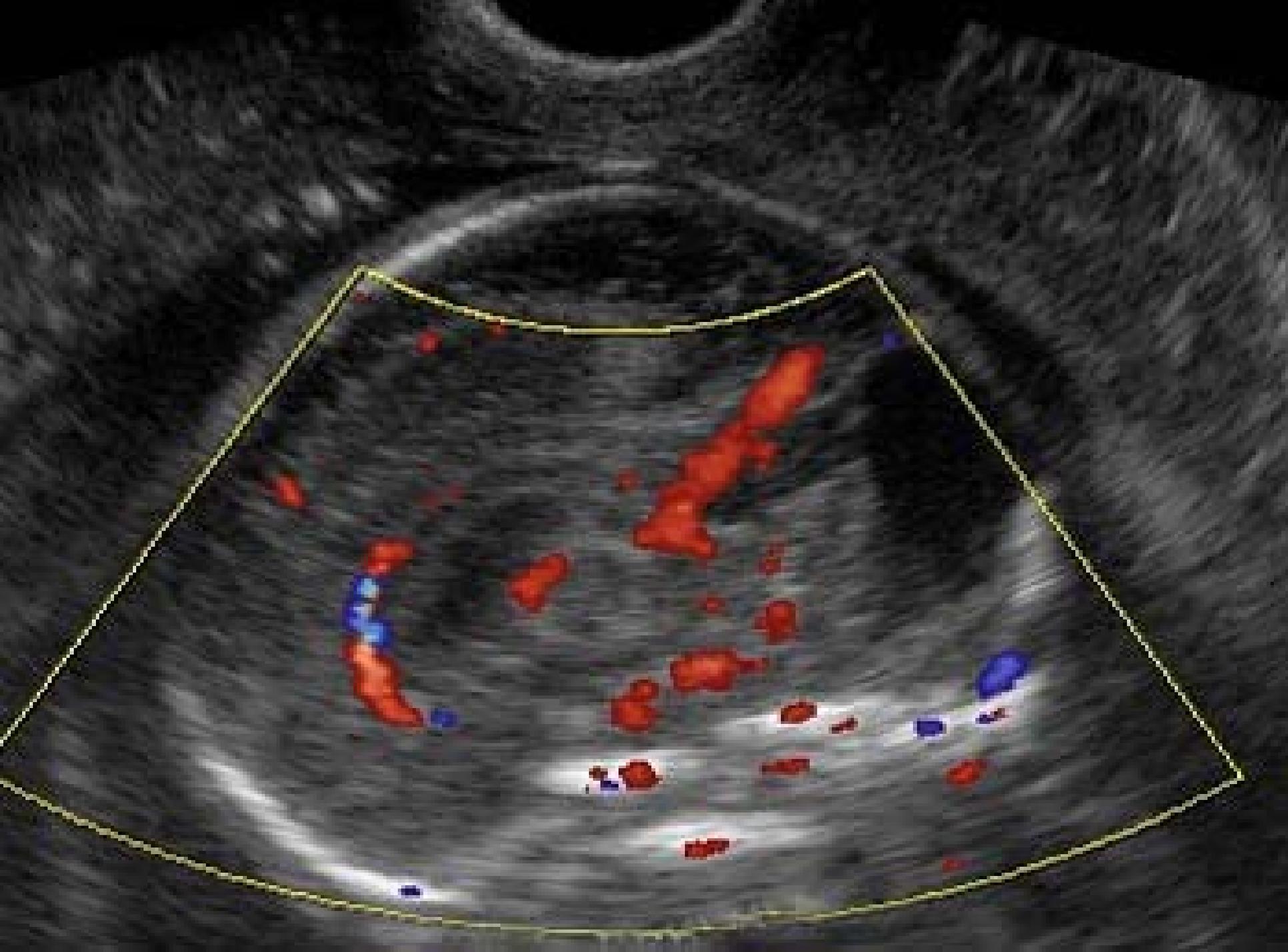


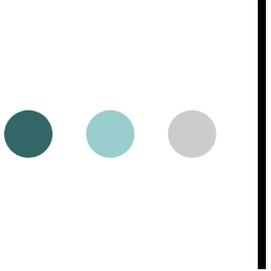












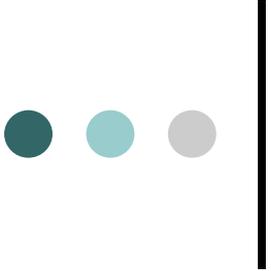
# *Associated Findings*

- CNS

- Ventriculomegaly
- Dysgenesis of corpus callosum
- Holoprosencephaly
- Encephaloceles, cephaloceles
- NTD

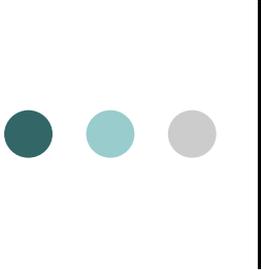
- Extra-cranial

- Cleft lip/palate
- Cardiac defects
- Polycystic kidneys



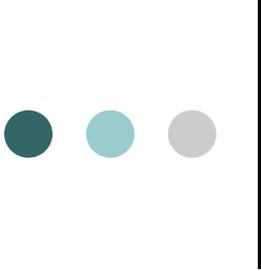
# Genetics

- The chromosomal abnormalities include trisomies 18, 13, 21 and Turner syndrome.



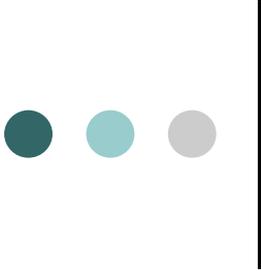
# Differential Diagnosis

- Mega Cisterna Magna
- Arachnoid Cyst (AC)
- Dandy-Walker Variant
- Persistent Blake Pouch Cyst
- Congenital Vermian Hypoplasia



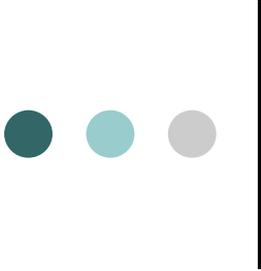
# Environmental Factor & Epidemiology

- Maternal diabetes.
- Alcohol.
- Early in utero infection.
  
