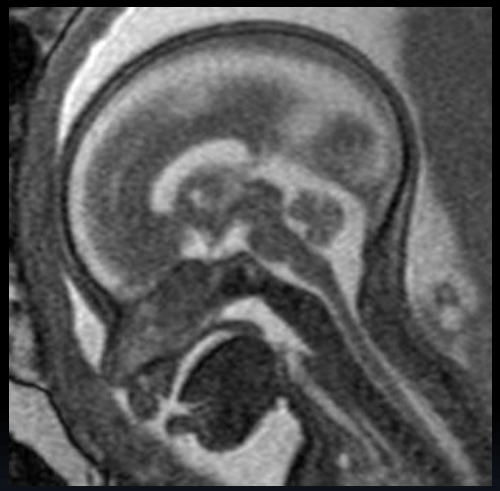
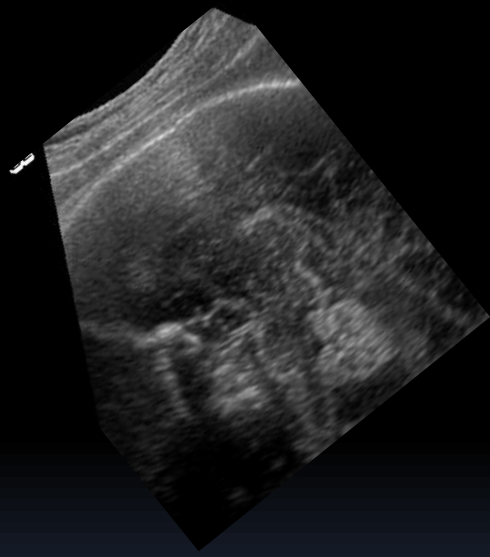
Panda SA
The Paediatric Neurology and Development Association of Southern Africa

Posterior Fossa Abnormalities in the Fetus





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א. וולפסון
E.WOLFSON
MEDICAL CENTER

Outline

- Assessment of normal development of the cerebellum and brainstem in utero
- Dandy-Walker spectrum and related disorders:
  - Dandy-Walker Malformation
  - Vermis hypoplasia
  - Cerebellar hypoplasia
  - Rhombencephalosynapsis
  - Blakes pouch cyst
  - Mega cisterna magna
  - Arachnoid cyst

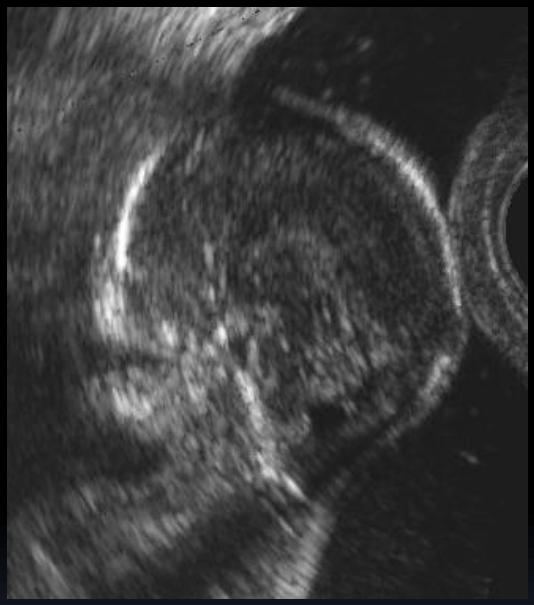
Outline

- Brain stem disorders and associated anomalies:
  - Joubert syndrome related disorders
  - Cobblestone malformation
  - Pontine cap dysplasia
  - Tectal Dysplasia
  - Brainstem Disconnection Syndrome

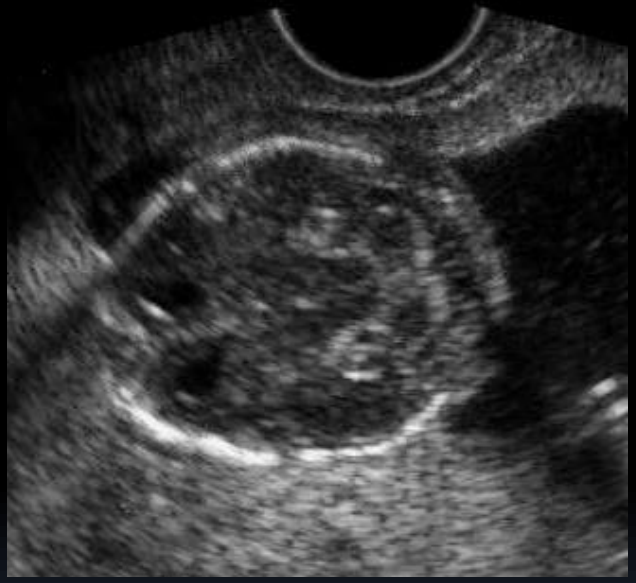
- The ultrasonographic evaluation should include multiplanar images of the cerebellum
- The axial plane is useful for determining the transcerebellar diameter, cisterna magna size and the cerebellar peduncles
- The coronal plane enables differentiation between the cerebellar hemispheres and the vermis
- The midsagittal plane allows depiction of the vermian lobules, fissures and shape of the fastigium; measurements of the vermian diameter and surface. This plane also permits evaluation of the size and shape of the tectum, pons, cisterna magna and tentorium
- Nomograms of the fetal midbrain-hindbrain have been established

Normal Cerebellum

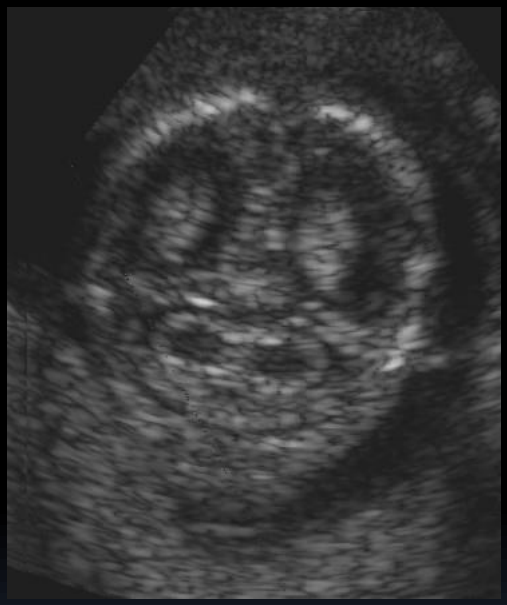
15 weeks



Sagittal

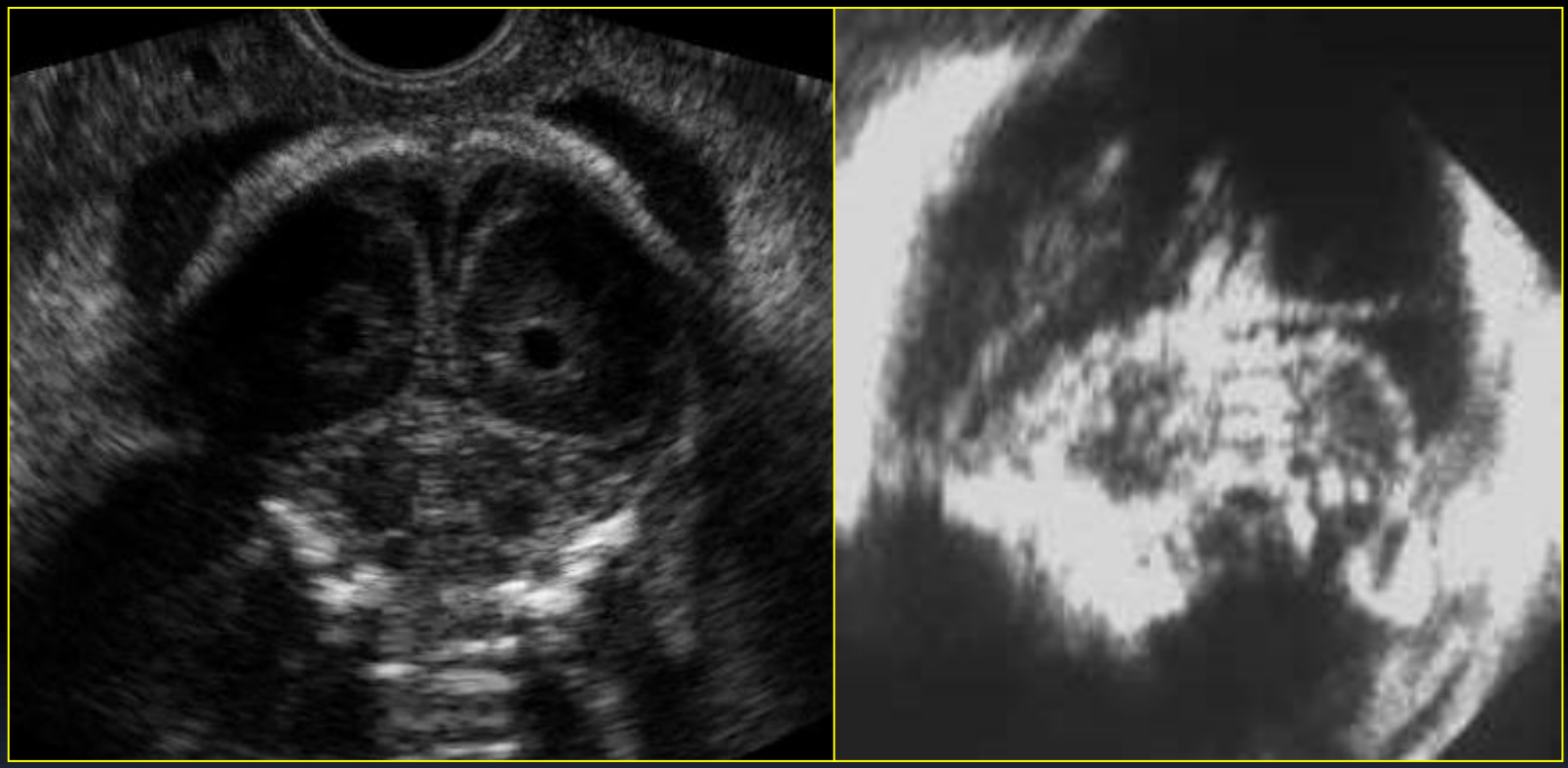


Axial



Coronal

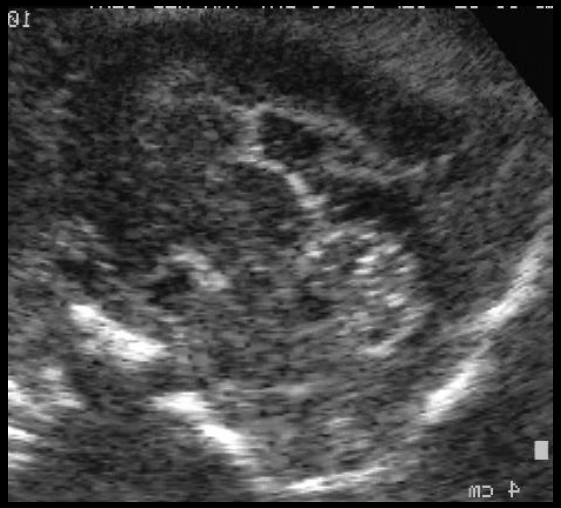
Normal Cerebellum – Coronal Planes



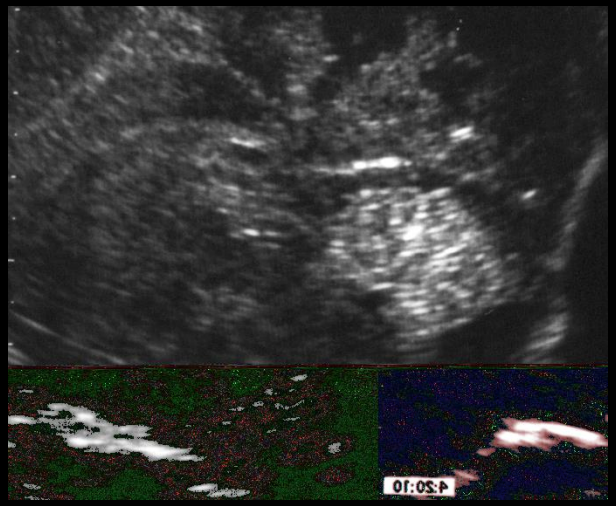
22 weeks

25 weeks

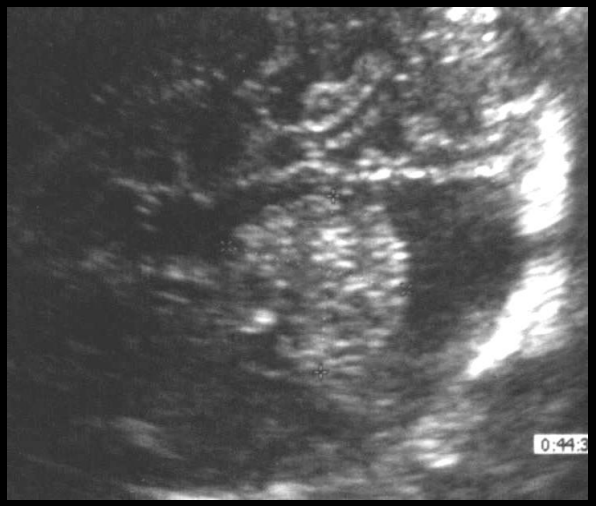
Normal Vermis – Sagittal Planes



21 weeks

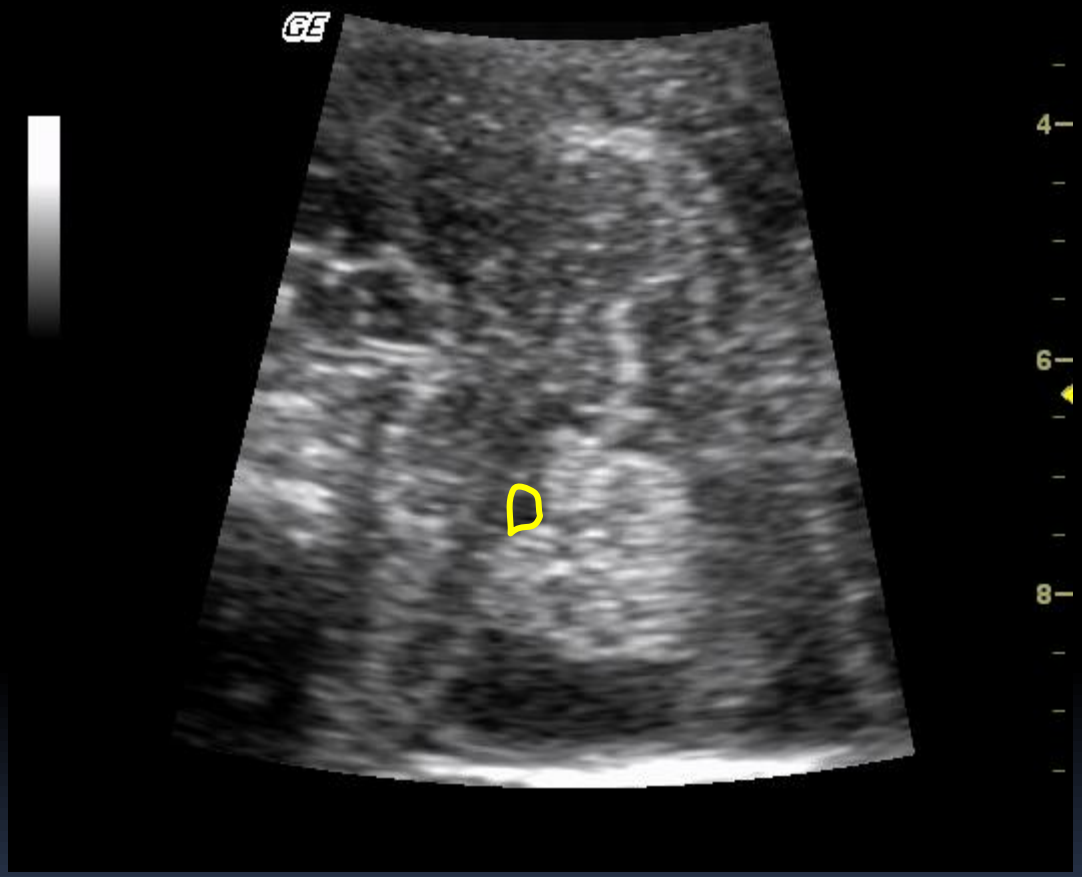


25 weeks



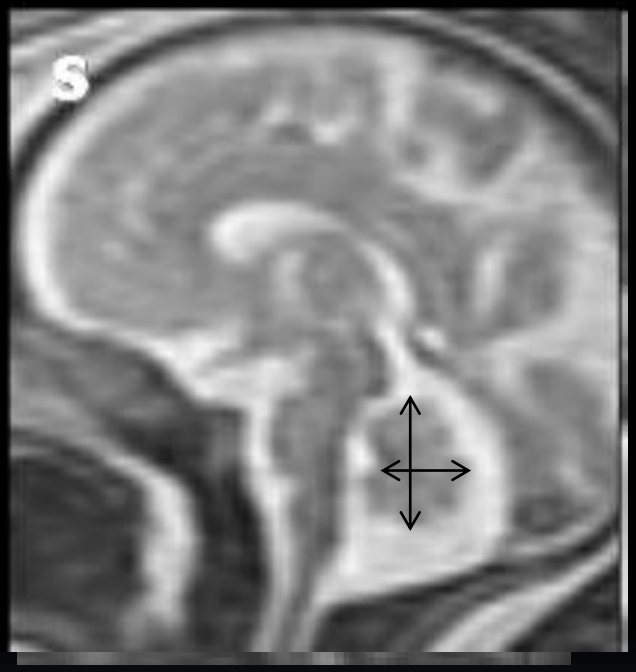
35 weeks

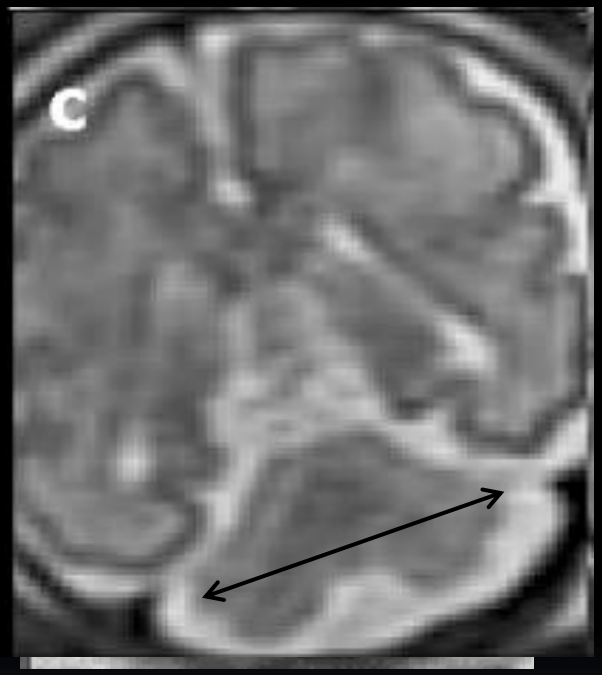
- The cerebellar vermis can be detected in the mid-sagittal plane as early as 18 weeks of gestation.
- The primary fissure is observed between 27 and 30 weeks of pregnancy. Some degree of differentiation between lobules is possible starting from 30-32 weeks of gestation.
- The fourth ventricle is uniformly observed as a triangular structure antero-caudal to the vermis. The inferior lobe of the vermis and the nodulus separate the fourth ventricle and the cisterna magna


GE
4
6
8

- MRI is highly accurate in illustrating the morphologic MRI biometry of cerebellar development .
- The cerebellar vermis is best assessed by MRI on direct midline sagittal images, and coronal images; the measurements should be compared with established norms.
- The cerebellar hemispheres are best assessed on nonoblique axial and coronal views.

