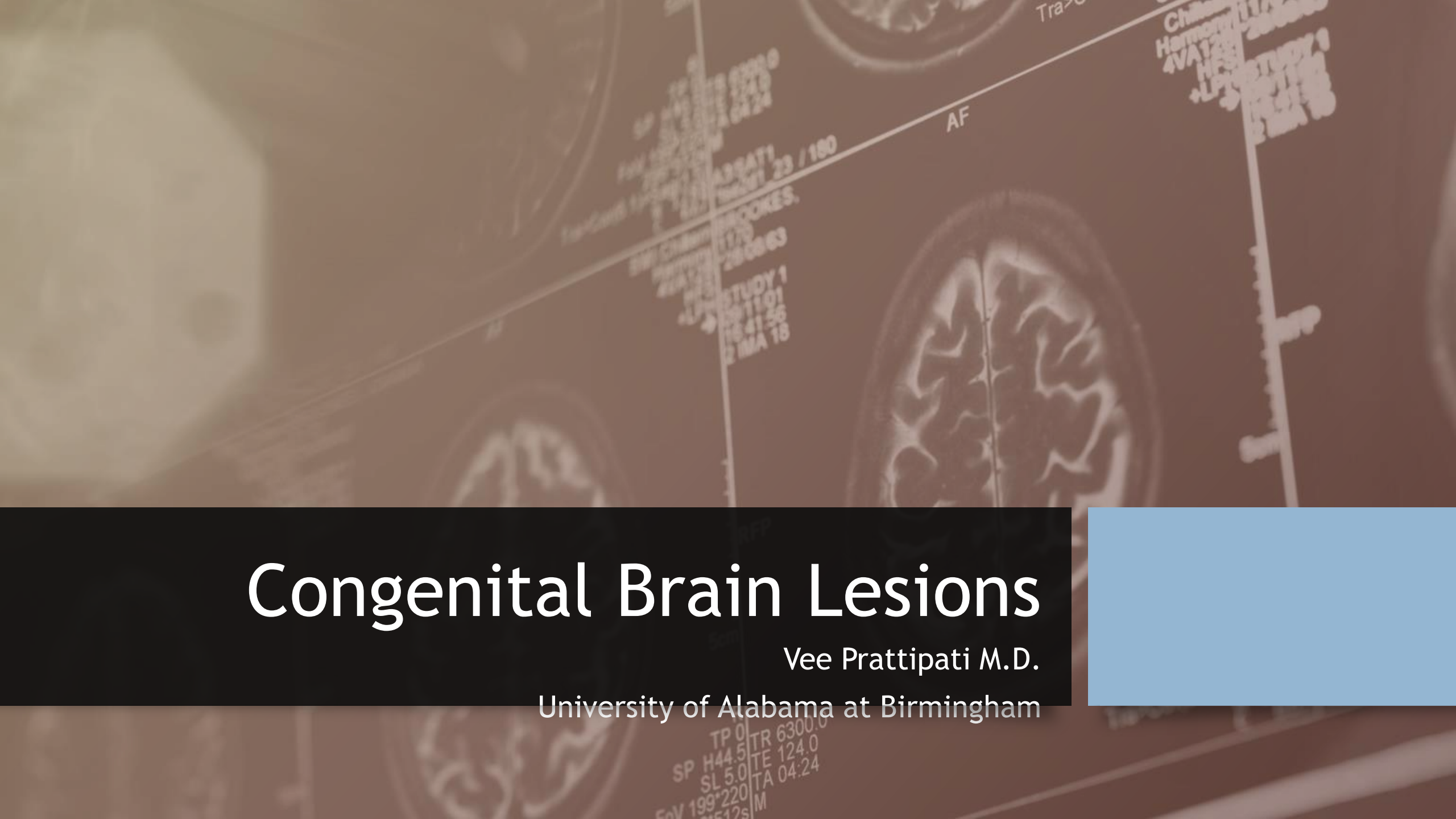




Congenital Brain Lesions

Vee Prattipati M.D.

University of Alabama at Birmingham

Overview

Recognition of most common posterior fossa malformations

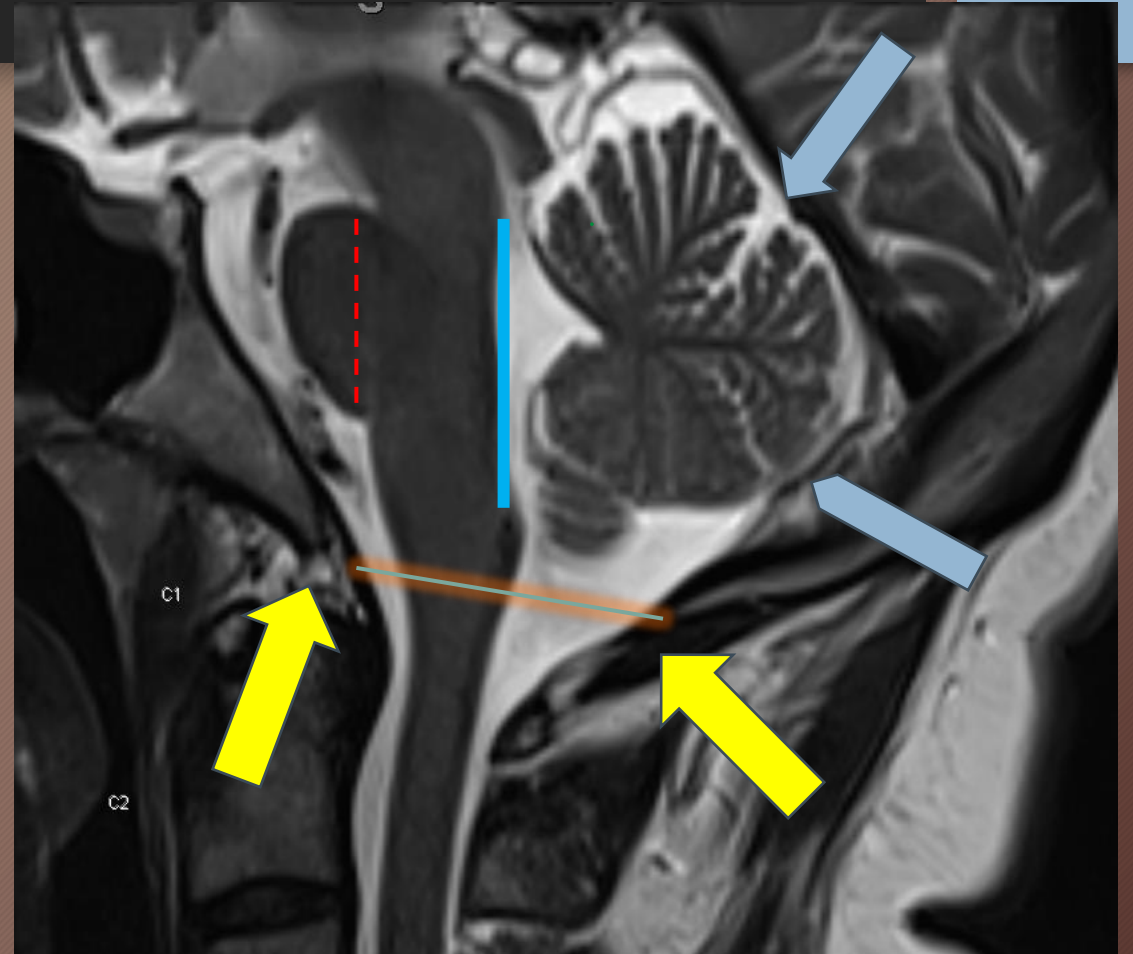
Understanding the spectrum of holoprosencephaly

Recognize the patterns of neuronal migration anomalies.

Congenital syndromes

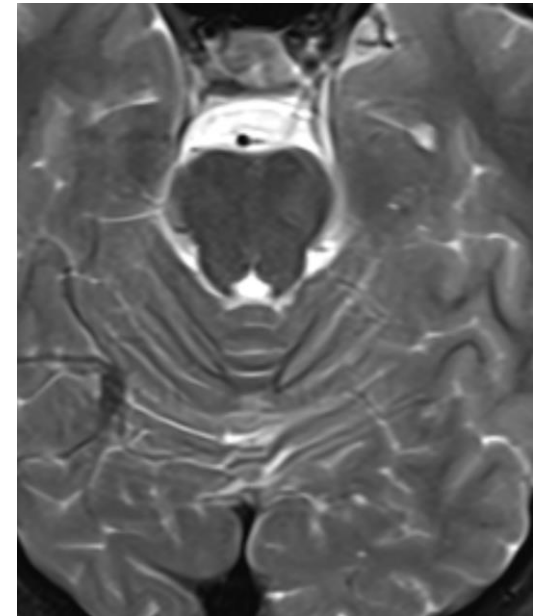
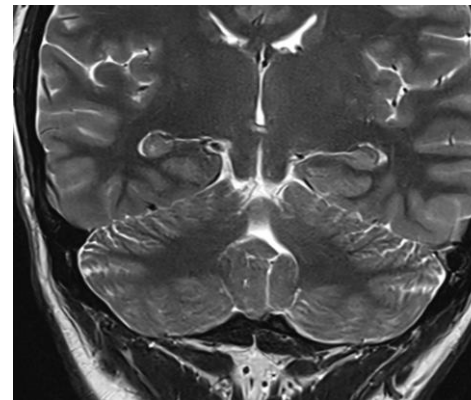
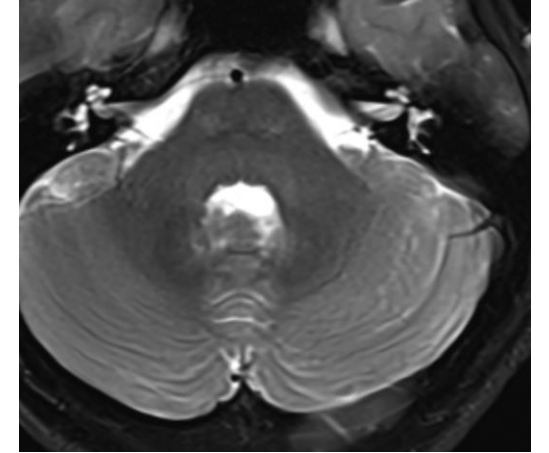
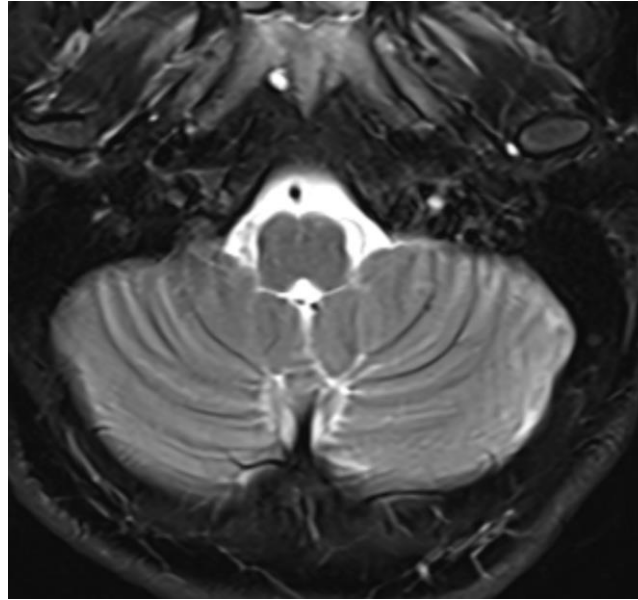
Posterior fossa anatomy

- Vermis
 - Primary Fissure
 - Prepyramidal Fissure
- 4th Ventricle
 - Fastigium angle
- Brain stem
- Pontine length= Midbrain + Medulla combined
- Alignment check
- Foramen Magnum

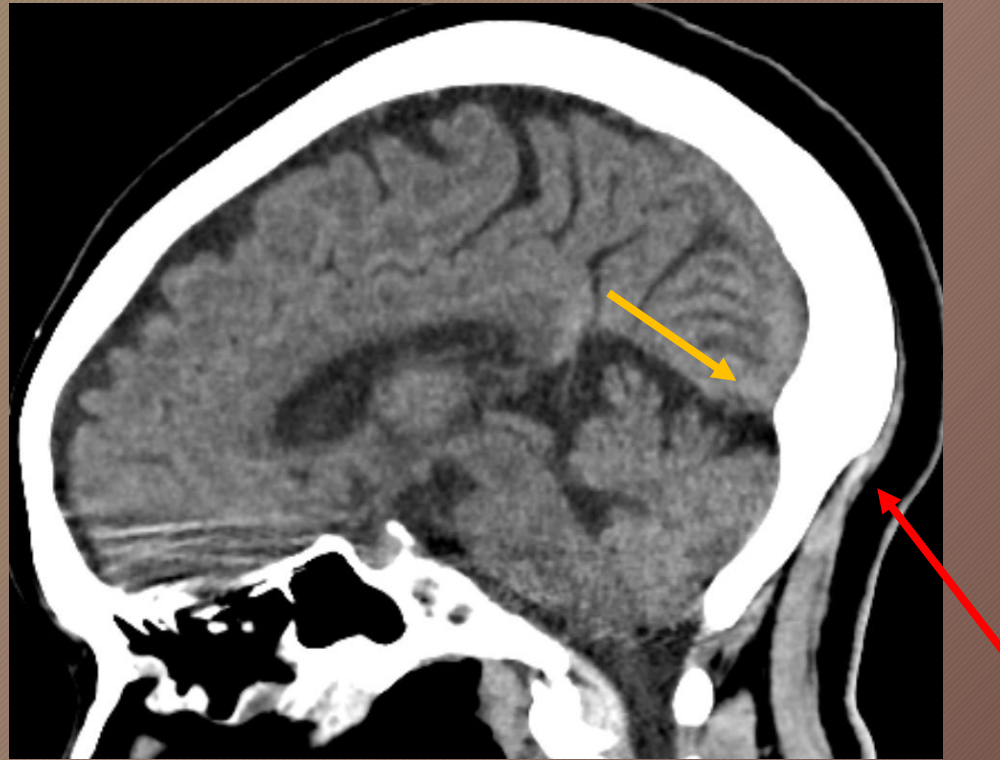


Anatomy

- Axial:
 - Onion like appearance
- Coronal:
 - fissures are radiating surrounding the nuclei



Torcular position



Posterior fossa malformations

1. Chiari I,II,III
2. Dandi-walker spectrum disorders
3. Rhombencephalosynapsis
4. Joubert syndrome

Chiari I Deformity ~~(malformation)~~

- Peg like tonsillar ectopia >5mm
- Syringomyelia-65%
- Scoliosis
- CVJ anomalies



Chiari I Deformity: Beyond 5 mm below the Foramen Magnum

James Ryan Loftus, MD • Catherine Wassef, MD • Shehanaz Ellika, MBBS, MD, DM



Chiari I Deformity ~~(malformation)~~

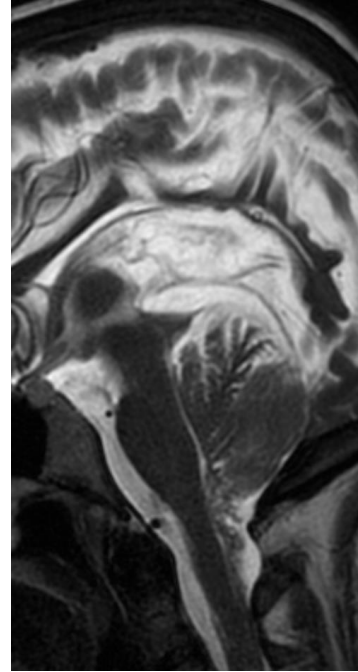
Symptoms

- Asymptomatic
- Neck pain
- Headache
- Numbness
- Ataxia
- Nystagmus
- Syringomyelia
 - CSF flow disruption at FM
 - Increased pressure leading to syrinx
 - MC in cervical spinal cord

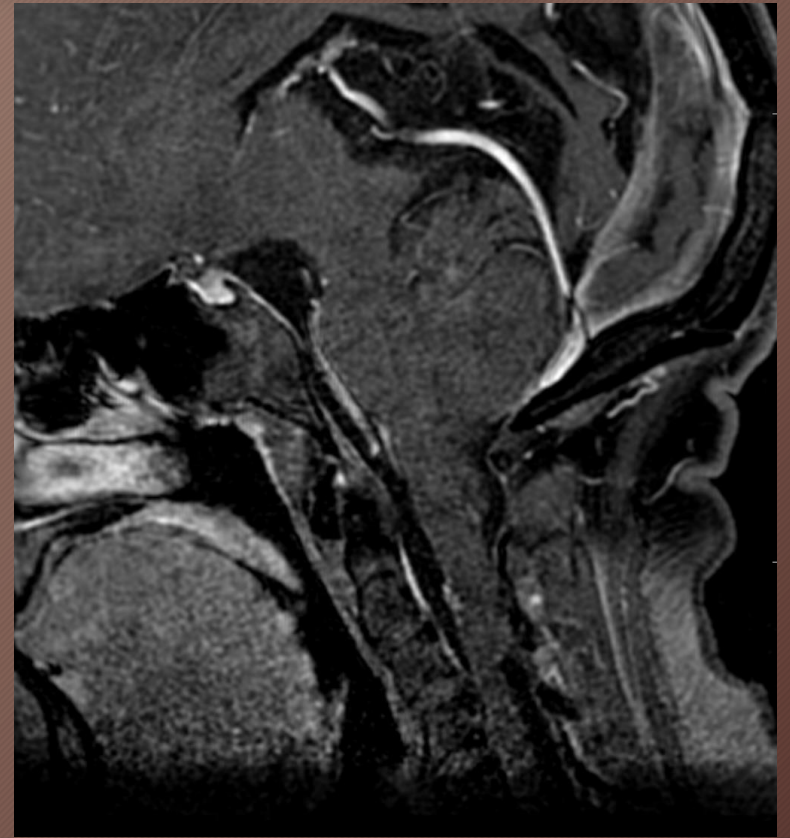
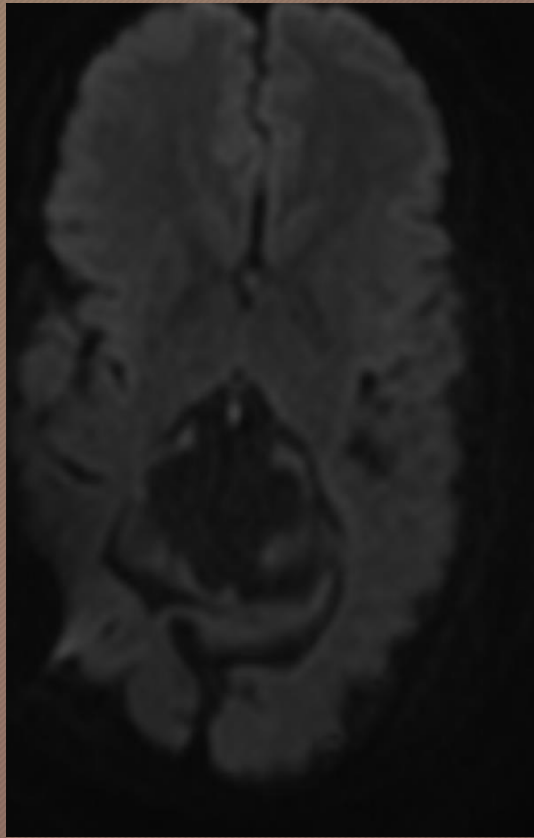
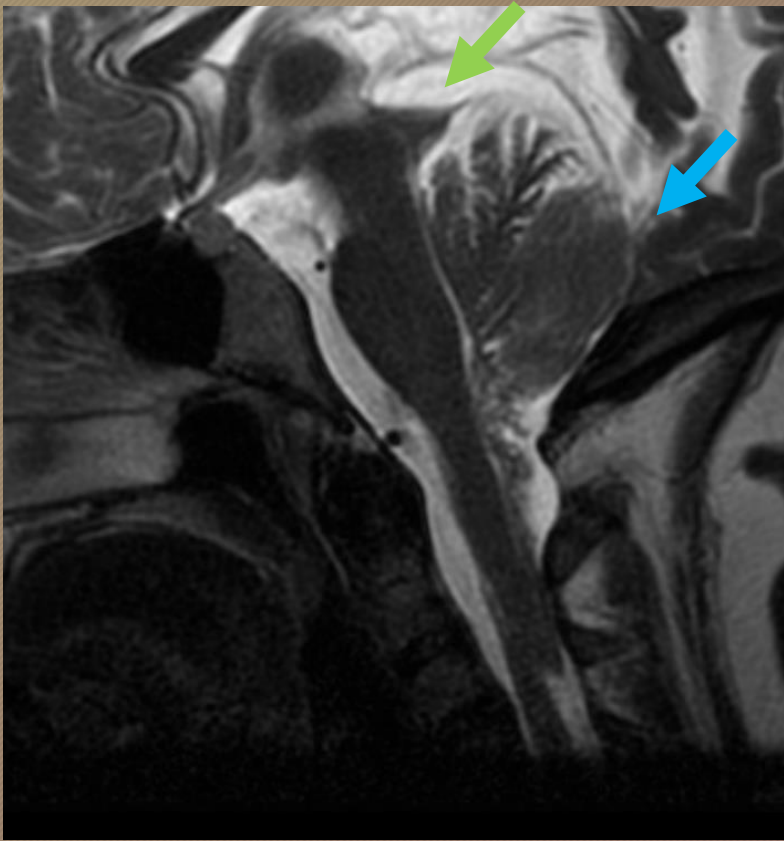