- 1:25000~1:35000 live birth.
- 10% of infantile hydrocephalus.



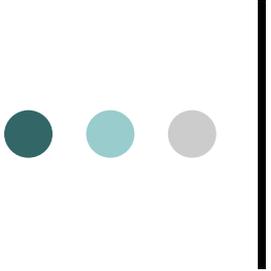
# Natural History & Prognostic

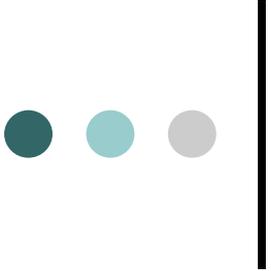
- The classic malformation is usually clinically manifest in the **first year** of life with symptoms of hydrocephaly and/or neurological symptoms. The **mortality is 24~40%** but with current neurosurgical techniques mortality has been decreasing.
- The intellectual development in the survivors is controversial. Intellectual deficit may be in the range of 40 to 70% of the cases. IQ: **35~50% normal**.
- The isolated defect seems to be associated with better prognostic.



# Management

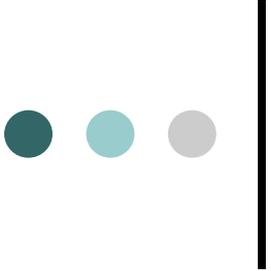
- There is no modification of standard obstetric management.
- All fetuses should be karyotyped.
- Cesarean delivery is indicated only if macrocrania is present.

- 
- Dandy-Walker continuum
  - **Chiari II malformation**  
Holoprosencephaly



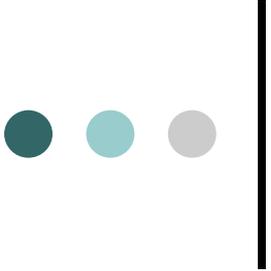
# *Abbreviations and Synonyms*

- Chiari II
- Arnold Chiari II
- Spinal bifida
- Open neural tube defect (ONTD)



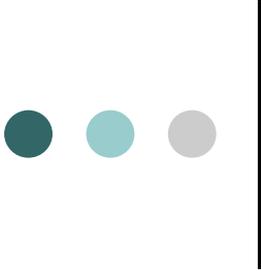
# History

- The Arnold-Chiari was first identified in 1883 by Cleland. It is characterized by the protrusions of hindbrain structures below the level of the foramen magnum. It can be associated with skeletal abnormalities and neurological dysfunction.
- The nickname “banana sign” has been applied to the deformity of the cerebellum.



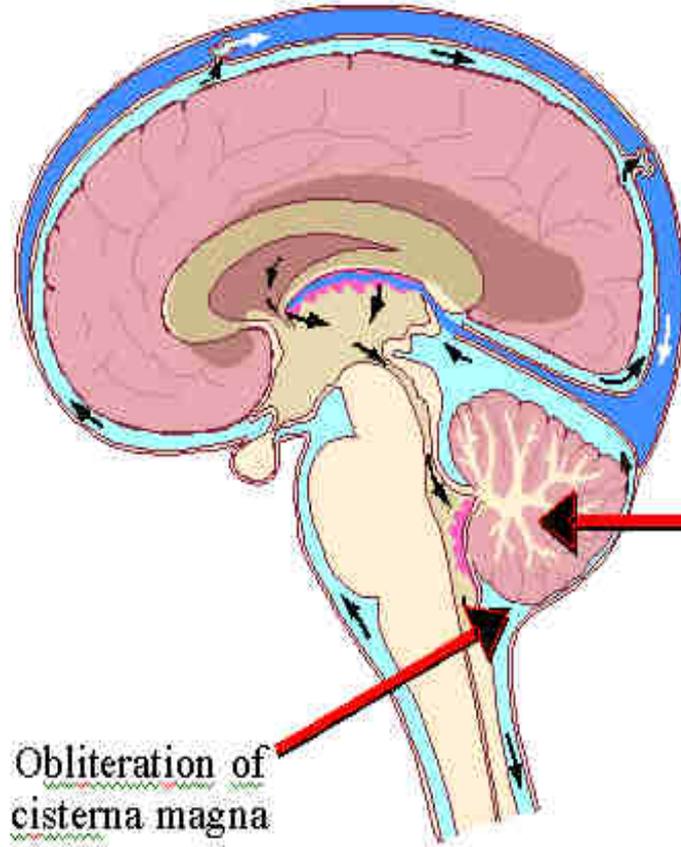
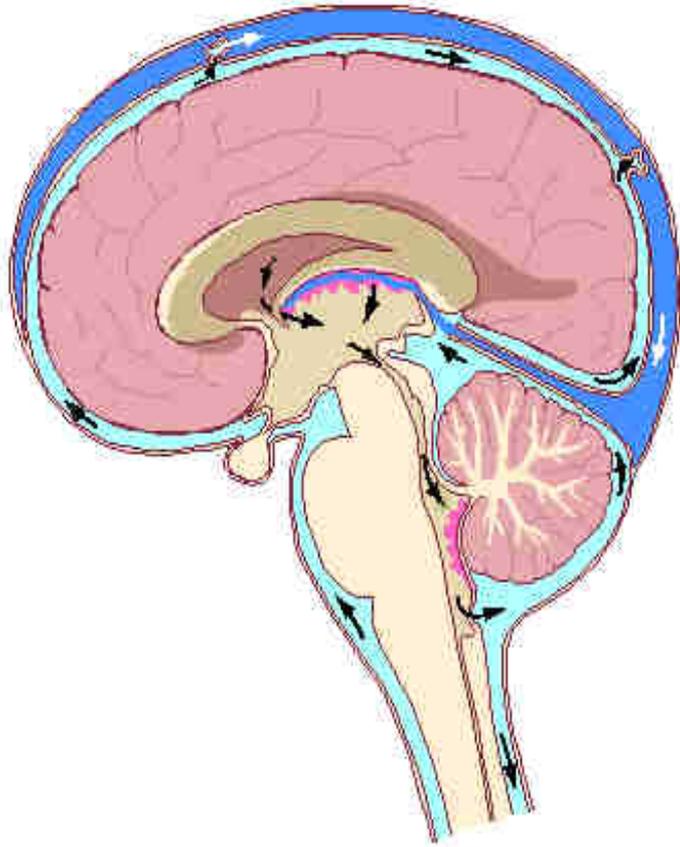
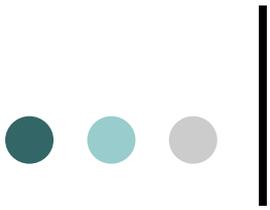
# Definitions