Cerebellar measurements





Vermis height
Vermis antero-posterior diameter

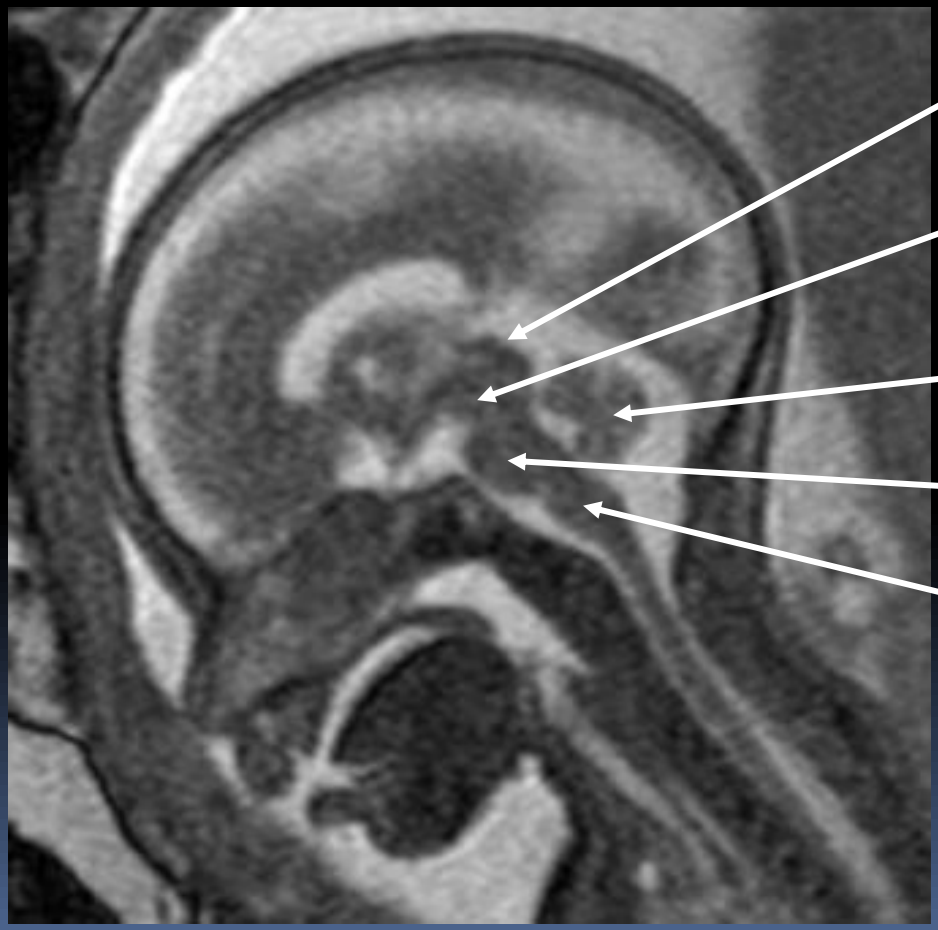
Transverse cerebellar diameter

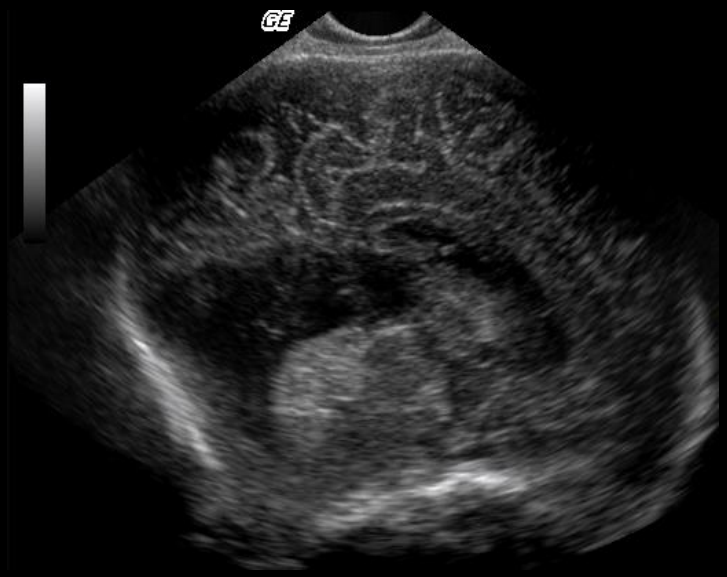
- Fetal MRI shows gestational age-specific changes in signal intensity in the normal development and maturation of the cerebellar hemispheres and brainstem
- The cerebellar cortex, dentate nucleus, tectum, dorsal pons, and medulla are T1-hyperintense and T2-hypointense
- The changes in signal intensity in the brainstem and cerebellum are not encountered until 20–23 weeks of gestation. By 26–27 weeks of gestation a three-layered pattern is noted in the cerebellar hemispheres corresponding to the cerebellar cortex, cerebellar white matter, and dentate nucleus

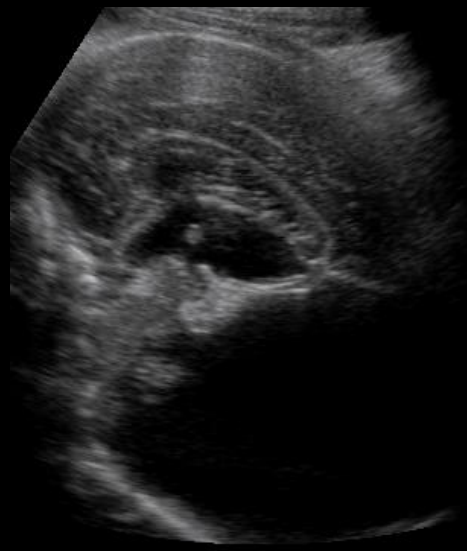
- Fetal brain MRI can show the fissures of the cerebellum depending on the gestational age
- The primary fissure is identified on sagittal images at 22 weeks, but the cerebellar surface is smooth
- From 24 to 29 weeks, foliation of the vermis and posterior lobes of the cerebellum is seen on sagittal images
- The cerebellar surface is smooth with the appearance of some indentations corresponding to the horizontal and secondary fissures on axial images
- The convoluted pattern of the cerebellum is well identified from 30 weeks on and is always seen beyond 33 weeks

Anatomy

Normal
23 weeks
GA







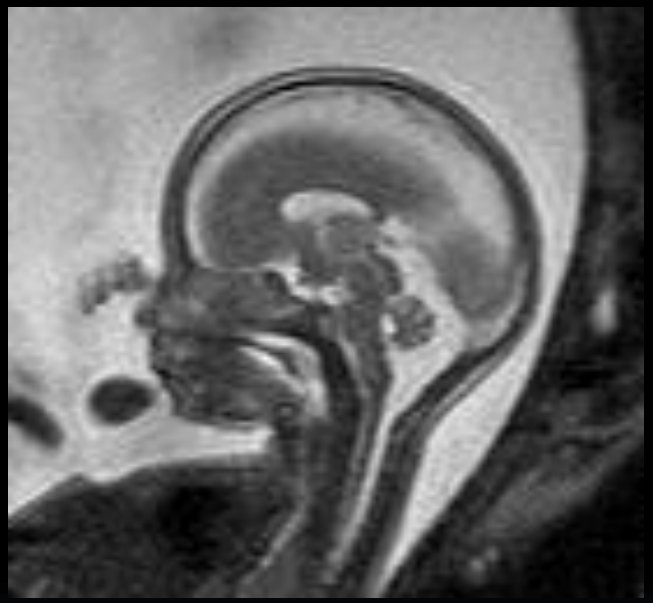
Dandy-Walker Spectrum Cystic Lesions of the Posterior Fossa

Definition of cystic lesions of the posterior fossa

- An anteroposterior diameter of the retrocerebellar fluid space larger than 10 mm (unless the fluid space-occupying lesion is not located in the midline)
- It may encompass different entities with very different prognoses

The Imaging Evaluation

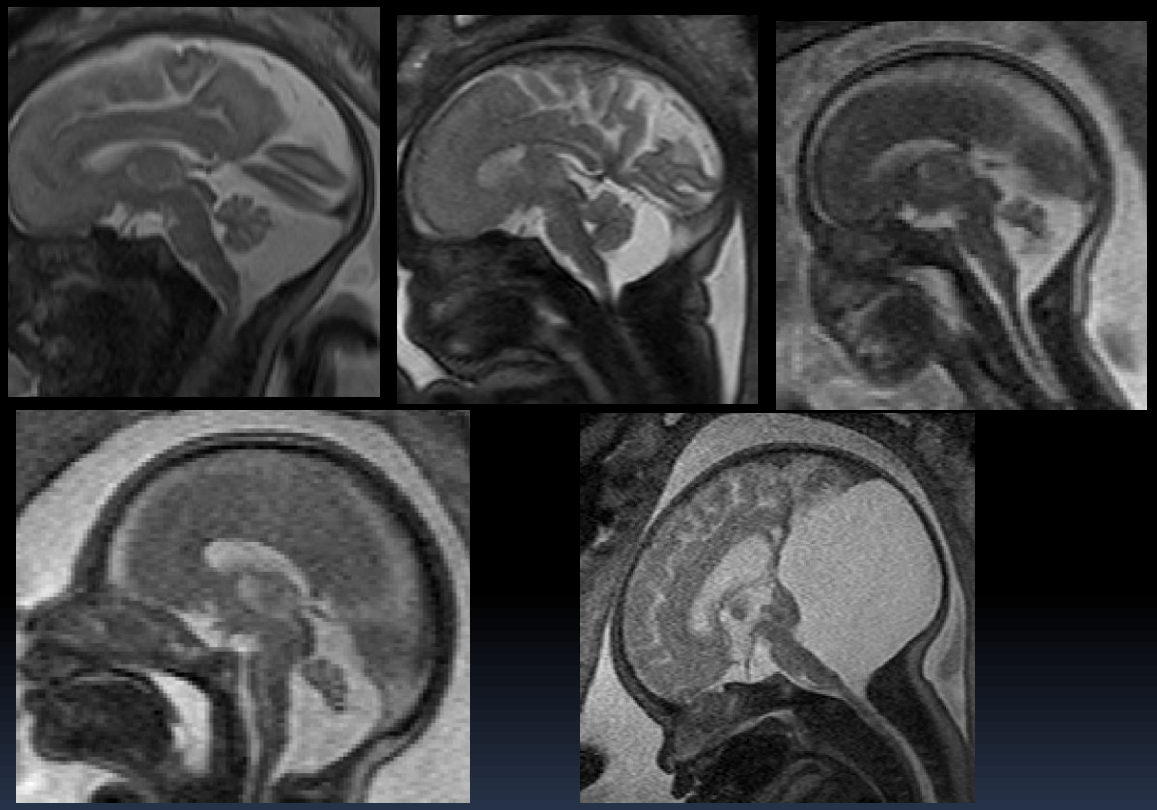
- The position of the torcula
- The orientation of the tentorium
- The vermis:
  - Axis
  - Foliation
  - Size
  - Morphology



- The echogenicity of the fluid-filled space
- The presence and location of the walls of Blake's pouch and the presence of septa within the lesion
- The mass effect on the cerebellum, the tentorium and the occipital vault

Cystic lesions of the posterior fossa

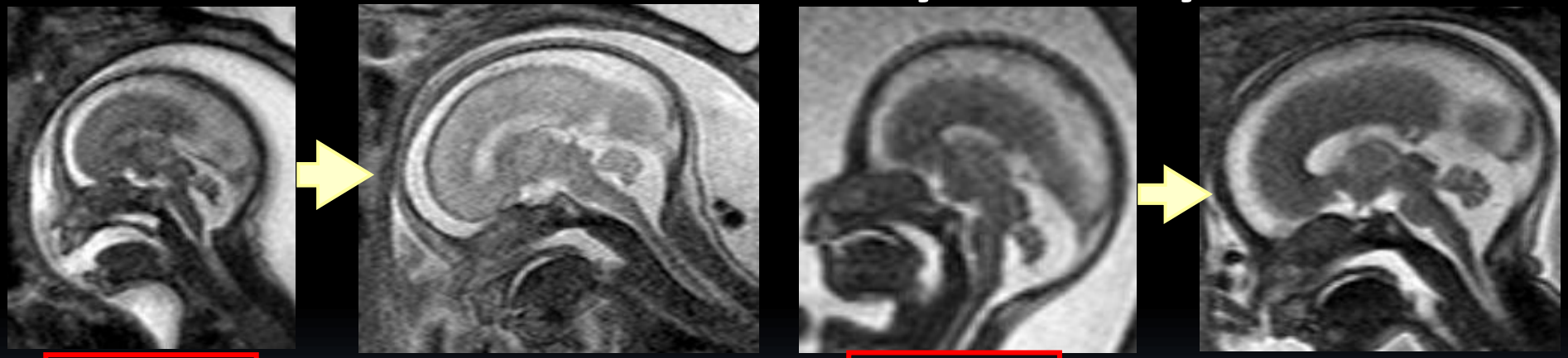
- Mega Cisterna Magna
- Arachnoid cyst
- Blakes Pouch
- Vermian hypoplasia
- Dandy Walker Malformation



Delayed rotation of vermis

brainstem-vermian angle
rhombic lip internalization

may decrease!
may be delayed!



19+5
40˚

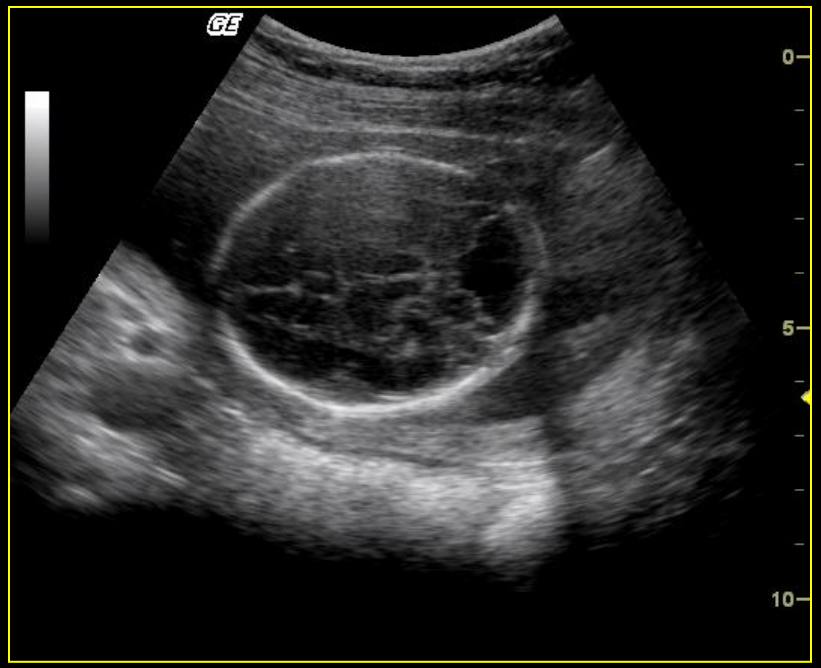
23+5

19+3
40˚

24+3
22˚

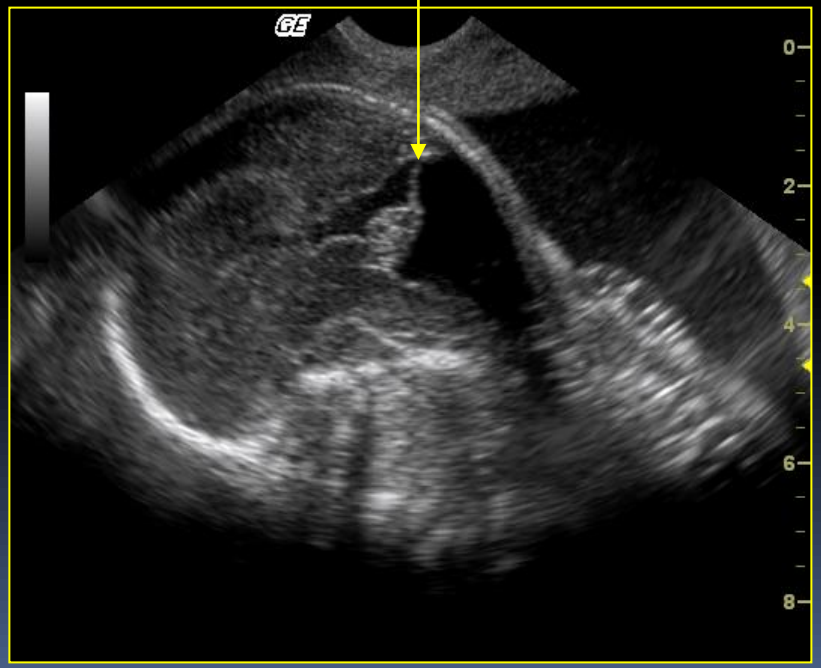
Dandy-Walker Malformation

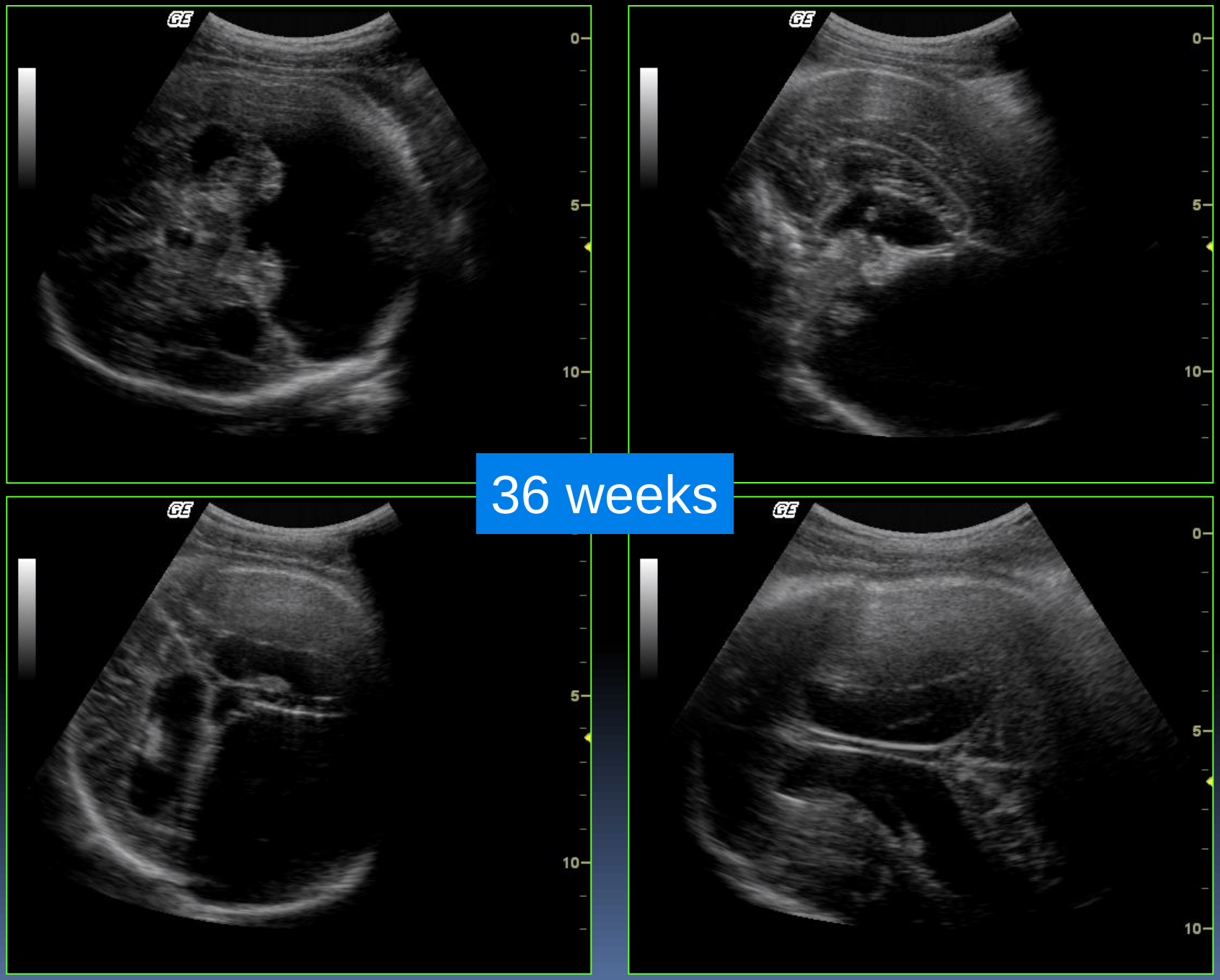
- Complete or partial agenesis of the vermis
- Cystic dilatation of the fourth ventricle
- An enlarged posterior fossa with upward displacement of lateral sinuses, tentorium and torcula
- Frequently associated with hydrocephalus