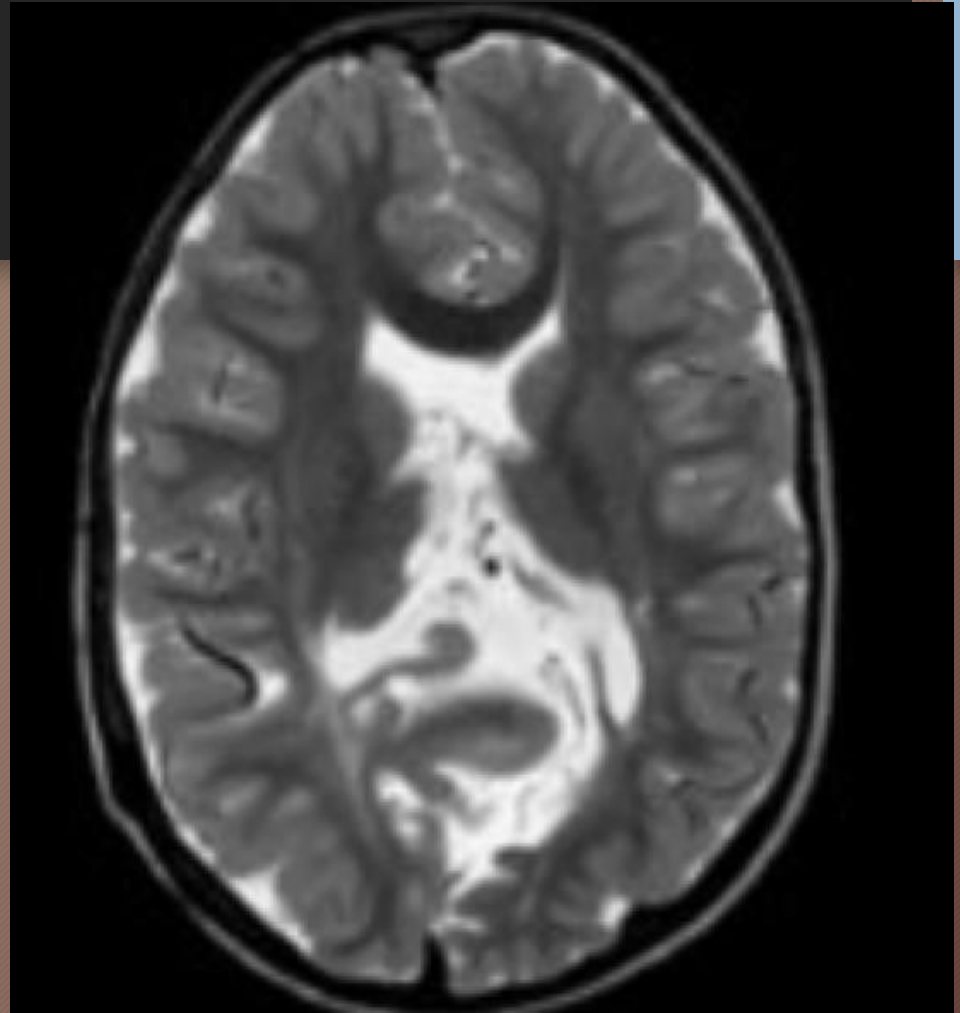
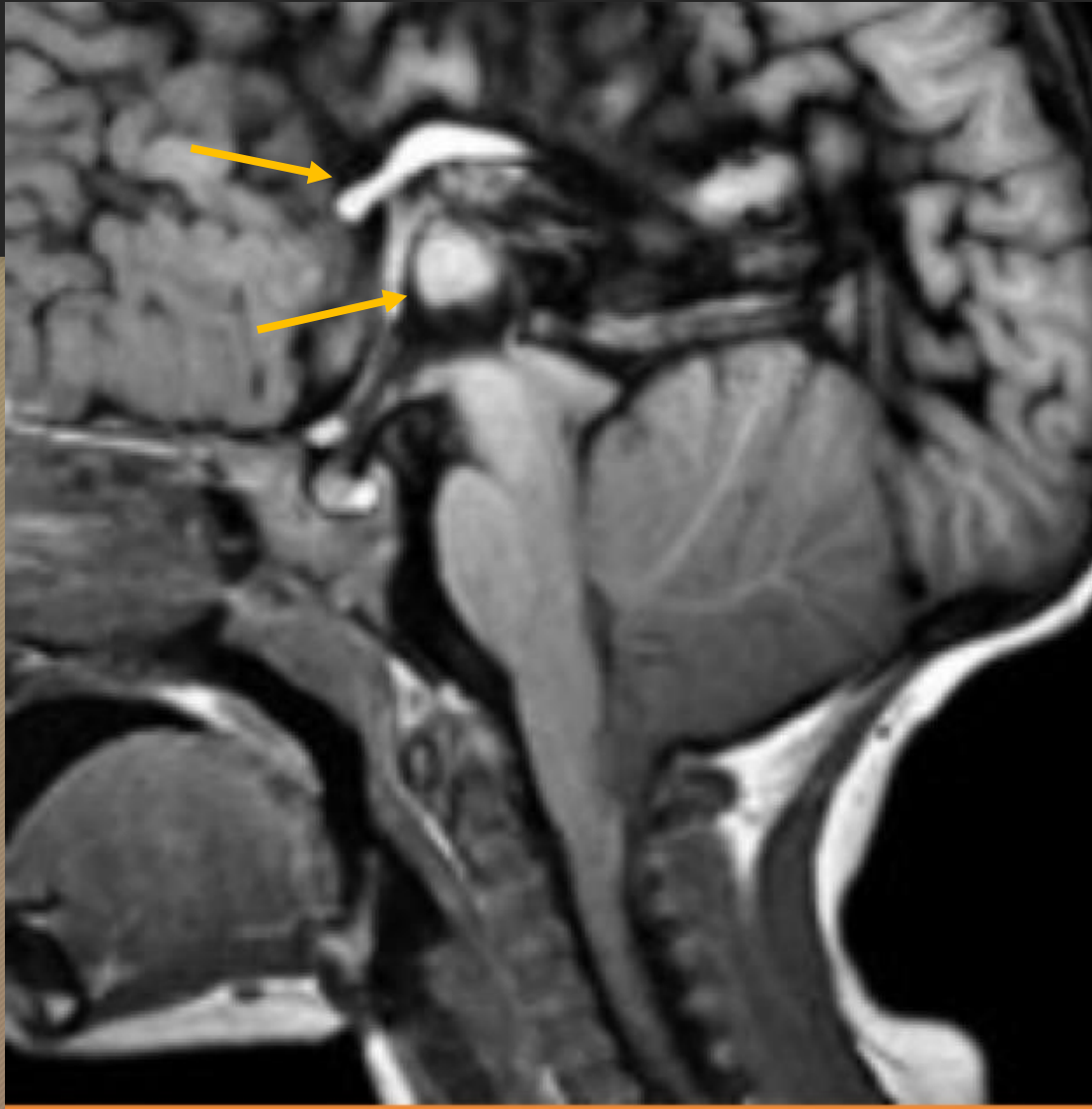
Chiari II malformation

- Open spina dysraphism => CSF leak => brain sagging => small posterior fossa with decreased brain development
- High incidence of associated cerebral malformations

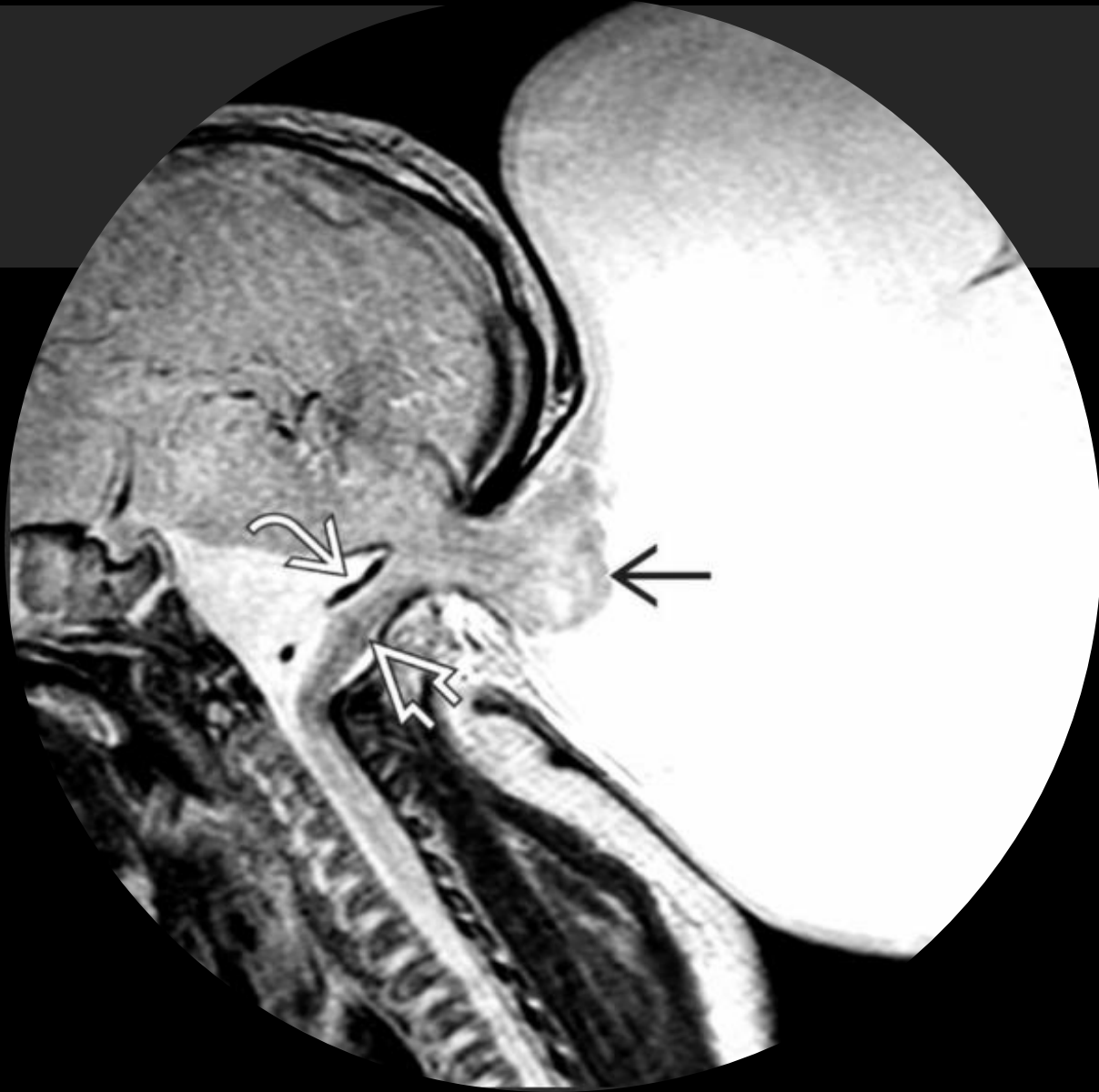


Chiari II





Falx insufficiency with interdigitation of gyri



Chiari III

- Type II+occipital-high cervical encephalocele
- No lumbosacral myelomeningocele
- No hydromyelia

Dandy-Walker “Continuum”

Posterior fossa malformations exist along a continuous range

Varies by degree of vermian hypoplasia and posterior fossa enlargement

Includes:

- 1. Classic Dandy-Walker malformation
- 2. Isolated Vermian Hypoplasia
- 3. Blake pouch cyst
- 4. Mega Cisterna magna

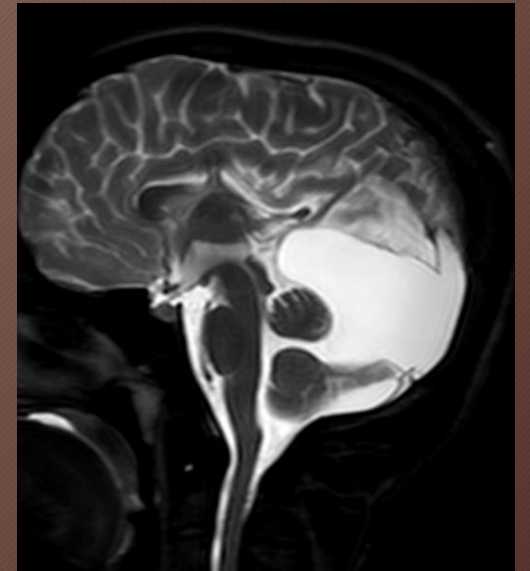
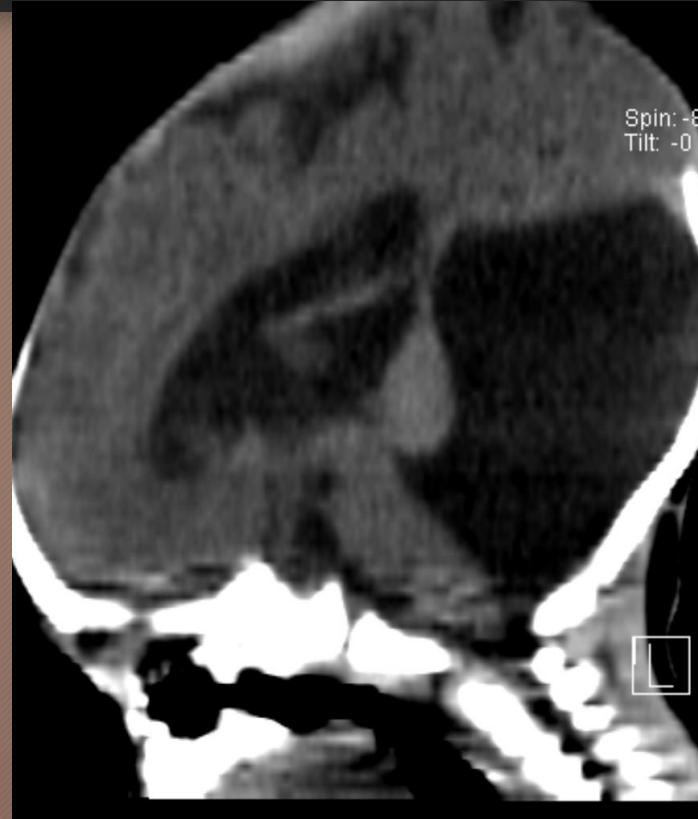
“Classic” Dandy-Walker

Hallmark Findings

- Cystic 4th ventricle dilation
- Vermian Hypoplasia
- Enlarged posterior fossa
- Elevated torcula/High Tentorium
- Upward-rotated vermillion remnant

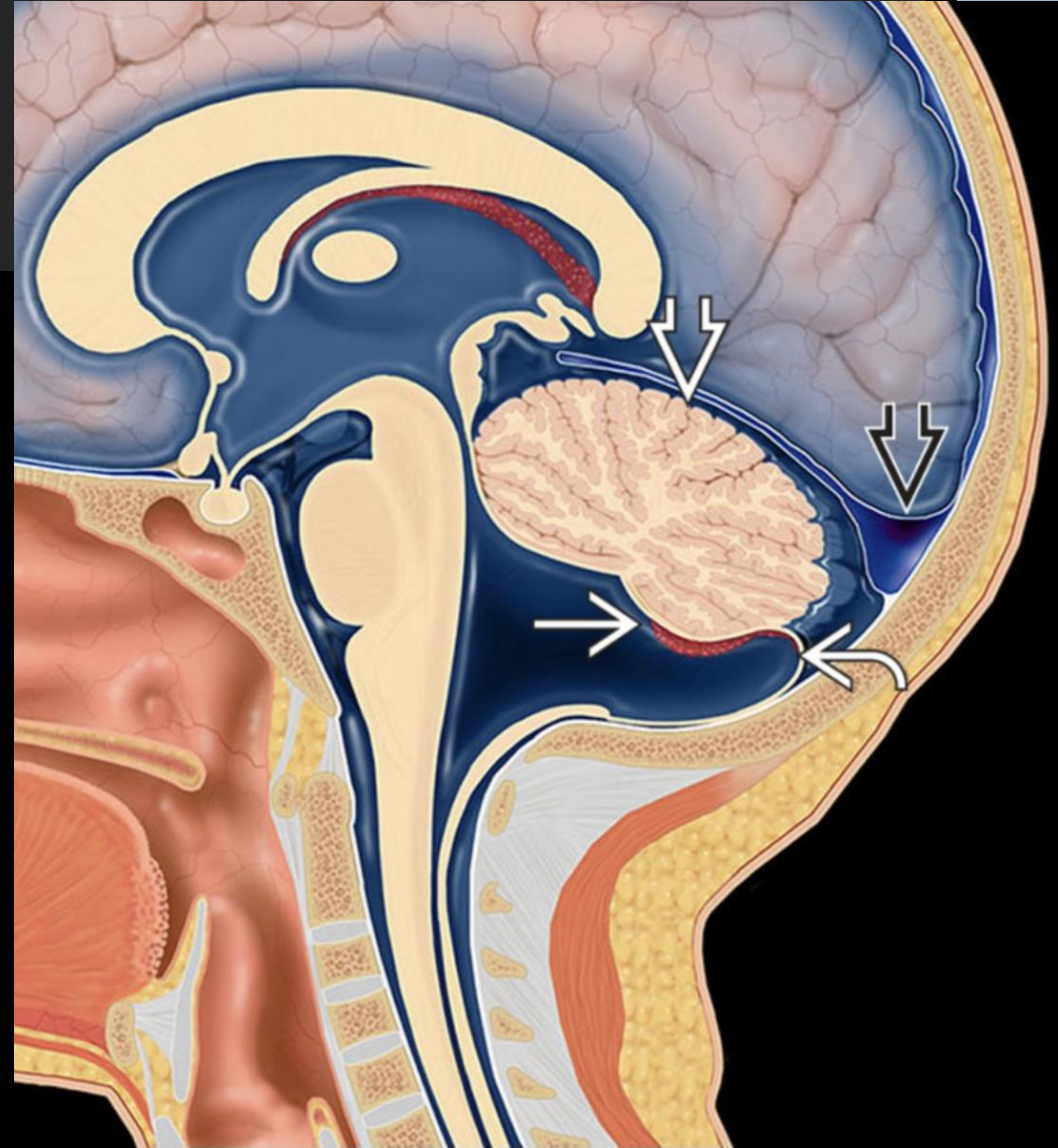
Associated Malformations (60% Cases)