- Symptomatic hindbrain herniation
  - Cerebellar vermis herniate through foramen magnum
  - 4<sup>th</sup> ventricle displaced inside neural canal
  - Tentorium pulled downward
  - Medulla displaced inferiorly and kinked
- **Virtually 100% associated with ONTD**



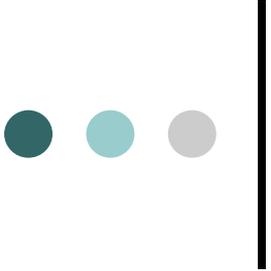
## *Three Types of Arnold Chiari*

- Type I: just a lip of cerebellum is downwardly displaced with the tonsils, but the fourth ventricle remains in the posterior fossa. This is mainly **an incidental CT discovery**.
- Type II: usually involved **in prenatal cases** and is a congenital deformity characterized by displacement of cerebellar tonsils, parts of the cerebellum, fourth ventricle, pons, and medulla oblongata through the foramen magnum into the spinal canal.
- Type III: **more severe form**, with large herniation of the posterior fossa content and myelomeningocele and hydrocephalus



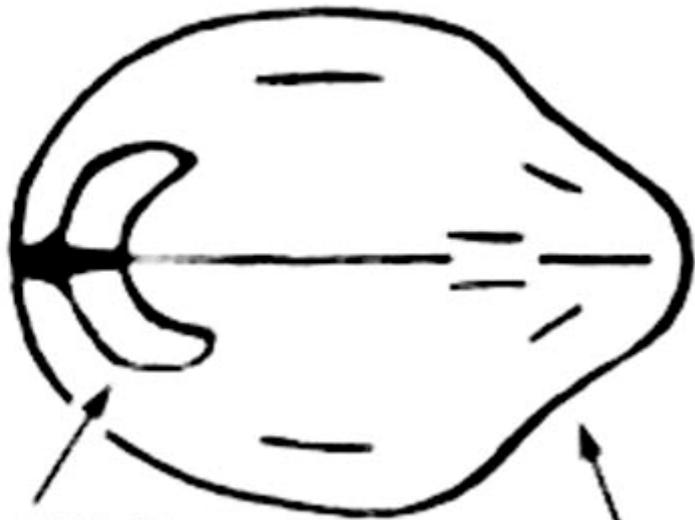
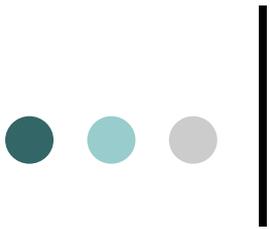
Downward displacement  
and hypoplasia of  
cerebellum

Obliteration of  
cisterna magna



# Imaging Findings

- Posterior fossa “banana sign”
  - Cisterna magna obliteration (most common)
  - CM is small or gone (<3mm)
- Calvarial “lemon sign”
- Ventriculomegaly (>10mm)
- ONTD (open NTD , spinal bifida)
  - 73% lumbar
  - 17% sacral
  - 9% thoracic
  - 1% cervical



