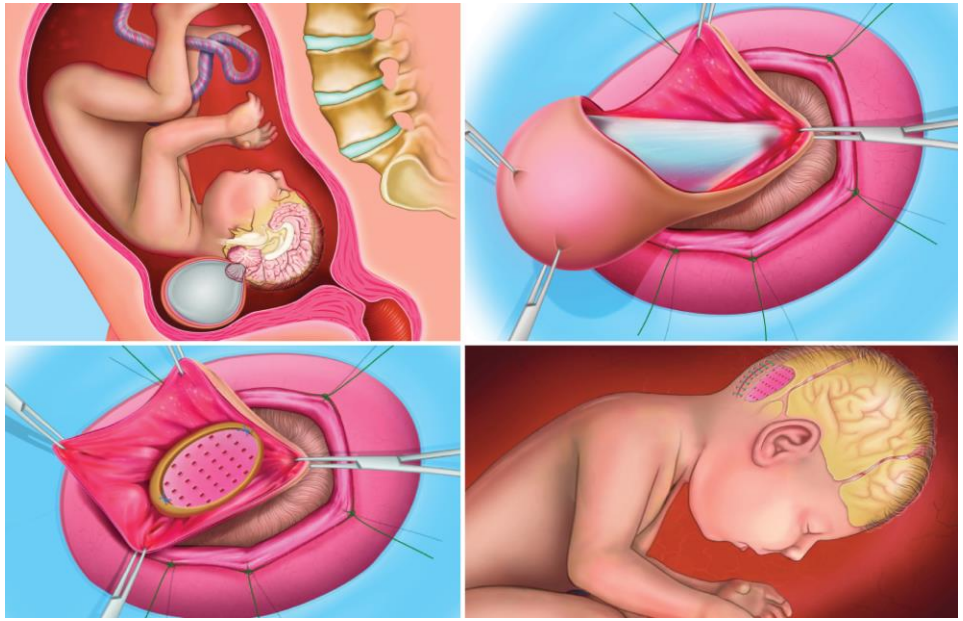
SEMINAR PEDIATRIC NEUROSURGERY: PEDIATRIC HYDROCEPHALUS, CRANIOPATHIES. SPINA BIFIDA AND OTHER DEVELOPMENTAL DISORDERS, PEDIATRIC BRAIN TUMORS

34484 Pathology of the nervous system

Neurosurgery



Prof. Vicente Vanaclocha
Prof. Pedro Roldan
Prof. Guillermo García-March
Prof. José María Gallego
Prof. Ricardo Prat
Prof. Francisco Verdú
vivava@uv.es
pedro.roldan@uv.es

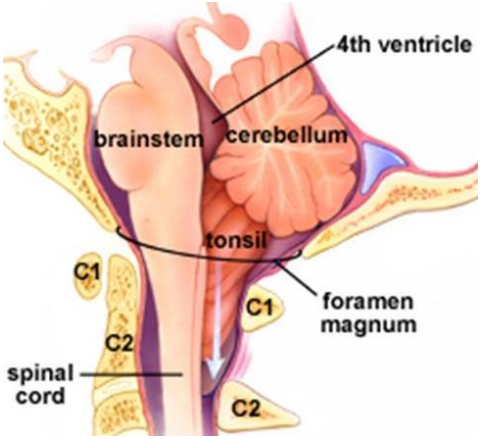
Key points to study

- Infantile hydrocephalus
- Craniopathies
- Spina bifida
- Pediatric age brain tumors
- Other developmental abnormalities
  - Chiari
  - Arnold-Chiari

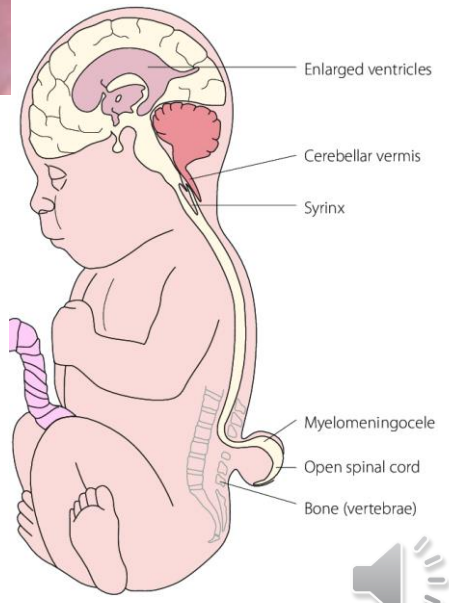
Craniopathy

Spina bifida

Hydrocephalus



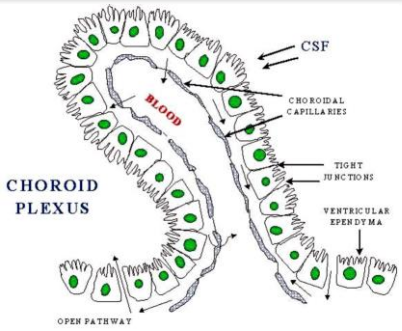
Chiari type I malformation

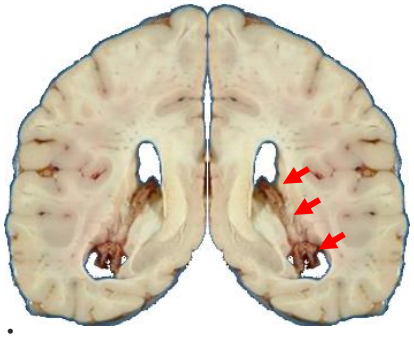


Arnold-Chiari type II malf.

Cerebrospinal fluid (CSF)

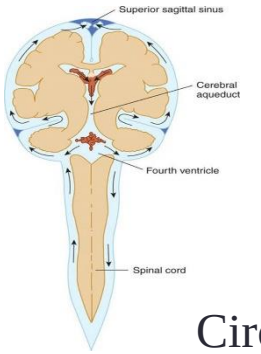
- **Production** ~ 200-500ml/day
  - Child 8 ml/h
  - Adult 20 ml/h
- **Total volume**
  - Newborn 20-50 ml
  - Baby 40-60 ml
  - Child 60-120 ml
  - Adult 120-200 ml
- **Functions**
  - Brain & spinal cord protection against blows
  - Transport of waste substances
  - Give space to avoid ⇑ICP
- **Circulation**
  - Ventricles ⇨ subarachnoid space ⇨ arachnoid villi ⇨ venous sinuses

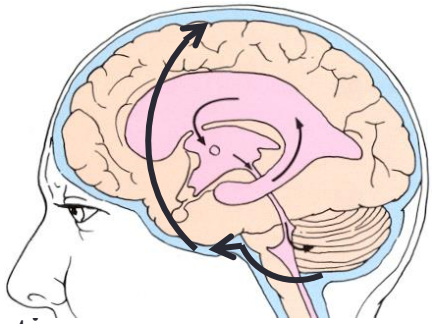




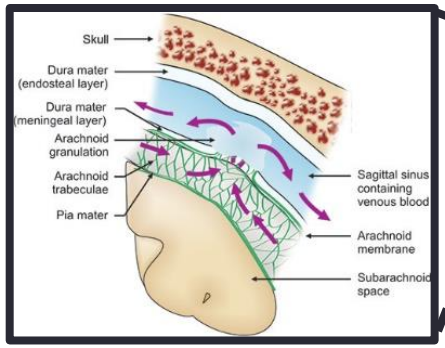
Production

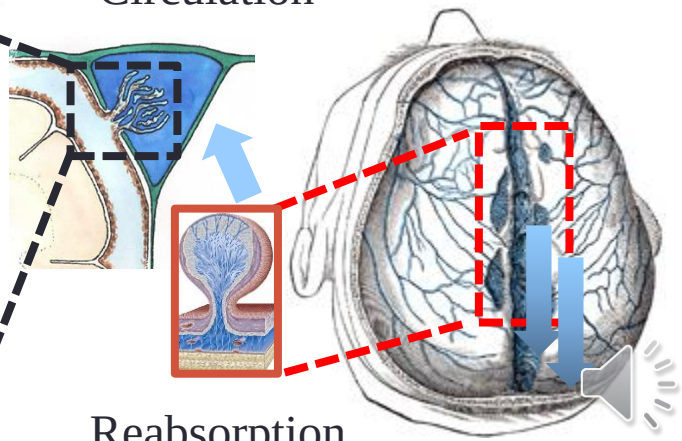
REMEMBER?