- Hydrocephalus
- Corpus callosum dysgenesis
- Polymicrogyria
- Cephaloceles
- Cognitive impairment depends on associated malformations (35-50% normal intelligence)

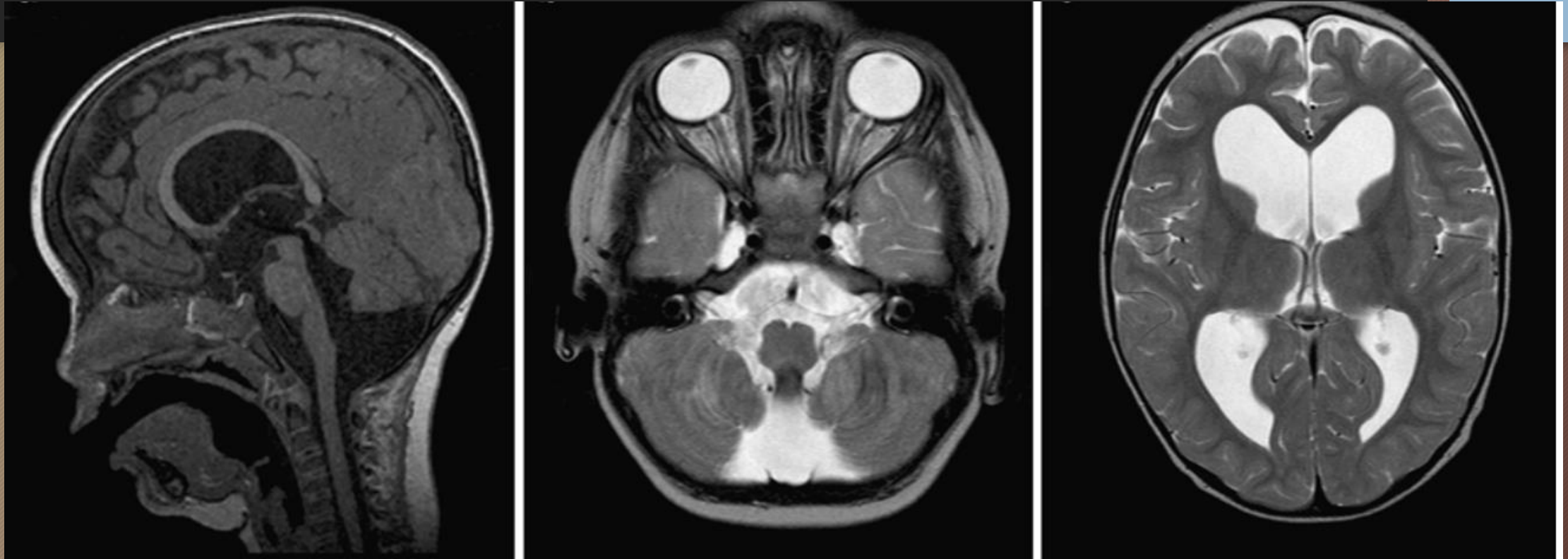


Blake's Pouch cyst

- Failure of regression of Blake's pouch due to non-perforation of foramen of Megendi
- Infra vermian cyst: communicates with 4th ventricle, not cisterna magna
- Posterior fossa may be enlarged

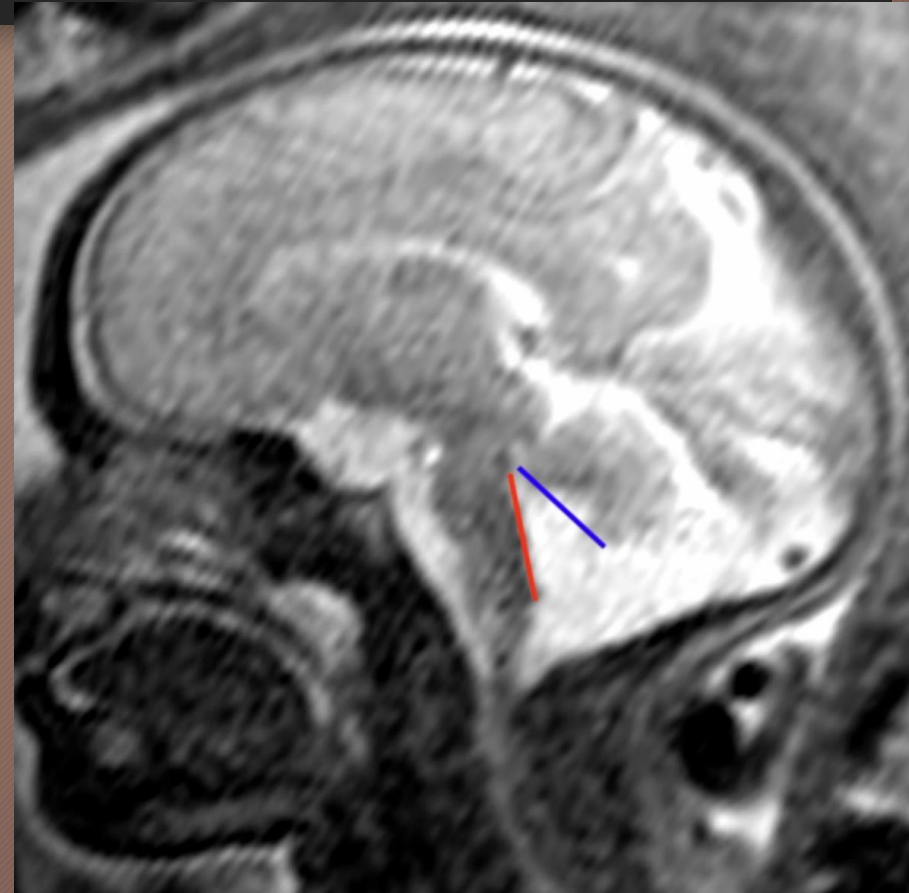
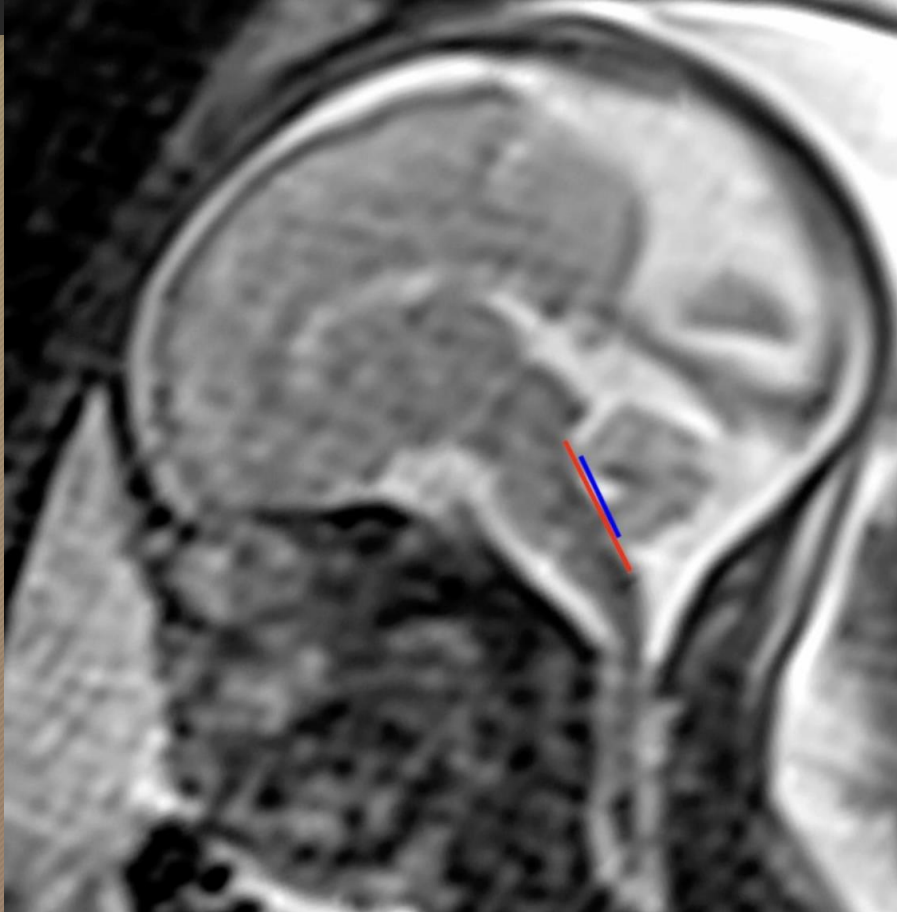


Blake Pouch Cyst (continued)



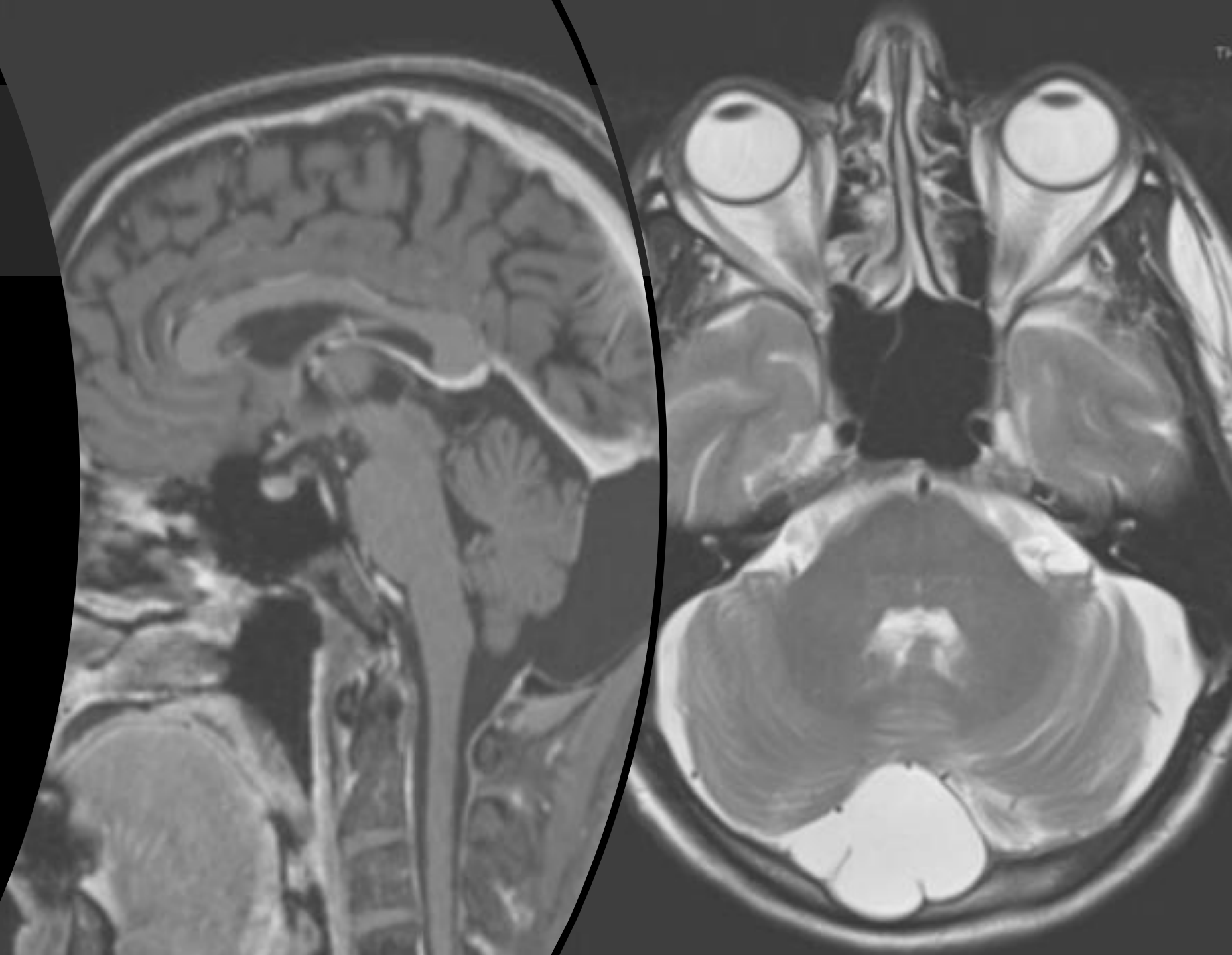
Childs Nerv Syst (2010) 26:1057-1064

Blake's Pouch Cyst (continued, tegmentovermian angle)

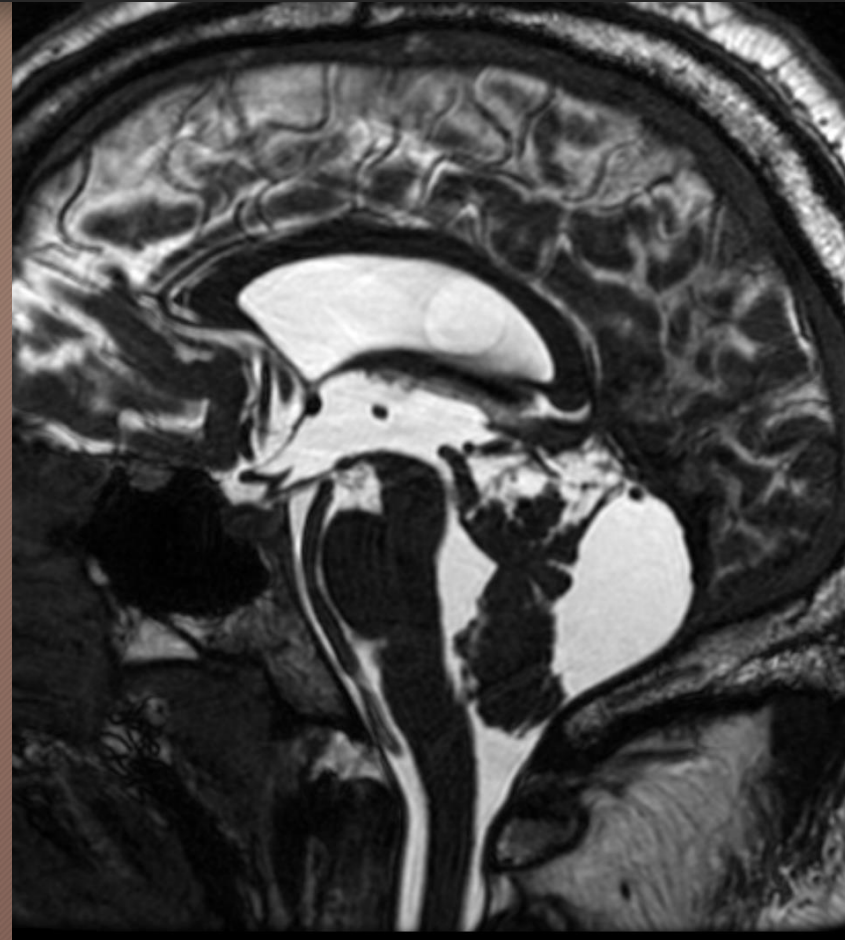
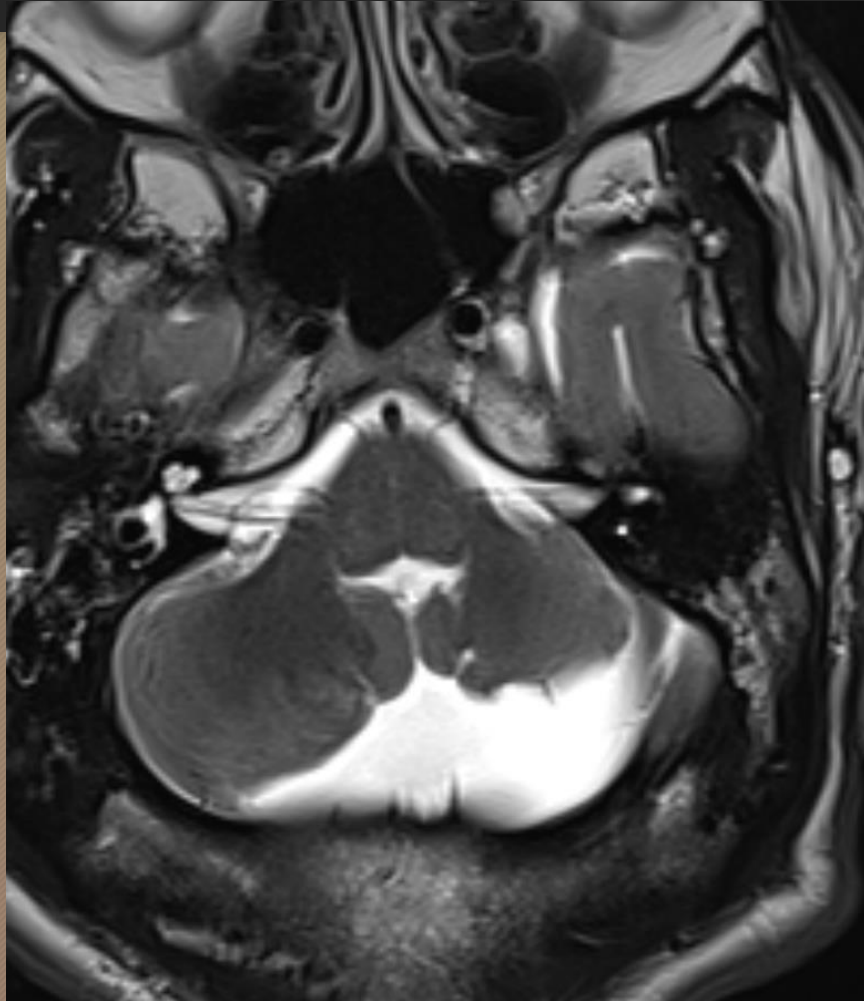