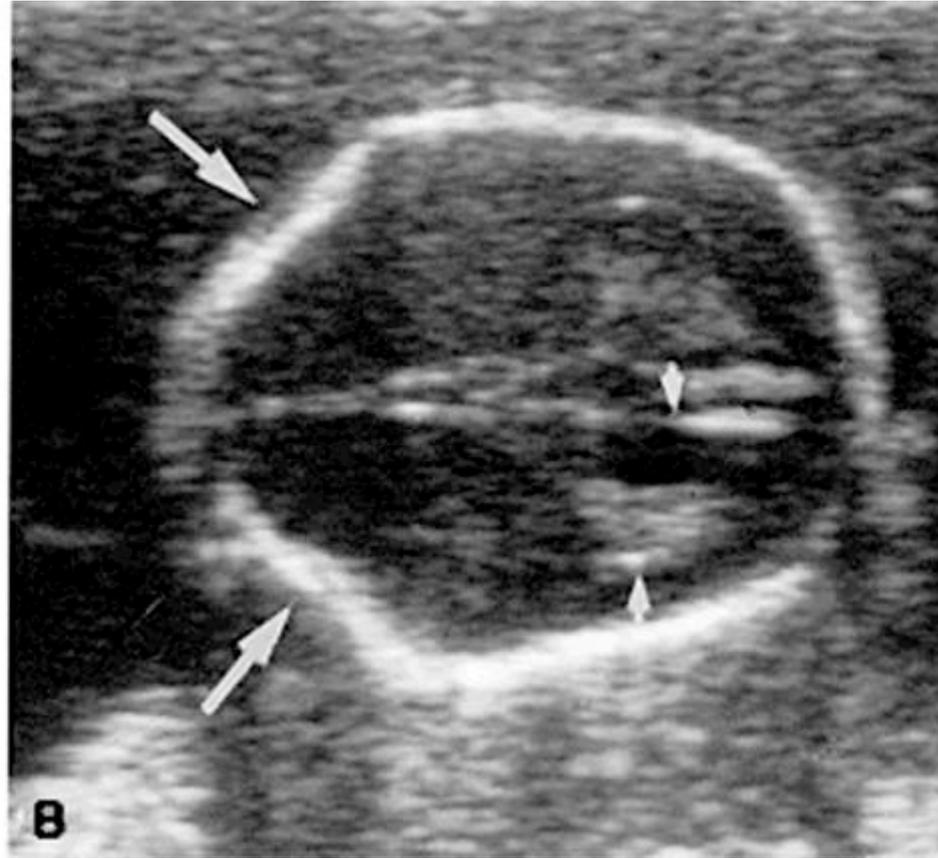
banana

sign

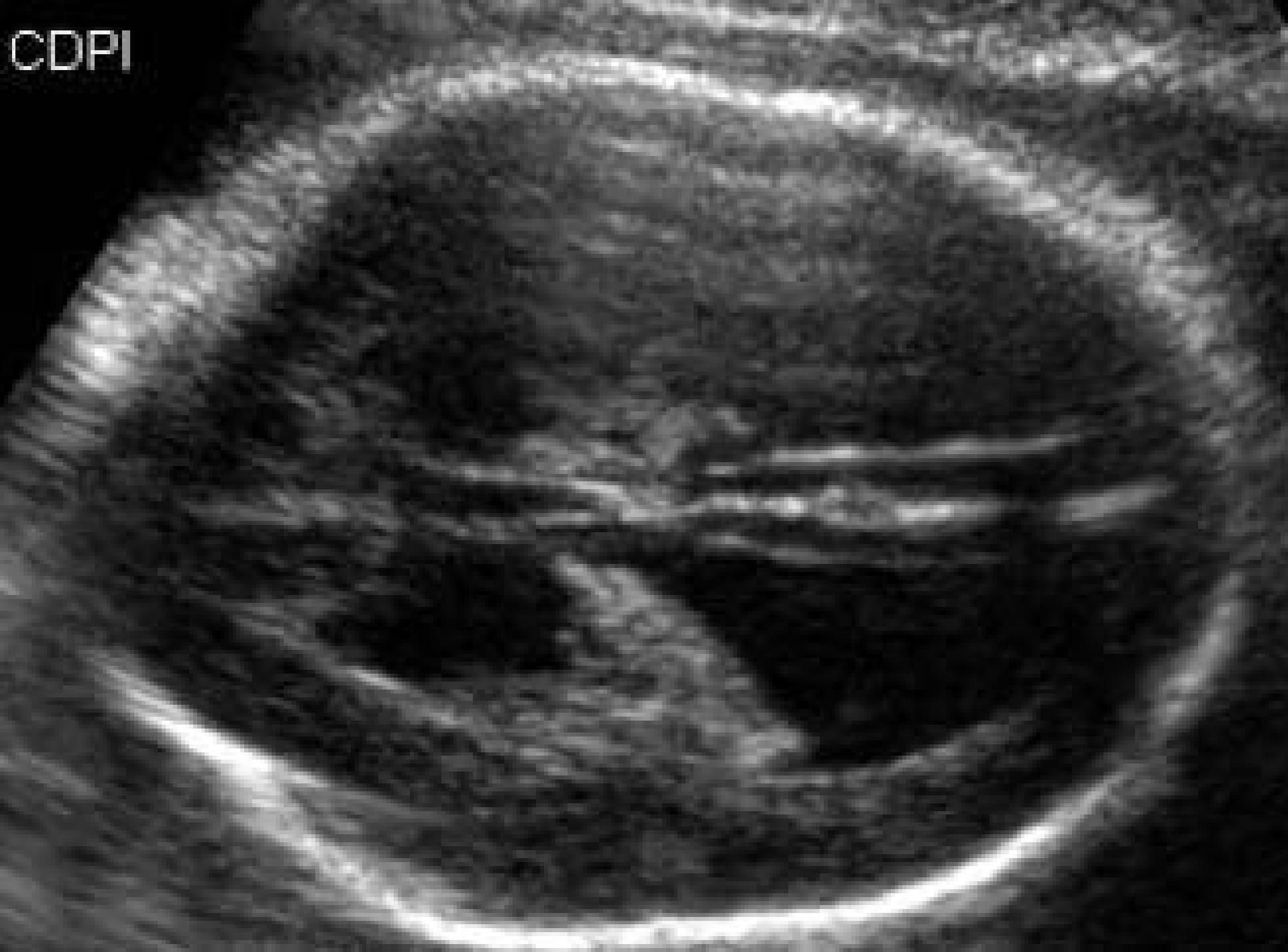
A

lemon

sign



B



CDPI