20 weeks

Elevated tentorium




GE
0
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10
GE
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36 weeks
GE
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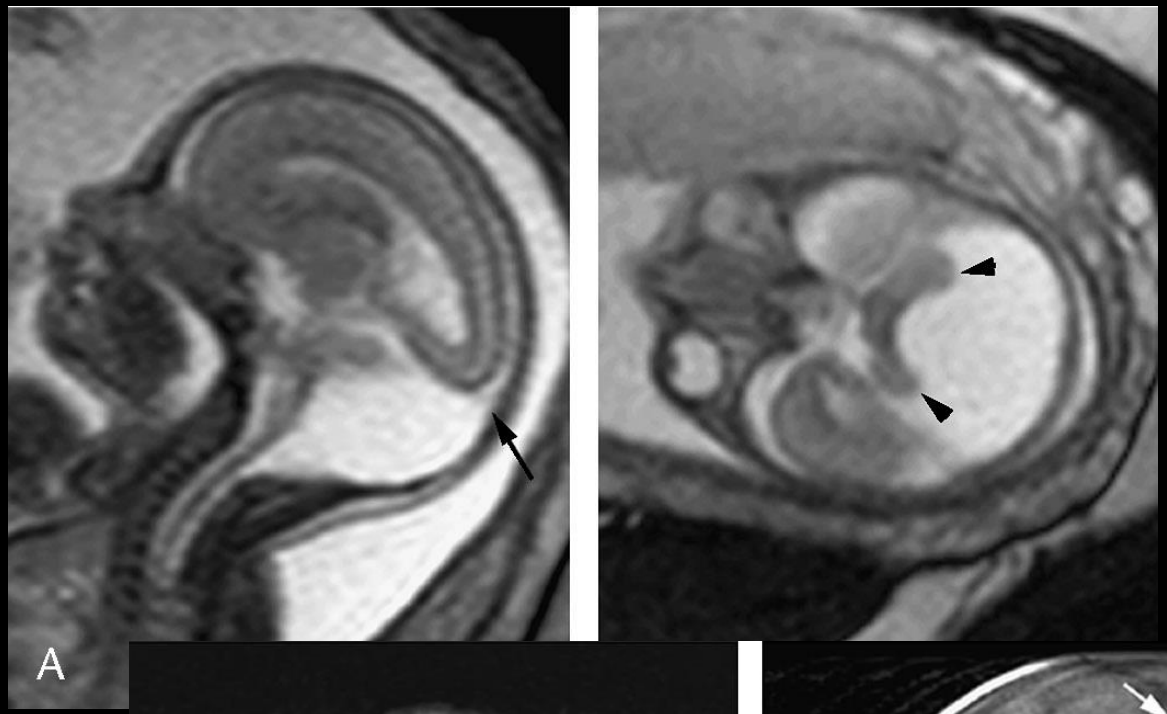
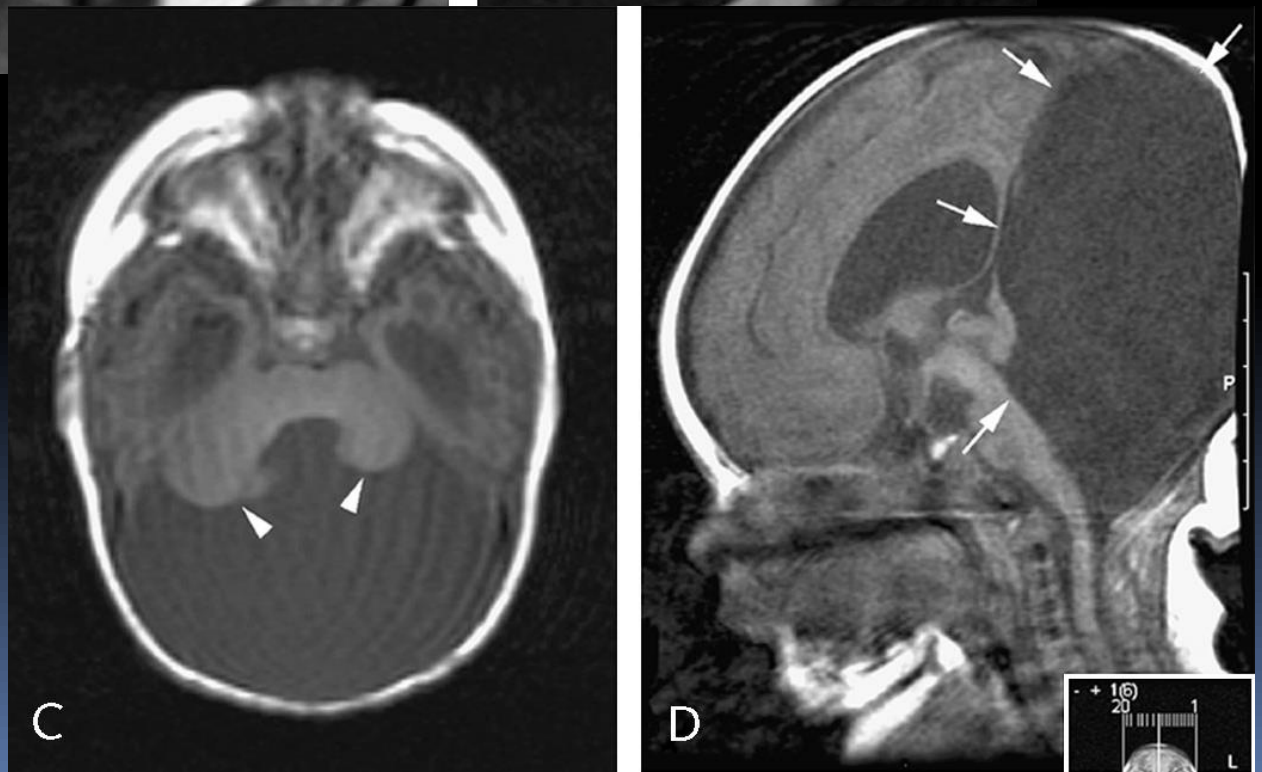
10
GE
0
5
10



22 weeks

postnatal

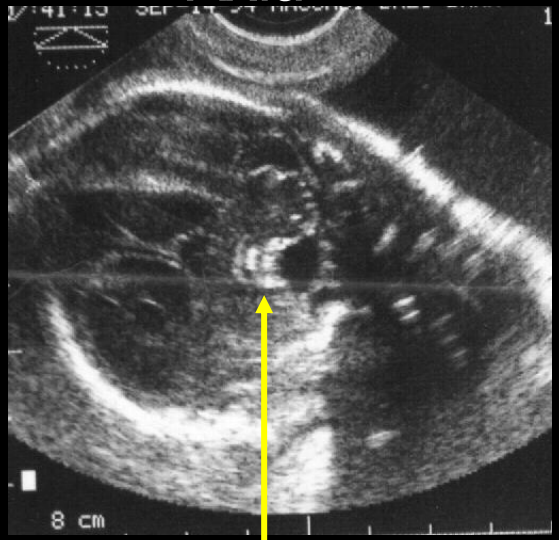


Cerebellar Vermis Hypoplasia

- Dandy–Walker variant is actually cerebellar vermis hypoplasia or partial vermis agenesis
- Frequently accompanied by other CNS and systemic malformations
- Part of multiple genetic syndromes
- Although the clinical heterogeneity in CVH is broad, the prognosis is often worse than for classic DWM

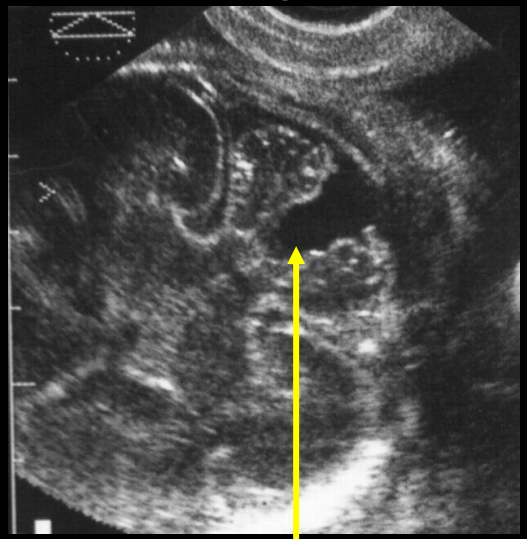
Cerebellar Vermis Hypoplasia

Axial



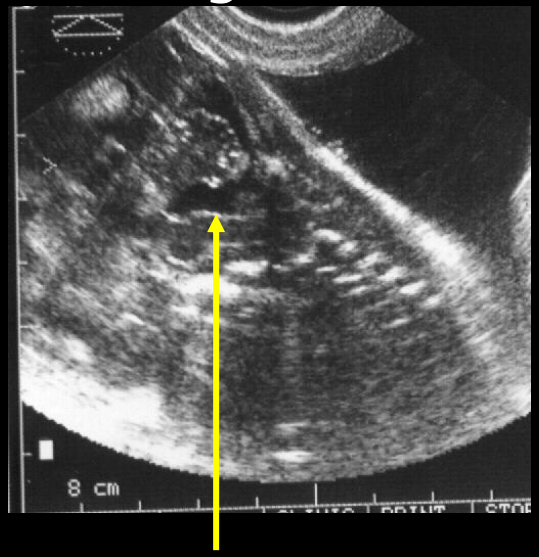
Superior vermis

Axial



Absent inferior vermis

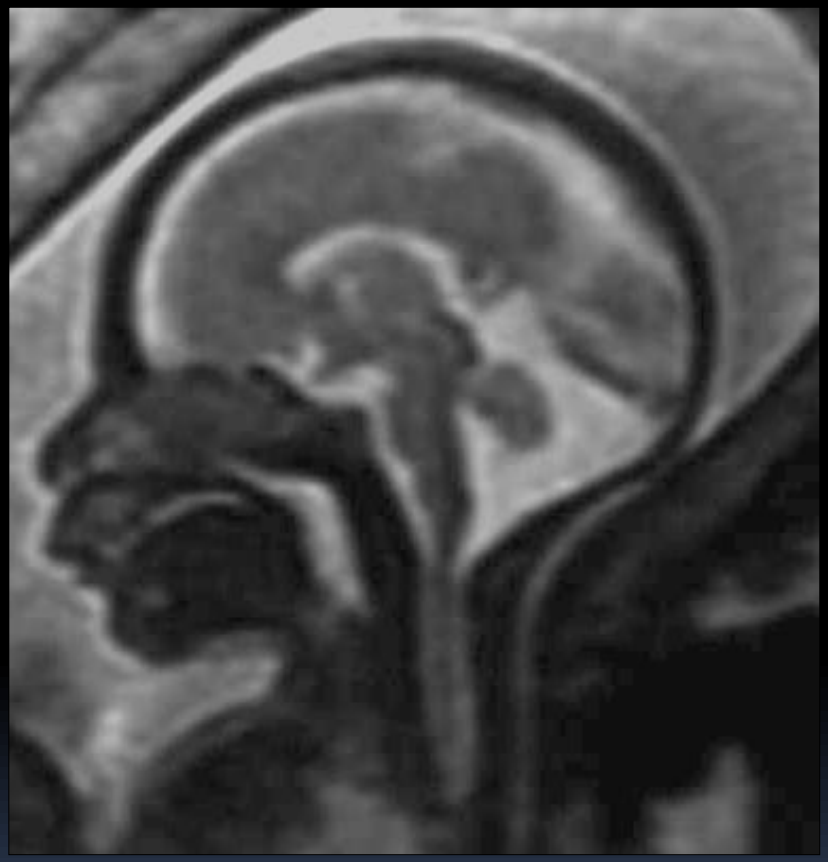
Sagittal



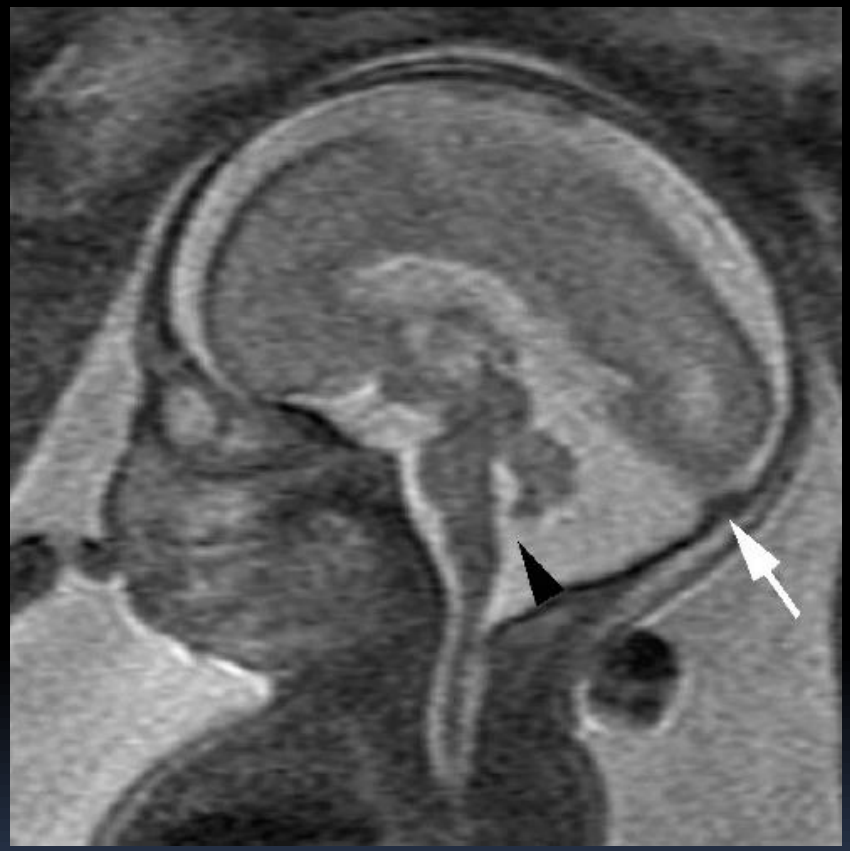
Communication between CM and 4th ventricle

29 weeks

Cerebellar Vermis Hypoplasia



24 weeks

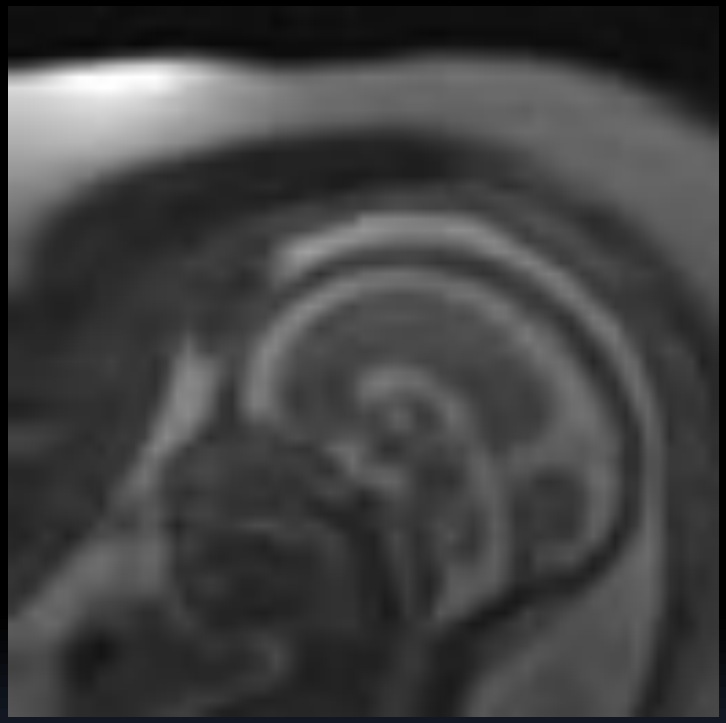


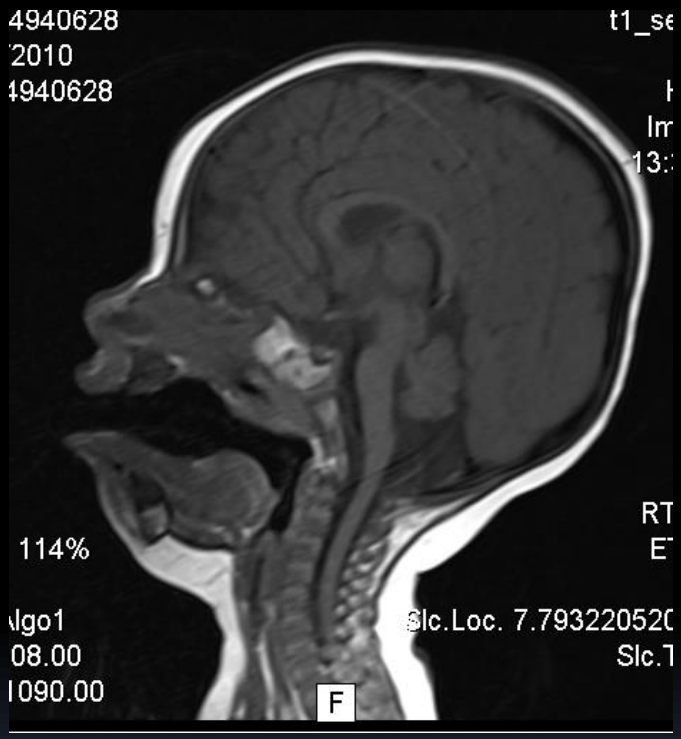
25 weeks

Cerebellar Hypoplasia

- Cerebellar hypoplasia has many causes including:
  - Chromosomal disorders
  - Genetic syndromes
  - Prenatal disruptions—eg, infection or ischaemia
- The vermis and both hemispheres can be equally small or might be hypoplastic in any combination
- Pontine hypoplasia and midbrain malformations are often, but not always associated with cerebellar hypoplasia