Mega Cisterna Magna

- Large retrocerebellar space
- Normal vermis and cerebellum
- 4th ventricle choroid plexus in normal location
- Posterior fossa usually normal to large
- Torcula normal
- Rarely associated malformations



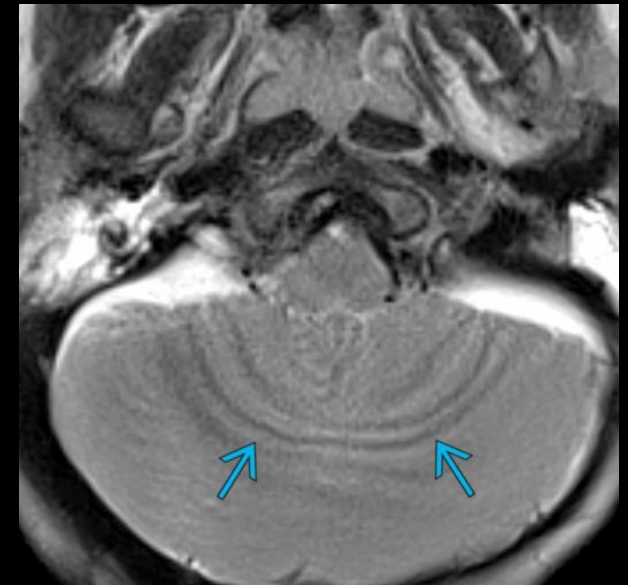
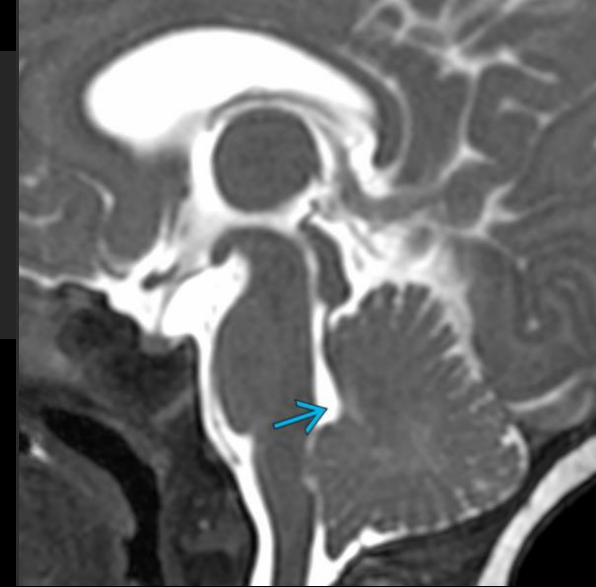
Arachnoid cyst (ddx)



Rhombencephalosynapsis

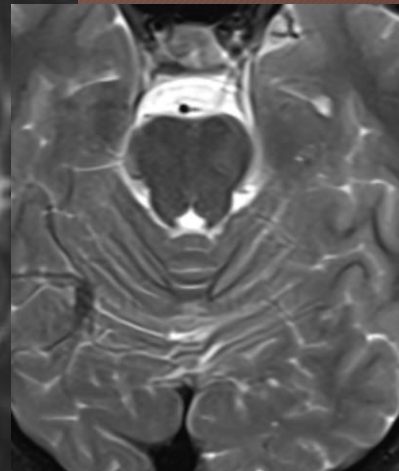
- Single lobed cerebellum
- Fusion of both cerebellar hemispheres, dentate nuclei and superior cerebellar peduncles
- Vermian agenesis
- Often associated malformations
- Poor prognosis

Look closely for RES in congenital aqueductal stenosis

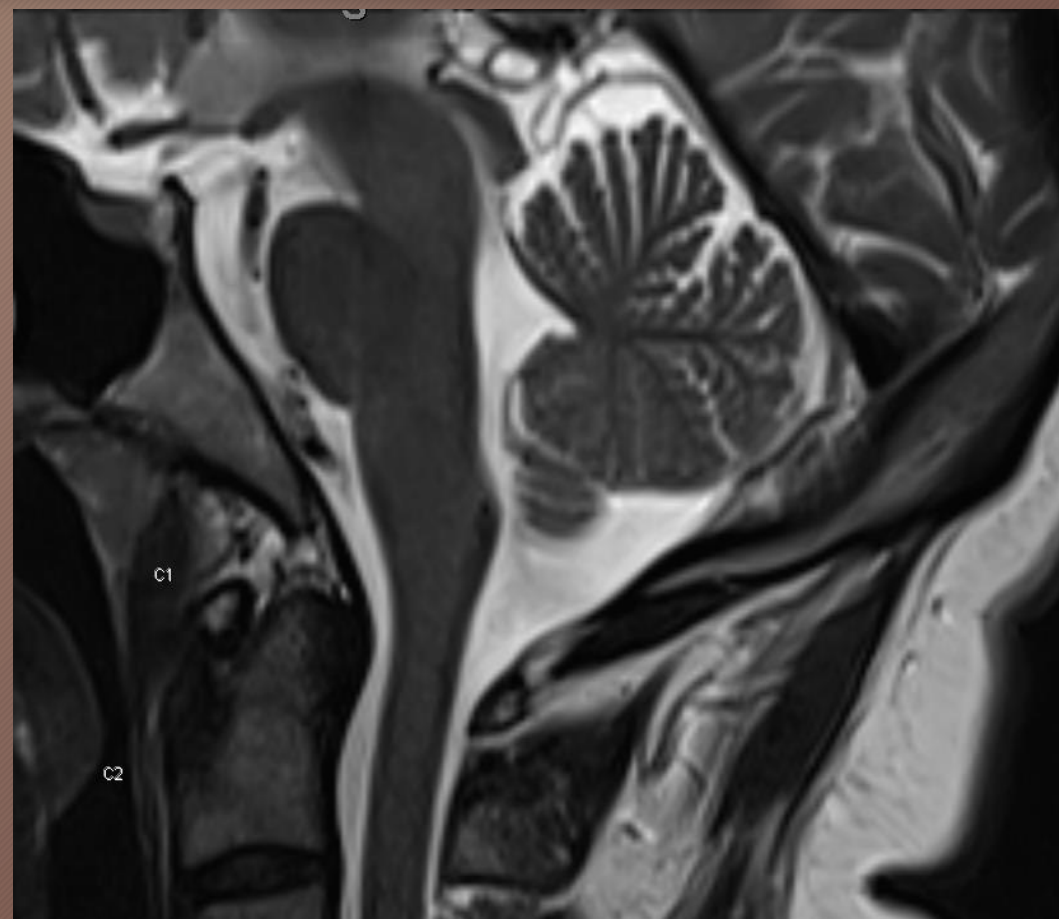


Joubert syndrome

- Vermian aplasia
- Episodic hyperpnea, abnormal eye movements, rhythmic tongue protrusion, ataxia, mental retardation
- Autosomal recessive
- **Molar tooth brainstem**, bat-wing or umbrella shaped 4th ventricle

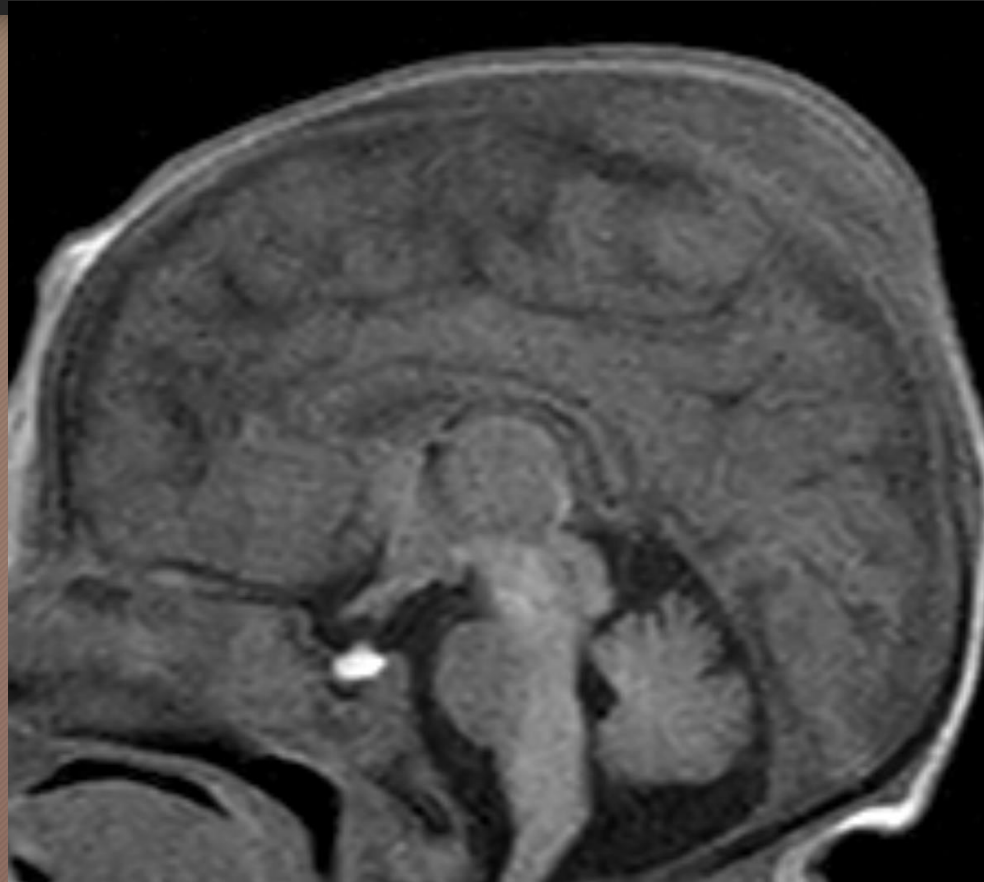
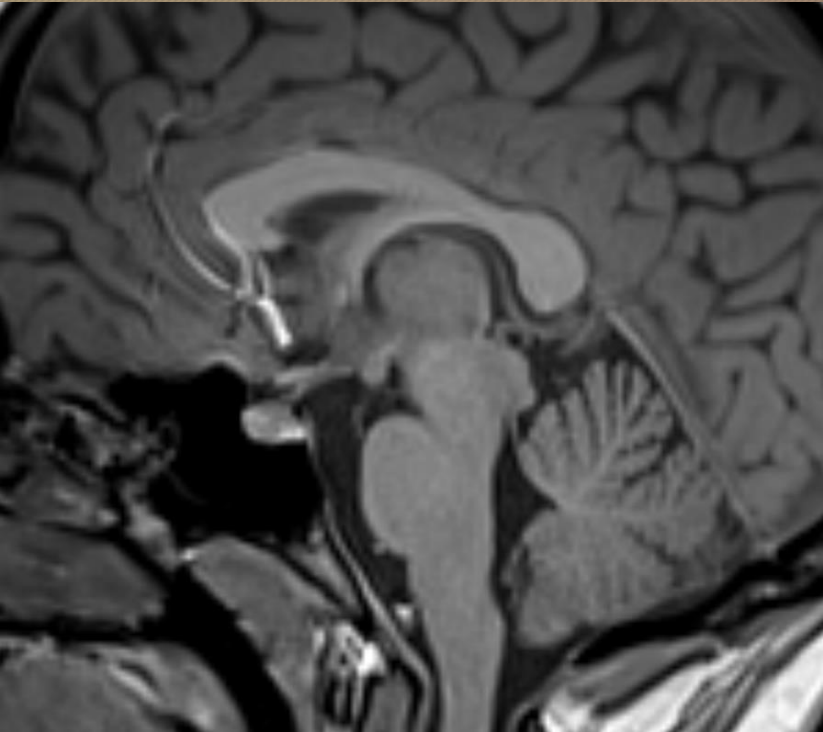


AJNR Am J Neuroradiol
32:1459 - 63



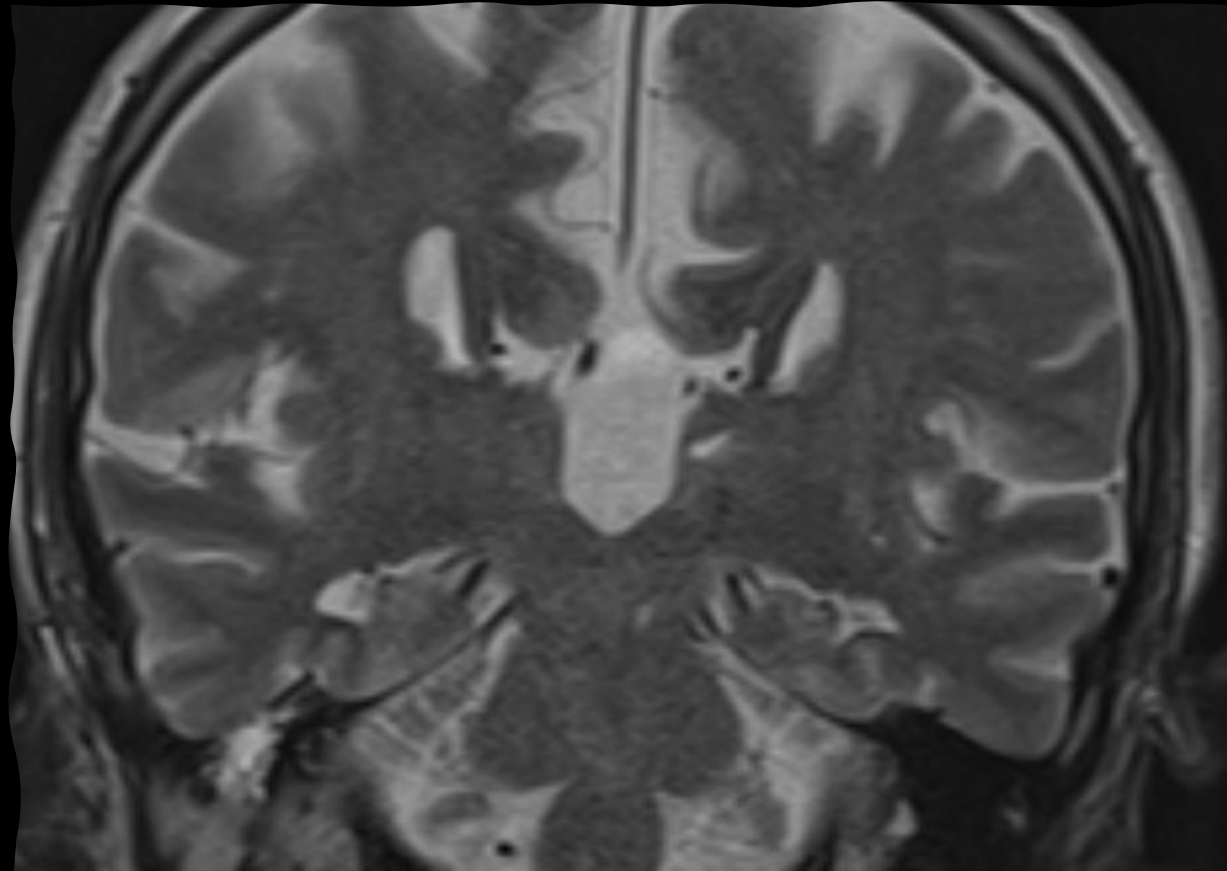
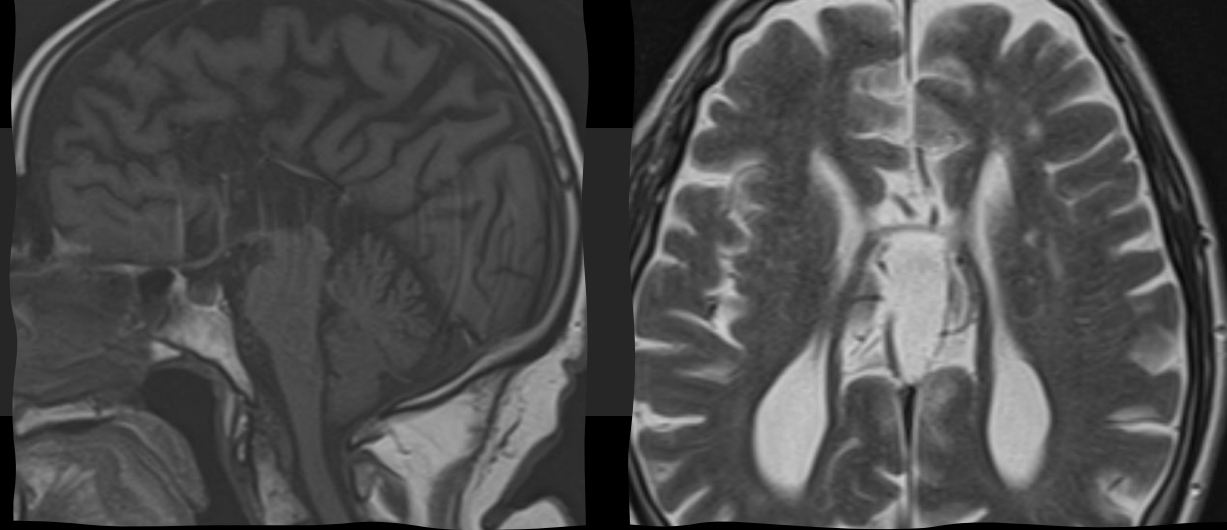
Corpus callosum