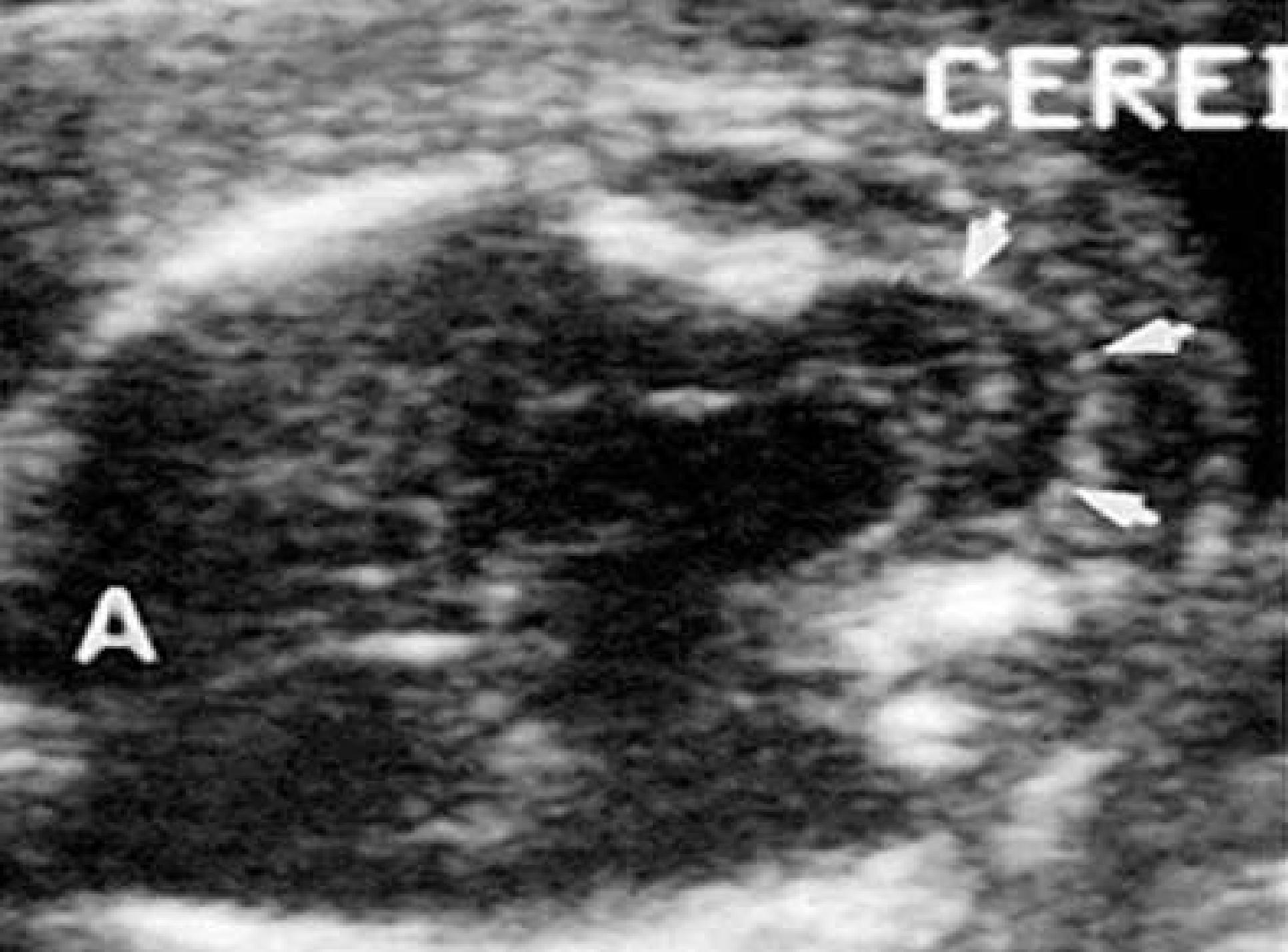


CDPI



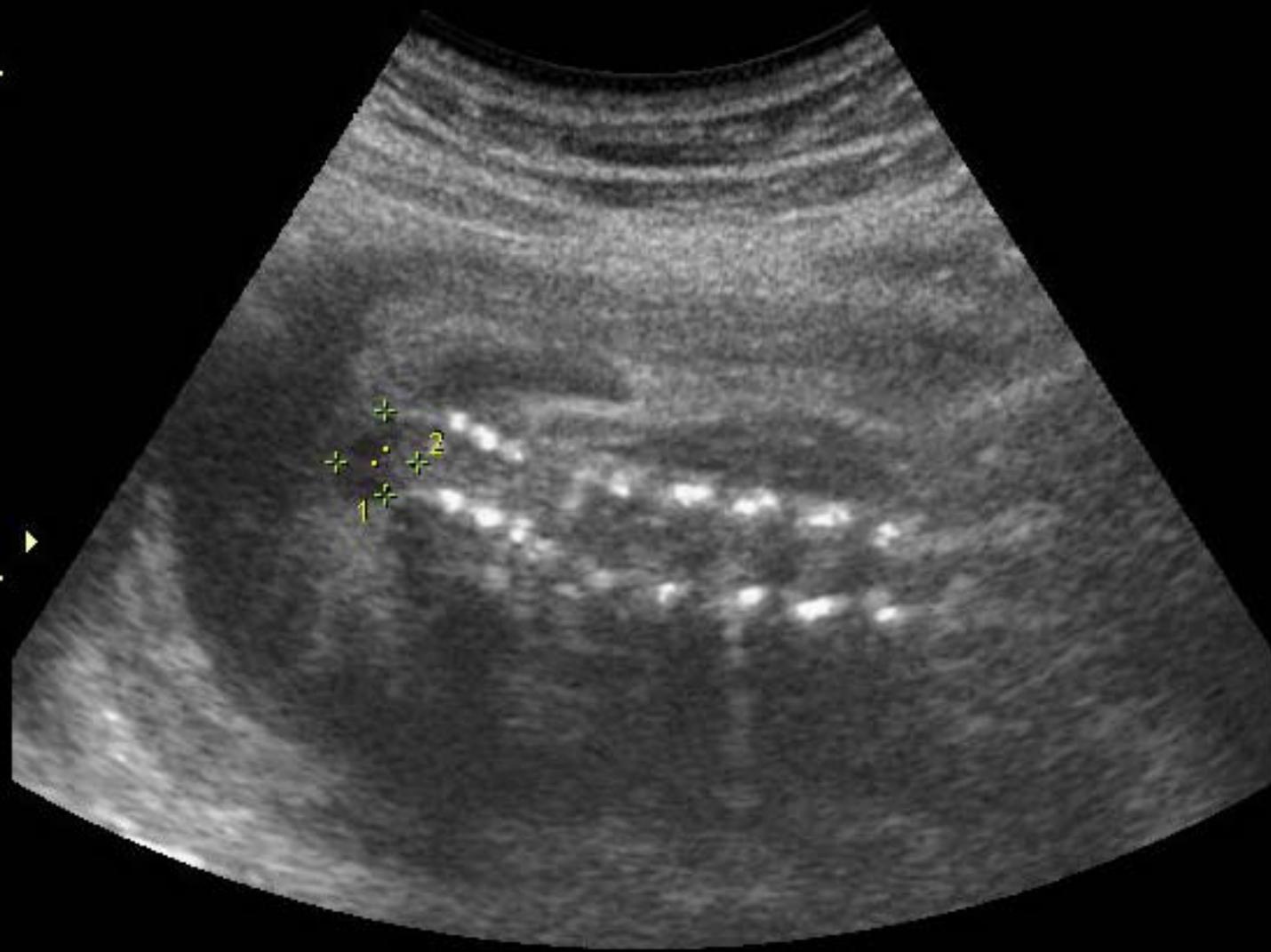
CERE

A



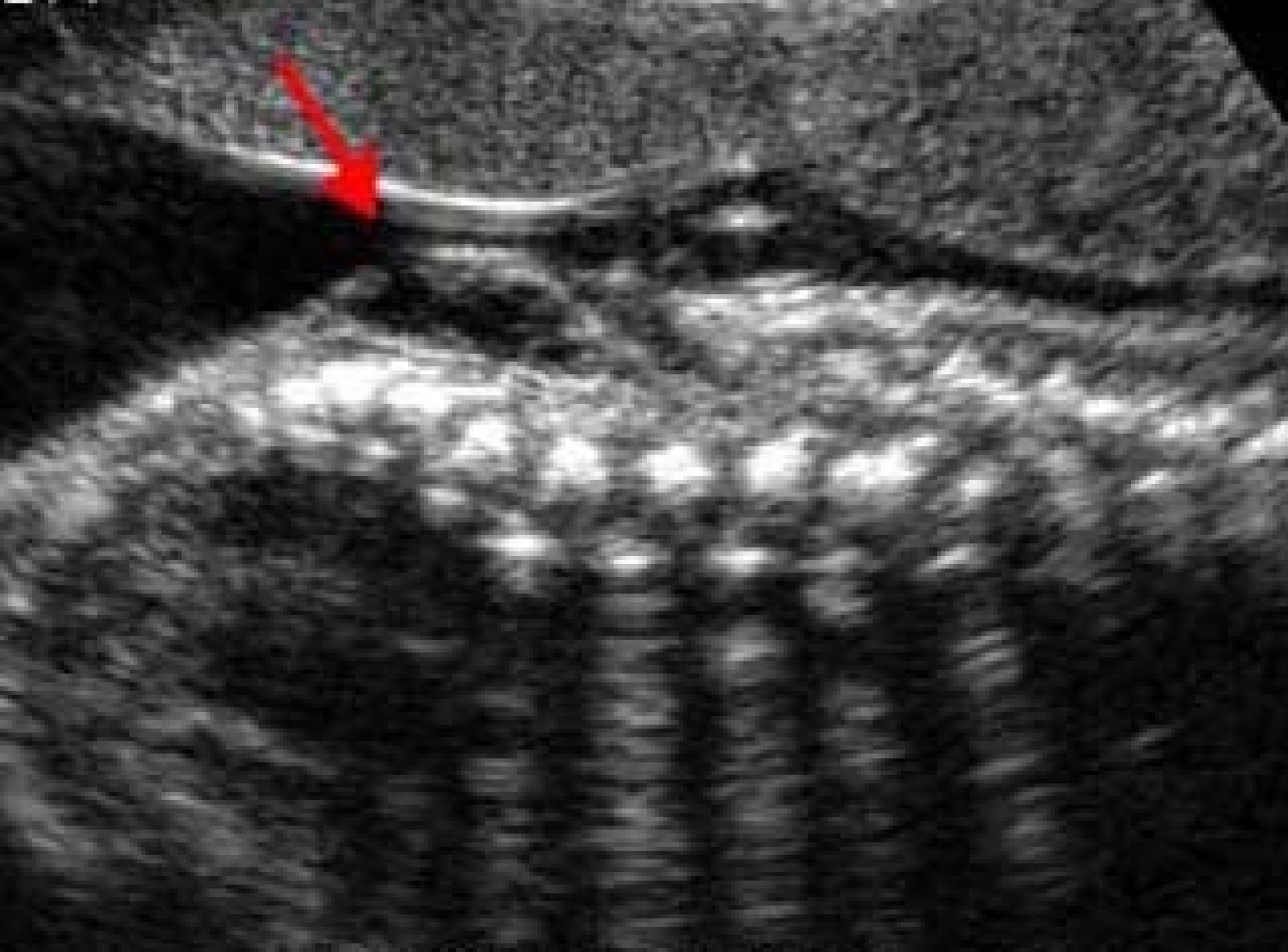