Circulation

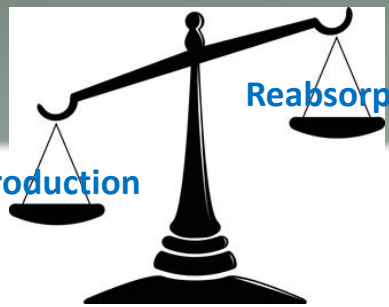


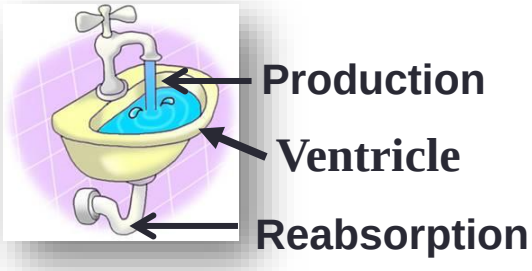


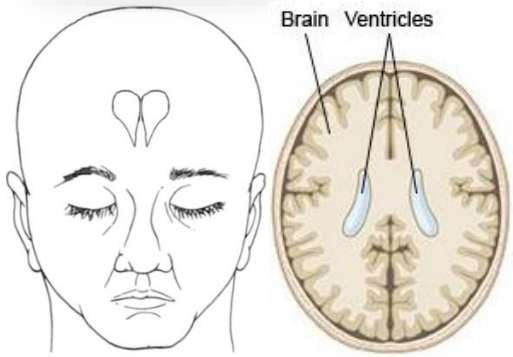
Reabsorption

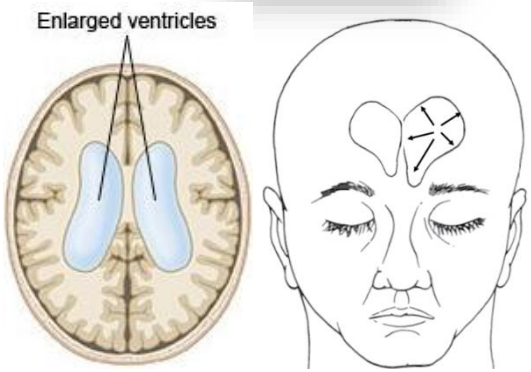
HYDROCEPHALUS

- Hydrocephalus = ⇑ intracranial CSF volume
  - Hydros = water
  - Kefalos = head
- **Causes**
  - ⇑ production (very rare)
  - Circulation block
    - The most frequent
  - ⇓ drainage (resorption)

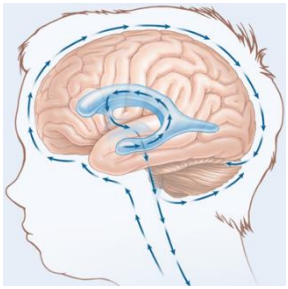




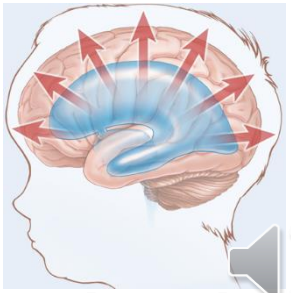




REMEMBER?



Normal

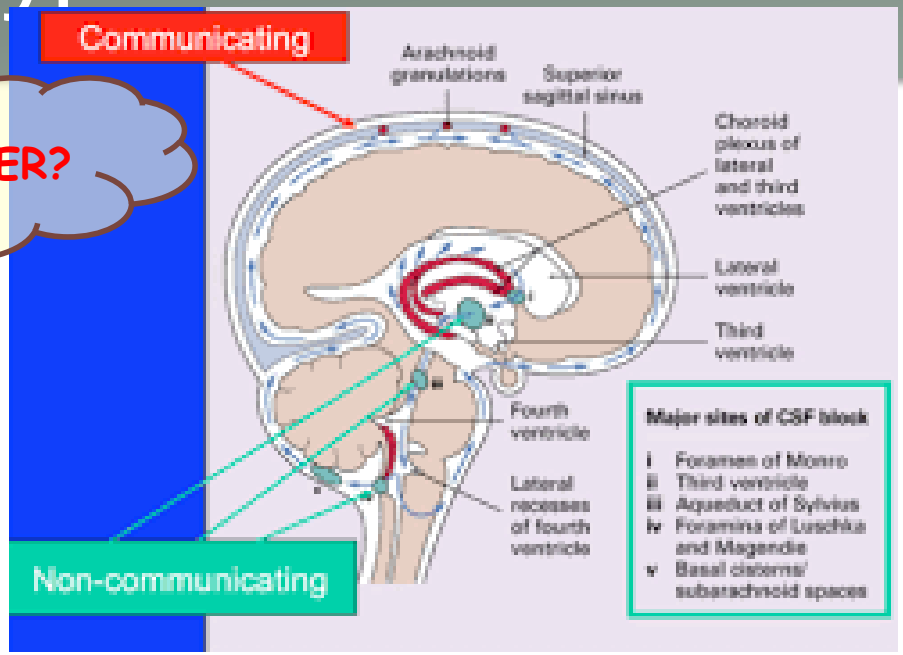


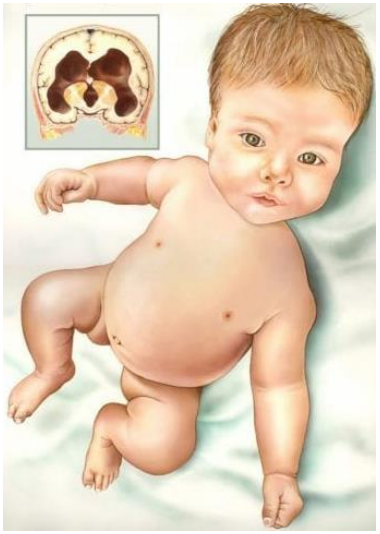
Hydrocephalus

Hydrocephalus: types

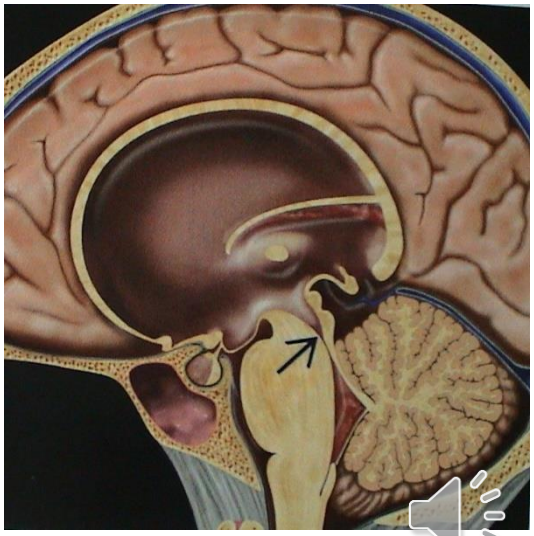
- According CSF flow
  - **Communicating**
    - CSF block in subarachnoid space
  - **Non-communicating**
    - CSF flow block inside ventricles
      - Foramen of Monro
      - Third ventricle
      - Aqueduct of Silvius
      - Sixth ventricle
- According to presentation
  - **Congenital**
    - Present from birth
    - Causes
      - Intraventricular hemorrhage (prematurity)
      - Silvius aqueduct stenosis
  - **Acquired**

REMEMBER?





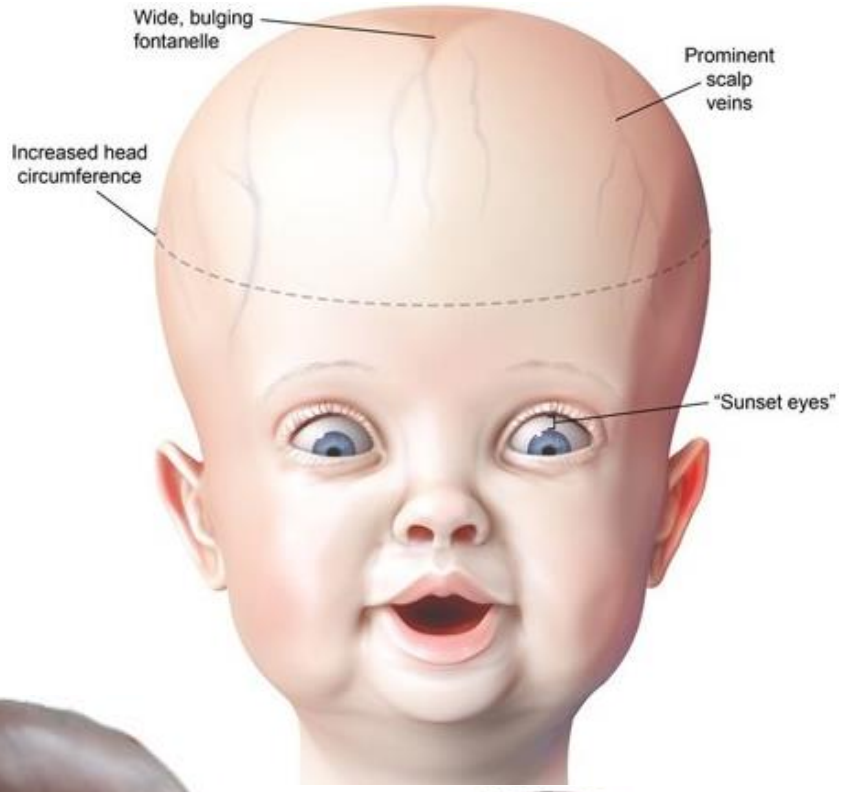
Prematurity

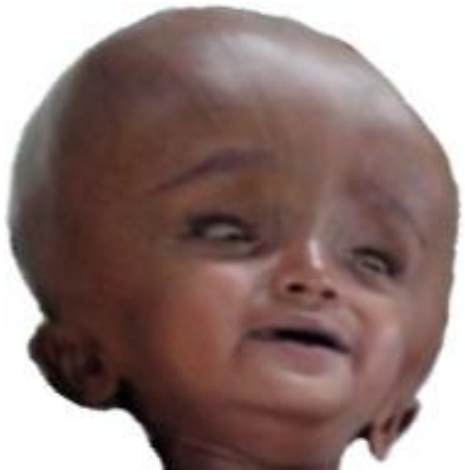


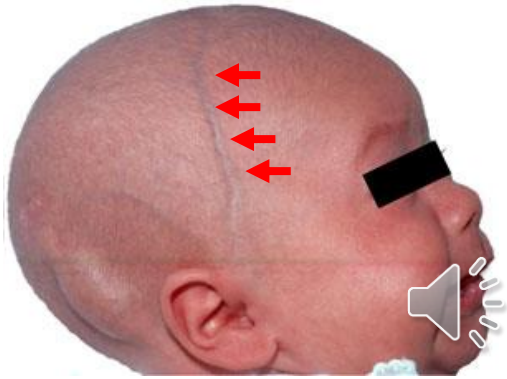
Silvius aqueduct stenosis

Hydrocephalus symptoms in newborn & baby

- **Baby** (sutures and fontanelles still open)
  - Irritability, crying
  - Poor feeding, lethargy
  - Bulging fontanelle
  - Prominent peri-cranial veins
  - Increased head circumference
  - ‘Setting sun’ eyes
  - Chronic hydrocephalus = psychomotor developmental delay
- **Child and adult**
  - Acute
  - Chronic