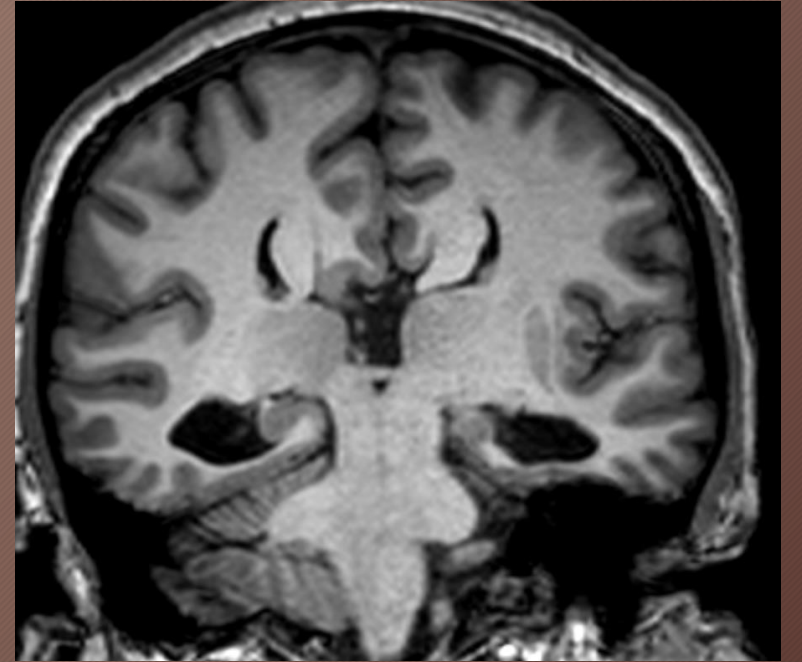
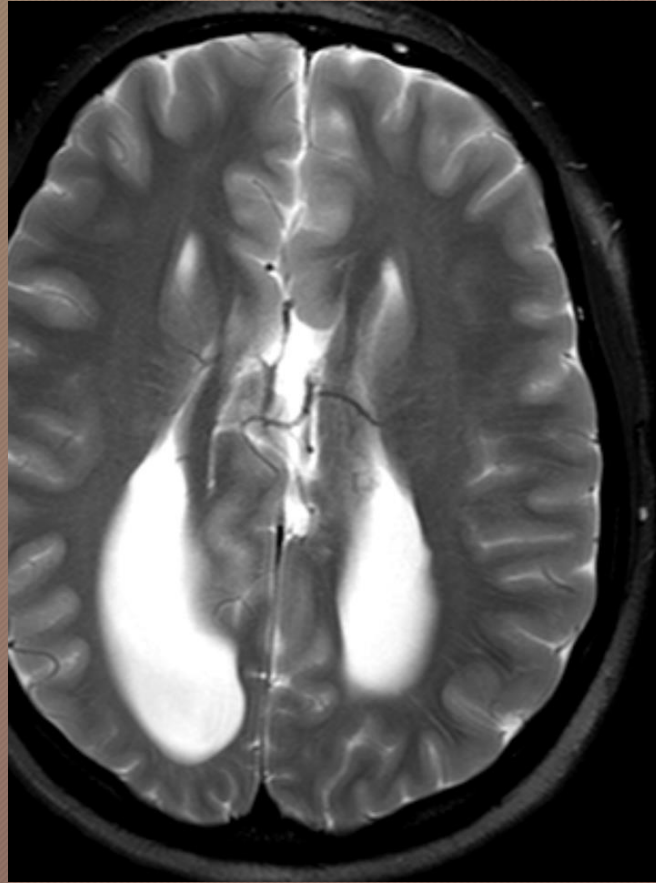
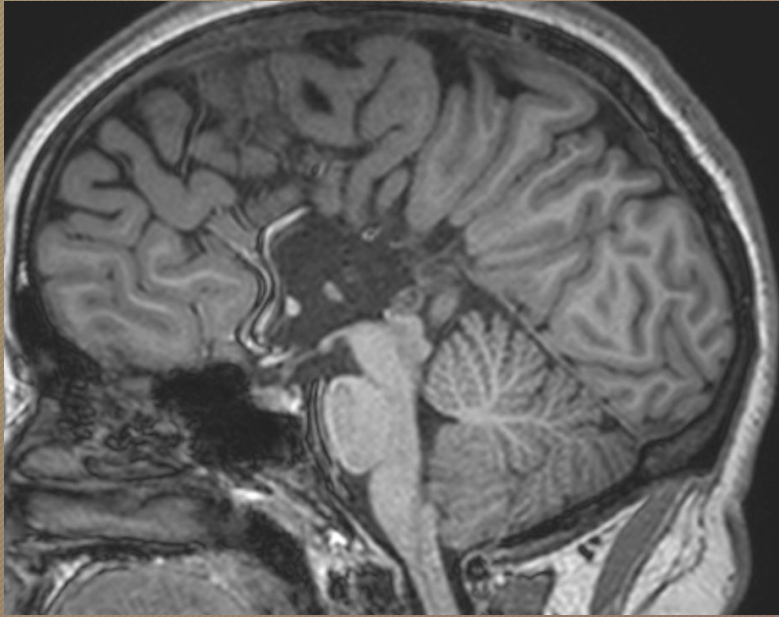
Normal variation in shape and size



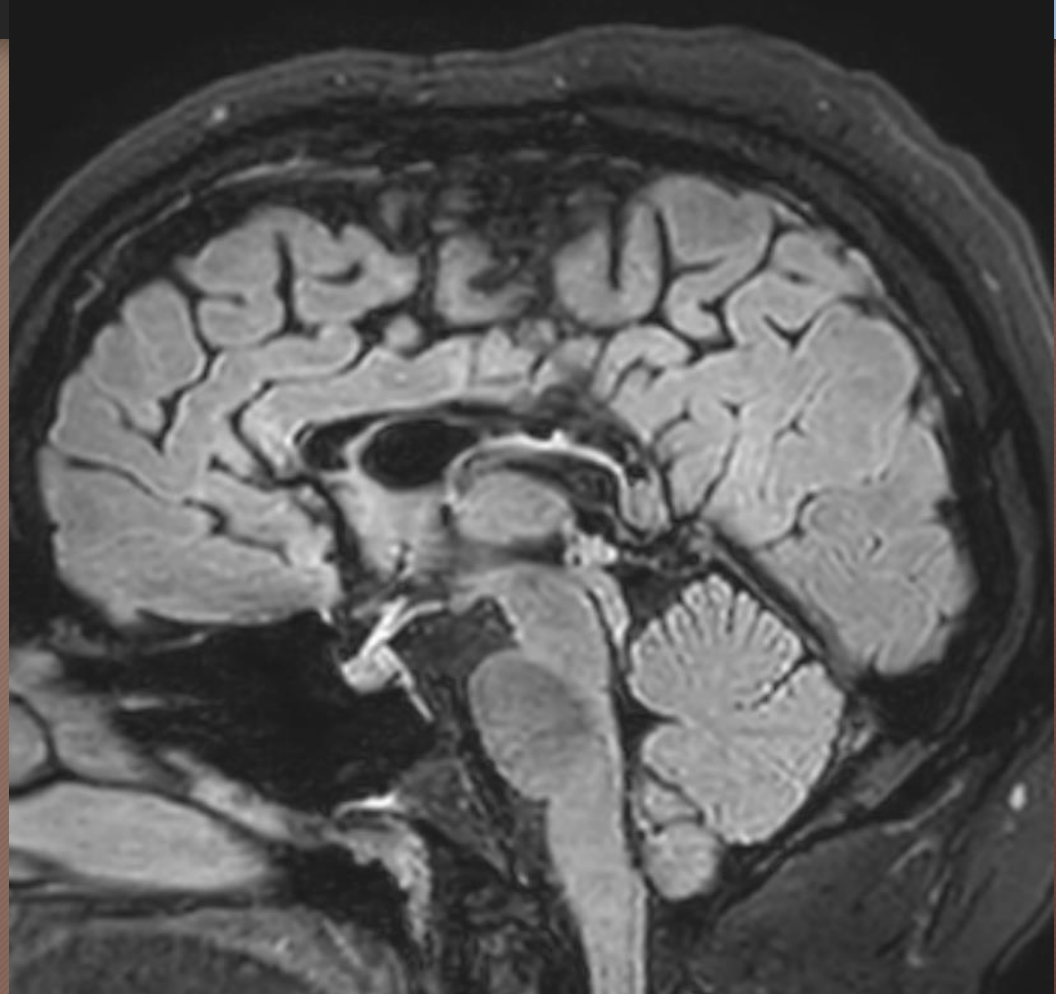
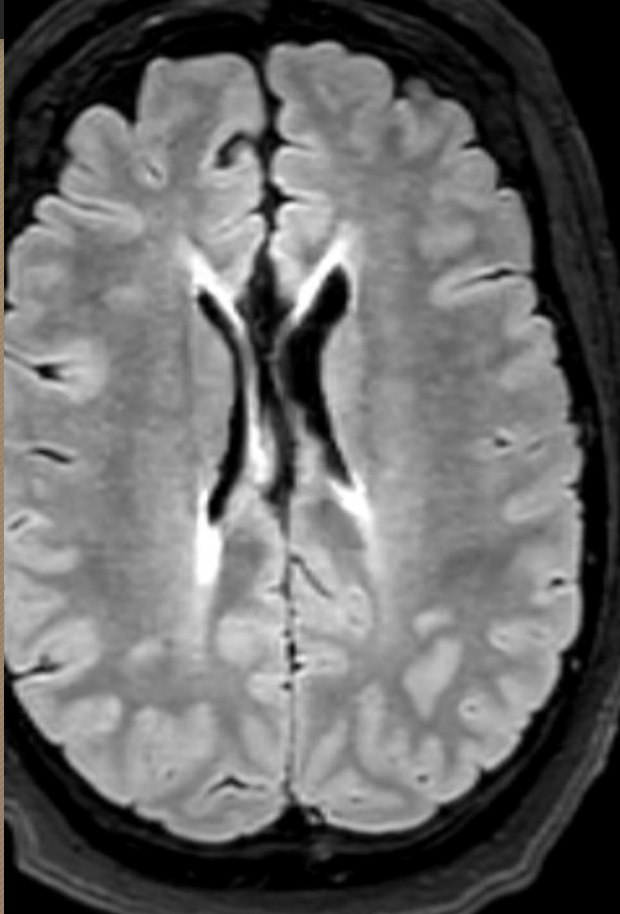
Callosal dysgenesis

- Trident shape of lateral ventricles (prost bundle)
- No inversion of cingulate gyrus
- High riding 3rd ventricle
- Lipoma
- In 50% associated with other CNS malformations (DW malformation, Chiari II, septo-optic dysplasia...)
- Seizures, developmental delay, microcephaly, hypothalamic-pituitary malfunction, hypertelorism



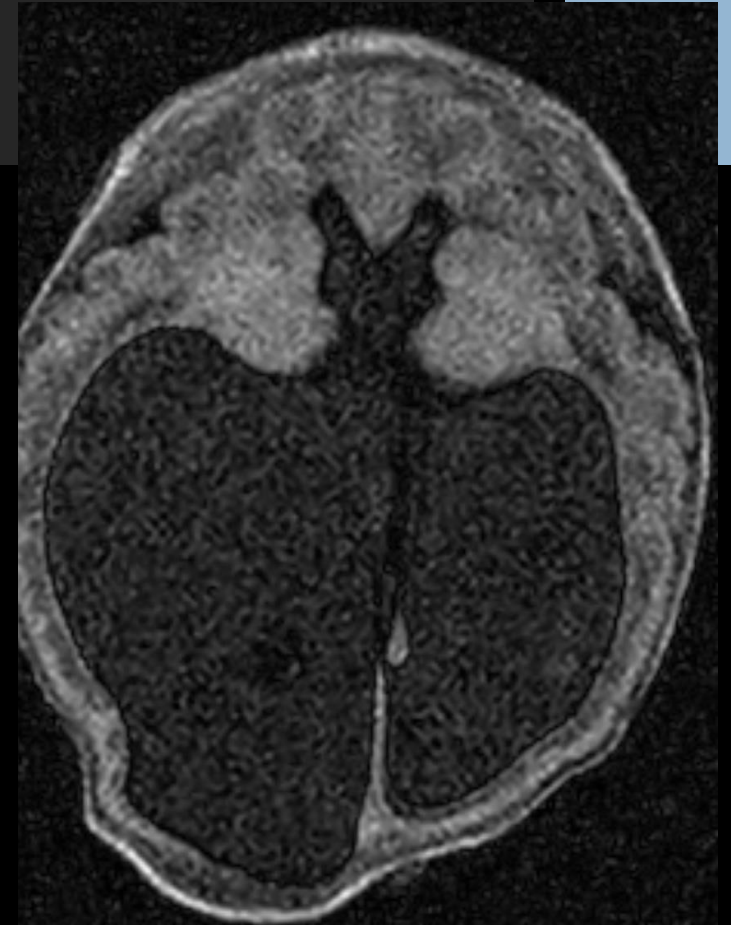
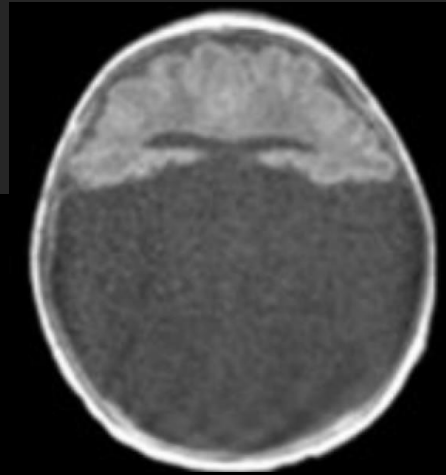


Corpus callosotomy

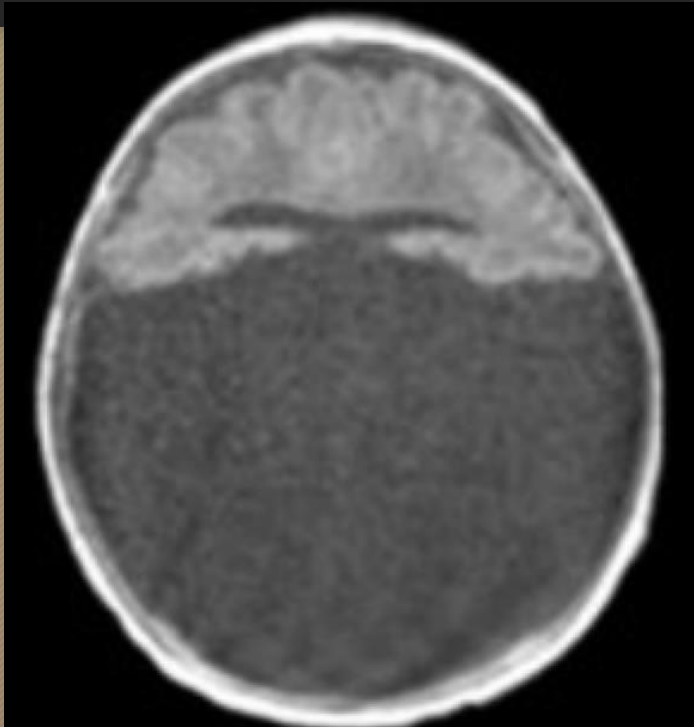


Holoprosencephaly

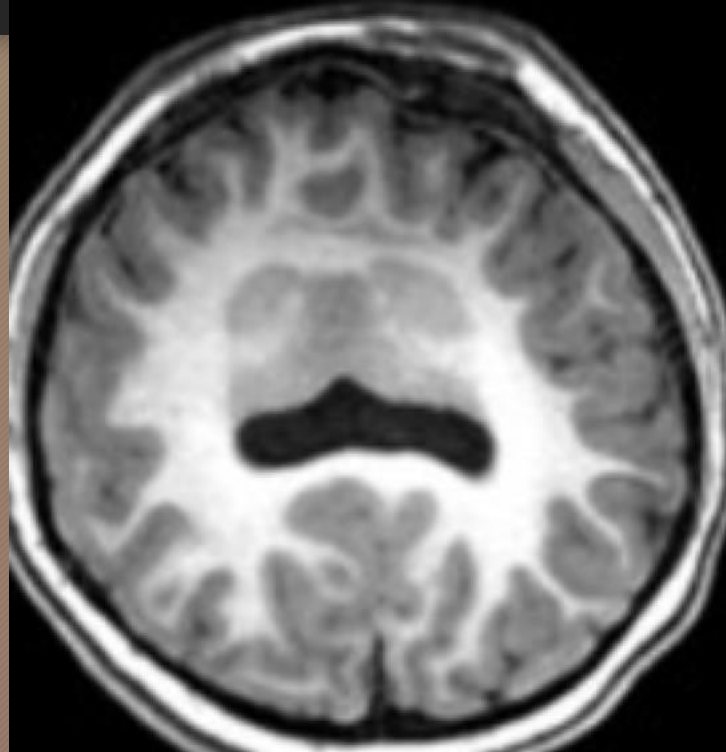
- Failure of midline cleavage
- Incomplete separation right and left cerebral hemispheres
- Alobar, semilobar, lobar
- Hypothalamic-pituitary dysfunction, seizures, mental retardation, dystonia
- Cyclopia, microcephaly



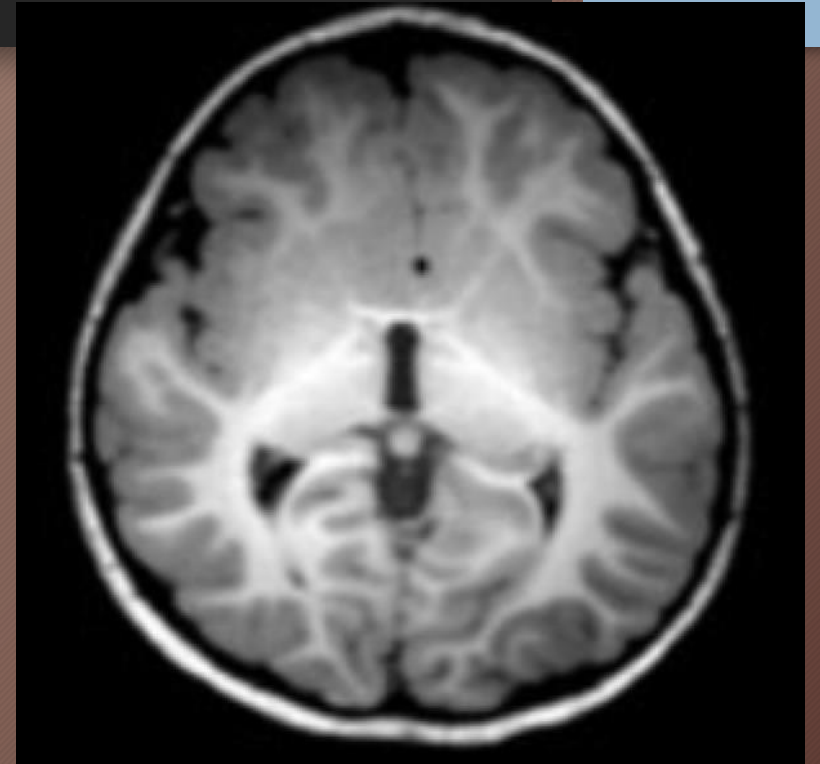
Holoprosencephaly (continued)



Alobar



Semilobar



Lobar

Cortical Development: Key Stages and Associated Malformations

Neuronal & Glial Proliferation (6-16 weeks)

Neoplasms

Balloon-cell overgrowth



Neuronal Migration (8-24 weeks)

Lissencephaly

Gray matter heterotopia



Cortical Organization (20 weeks-brith)