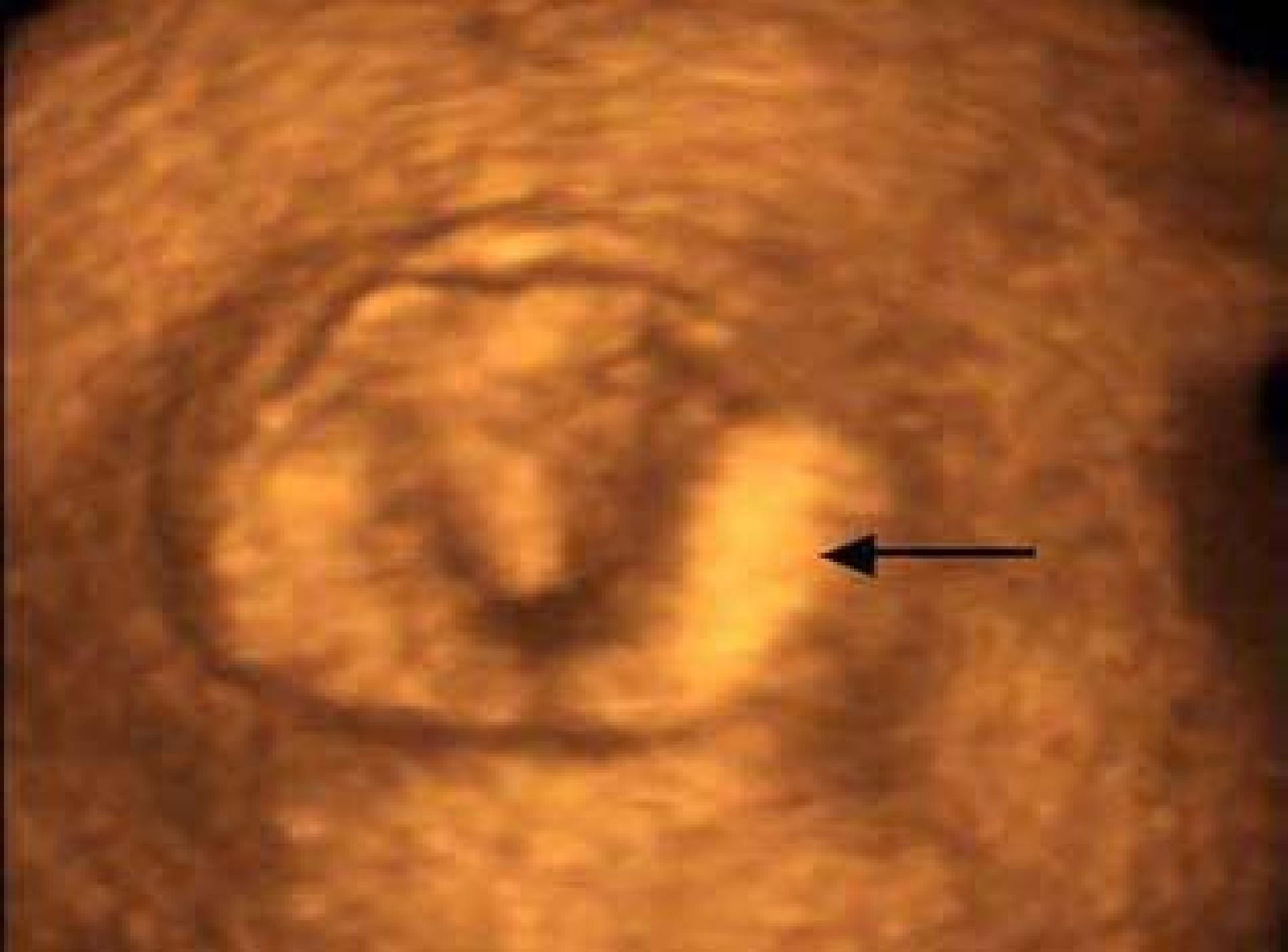
Routine  
Har-mid  
Pwr -1  
Gn 6  
C7 / M7  
P3 / E1  
MI 1.1