Increased newborn cranial circumference

Normal

Hydrocephalus

Hydrocephalus clinical features

Macrocephaly neonatal period

Childhood macrocephaly

Sun setting eyes

Convergent strabismus

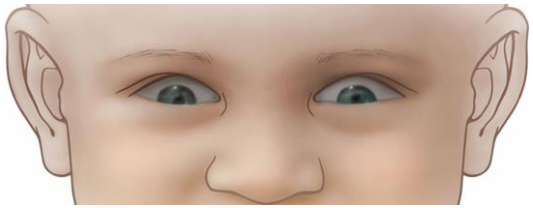
Bulging fontanelle

Hydrocephalus clinical features: child and adult

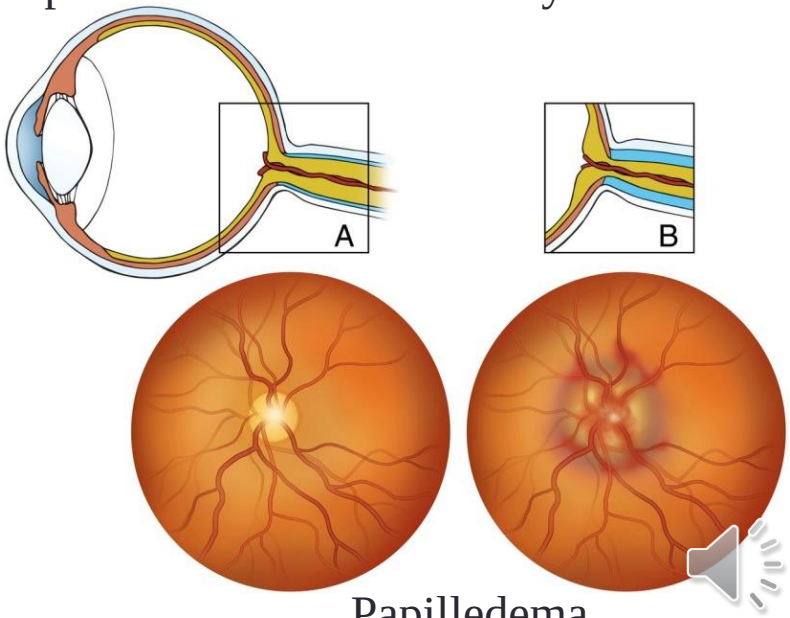
- **Acute**: intracranial hypertension
  - Headache, nausea, vomiting
  - Papilledema
  - Fourth cranial nerve palsy
  - Gait disorders
  - Parinaud syndrome
    - Paralysis upwards vertical conjugate gaze
- **Chronic**
  - Papilledema → optic nerve atrophy → blindness
  - Gait disturbances (spastic paraparesis)
  - Upper limb dysmetria
  - Endocrine disturbances

Spastic paraparesis

Sixth cranial nerve palsy



Parinaud syndrome



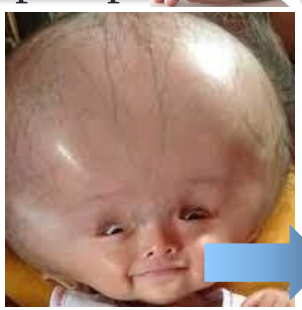
Papilledema

Hydrocephalus diagnosis

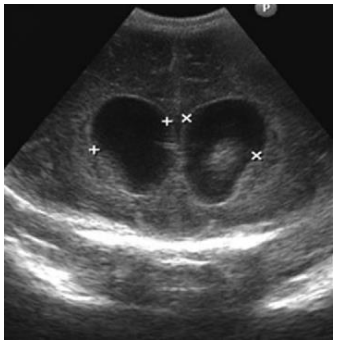
- **Baby**
  - Increased cranial perimeter
  - ‘Waterfall pot’ Macewen's sign
    - Skull percussion sound
  - Transillumination
  - Trans-fontanelle echography
- **Infant, child and adult**
  - Baby: skull x-ray
    - Acute: suture diastasis
    - Chronic: in the skull digitiform impressions and enlargement, erosion, or decalcification sella turcica
    - CT-scan/MRI

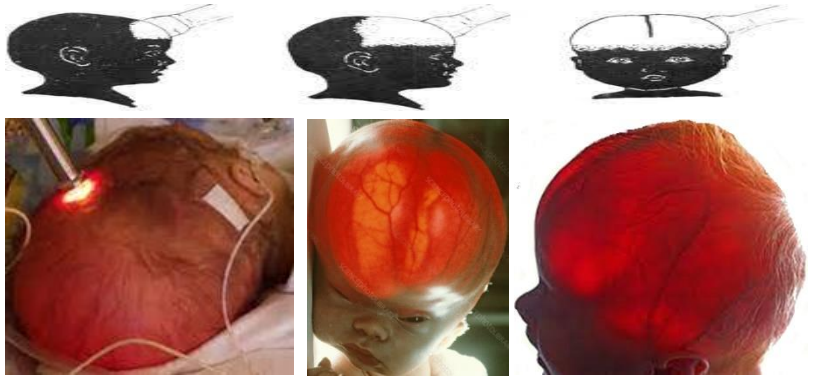
Tap, tap!