Cerebellar Hypoplasia





Prenatal MRI

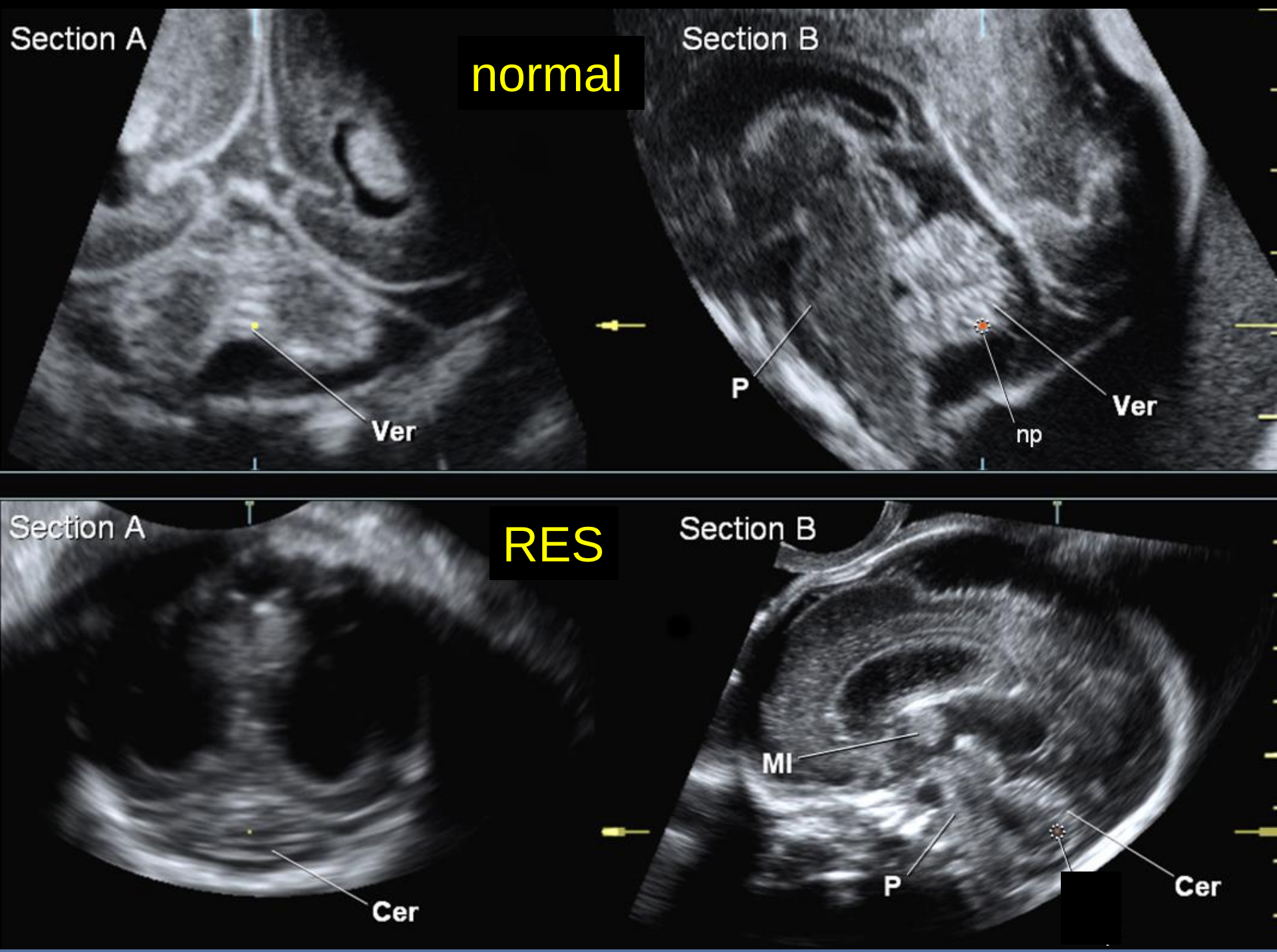
Post natal MRI

Rhombencephalosynapsis

- Midline fusion of the cerebellar hemispheres
- Absence of the incisura cerebelli posterior
- Fusion of the dentate nuclei
- Absence of the vermis
- Convergence of the middle and superior cerebellar peduncles

Rhombencephalosynapsis

- Clinical presentation is highly variable
- The clinical severity is determined by the presence of associated supratentorial malformations: hydrocephalus, callosal dysplasia, brain atrophy
- Usually sporadic


Section A
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Section B
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Section A
RES
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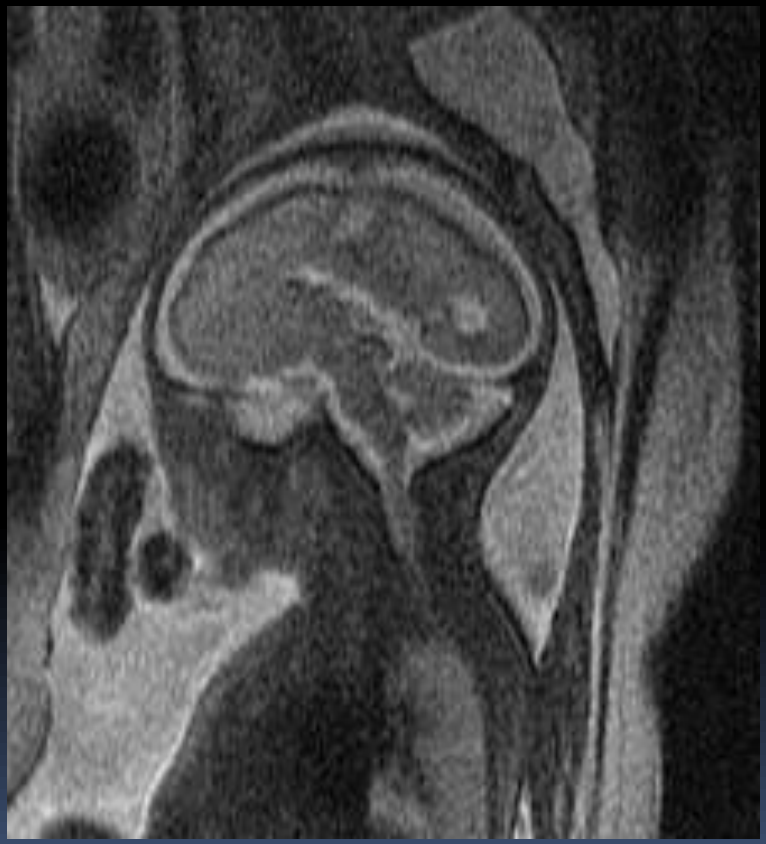
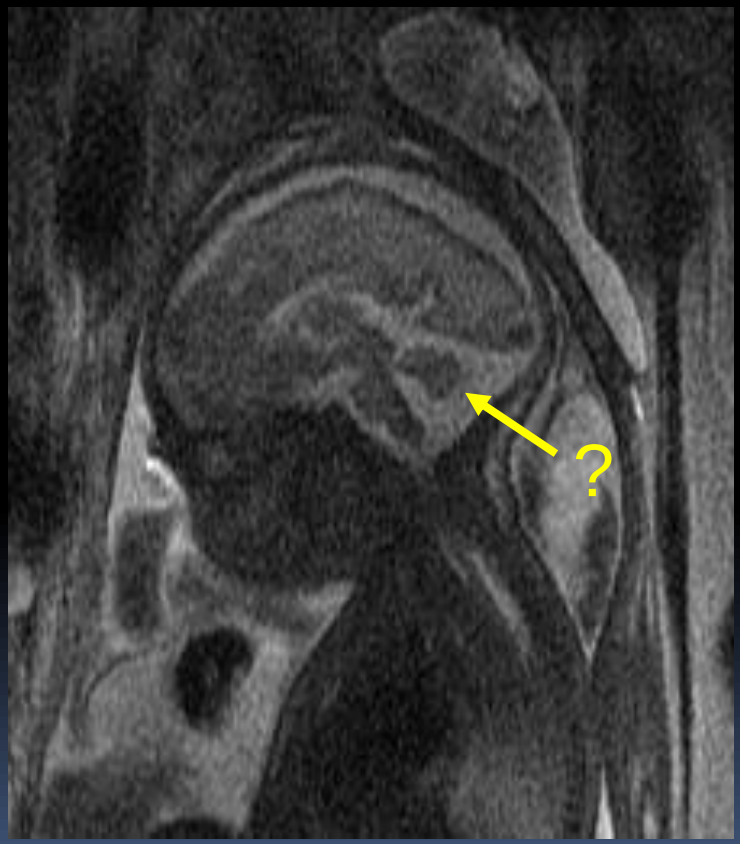
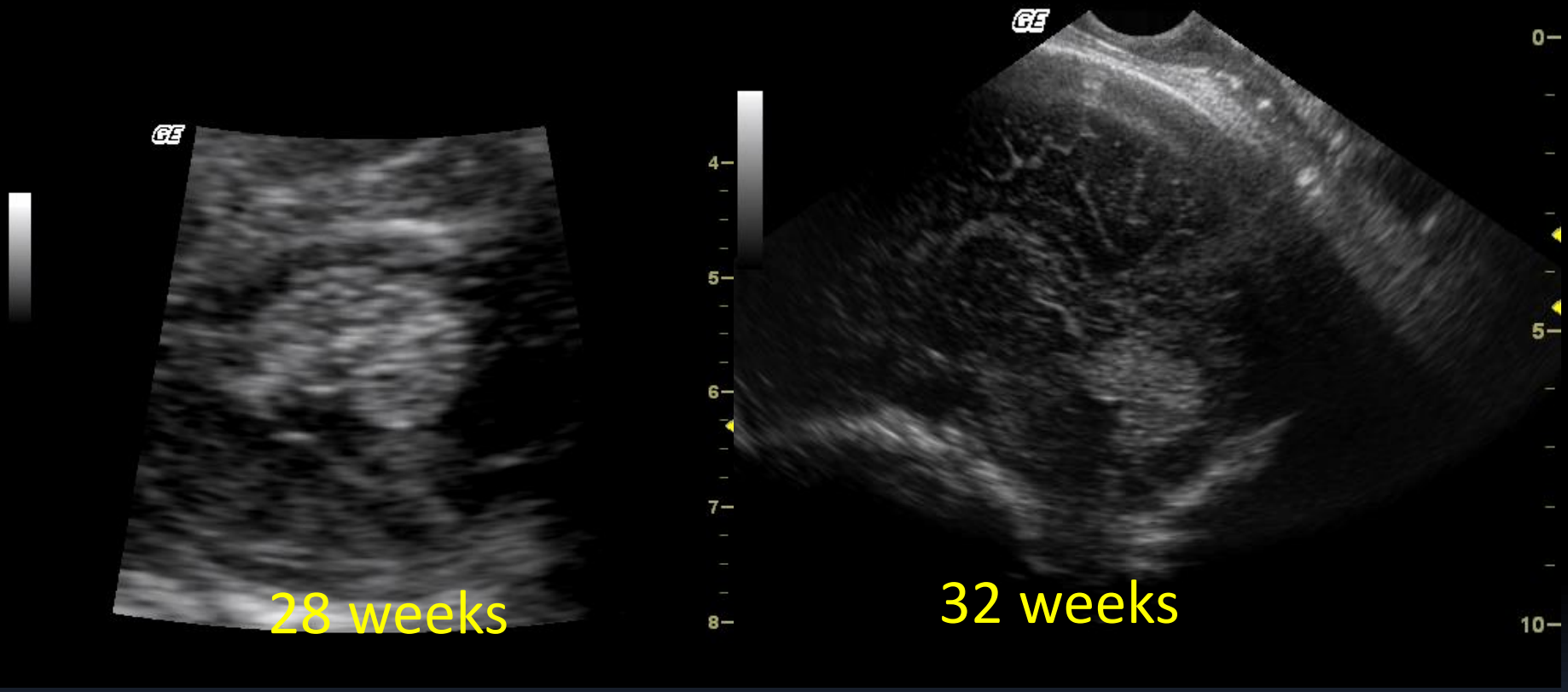
Cer

Blakes Pouch Cyst





27 weeks susp. inferior vermis hypoplasia



Neurologic examination: normal

Closure of the cerebellar vermis: evaluation by second trimester US

Percentage of Vermes Open versus Fetal Age at Time of First Scan

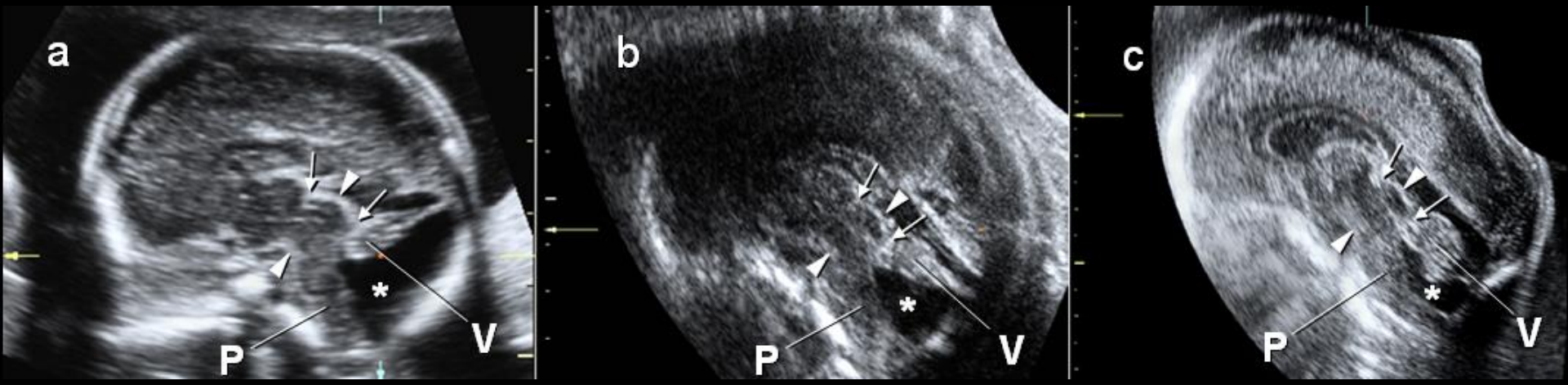
Gestational Age (wk)	Total No. of Fetuses (n = 897)	No. with Open Vermis
13.5	6	4 (67)
14	43	24 (56)
14.5	88	36 (41)
15	136	32 (23)
15.5	118	17 (14)
16	171	22 (13)
16.5	121	5 (4)
17	85	5 (6)
17.5	47	2 (4)
18	29	0 (0)
18.5	10	0 (0)
19	15	0 (0)
19.5	14	0 (0)
20	5	0 (0)
20.5	1	0 (0)
21	3	0 (0)

Note.—Percentages in parentheses. Open vermis defined as a communication between the fourth ventricle and the cisterna magna.

CONCLUSION:
The prenatal diagnosis of “Dandy-Walker variant” should not be made before 18 weeks gestation because the development of the cerebellar vermis may be incomplete at that time.