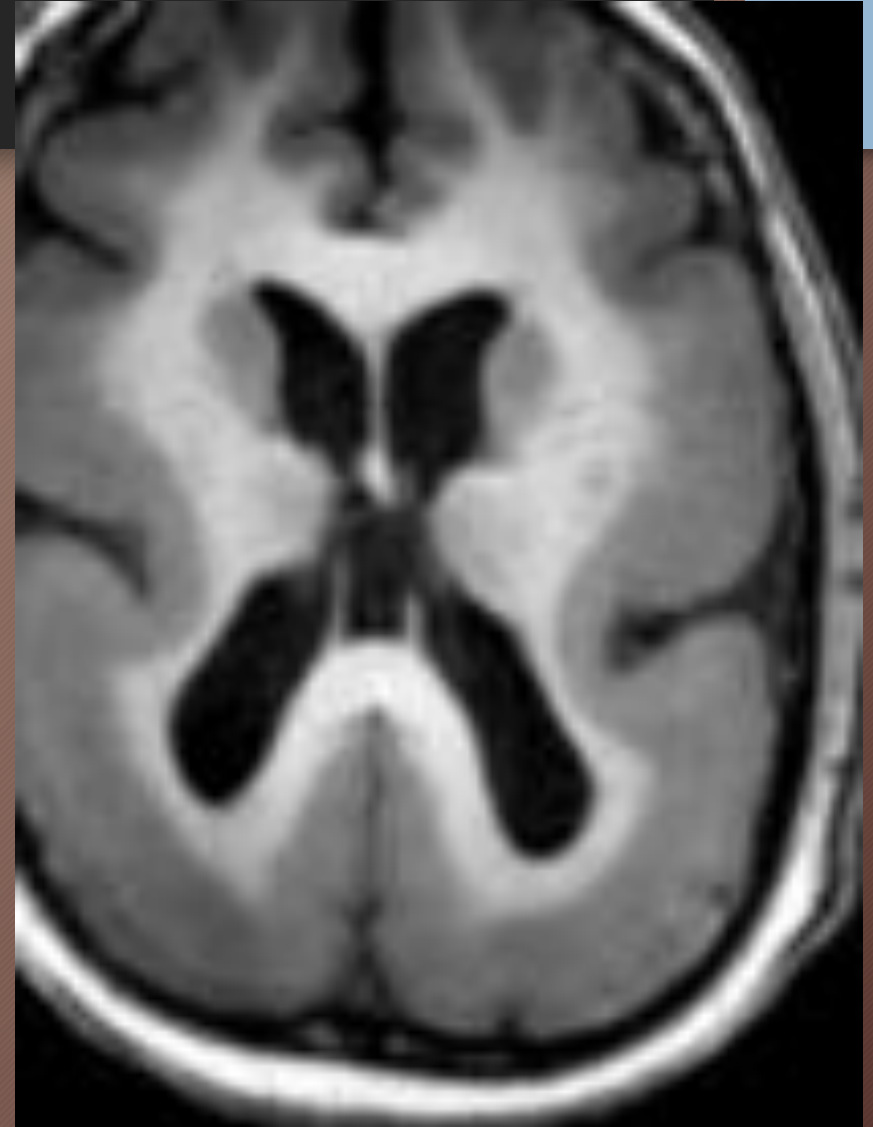
Polymicrogyria

Schizencephaly

Focal cortical dysplasia (FCD)

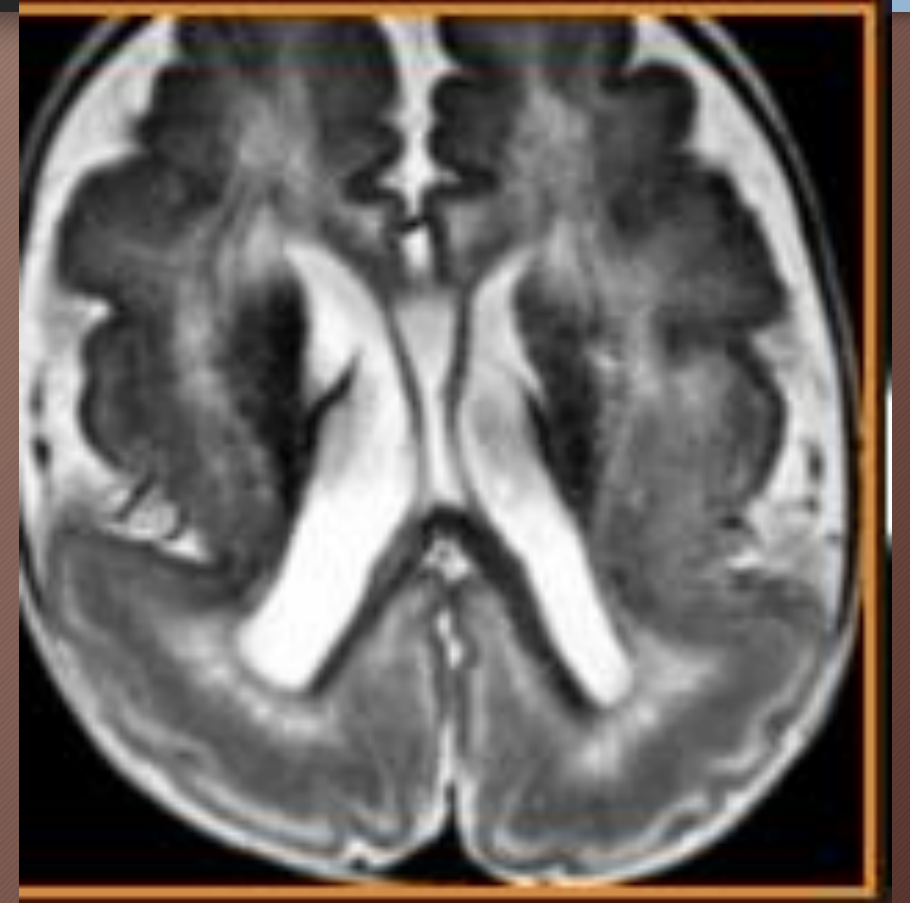
Lissencephaly

- Arrested neuronal migration
 - Agyria
- Lissencephaly type 1
 - Classic pachy/agyria complex
 - X-linked
 - CMV or toxic insult
- Type 2: cobble stone



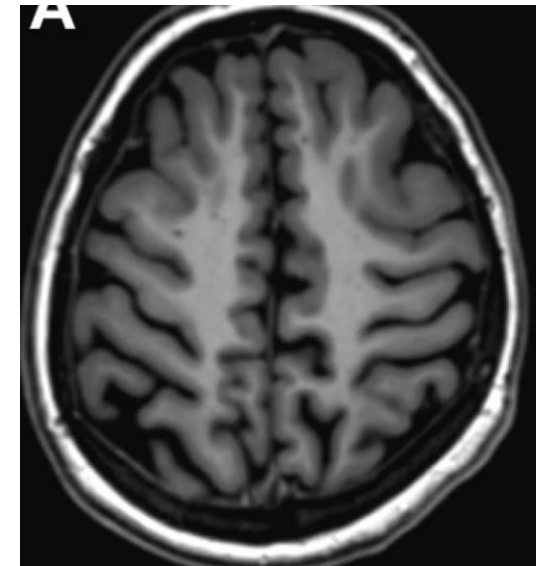
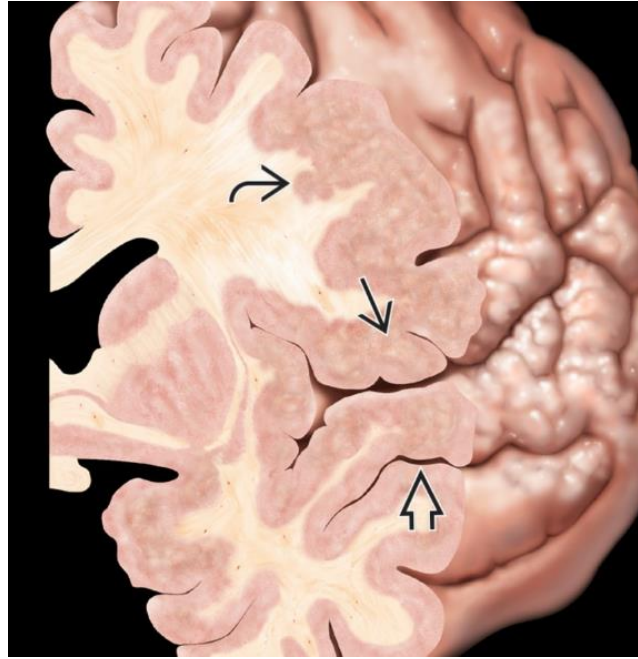
Lissencephaly MRI

- Type 1(classic)
 - Hour-glass shape of brain
 - 3 visible layers
 - Outer cortex
 - Cell sparse zone
 - Deeper cortical layer
 - Appearance of band heterotopia
- In preterms be careful!
 - Preterm brain is normal agyric until 26 weeks gestation



Pachygyria

- Cortical malformation
- 2nd half of 2nd trimester insult
- Secondary to congenital, TORCH, ischemic or toxic etiologies
- MRI
 - Thickened cortex, flattened gyral pattern
 - Normal cortex-white matter junction



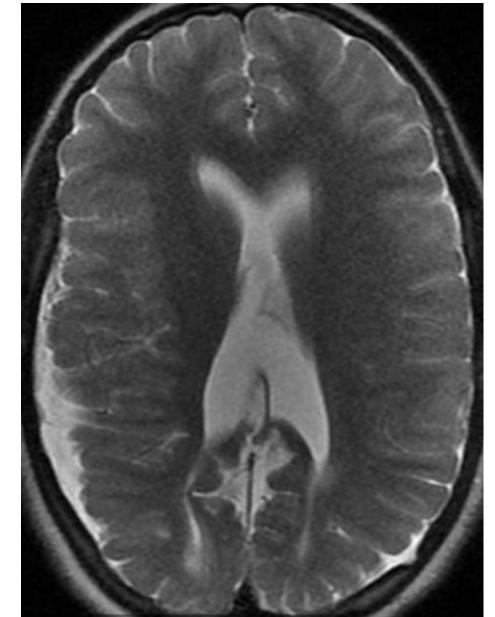
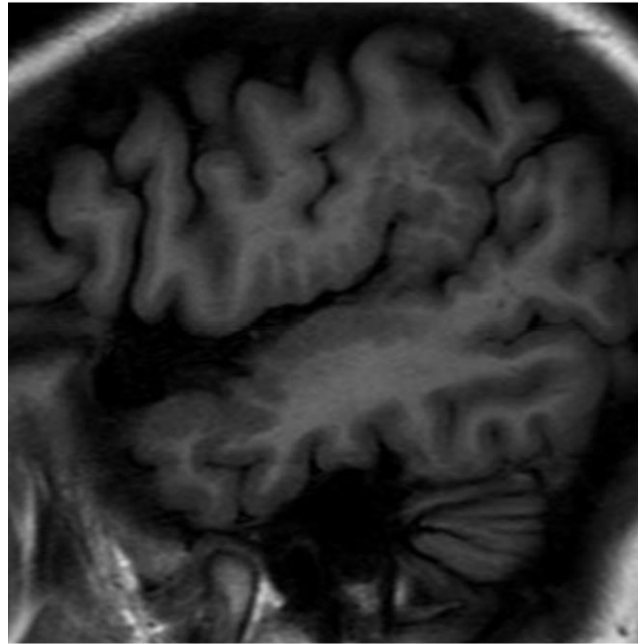
Polymicrogyria

Clinical/Developmental Features

- Cortical malformation: abnormal migration + organization
- Perisylvian predominant (~80%)
- Bilateral → often syndromic
- Seizures (40-60%)
- Developmental delay +/- motor deficits

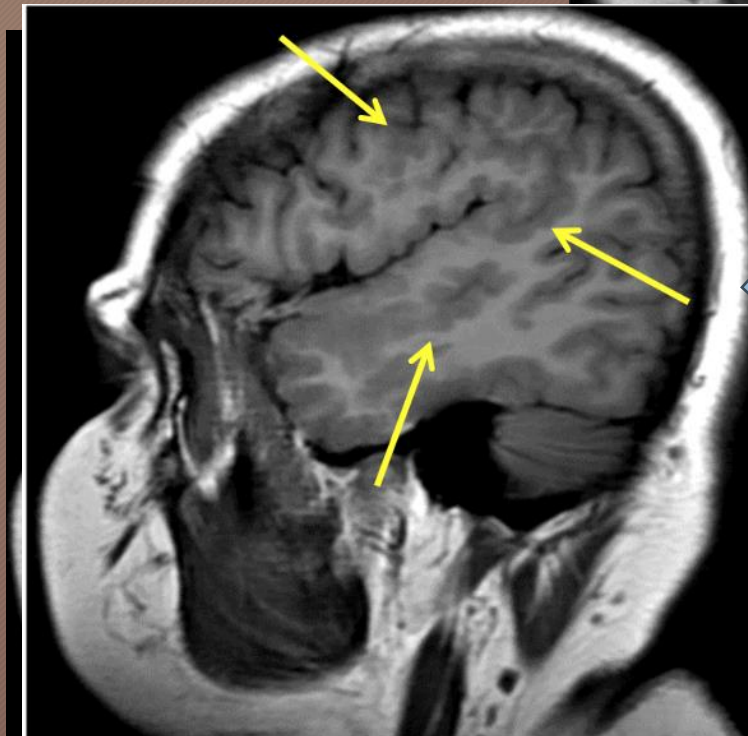
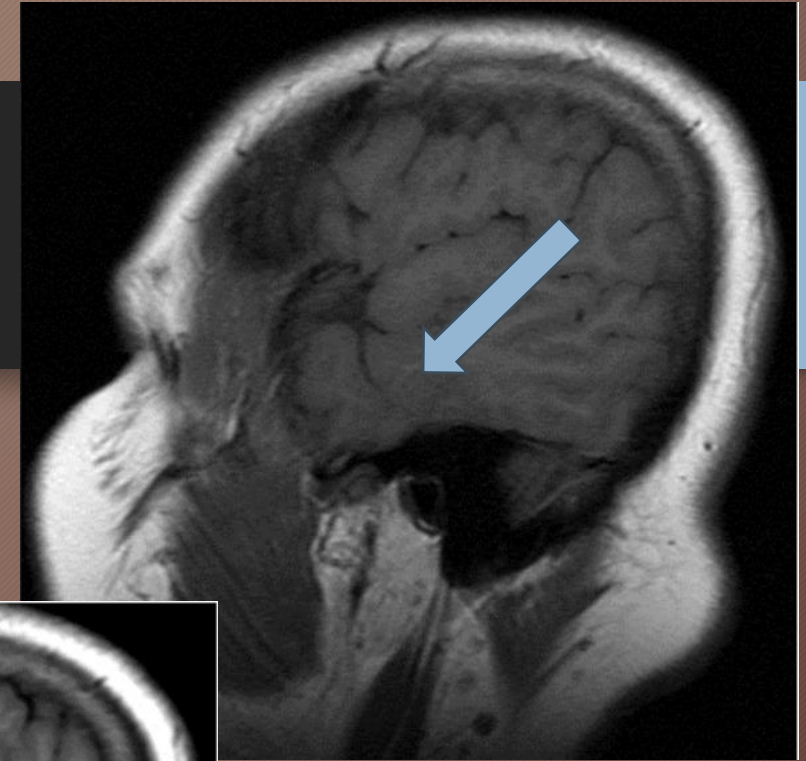
MRI Characteristics

- Excess gyral infolding with numerous tiny sulci
- Cortex thickened or normal thickness
- Irregular GM-WM junction
- Prominent overlying vein (“cortical vein sign”)
- May mimic pachygyria (overlap in the malformation spectrum)



Polymicrogyria

- Abnormal late migrational anomaly
- Irregular, nodular cortex and shallow sulci
- MC involves perisylvian region
- Polymicrogyria lines schizencephaly clefts.
- Check for underlying CMV or metabolic or syndromic diagnosis.

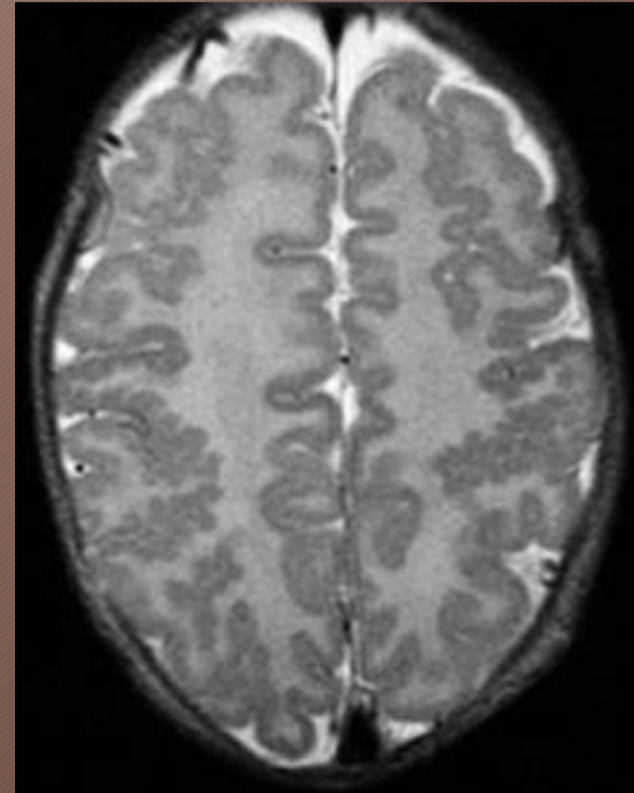


Pachygyria vs. Polymicrogyria

Thickened cortex



Multiple small gyri



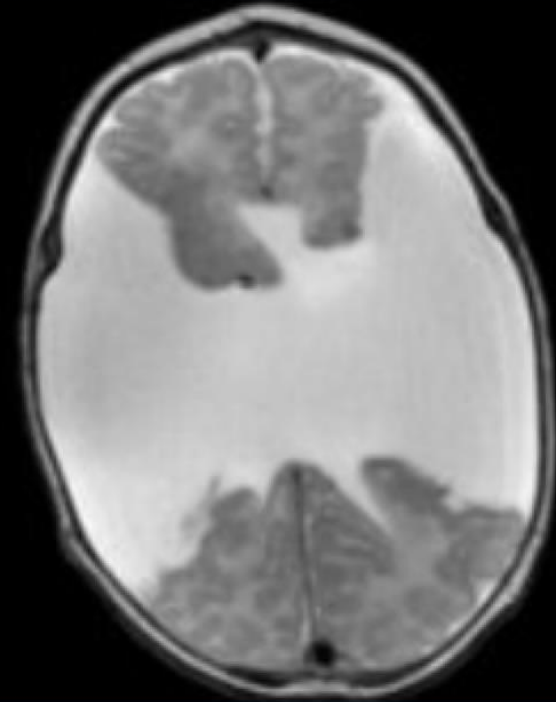
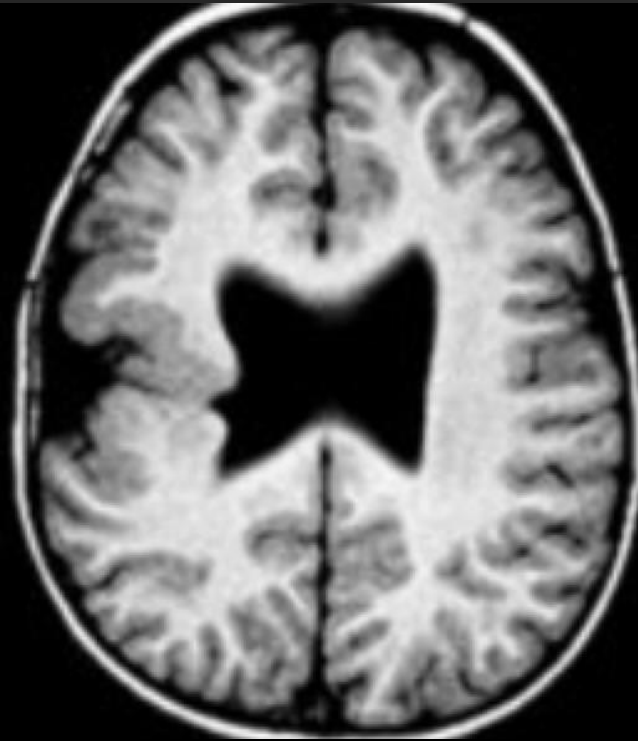
Schizencephaly

- Seizures
- Watershed ischemia in utero, infection, maternal trauma, toxins and genetic etiologies
- Transmantle CSF cleft extending from ventricle to cortical margin.