Cranial circumference



Waterfall pot sign





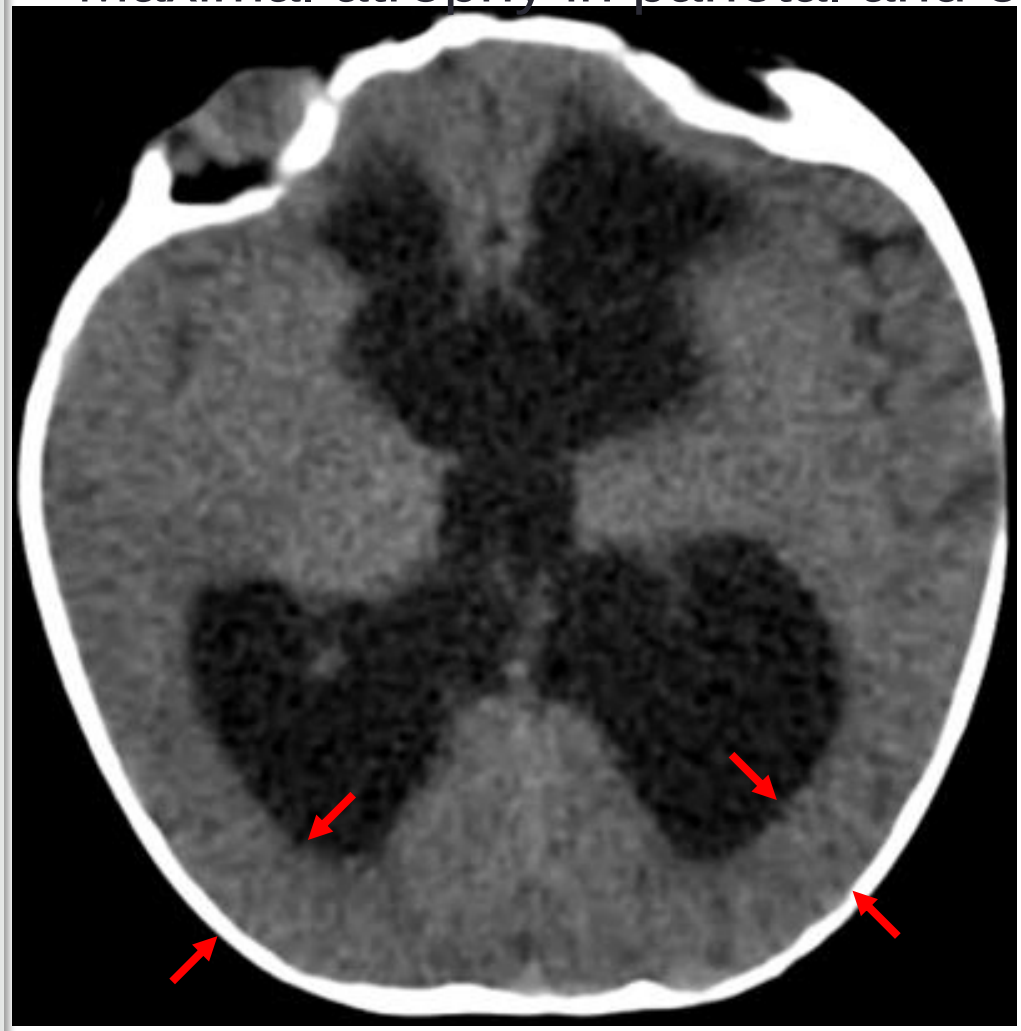
Transillumination

Transfontanelle echography

Skull x-ray

CT-scan/MRI neonatal hydrocephalus: brain atrophy

- Maximal atrophy in parietal and occipital lobes

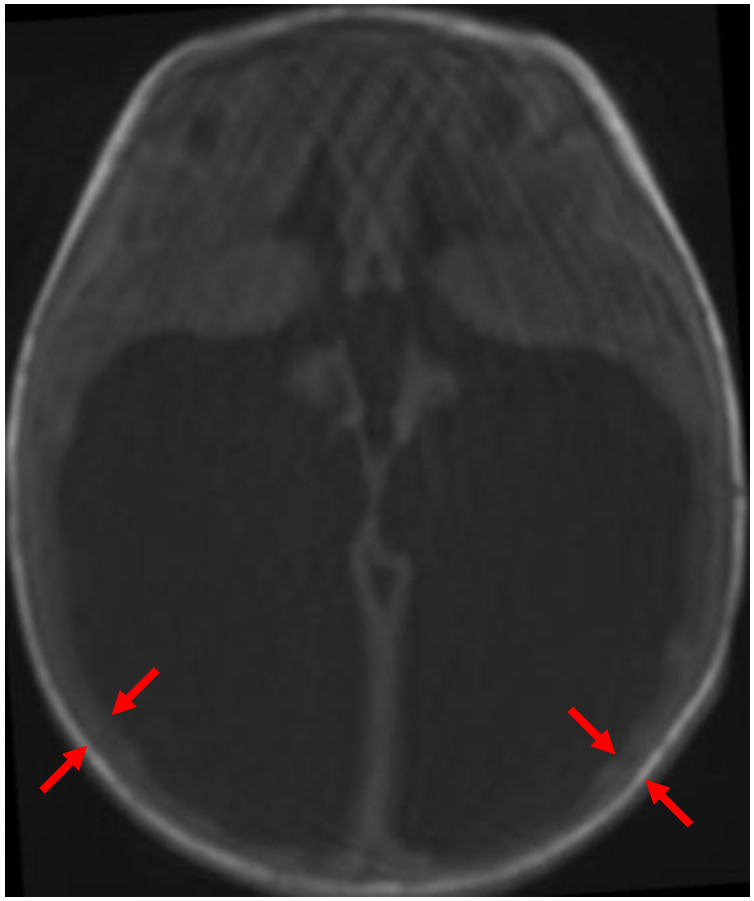


CT-scan

MRI

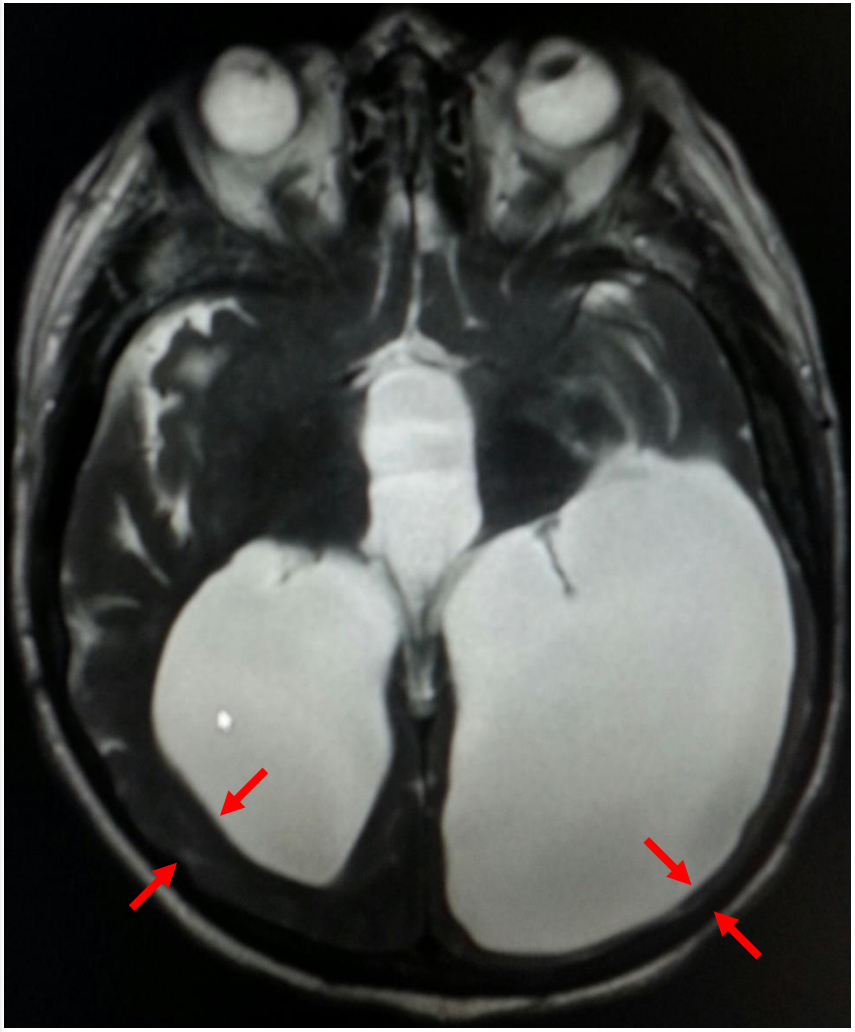
CT-scan neonatal hydrocephalus: brain atrophy

- Ventricular system dilatation + lack of nervous tissue development = brain atrophy = psychomotor retardation
- Increased head circumference



MRI neonatal hydrocephalus: brain atrophy

- Maximal atrophy in parietal and occipital lobes

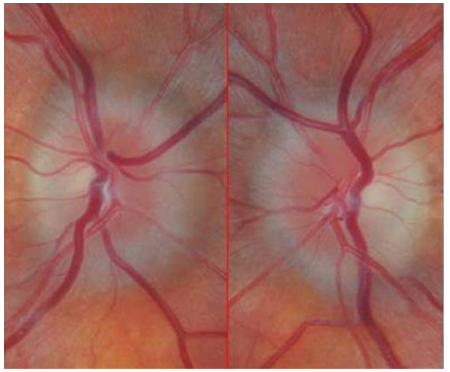


CT-scan child and adult: moderate ventricular size increase without brain atrophy

- Closed skull = skull expansion impossible = intracranial hypertension

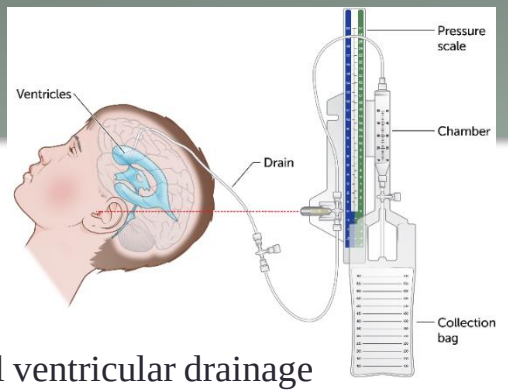




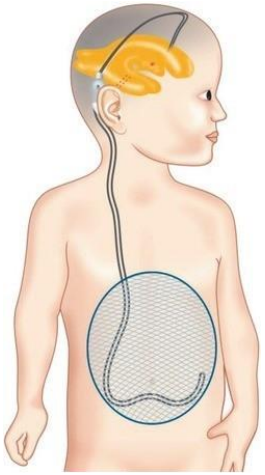


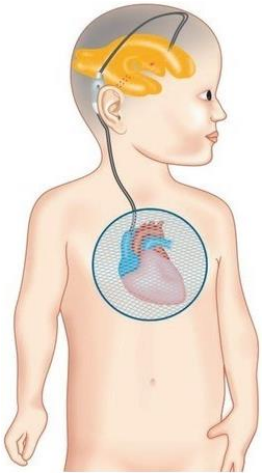

CSF drainage options

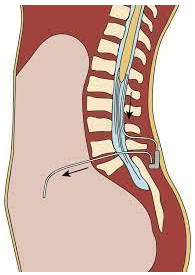
- **External ventricular drainage**
  - Temporary measure
  - In intraventricular hemorrhage (prematurity) or infection
- **Permanent CSF diversion (shunt)** ~ 80% cases
  - Ventricleperitoneal shunt
  - Ventricleatrial shunt
  - Lumboperitoneal shunt
  - Ventriclepleural shunt
- **Ventriculostomy** ~ 15% cases
  - Third ventricle floor opening = CSF flows out directly into the subarachnoid space
  - Indicated in CSF obstruction at third ventricle, aqueduct of Sylvius, or posterior fossa