Bromley B, et al. Radiology 1994

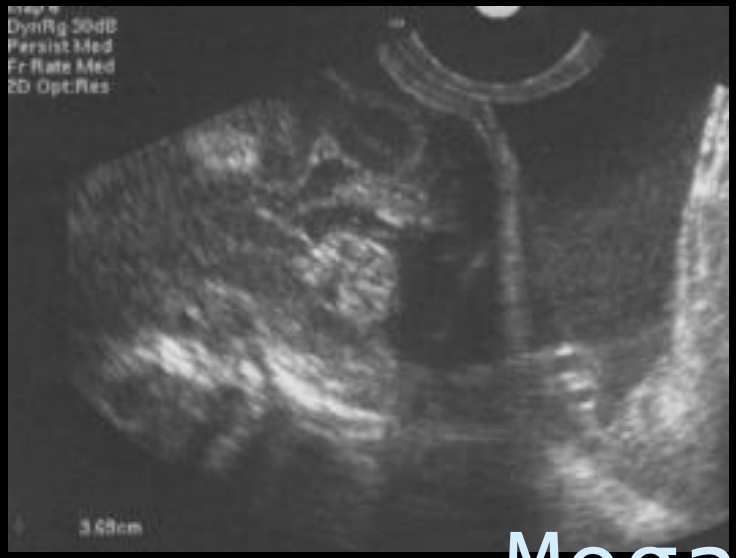
Dandy Walker Spectrum

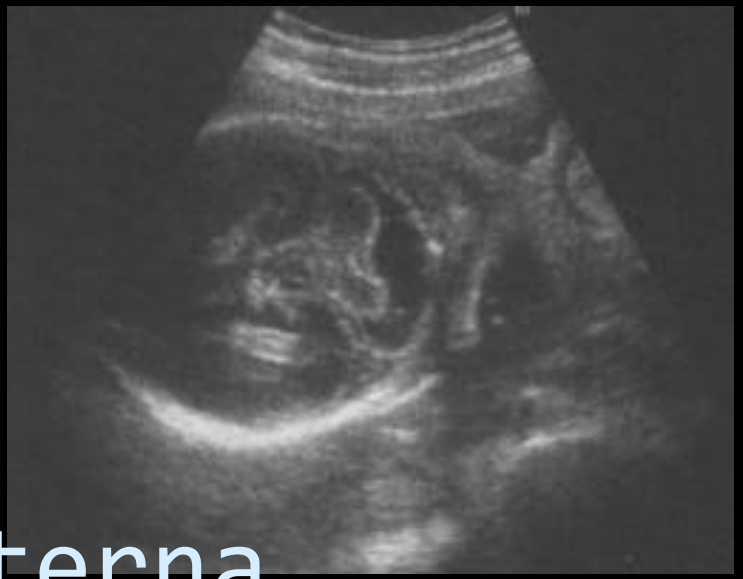


DWM VH BPC

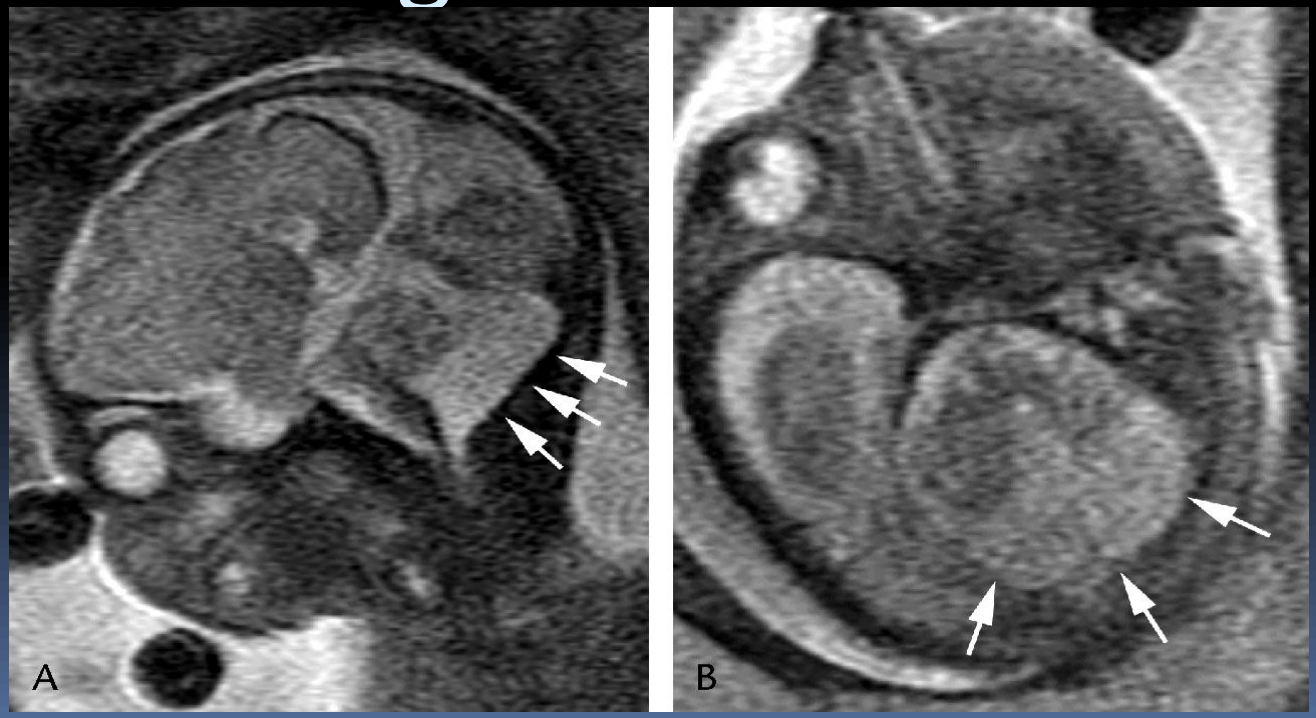
Mega Cisterna Magna

- Expansion of the cisterna magna with a morphologically intact vermis
- Common condition
- Most children are detected incidentally
- Developmental delay is uncommon and is determined by associated malformations





Mega Cisterna



Posterior Fossa Arachnoid Cyst

- Benign CSF like fluid collection that develops between the layers of the arachnoid membrane
- They do not communicate freely with the ventricular or subarachnoid space
- No recognized association with supratentorial congenital abnormalities
- Most are isolated and sporadic

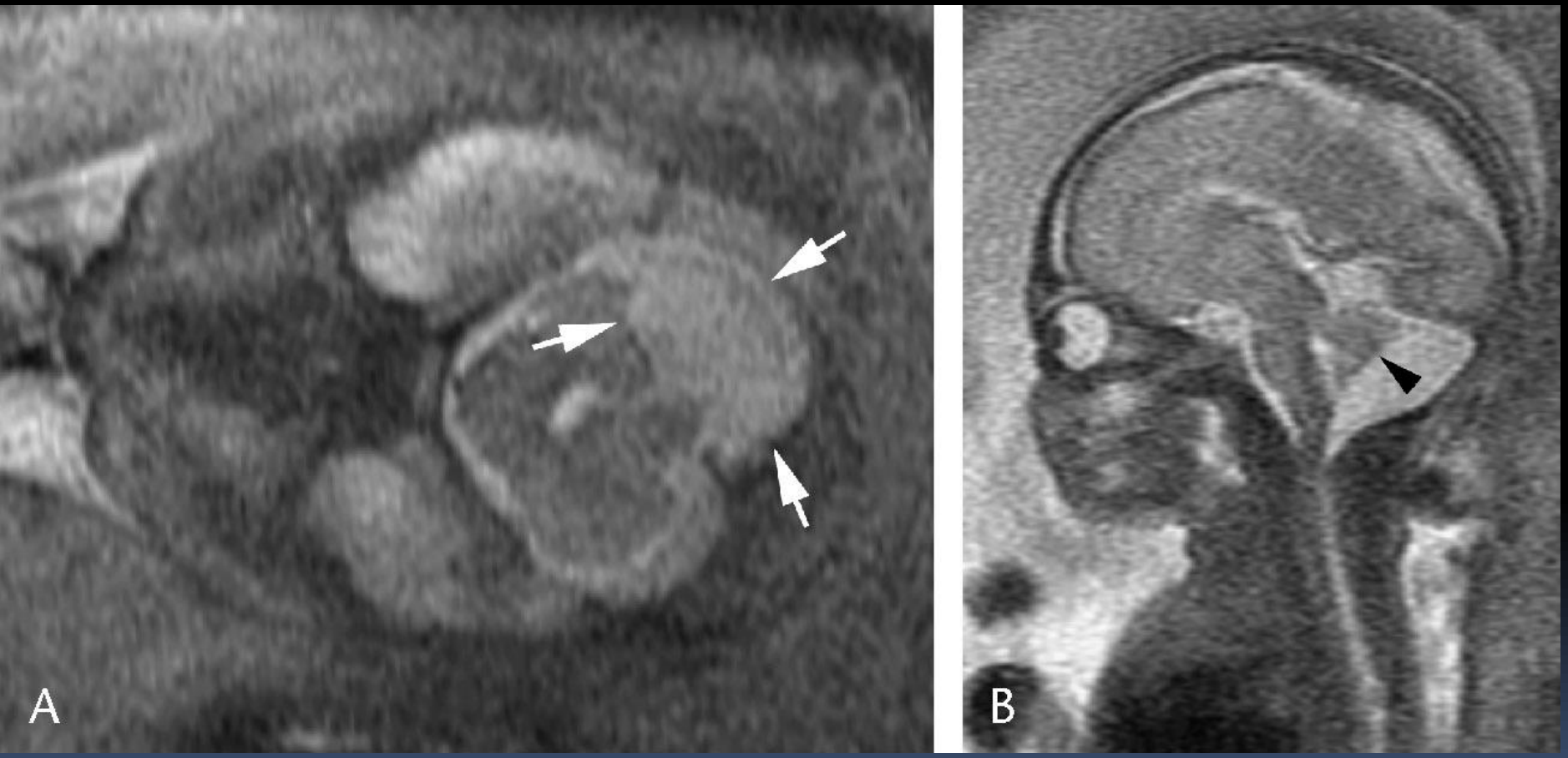
Posterior Fossa Arachnoid Cyst

- The clinical presentation depends on the size, age of the patient, location and presence or absence of complications (hemorrhage)
- Hydrocephalus may be present in 30-100% of patients
- Ataxia and calvarial asymmetry are common

Posterior Fossa Arachnoid Cyst

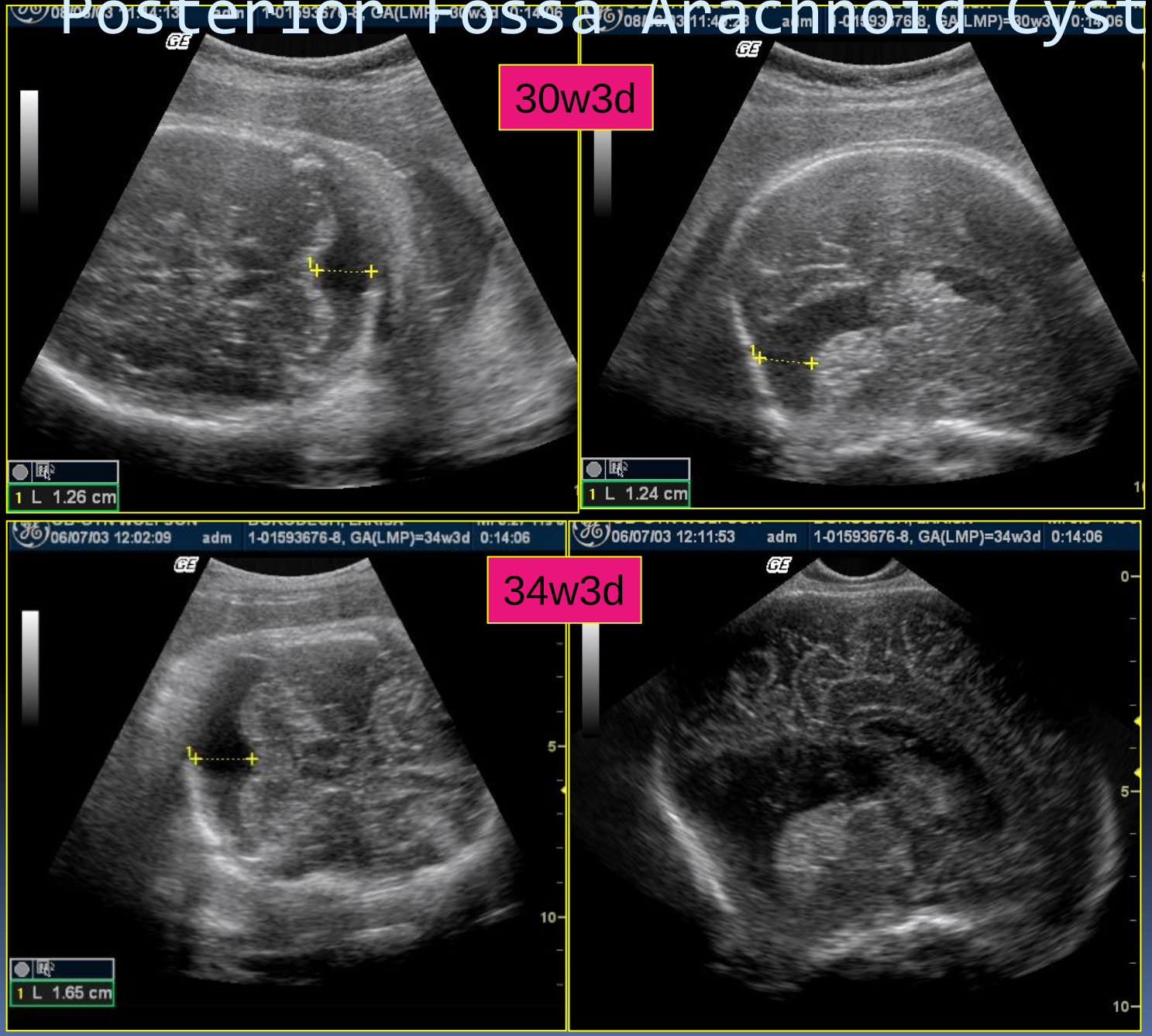
- Criteria for diagnosis:
  - The cyst must be of CSF density
  - There should be no associated mass lesion or
  - enhancement
  - The surrounding brain must be normal
- The vermis and cerebellar hemispheres may be compressed
- Treatment consists of surgical resection of the cyst wall and/or shunting

Posterior Fossa Arachnoid Cyst



27 weeks

Posterior Fossa Arachnoid Cyst

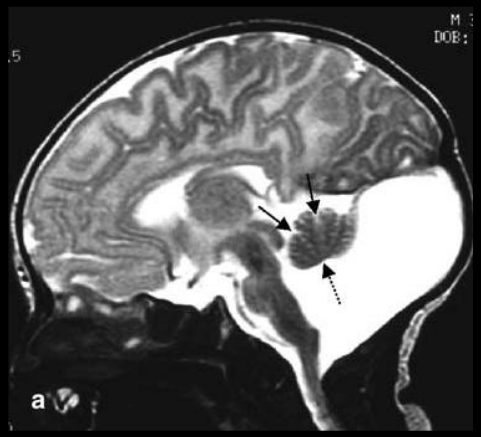


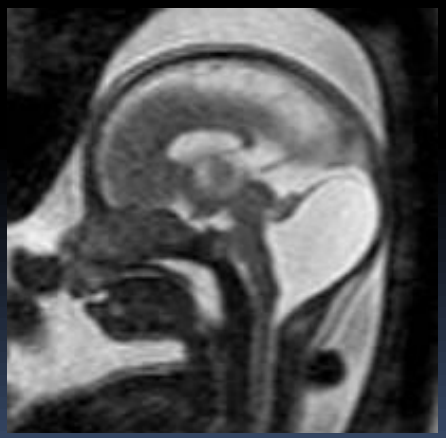
Outcome of Posterior Fossa Cystic Lesions

- Mega cisterna magna has a favorable outcome in 92–100% of cases
- Isolated Blake's pouch cysts have no consequences on neurodevelopment. The main concern is to rule out inferior and posterior vermian agenesis
- Arachnoid cysts are sporadic and incidentally discovered in 2.6% of the pediatric population
- The great majority remain stable
- If the cyst interferes with CSF circulation, surgery may be required; when possible, endoscopic fenestration is performed

Outcome of Posterior Fossa Cystic Lesions

- The overall rate of abnormal neuro-developmental outcome in children with a prenatal diagnosis of DWM is 58.2% and varies from 0–100%
- Prognosis depends on associated CNS abnormalities and whether there is early onset of hydrocephalus
- When isolated, it may be asymptomatic or manifest with: hypotonia, hydrocephalus, intellectual disability, and epilepsy
- Cerebellar signs are less frequent
- Neurodevelopmental outcome can be normal when the vermis is normally lobulated





Cerebellar Disorders Associated with Brain Stem Anomalies

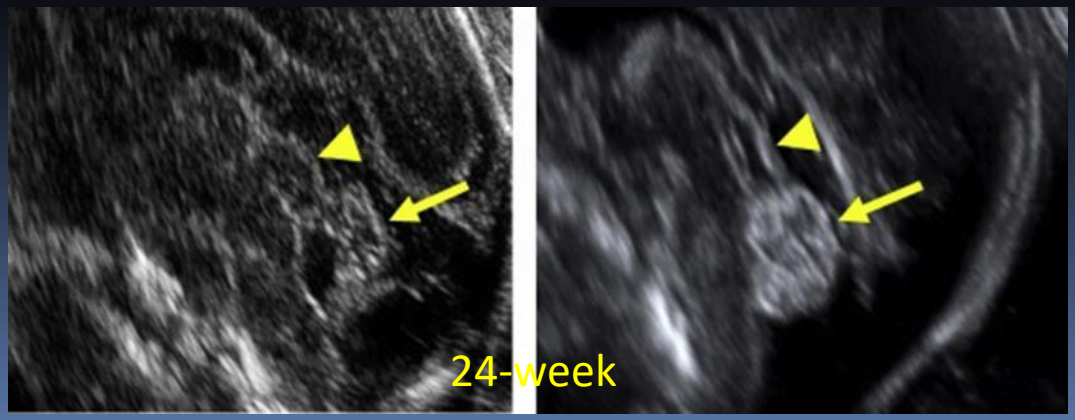
Courtesy of Dr Michelle Fink, RCH Melbourne, Australia

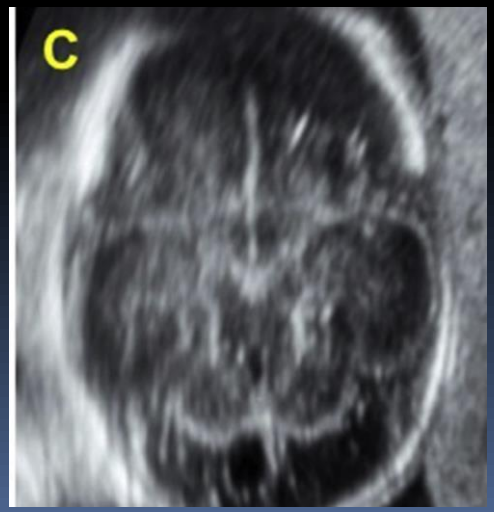
Joubert Syndrome Related Disorders

- Rare inherited cerebellar ataxia-1:100.000 births
- The pathogenesis-defective function of primary cilia (ciliopathy) which affects brain, kidney, liver retina and tubular bone development
- The molar tooth sign is pathognomonic for diagnosis of JSRD
- The MTS can be diagnosed prenatally by US and MRI especially in the 3rd trimester, but a partial MTS has been described as early as 12 GW.