D1 0.837cm  
D2 0.814cm  
D1/D2 103%



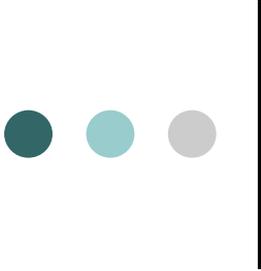






CDPI





# Special Concerns

## ○ ONTD

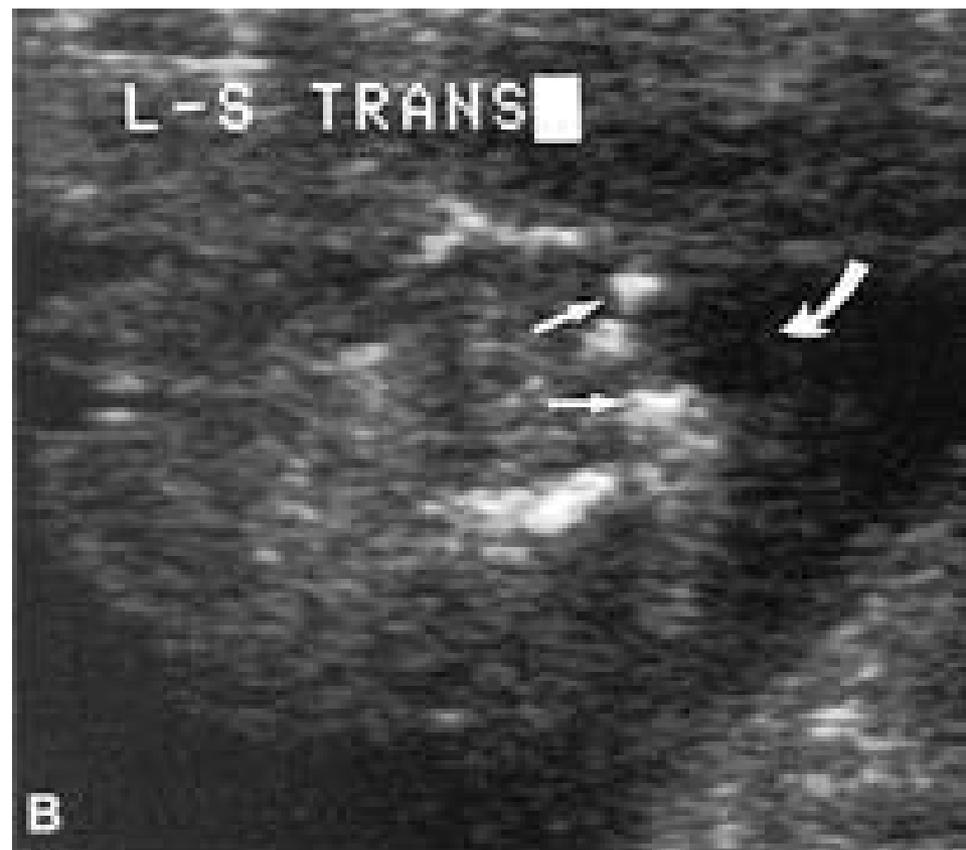
- Bony dorsal arch defect + neural content exposure
- “U” shaped vertebra on axial view

## ○ Meningocele sac

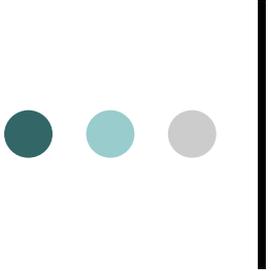
## ○ Myelomeningocele

## ○ Associated findings

- 24% (1/4) clubfoot
- 40% fetus with additional anomalies (67% aneuploid fetuses with other anomalies)
- Scoliosis & kyphosis

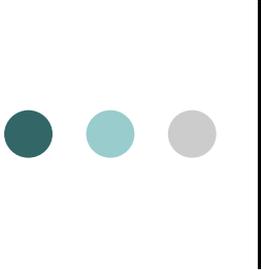






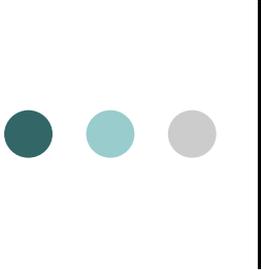
# *Differential Diagnosis*

- Sacrococcygeal teratoma
- Aqueductal stenosis
  - Moderate to severe hydrocephalus
  - atria > 15mm & dangling choroid plexus
- DWC
- Isolated frontal bone concavity
  - Seen in 1% of normal fetuses
  - 3<sup>rd</sup> trimester resolve
  - Normal CM