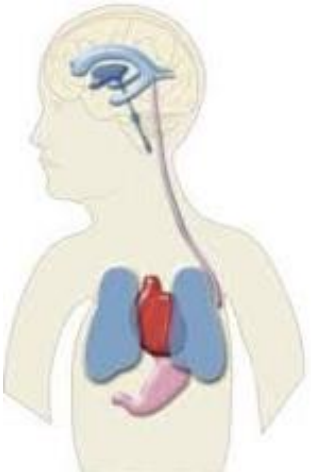


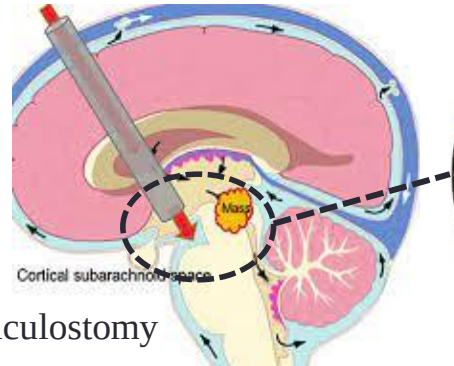
External ventricular drainage


Ventricle-peritoneal

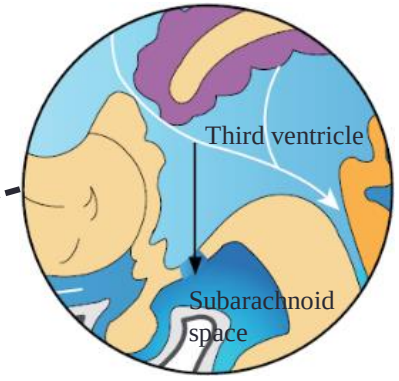

Ventricle-atrial

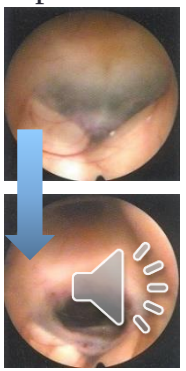

Lumbo-peritoneal


Ventricle-pleural



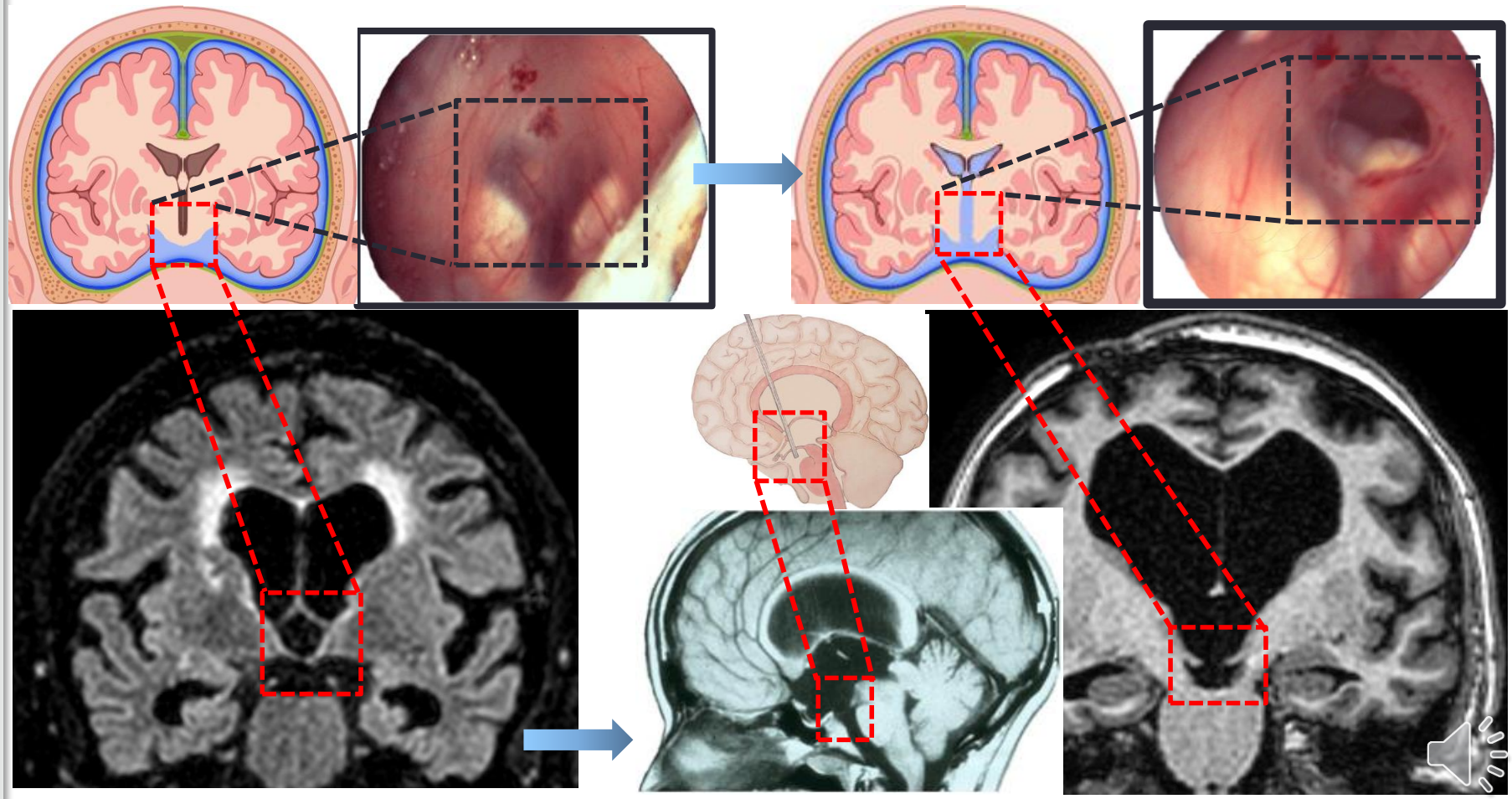
Ventriculostomy





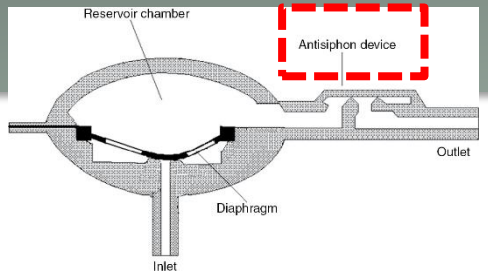
Ventriculostomy third ventricle floor

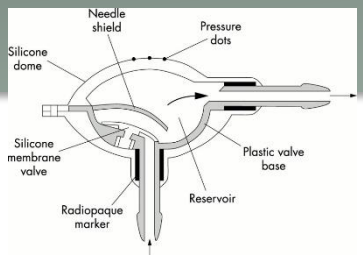
- Few indications
- Ideal: NO need for CSF shunt = no reinterventions

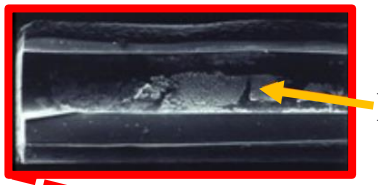


CSF shunting possibilities

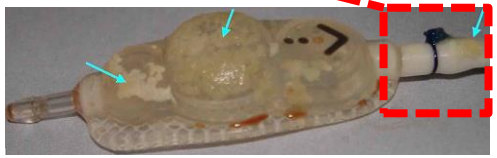
- **Inadequate drainage**
  - Excessive abdominal pressure
  - CSF shunt colonisation by bacteria
- **Adequate drainage**
- **Excessive drainage**
  - CSF shunt without anti-siphon system

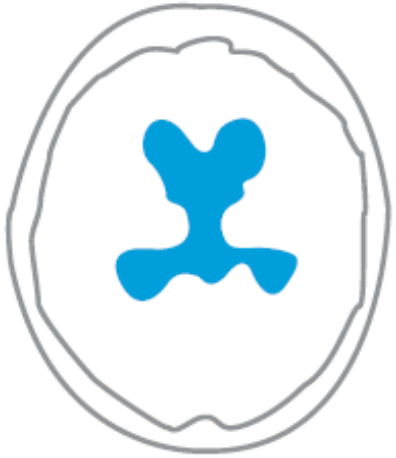






Bacteria





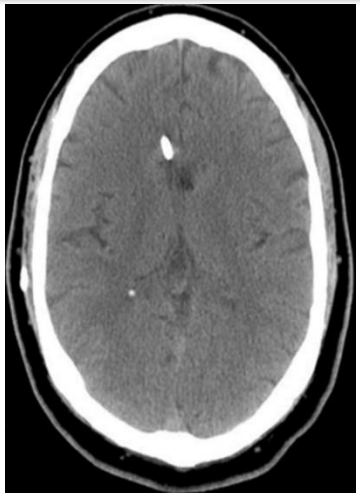
Inadequate drainage

Adequate drainage

Excessive drainage

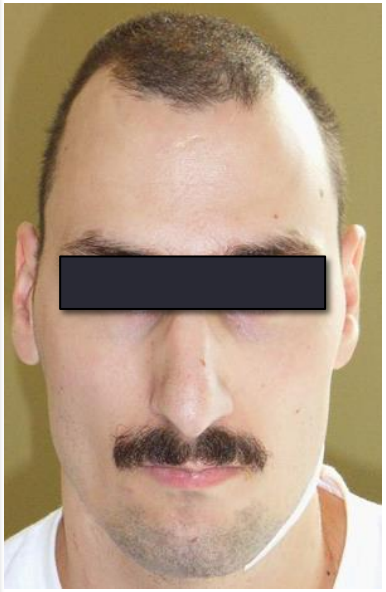
Excessive CSF drainage: consequences

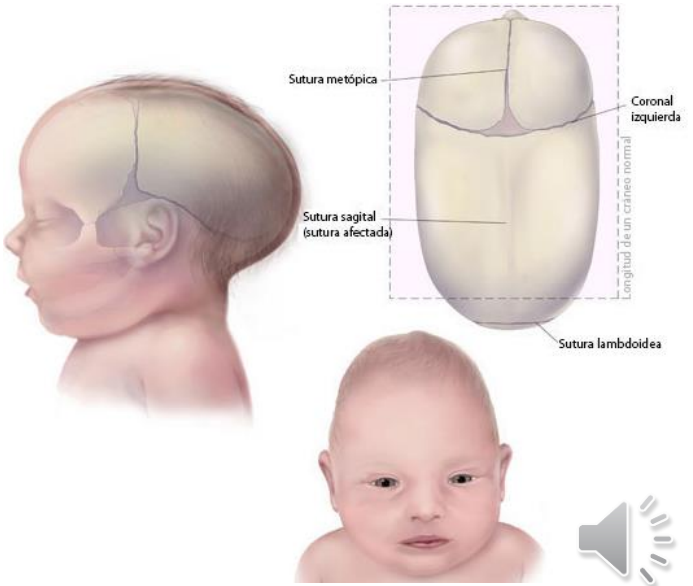
- Ventricular collapse
  - Lower tolerance to ↑ICP
- Chronic subdural hematomas
  - Uni or bilateral
- Secondary craniosynostoses