Joubert Syndrome Related Disorders

- The molar tooth sign Is observed in lower axial views at the level of the midbrain:
  - Thickened and elongated superior cerebellar peduncles (SCP)
  - Narrow pontomesencephalic junction
  - Deep interpeduncular fossa
  - Absent or dysplastic vermis

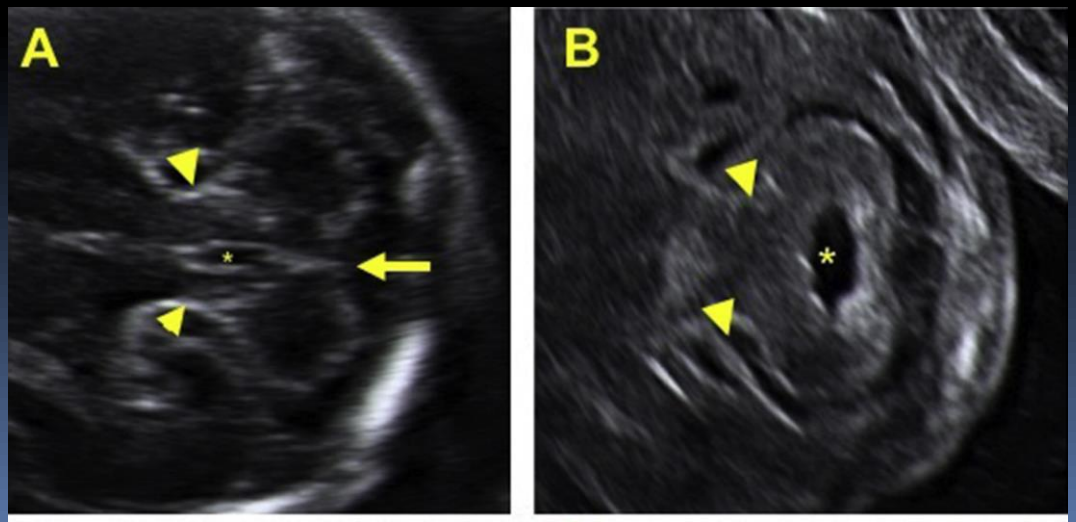




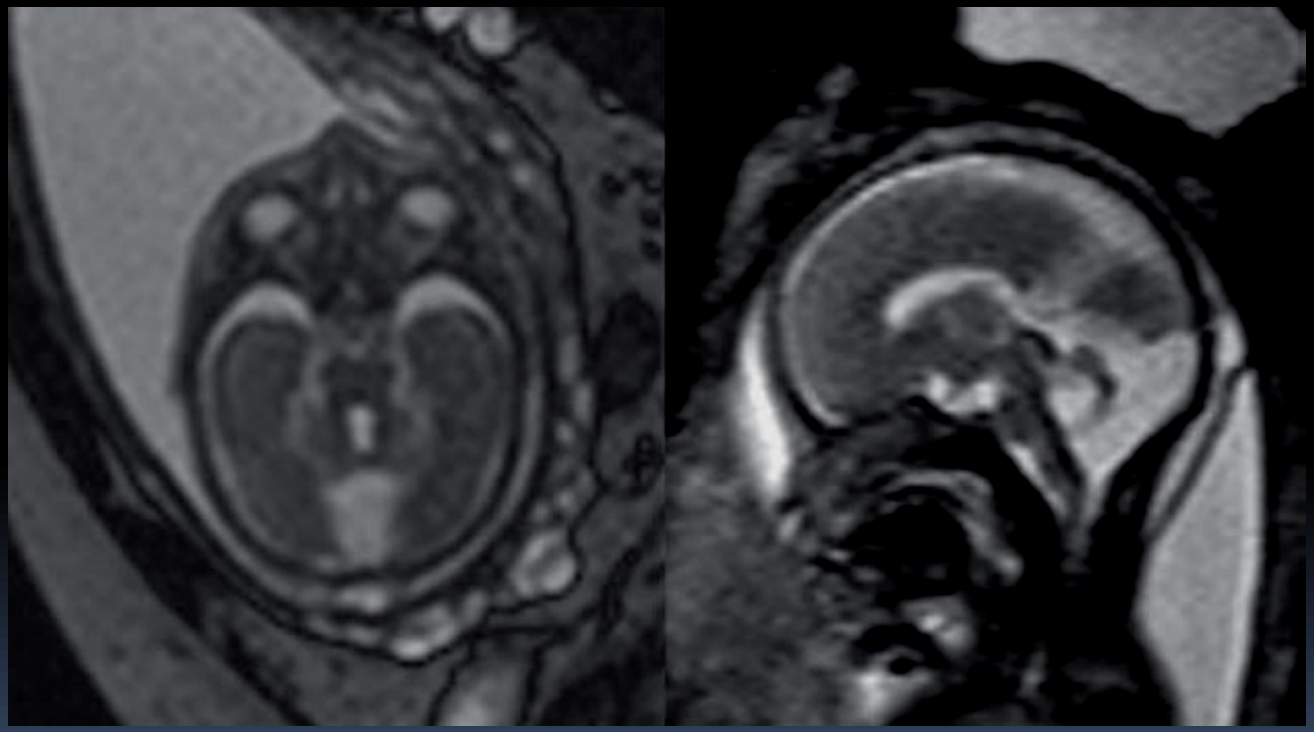
20-week

Joubert Syndrome Related Disorders

- Indirect signs:
  - Abnormal morphology of the 4th ventricle (4v) in axial views-elongated antero-posteriorly rather than latero-laterally – inverted proportions)
  - Enlarged and quadrangular 4v, and flat fastigium on sagittal views.
  - Vermian dysplasia identified by a small biometry and lack of the primary and secondary fissures



Joubert Syndrome Related Disorders-MRI



Joubert Syndrome Related Disorders-Clinical Features

- Hypotonia
- Abnormal breathing pattern
- Ocular motor apraxia
- Ataxia
- Intellectual disability of varying degrees
- Chorioretinal coloboma
- Polydactyly
- Cystic or echogenic kidneys
- Leber amaurosis

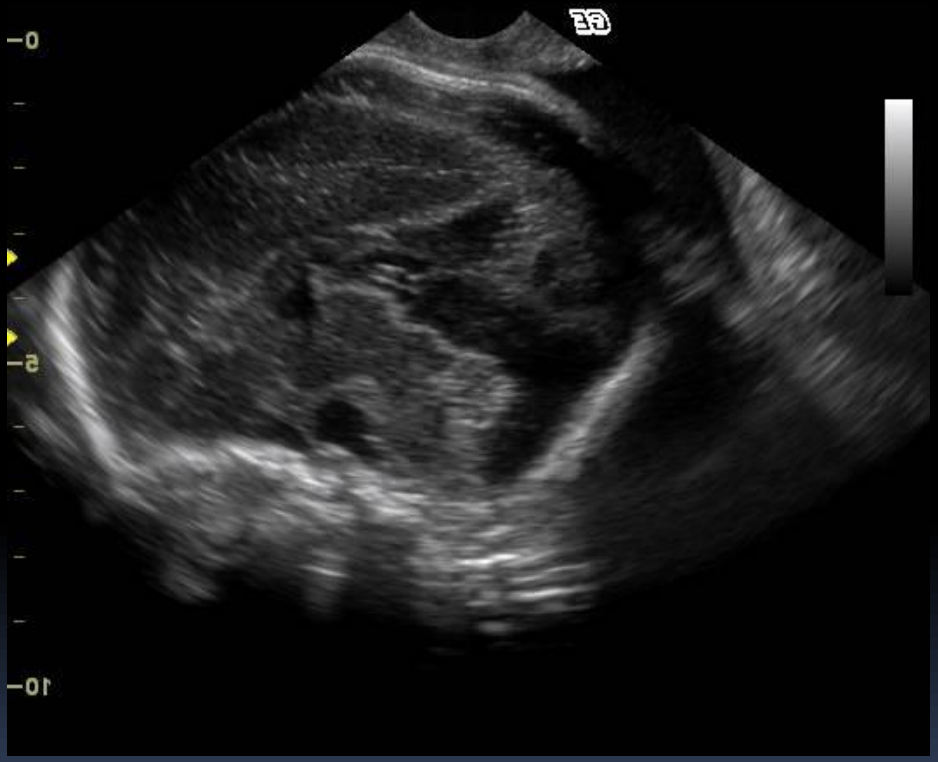
Joubert Syndrome Related Disorders-Associated brain Malformations

- Polymicrogyria
- Agenesis of corpus callosum
- Cortical and brainstem heterotopia
- Pontine and medullary tegmental cap dysplasia

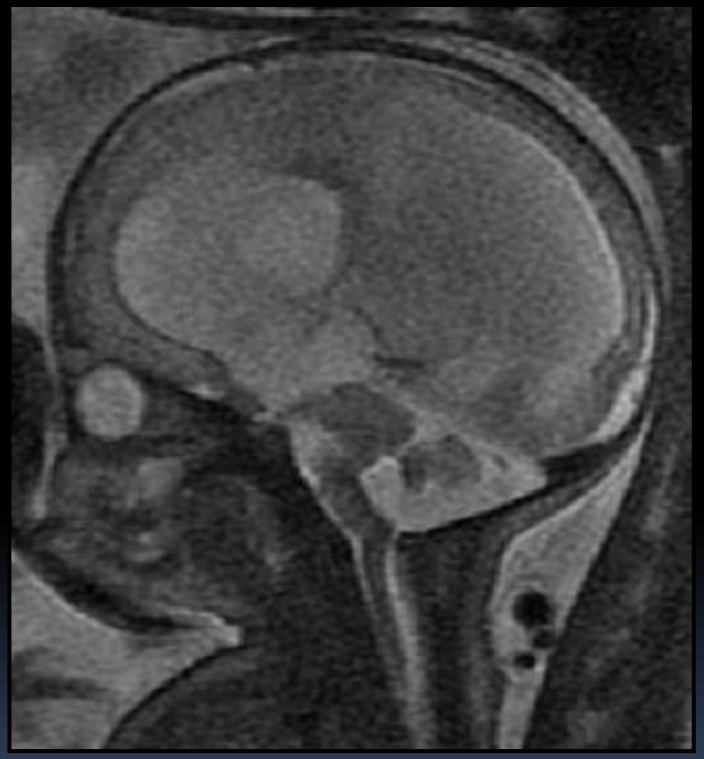
Cobblestone Malformation
Walker-Warburg Syndrome

- Autosomal recessive disorder
- Cobblestone malformation, kinked brainstem, cerebellar cysts, retinal dysplasia and congenital muscular dystrophy
- Associated abnormalities: cleft lip and palate, occipital encephalocoele, microophthalmia, corneal opacities
- Children present with progressive hydrocephalus and severe neurologic dysfunction: profound mental retardation, intractable seizures, hypotonia