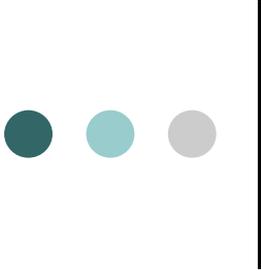
# Genetics & Epidemiology

- 4% aneuploid rate with spinal bifida
  - Trisomies 13, 18
- 0.4 : 1000
- 3% of all spontaneous abortion
- 1~2 % recurrent rate



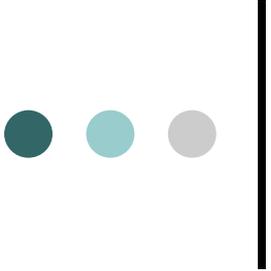
# Clinical Issues

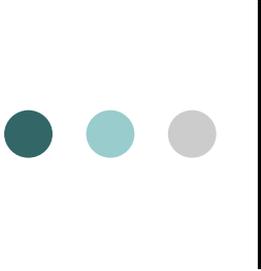
- Most common : maternal AFP↑
- Nature history & prognosis
  - High morbidity and mortality (35% 5yrs)
  - 50% IQ>80
  - Intrauterine findings ≠outcome
  - Obstructive hydrocephalus
  - Musculoskeletal dysfunction (25% lower ext)
  - GI or GU dysfunction



# Treatment

- Cesarean section
- Immediate post-natal ONTD surgery
- In utero surgery in clinical trial
- Preventive treatment with folic acid
  - 4mg/day reduces recurrence by 70%

- 
- Dandy-Walker continuum
  - Chiari II malformation
  - **Holoprosencephaly**

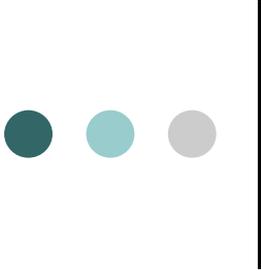


## ***Psalms chapter 115 : 13~16***

***He will bless them that fear the LORD,  
both small and great. The LORD shall  
increase you more and more, you and  
your children. Ye are blessed of the  
LORD which made heaven and earth.  
The heaven, even the heavens, are the  
LORD's: but the earth hath he given to  
the children of men.***



**QUIZ TIME**



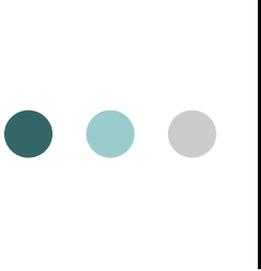
Dandy-Walker Continuum 最常合併的基因異常是？

- A. Turner syndrome
- B. Trisomy 18
- C. Trisomy 21
- D. Trisomy 13



# 如何區分DWC & Chiari II? 何者為非

- A. 看小腦的vermis是否完整。而DWC的vermis通常是absent or severely hypoplastic
- B. 學理上DWC的tentorium是elevated，而Chiari II的則是downward displacement
- C. DWC幾乎100%合併ONTD，例如spinal bifida



下列何者為Chiari II的典型特徵？

- A. Banana sign
- B. Lemon head sign
- C. Severe hydrocephalus
- D. 100% ONTD

# 婦產部每月晨會超音波教學

Section 1	First Trimester (2)	(已完成)
Section 2	Fetal Brain Develop (2)	(已完成)
Section 3	Fetal Spine (2)	
Section 4	Face and Neck (2)	
Section 5	Chest (2)	
Section 6	Fetal Heart (5)	
Section 7	Abdominal Wall & GI (2)	
Section 8	GU system (2)	
Section 9	Muscular System (3)	
Section 10	Placenta (1)	
Section 11	Umbilical Cord (1)	(已完成)

- I. 每月第 3 週或第 4 週的週五上午 7:15~7:55 為部內晨會超音波教學，一個完整的教學共 24 個月。
- II. 每週二、四上午陳志堯醫師於 7 樓超音波實際操作教學。

# 婦產部中正七樓超音波負責實地操作教學並教學之醫師時間表

參加者: 平常日(W1~W5)由楊明智主任團隊住院醫師及超音波室之產科住院醫師參加學習 (共三位)

	W1	W2	W3	W4	W5	W6
第一週	V 江美美小姐 H 陳立珣小姐 張家銘醫師	V 陳志堯醫師 H 楊明智主任 (抽羊水教學)	V 陳怡仁醫師 (乳房超音波) H 陳立珣小姐	V 陳志堯醫師 (胎兒高層次) V 洪正修主任		江美美小姐
第二週	V 江美美小姐 H 陳立珣小姐	V 陳志堯醫師 H 楊明智主任 (抽羊水教學)	V 陳怡仁醫師 (乳房超音波) H 江美美小姐	V 陳志堯醫師 (胎兒高層次) V 洪正修主任		舒麗萍老師 案例教學 兩位產科住院醫師受訓
第三週	V 陳立珣小姐 H 江美美小姐 張家銘醫師	V 陳志堯醫師 H 楊明智主任 (抽羊水教學)	V 陳怡仁醫師 (乳房超音波) H 陳立珣小姐	V 陳志堯醫師 (胎兒高層次) V 洪正修主任		陳立珣小姐
第四週	V 陳立珣小姐 H 江美美小姐	V 陳志堯醫師 H 楊明智主任 (抽羊水教學)	V 陳怡仁醫師 (乳房超音波) H 江美美小姐	V 陳志堯醫師 (胎兒高層次) V 洪正修主任		徐慧玲小姐

V 代表 V730 高層次超音波

H 代表 HP 一般胎兒及婦科超音波

打框者時段為臨床實際操作示範

☪ 將定期(每 1~2 個月)針對至 7F 超音波室之住院醫師實施考試