Ventricular collapse

Chronic subdural hematoma uni- or bilateral

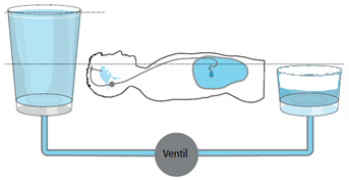


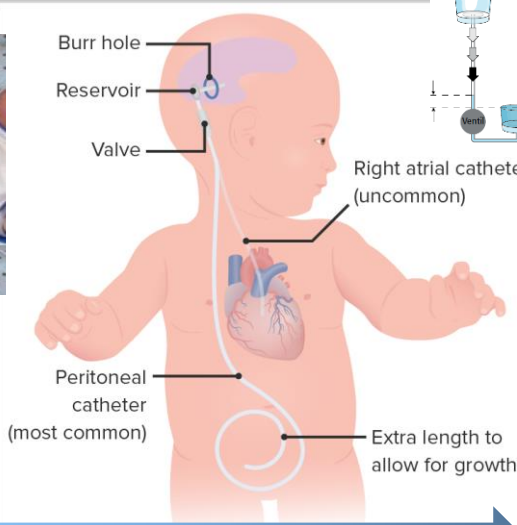


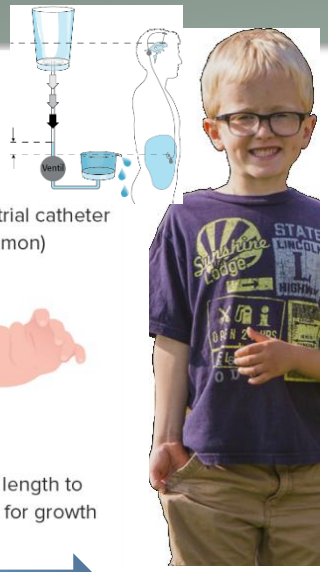
Secondary craniosynostoses

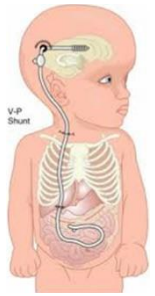
Infantile hydrocephalus problems

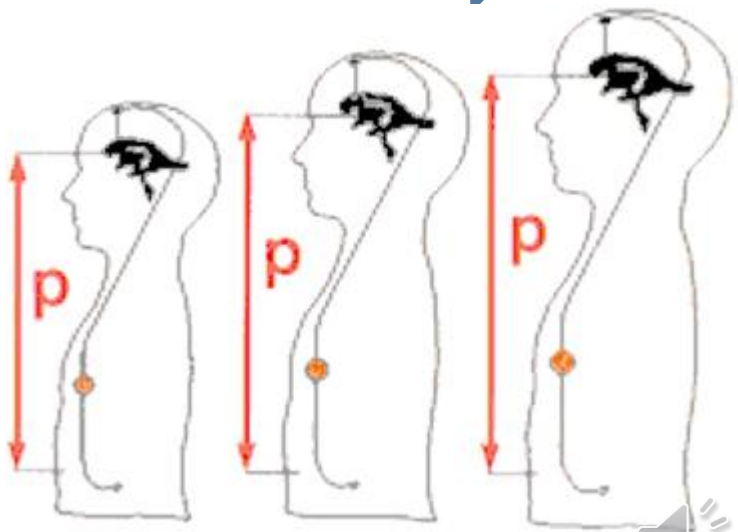
- **Patient growth**
  - Prevented by a sufficiently long peritoneal catheter
    - Impossible in ventriculoatrial shunt
  - Hydrostatic pressure changes with increased patient height
- **Intracranial and abdominal pressure changes**
  - Baby = decubitus,
    child = standing position
    - Siphon effect?
  - Valve that works in infant may not be suitable for child / adolescent / adult
- **Result** = frequent CSF shunt replacement in children





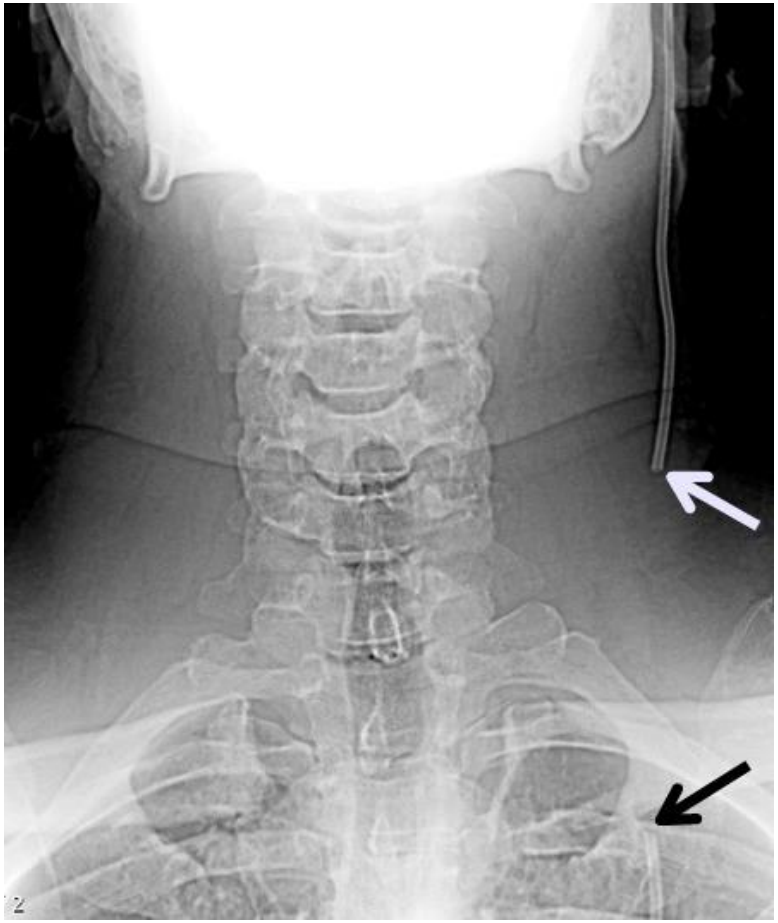






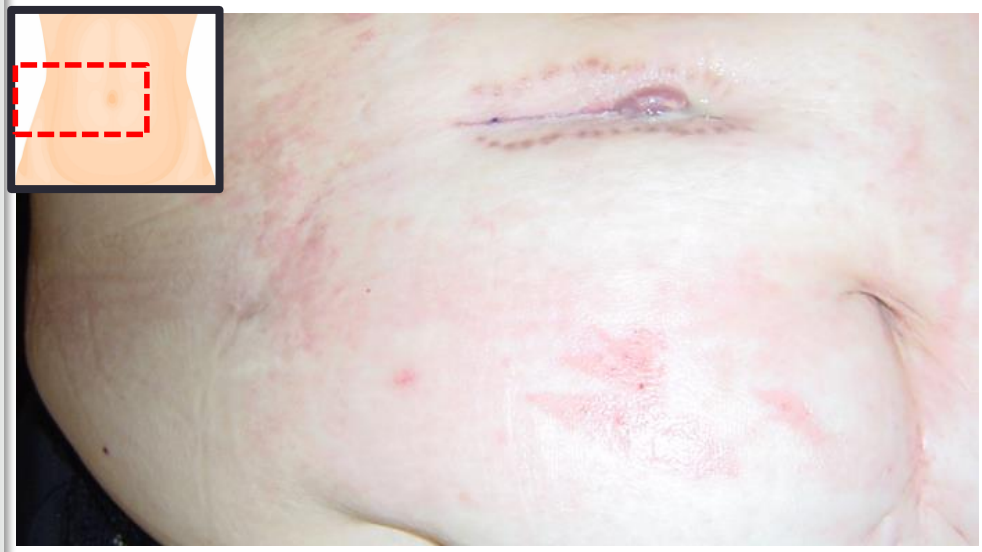
CSF shunt complications: catheter rupture

- Common in children growing up
- ↑ at the clavicle level



CSF shunt complications: shunt infection (1)