Walker-Warburg Syndrome

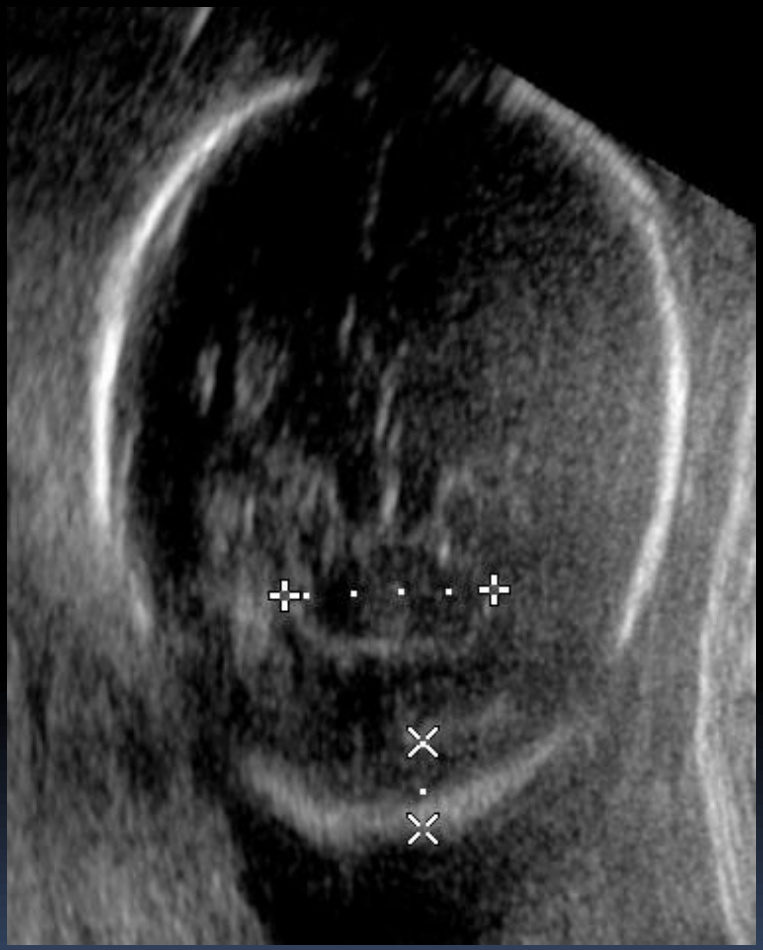


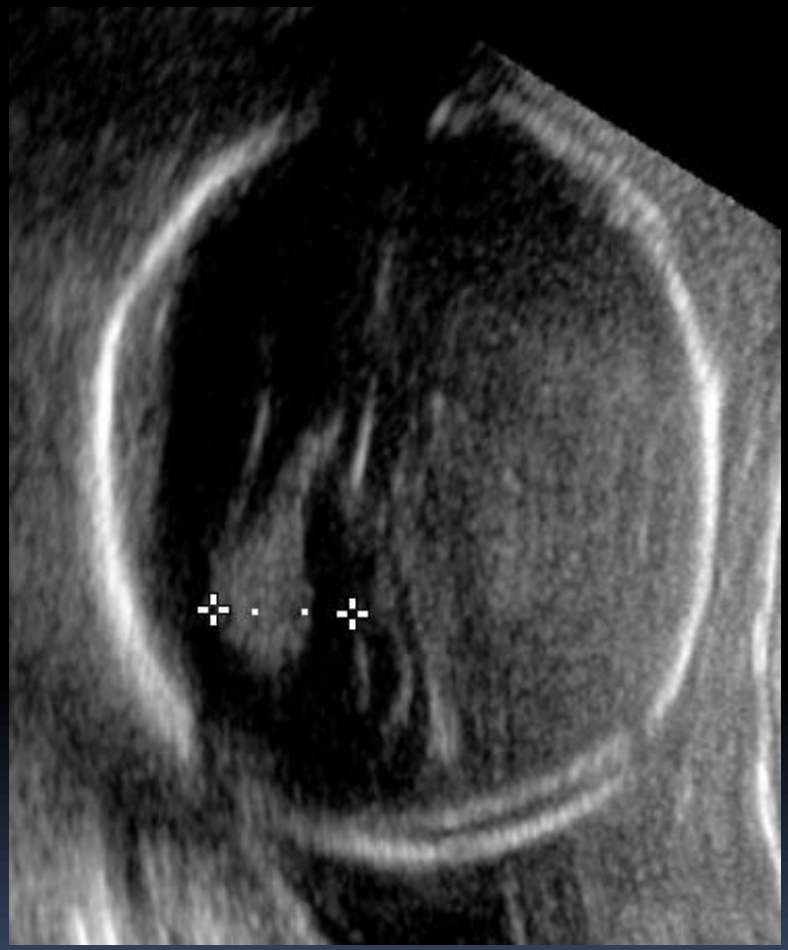
29 weeks



34 weeks

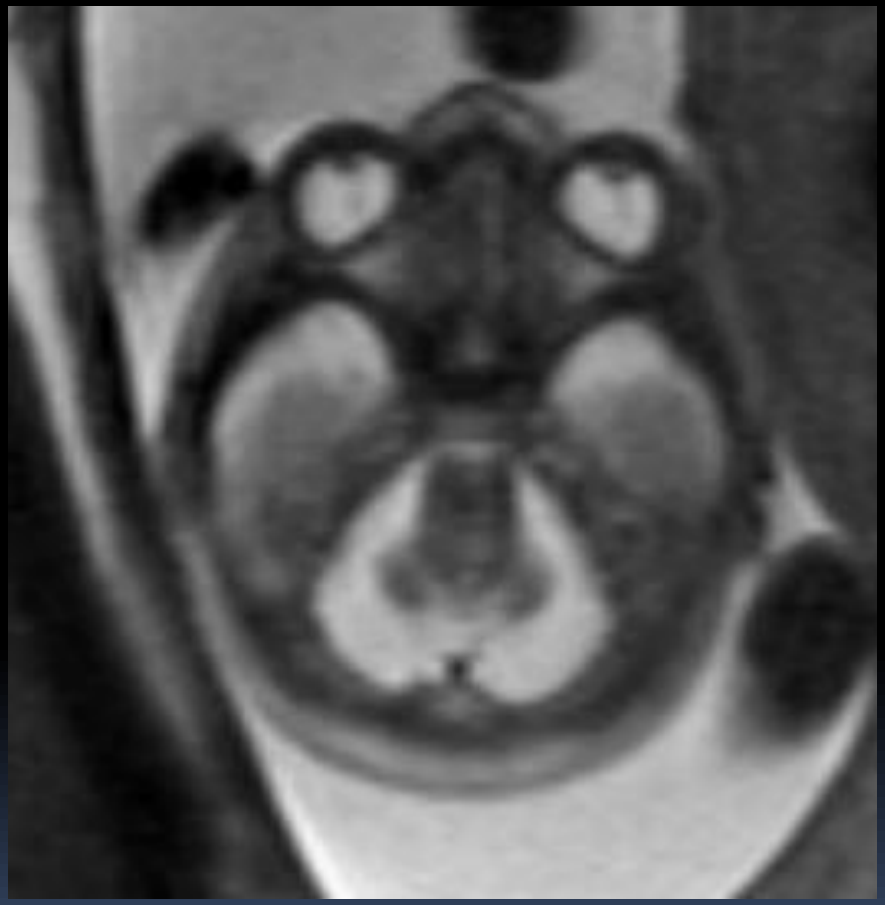
Pontine Cap Dysplasia



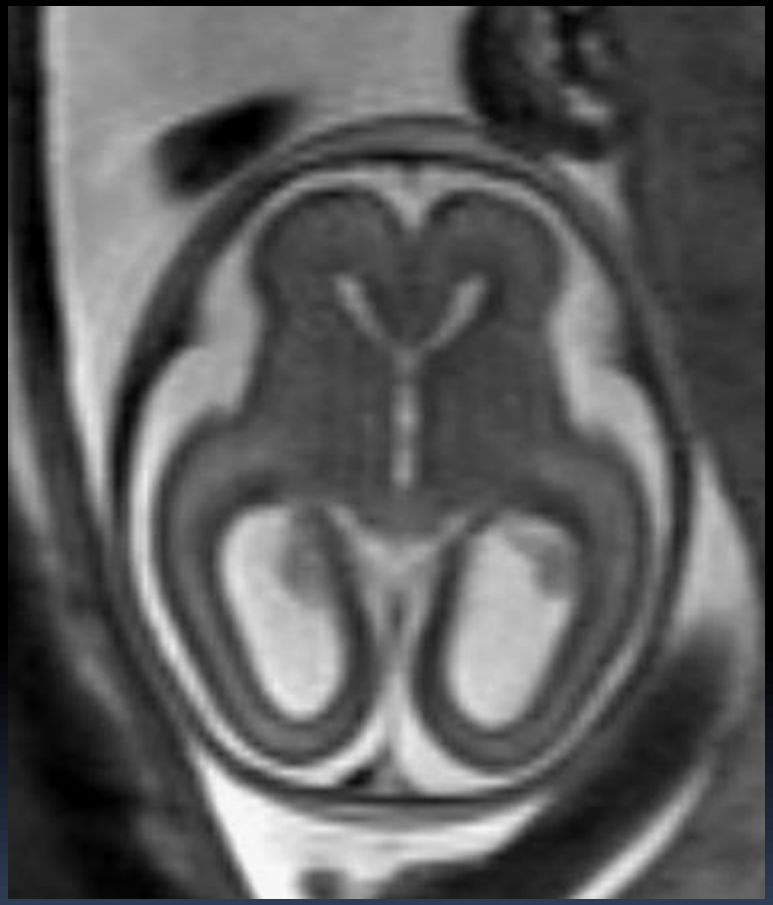


Cerebellum 18 mm
Nuchal fold 7.5 mm

Lat Vent 12 mm

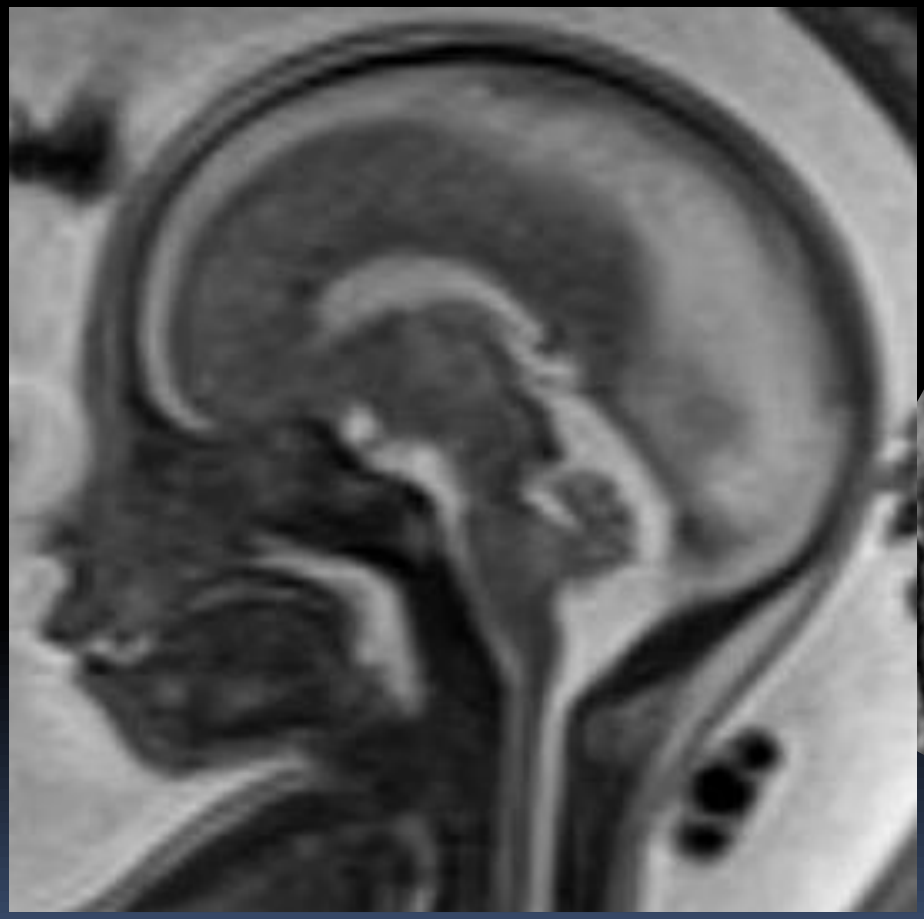


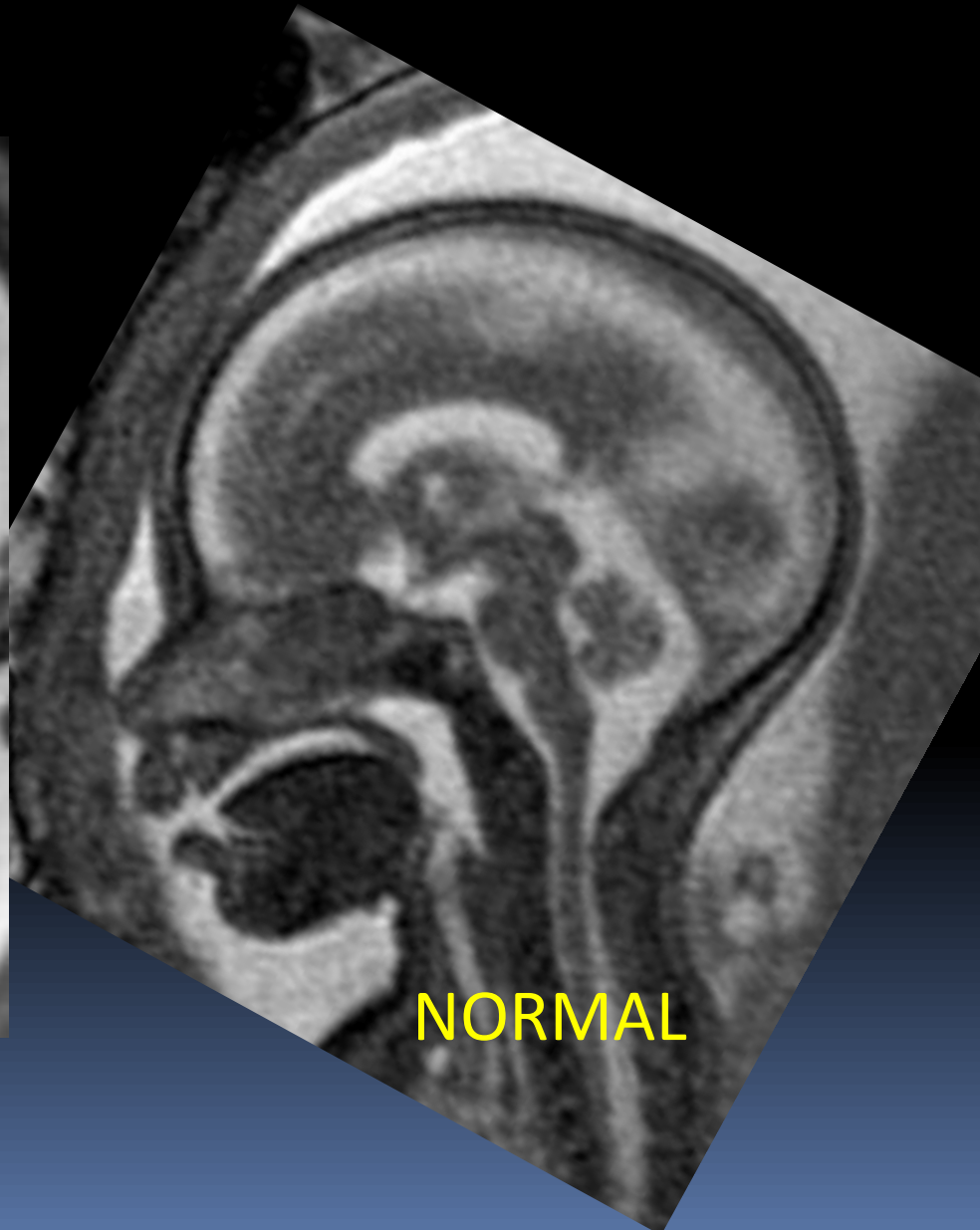
Very hypoplastic cerebellum



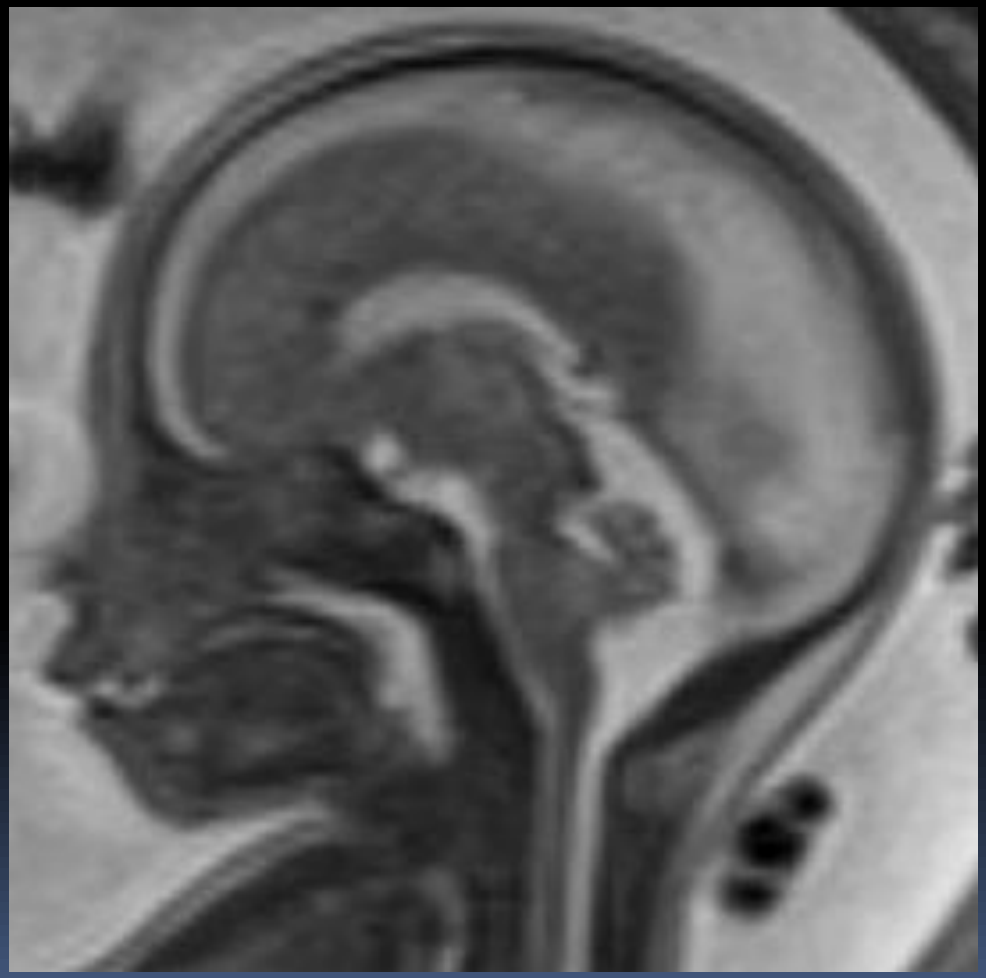
VM

Pontine Cap Dysplasia





Pontine Tegmental Cap Dysplasia

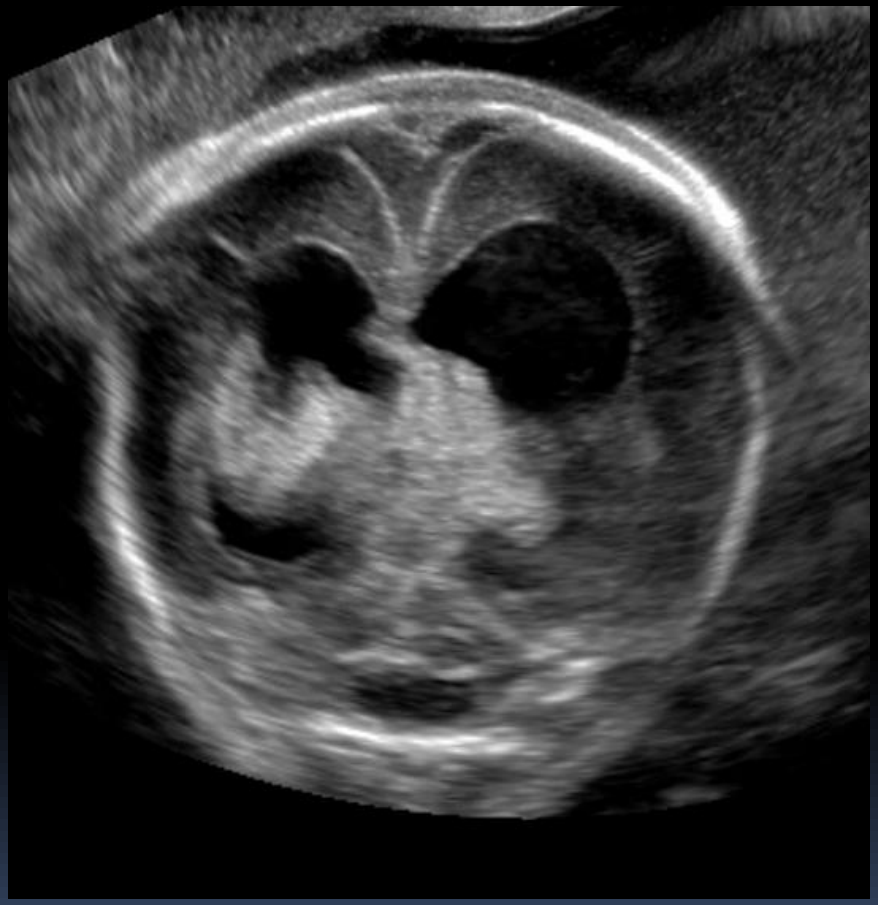


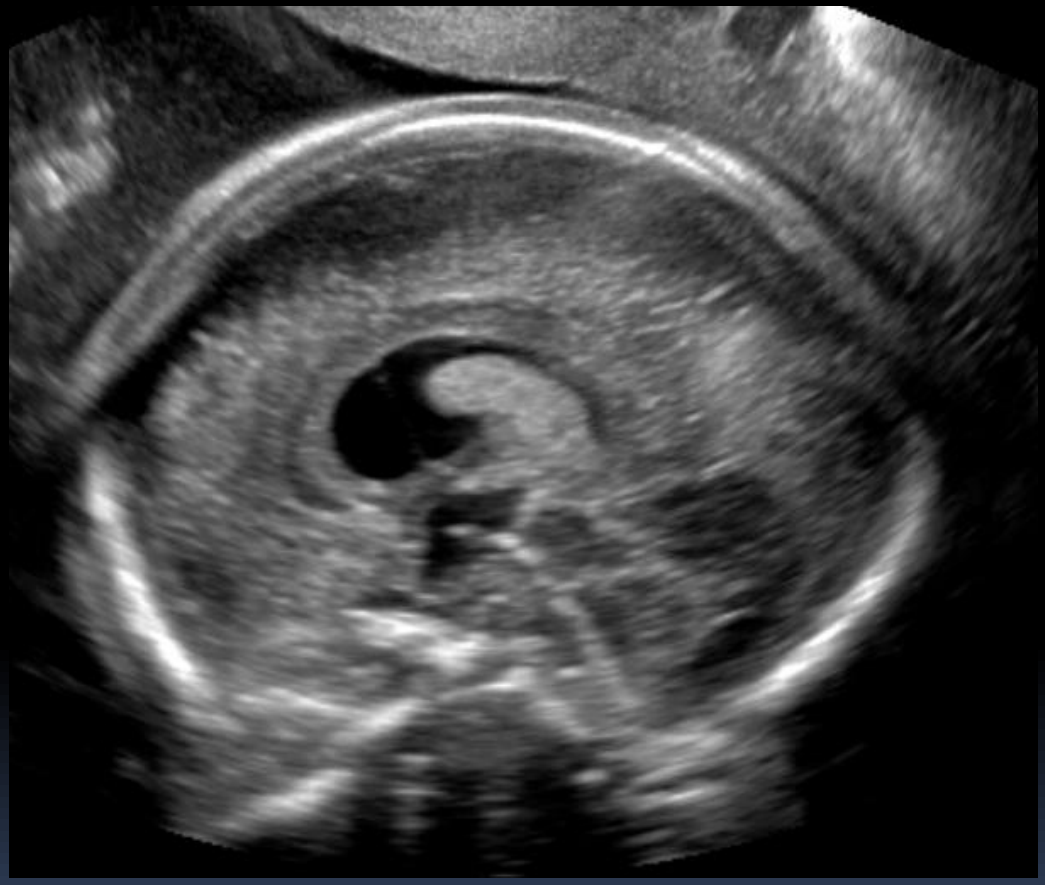
- Dorsal pontine protruberance into V4 (= ectopic transverse pontine fibers)
- Hypoplastic pontine belly
- Elongated midbrain
- Can be associated with Joubert syndrome

Pontine Tegmental Cap Dysplasia

- Multiple cranial neuropathies
  - Cochlear nerves (deafness)
  - Facial nerve
  - Trigeminal sensory nerve with corneal anesthesia
  - Glossopharyngeal nerve causing swallowing disorders necessitating gastrostomy
- Ocular movement abnormalities
- Speech impairment / mutism
- Ataxia
- Developmental delay / failure to thrive
- Associations:
  - Rib and / or vertebral malformations
  - Congenital heart defect
  - Renal anomalies