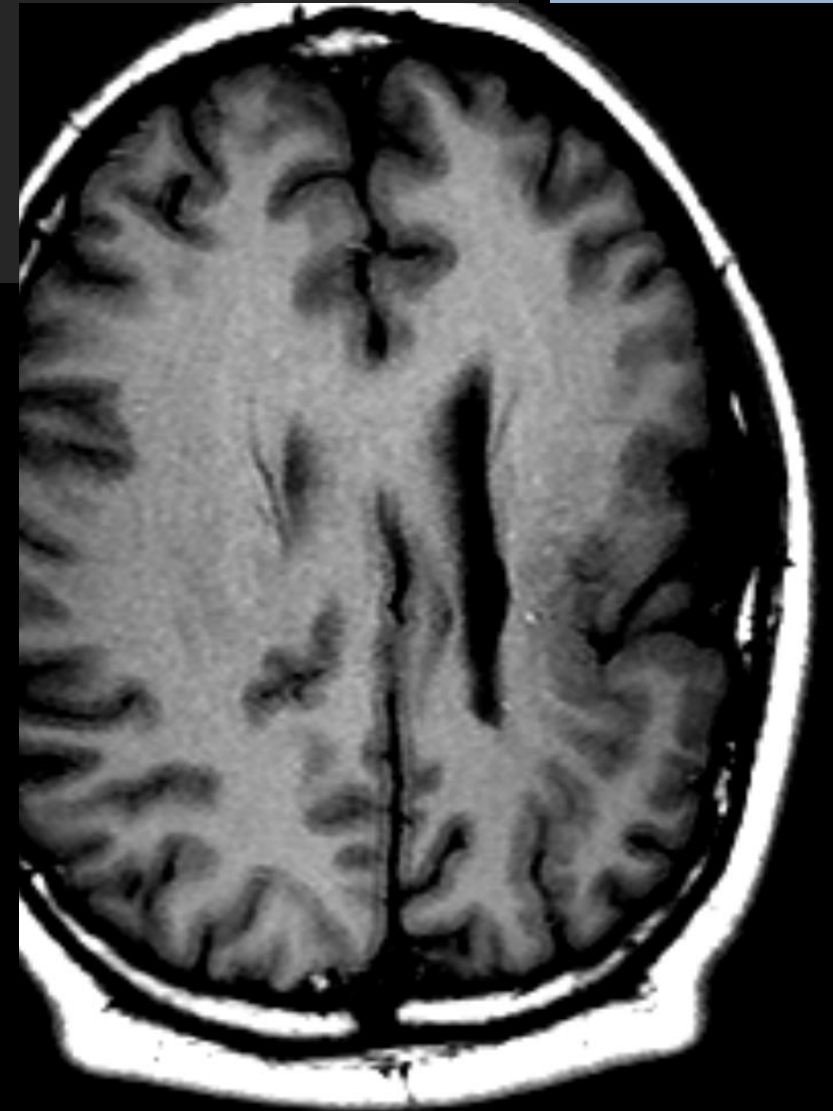
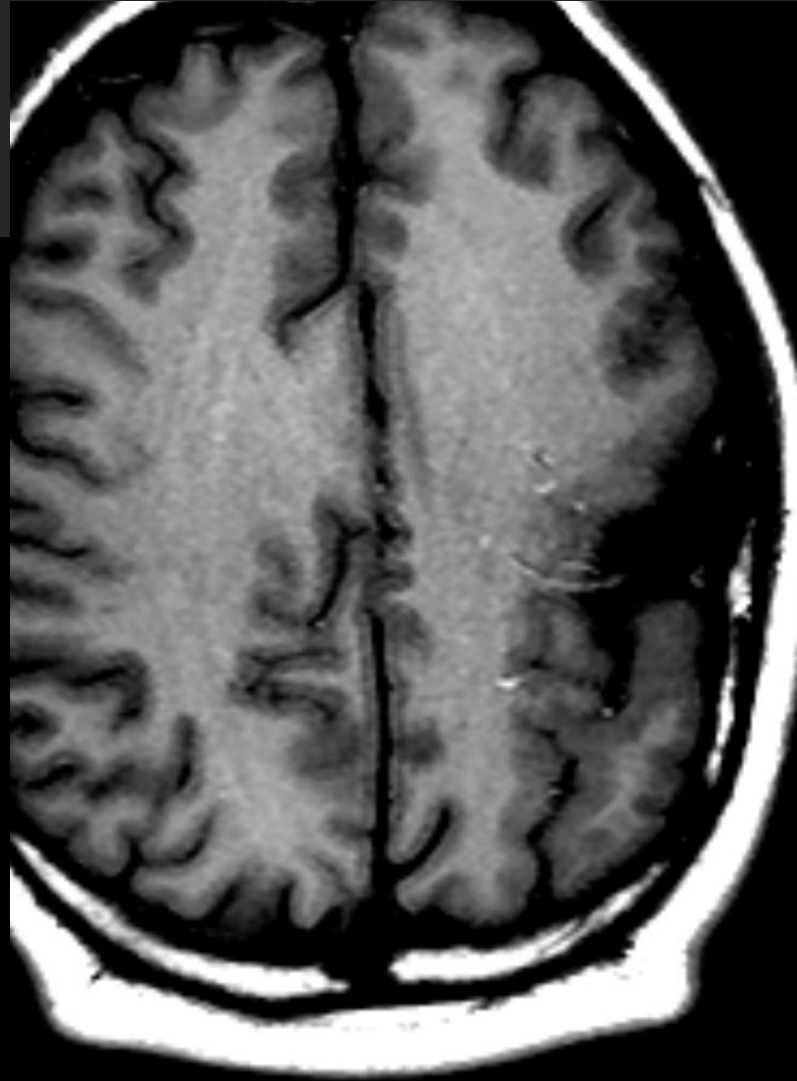
Schizencephaly

- Cortical migrational abnormality
 - Cleft extending from cortex to ventricular surface
 - Lined by PMG
- Closed lip
 - Close approximation of cleft
 - Nipple on ventricular surface
- Open lip
 - Wedge gap
 - 70% association with SOD



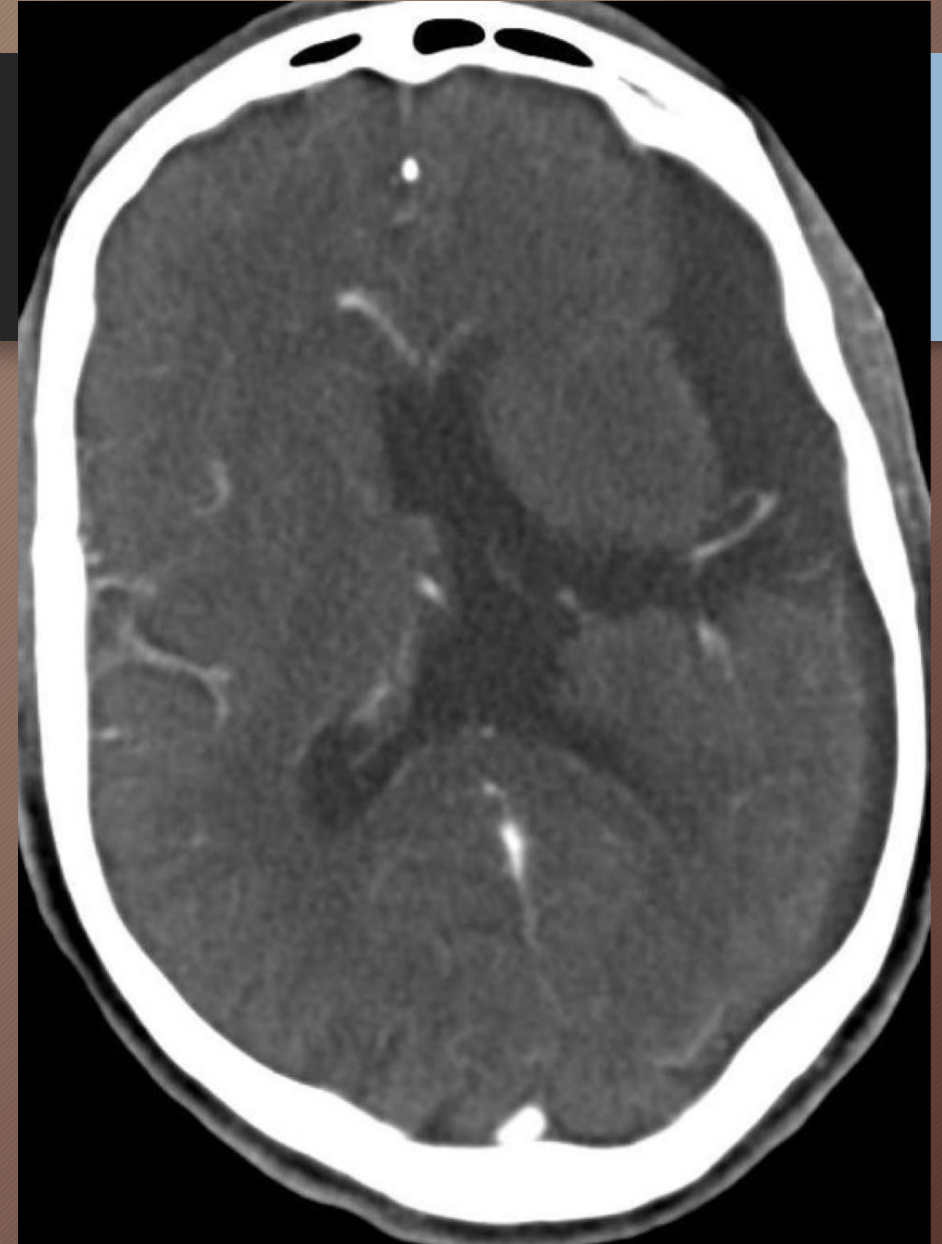
Closed lip Schizencephaly

- Cleft lined by
 - Pia along lateral margin
 - Ependyma medially
- Cortical veins can traverse the cleft



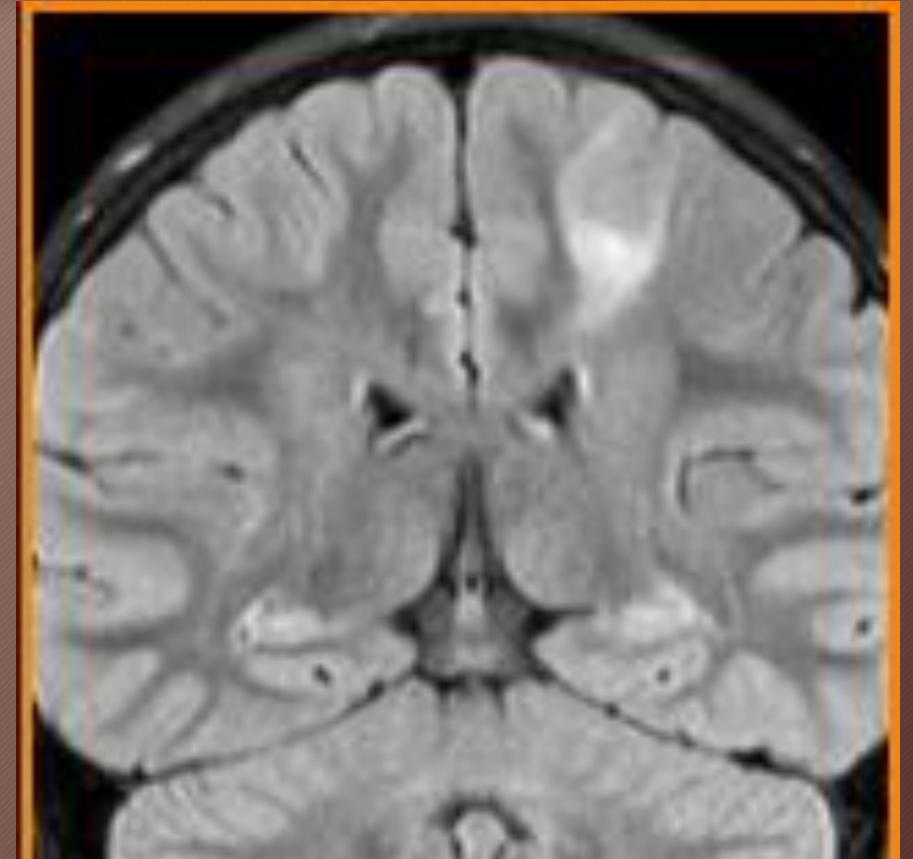
Open lip Schizencephaly

- Can be bilateral
- Absence of septum pellucidum (spectrum of SOD)-70% cases.
- Volumetric MRI imaging helpful
- Check for midline structures: CC and hypothalamic pituitary axis.



Cortical dysplasia-Balloon type

- MRI findings:
 - Gyral expansion
 - Cortical thickening with blurring of the G-W jxn
 - T2 signal abnormality in the underlying white matter signal can extend in curvilinear manner to ventricular surface



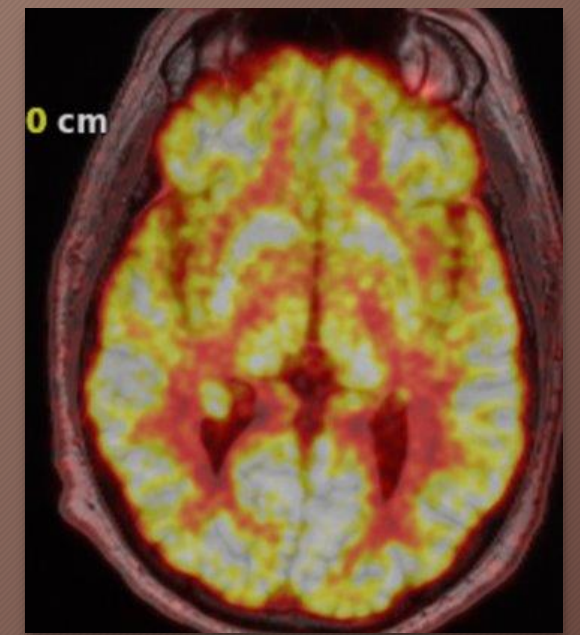
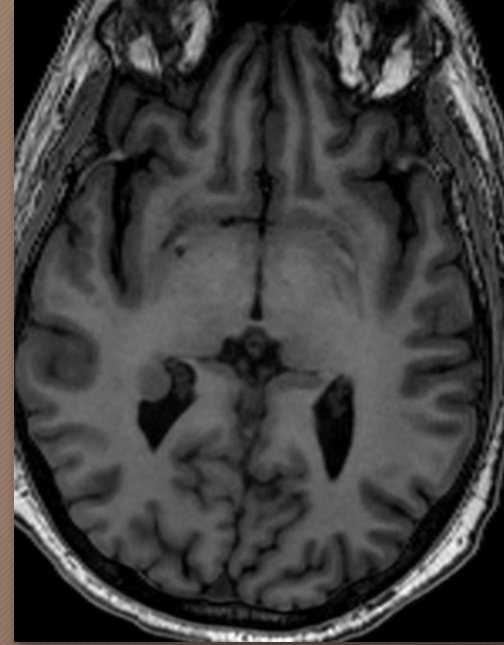
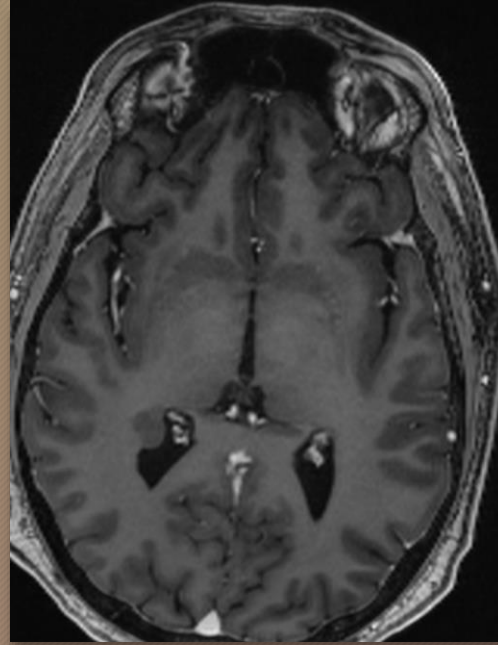
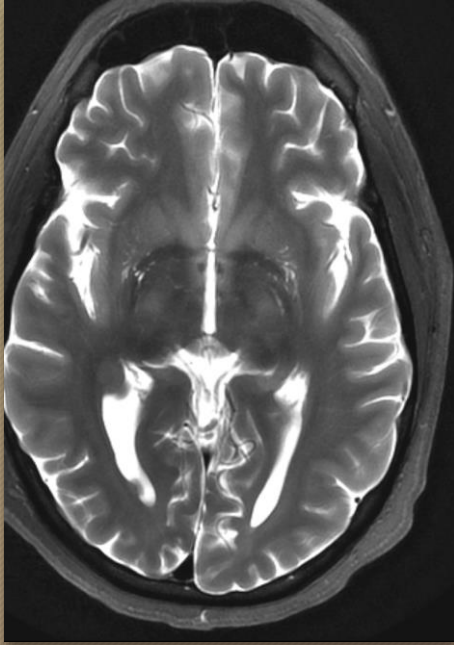
Cortical dysplasia-Balloon type

Almost all have
refractory partial
epilepsy

Lesions pathologically
and radiologically to
cortical tubers of TS
and transmantle
dysplasias.