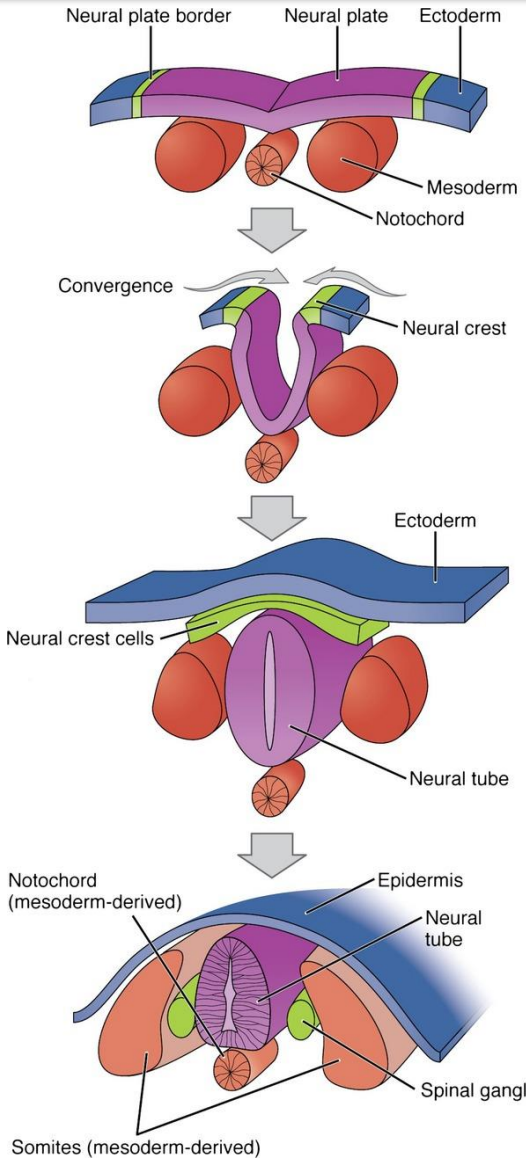
- Shunt = foreign body = ease of infection



Dysraphism

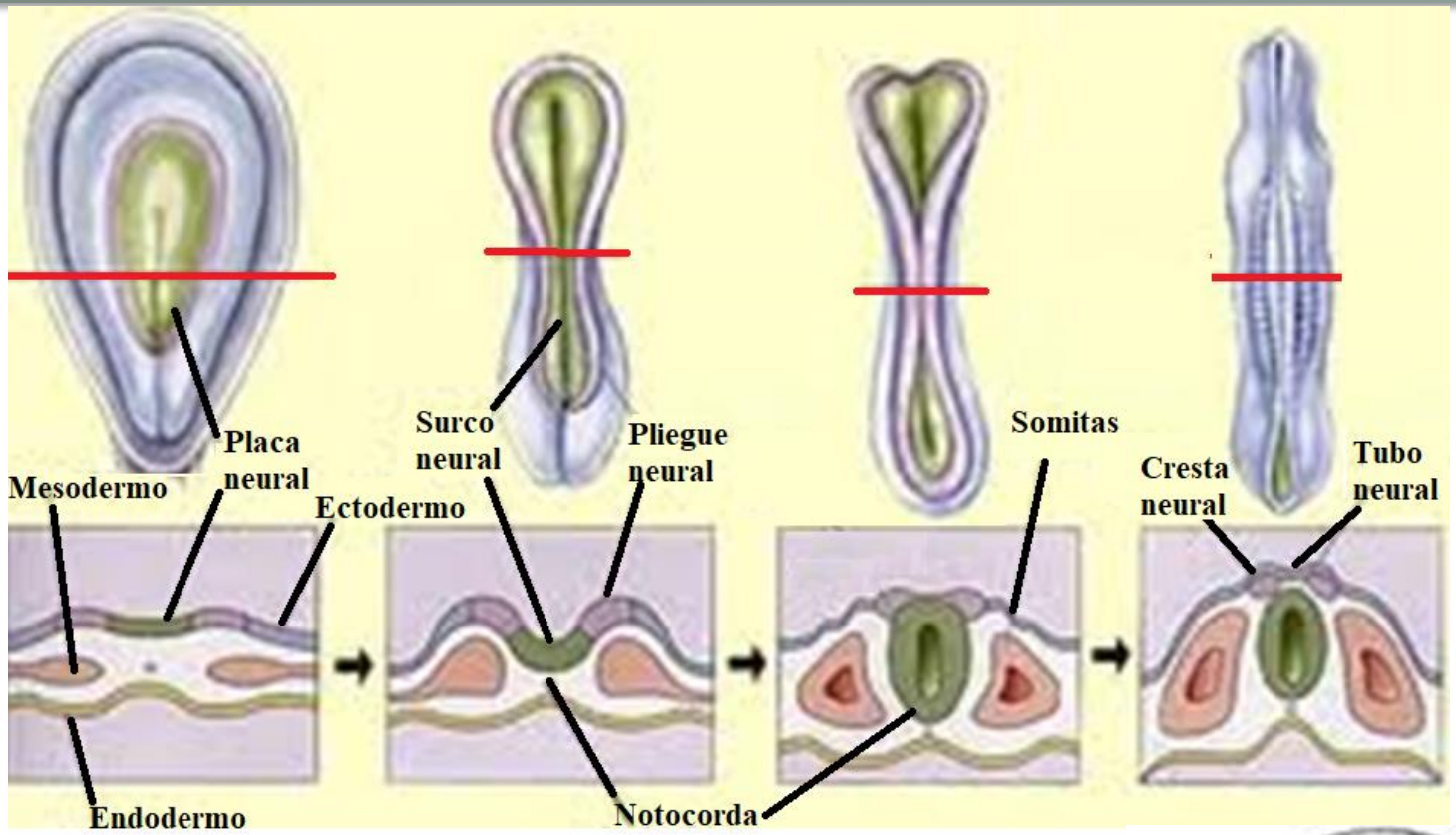
REMEMBER?

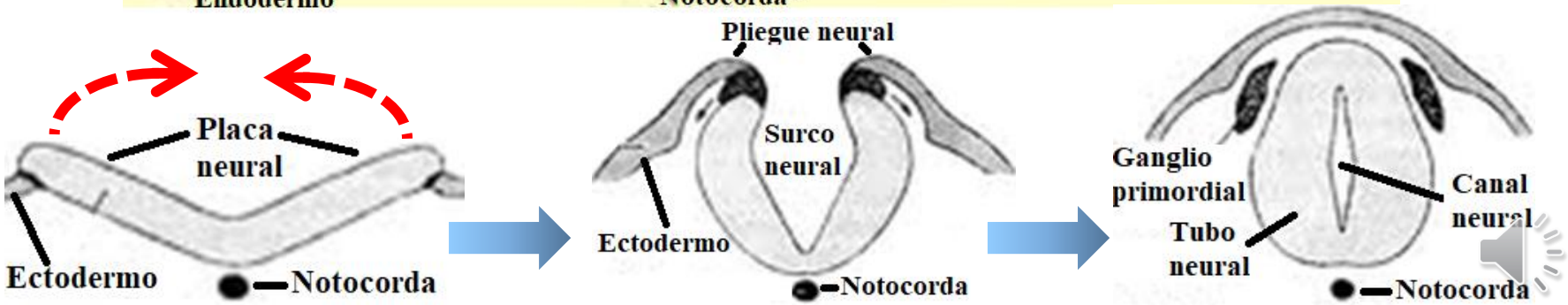
- Dysraphism: midline malformation
- **Midline fusion and developmental** defects
  - Skull
  - Spine
- **Congenital**
  - Some have genetical basis
- **May involve** soft tissue, bone structures and/or nervous tissue



Neural tube closure

REMEMBER?