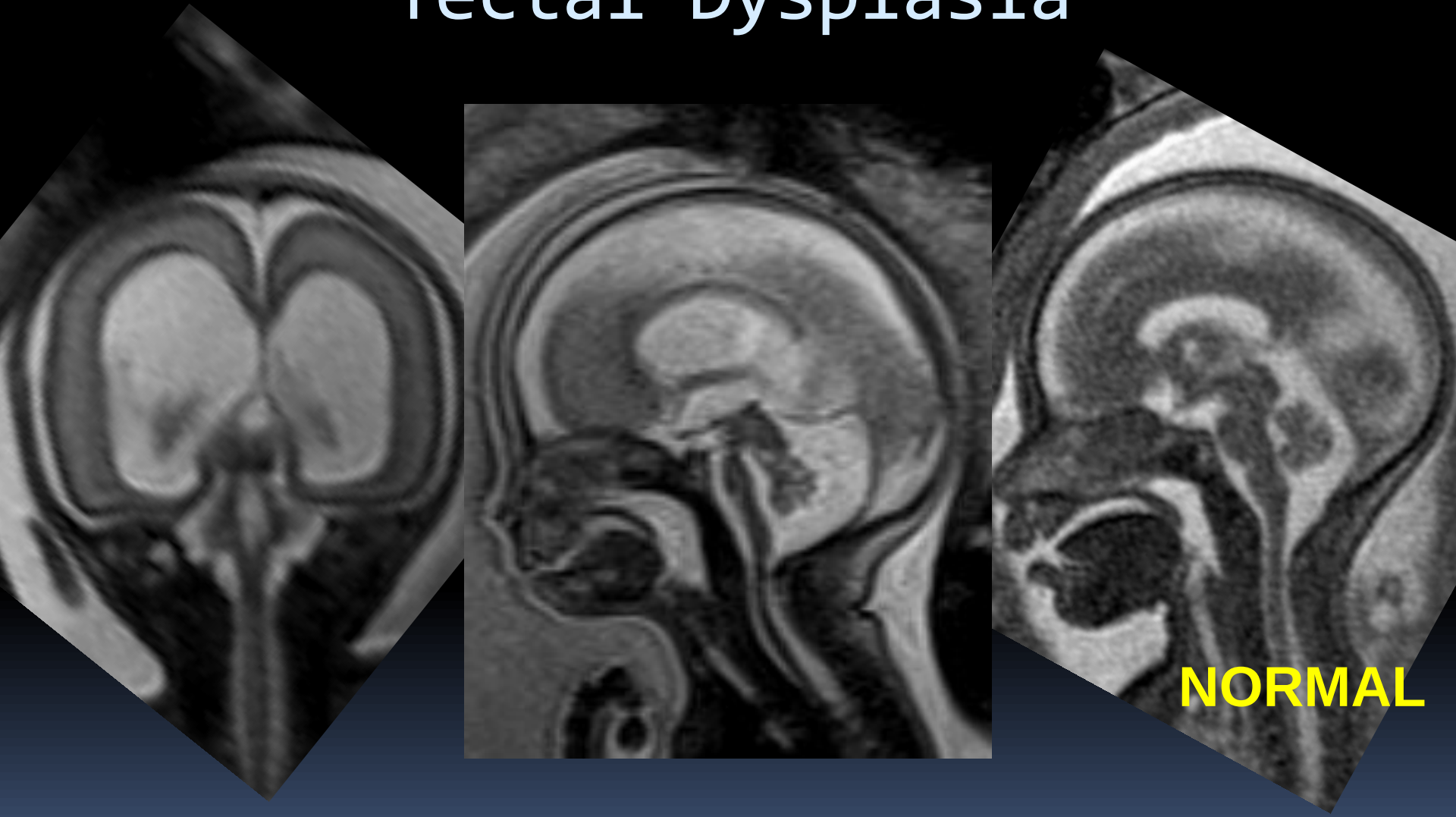
Tectal Dysplasia



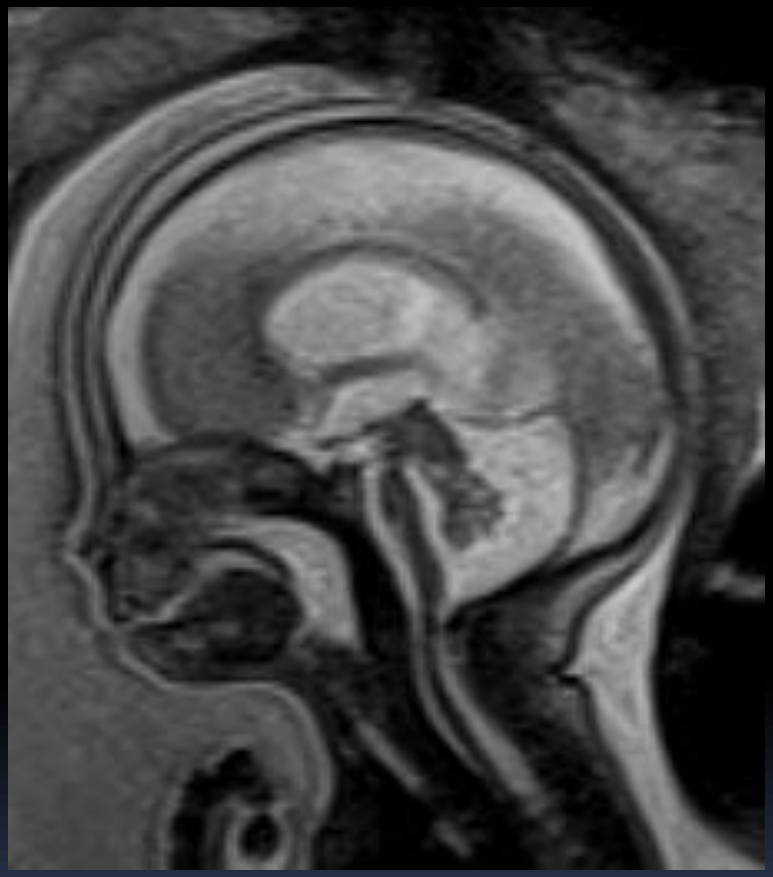


US images: Dr Amanda Sampson
24 weeks

Tectal Dysplasia



Tectal Dysplasia

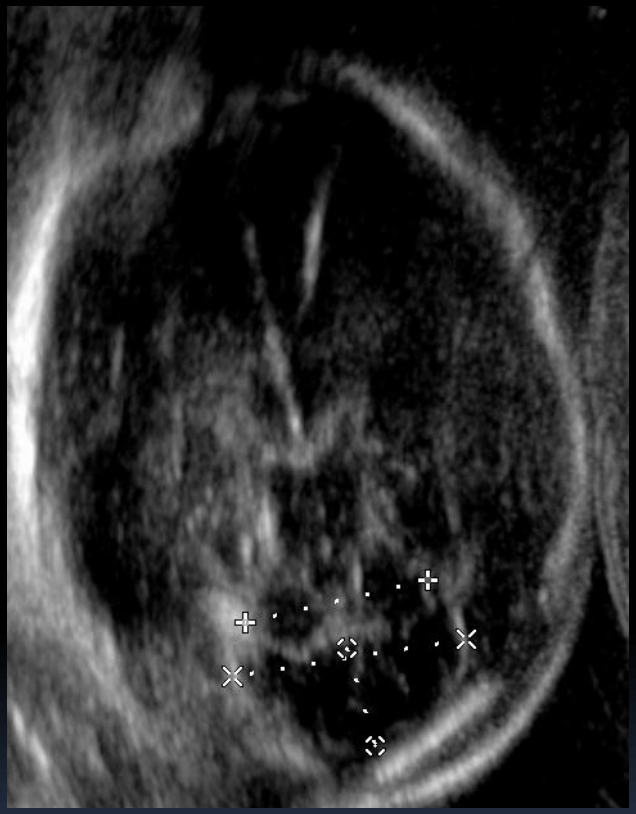


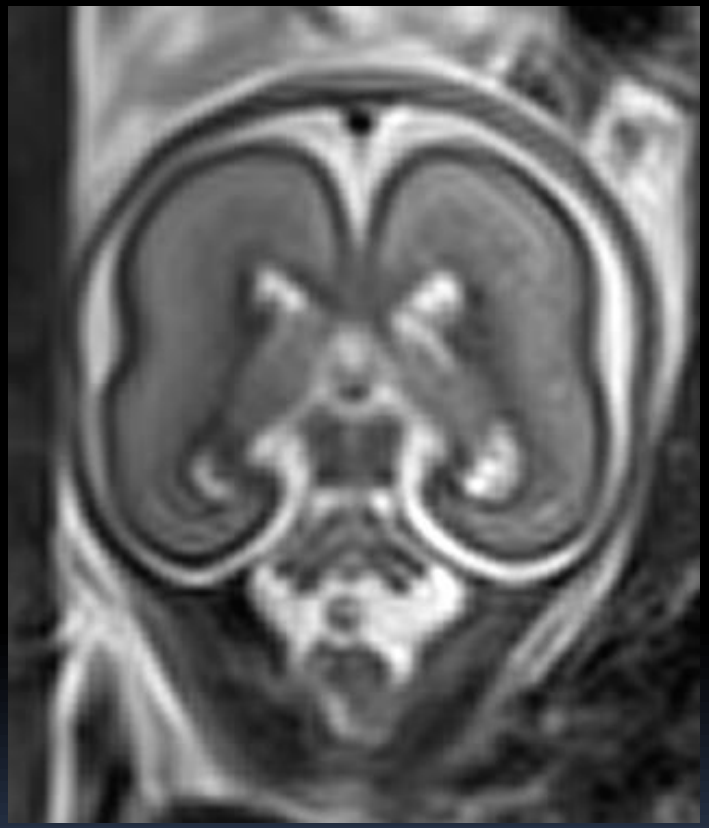
- Hypoplastic midbrain and pons
- Bulky overhanging tectal plate
- Severe VM
- Delayed sulcation
- Enlarged quadrigeminal plate
- Mechanism not known

Tectal Dysplasia in Delleman Syndrome

- Oculocerebrocutaneous syndrome, a disorder characterized by a colobomatous ocular malformation, skin appendages, and cerebral malformations:
  - Unilateral frontal PMG with PVNH
  - Agenesis of the corpus callosum
  - Large dysmorphic tectum
  - Absent cerebellar vermis
  - Small cerebellar hemispheres

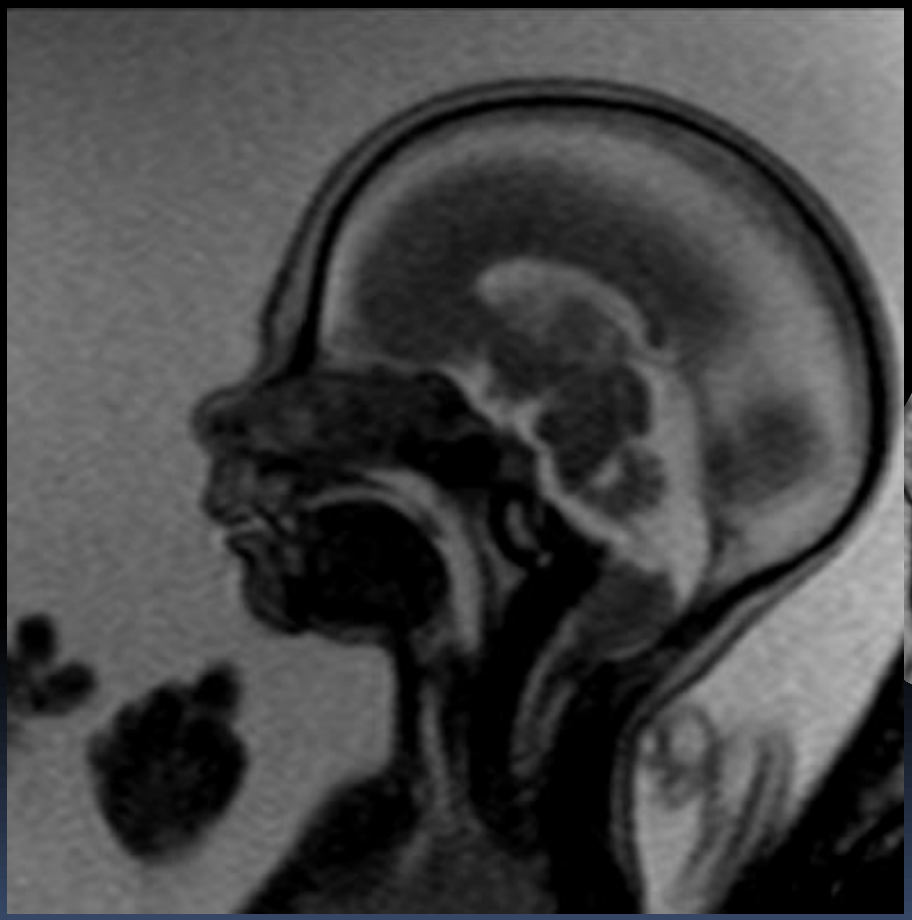
Brainstem Disconnection Syndrome

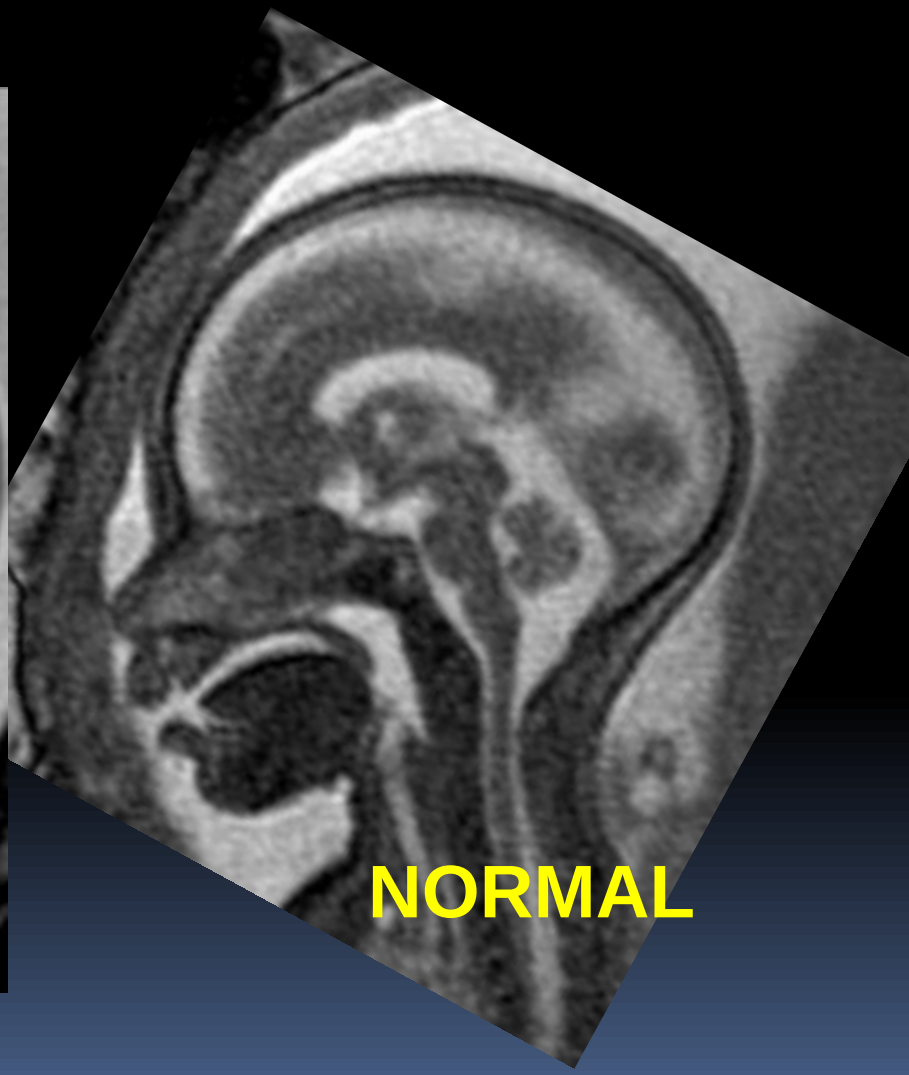




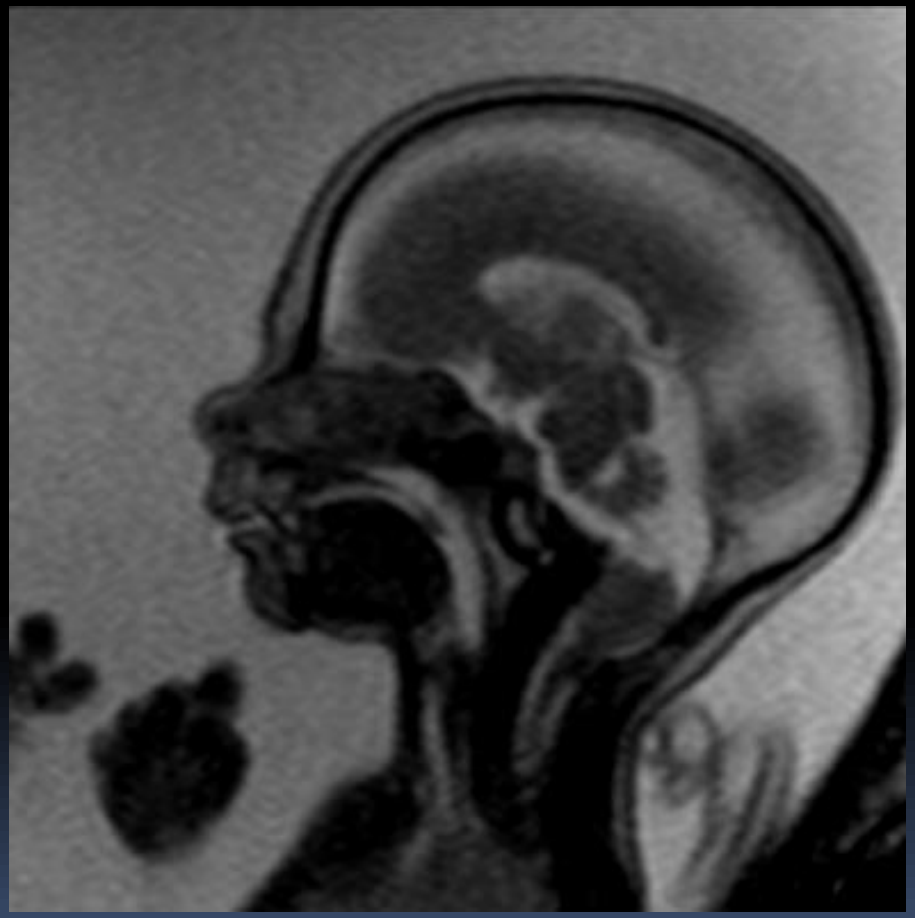
Cerebellum 15.5 mm
21 w

Brainstem Disconnection Syndrome



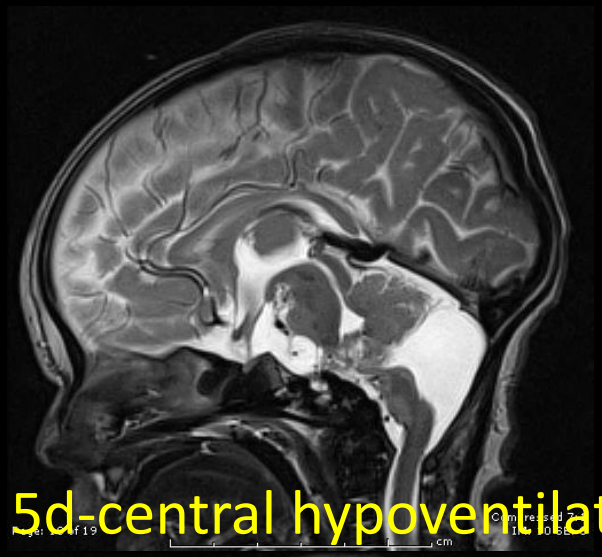


Brainstem Disconnection Syndrome

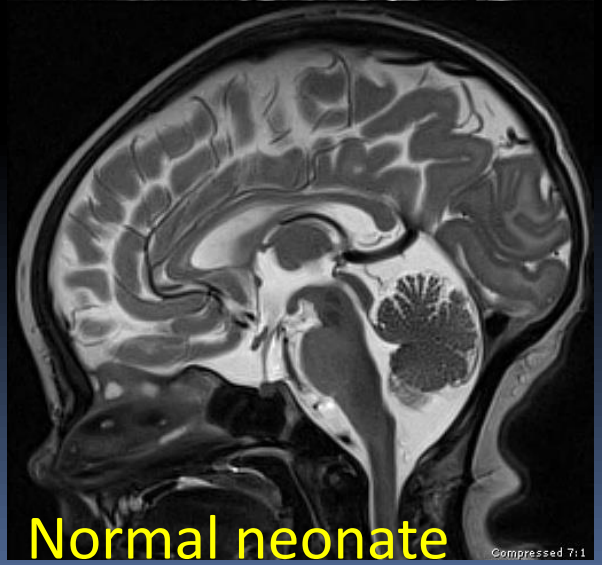


- Stubby dysmorphic midbrain
- Absent pons and normal medulla
- Only faint strand connecting midbrain to medulla

Brainstem Disconnection



5d-central hypoventilation



Normal neonate

- Disconnection can be
  - Between the midbrain and pons (pontomesencephalic) or between the pons and medulla (pontomedullary)
- Associated with vermian and cerebellar hypoplasia
- ? Genetic ? injury related
- Fatal

Conclusions

- Posterior fossa anomalies can be accurately diagnosed in utero by dedicated ultrasonography and MRI
- The diagnosis of vermis hypoplasia should be deferred into the third trimester since delayed closure of the 4^{th} ventricle and persistent Blakes pouch cyst may simulate vermis hypoplasia

Conclusions

- Cerebellar anomalies are often associated with:
  - Malformations of cortical development
  - Brainstem anomalies
  - Systemic anomalies
- The prognosis frequently depends on the associated anomalies
- Isolated enlargement of the posterior fossa carries an excellent prognosis