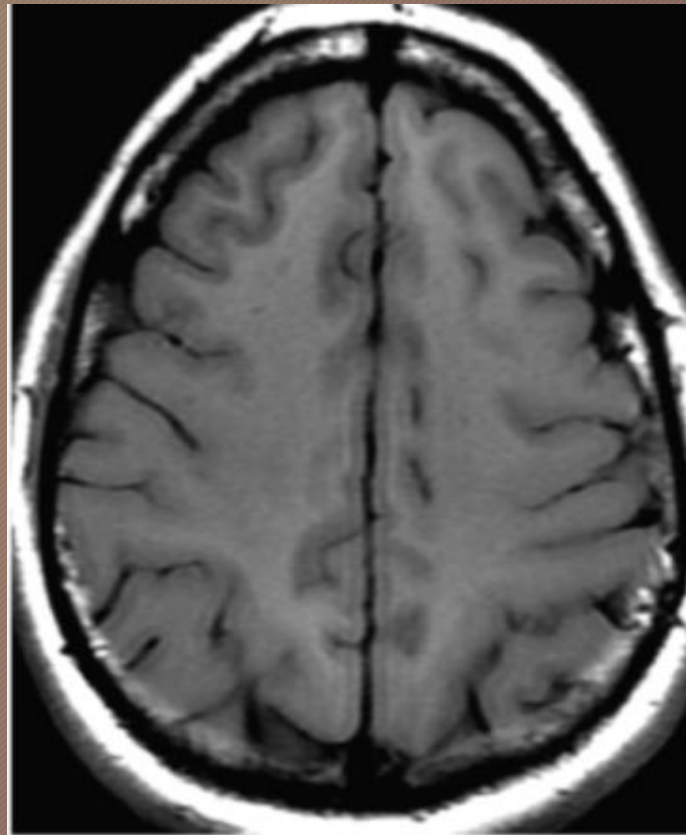
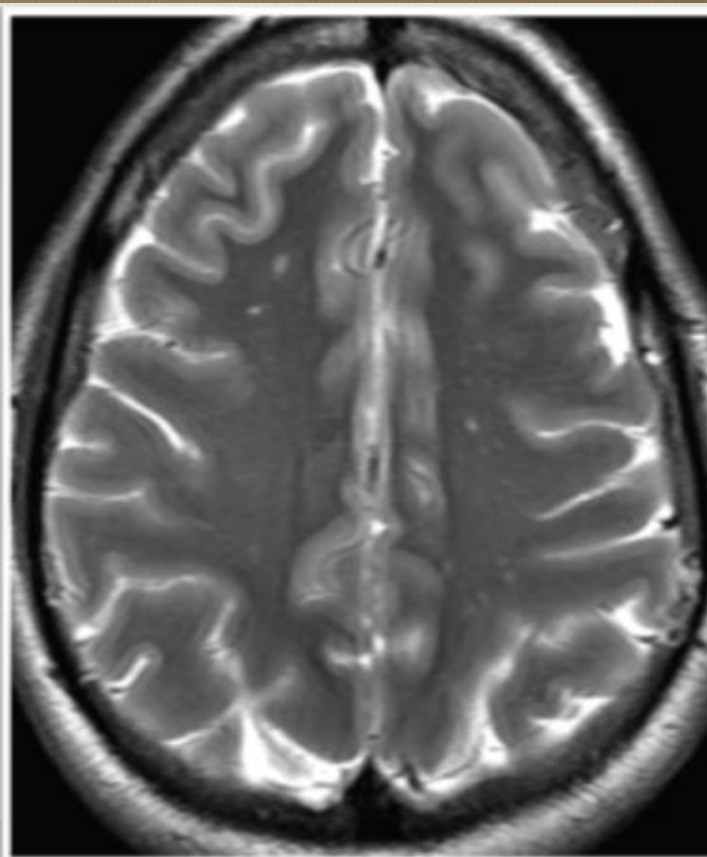
Gray matter Heterotopias

- Related to abnormal developmental cortical migration
 - Normal neurons in abnormal locations
 - Arrest of normal neuronal migration at any point from germinal matrix, along radial glial fibers to cortex
- 3types
 - Subependymal
 - Subcortical
 - Band



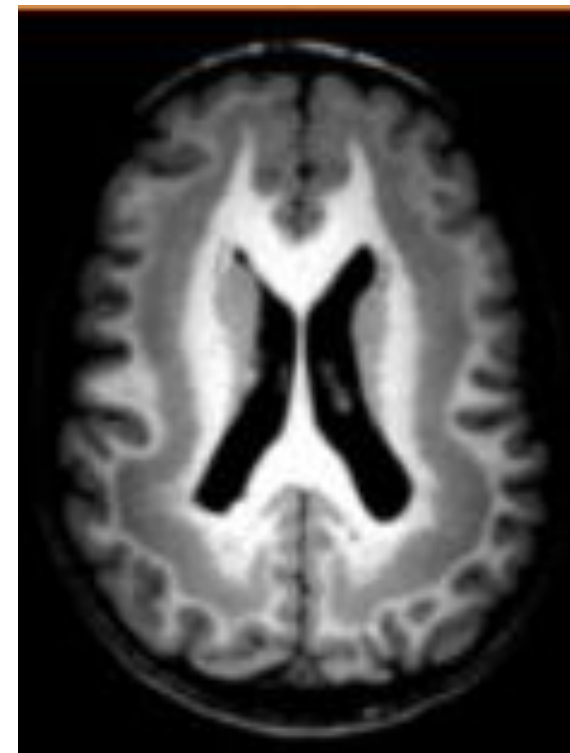
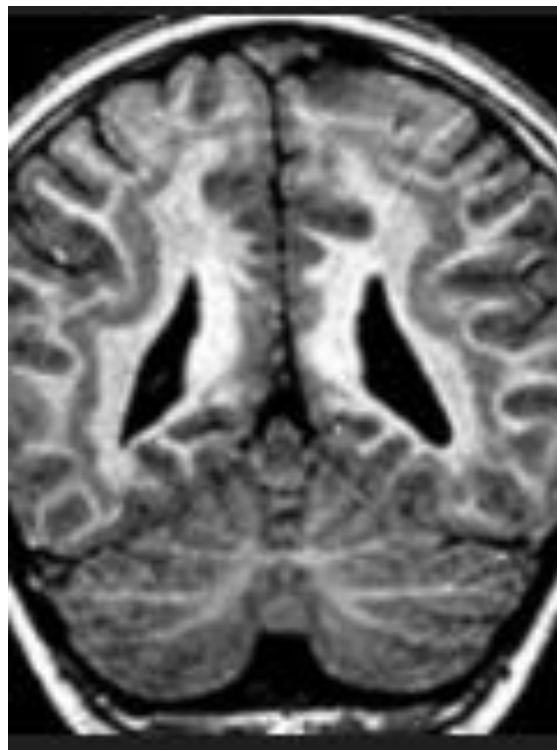
Subependymal Heterotopia

Diffuse subcortical laminar heterotopia



Double cortical sign

Band Heterotopia



Hemispheric abnormalities



Volume asymmetry

Determine which hemisphere is abnormal



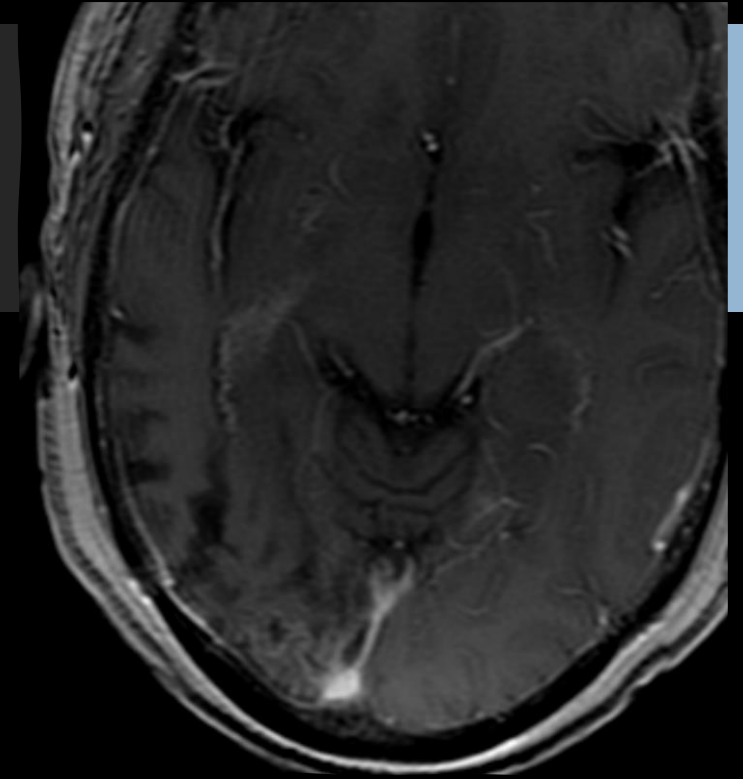
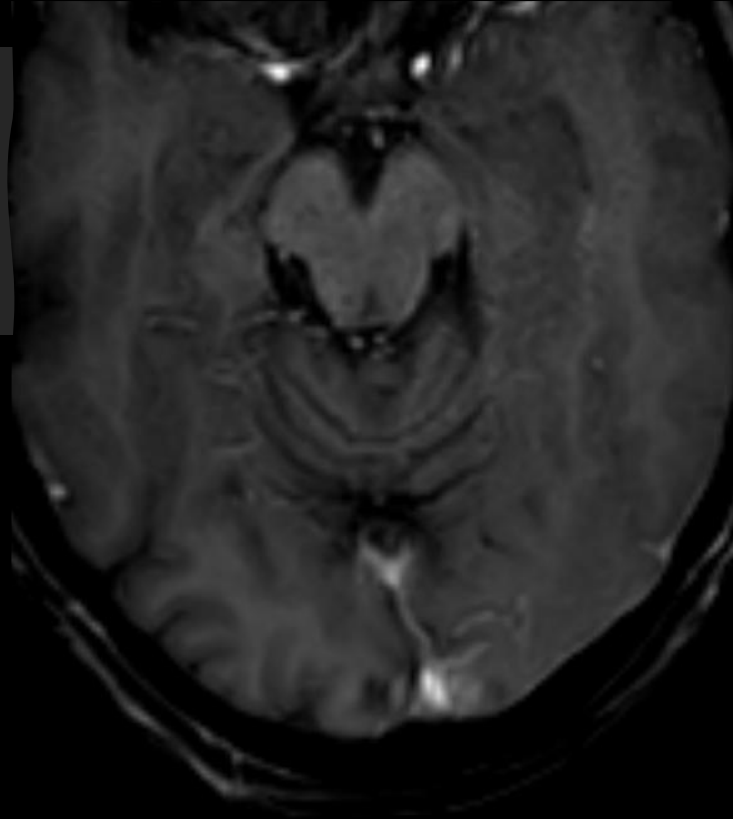
If smaller hemisphere abnormal

Phakomatosis: Sturge Weber
Post-infectious or traumatic: Rasmussen



If larger hemisphere abnormal

Hemimegalencephaly
Acute Rasmussen



Sturge Weber syndrome

- Anomaly of venous drainage of cerebral hemisphere
 - Pial and choroidal angiomas develop
 - Leptomeningeal and prominent
 - Choroidal enhancement
 - Cerebral atrophy
 - Gyriform calcification develop

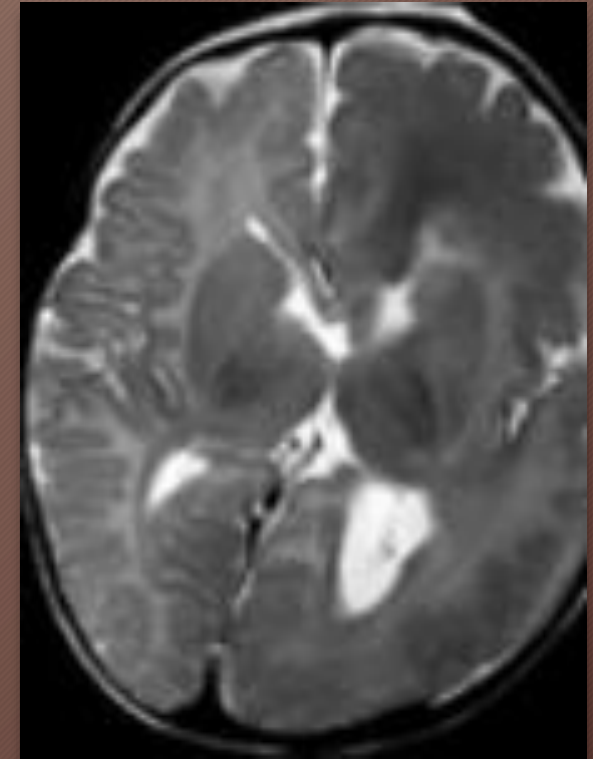
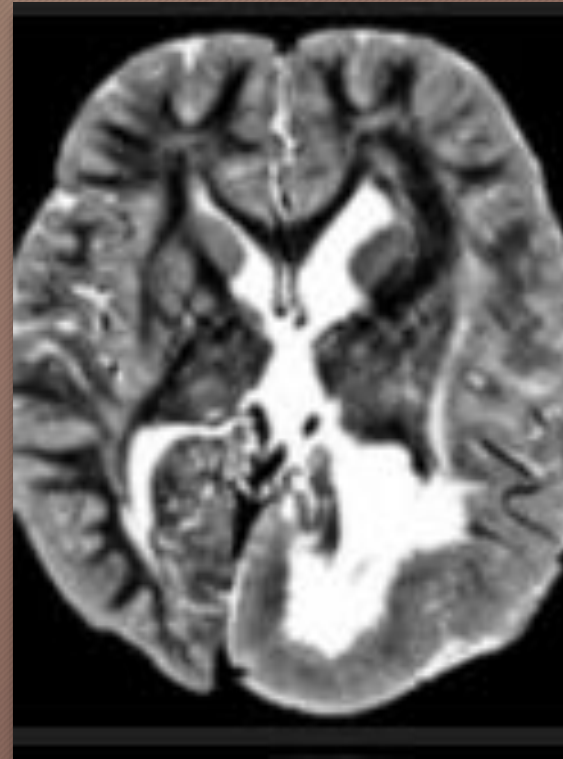
Hemimegalencephaly

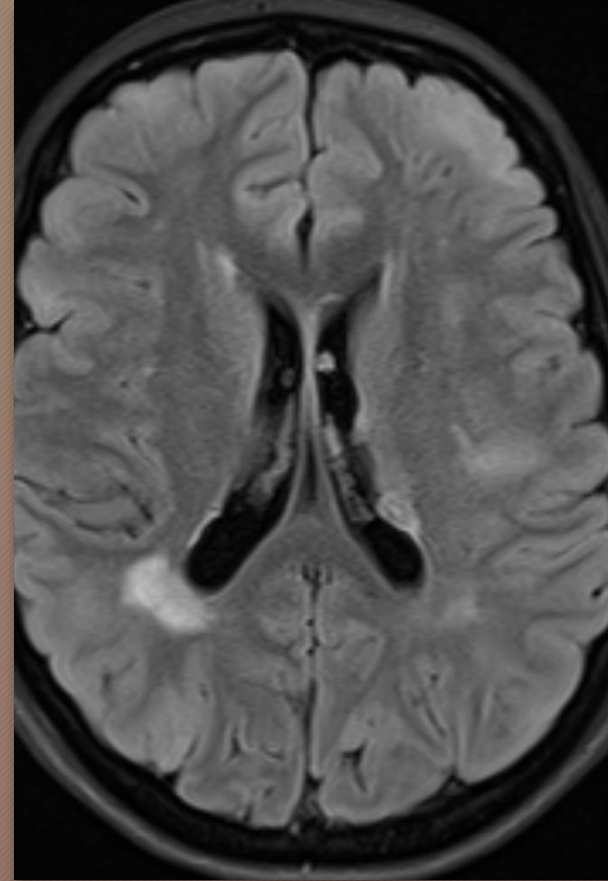
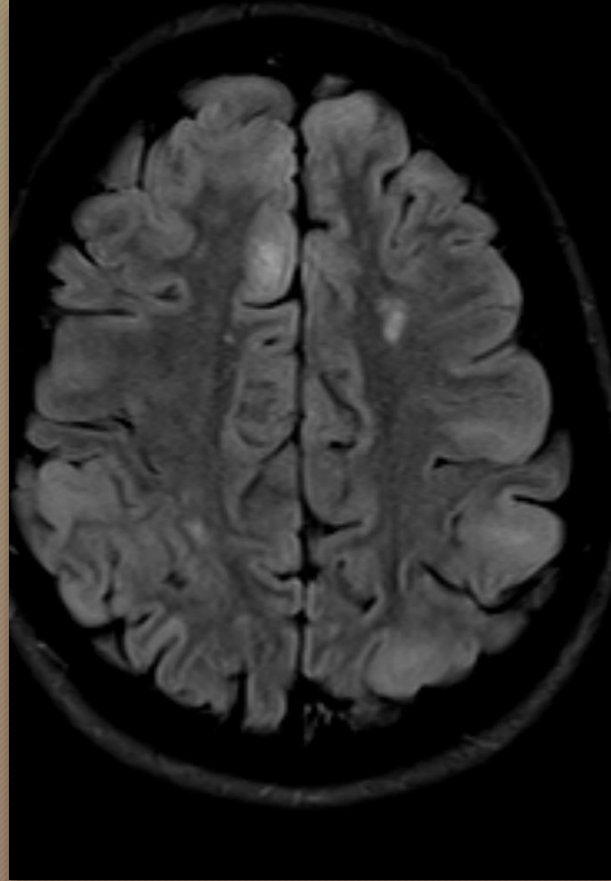
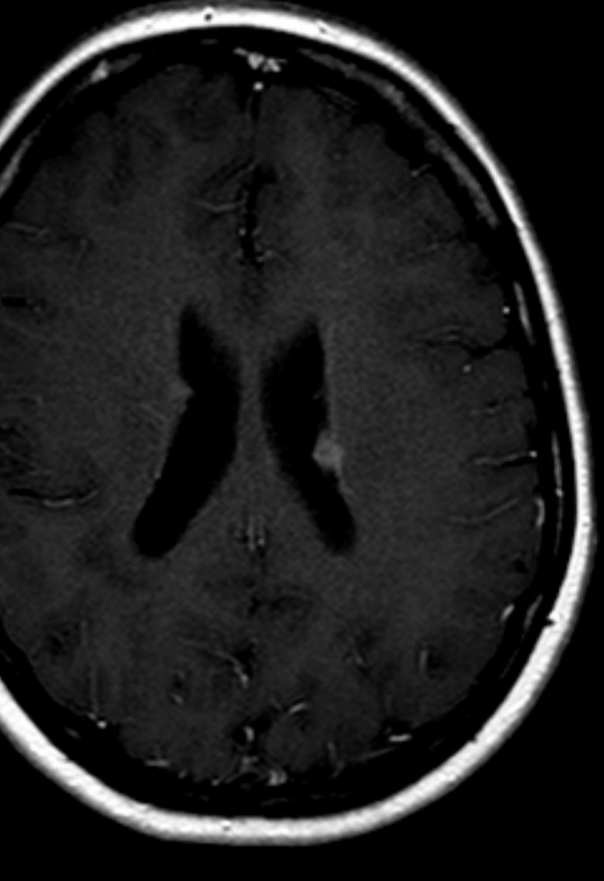
- Hamartomatous overgrowth of part or all of cerebral hemisphere
- Defective cellular organization and migration
- Refractory seizure activity : require hemispherectomy



Hemimegalencephaly

- MRI
 - Large dysplastic cerebral hemisphere
 - Extensive dysplasia
 - PMG and pachygyria with thickened cortices
 - Increased myelination
- 30% syndromic
 - Klippel Trenaunay Weber
 - Proteus
 - Hypomelanosis of Ito
 - Epidermal Nevus syndrome
 - NF1





Tuberous sclerosis

Thank you!

- vprattipati@uabmc.edu