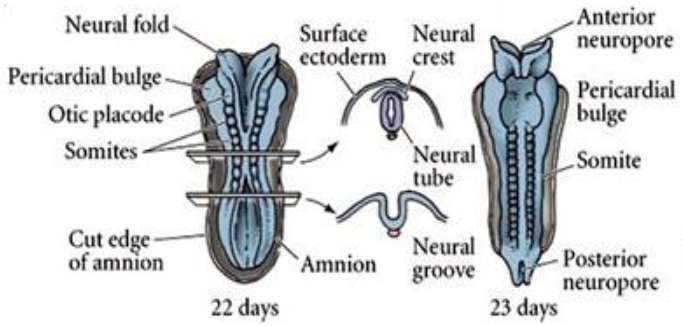


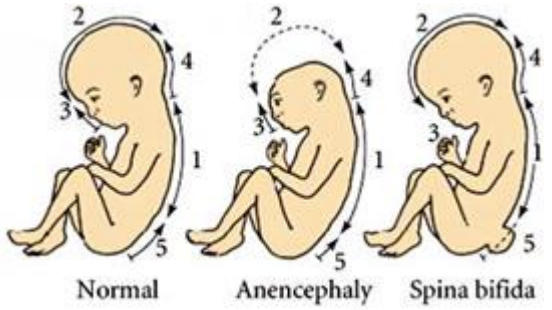


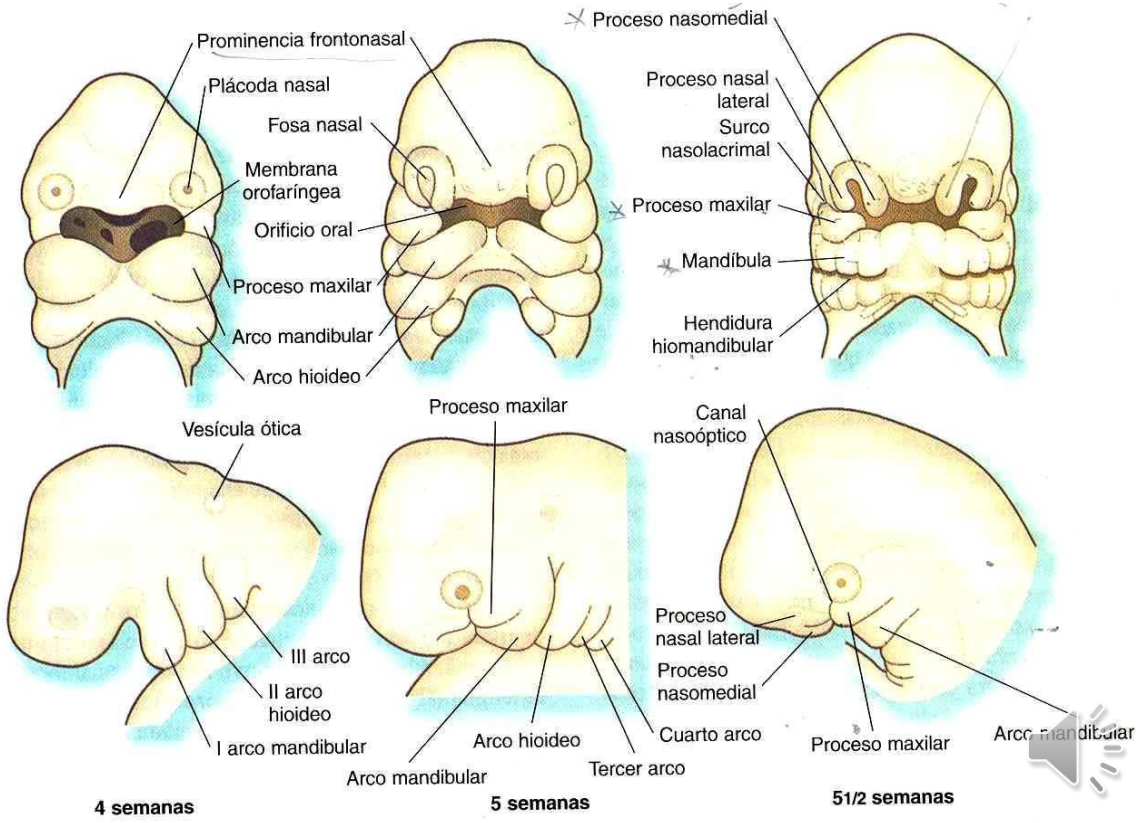
Review embryology: < 8 weeks

- **4th week**: Neural tube closure
  - Anencephaly
  - Myelomeningocele
- **5th week**: Closure anterior middle line & pontine flexure development
  - Arrinencephaly
  - Arnold-Chiari malformation

REMEMBER?

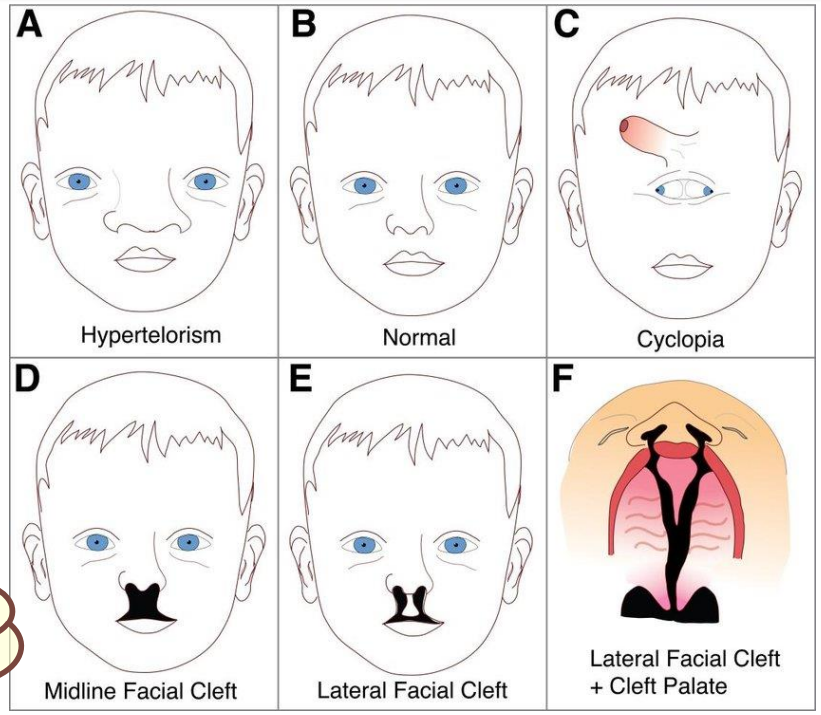






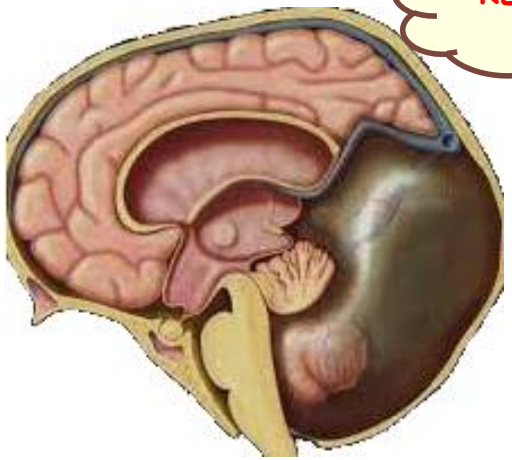
Embryological development < 8 weeks

- **6th week**: rostral membrane & first visceral arcs development
  - Dandy-Walker syndrome
  - Craniofacial malformations
  - Cleft palate & harelip



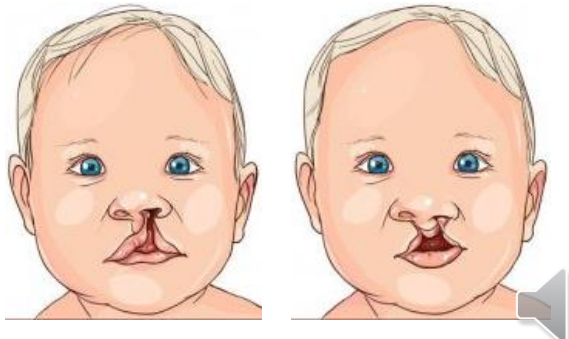
Craniofacial malformations

REMEMBER?



Dandy-Walker syndrome

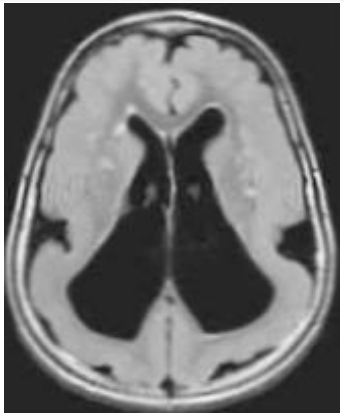
Cleft palate



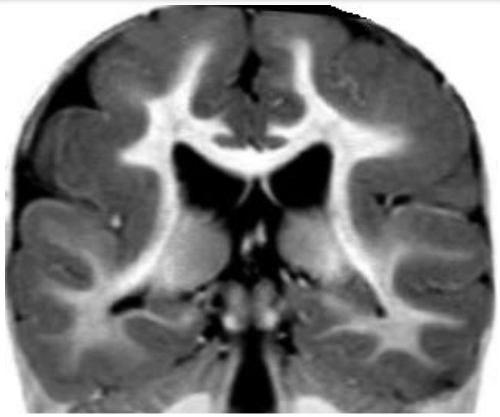
Harelip

Fetogenesis > 10th week

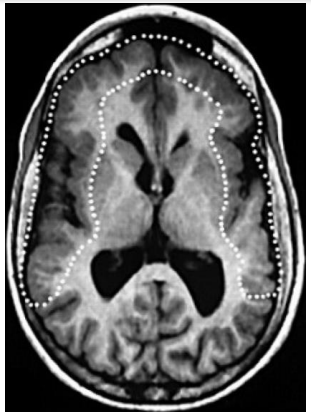
- **Neuronal migration defects**
  - Agyria
  - Pachygyria
  - Microgyria
  - Periventricular heterotopias
- **Commissure defects**
  - Agenesis corpus callosum



Agyria

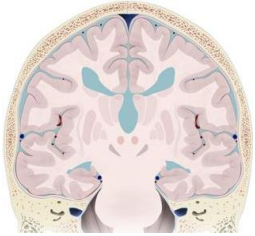


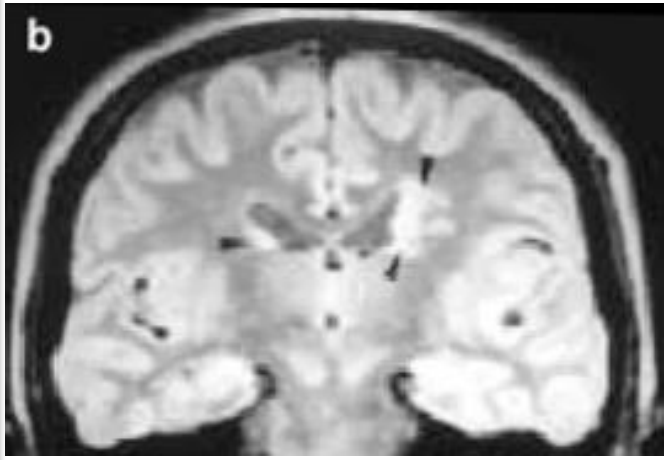
Pachygyria



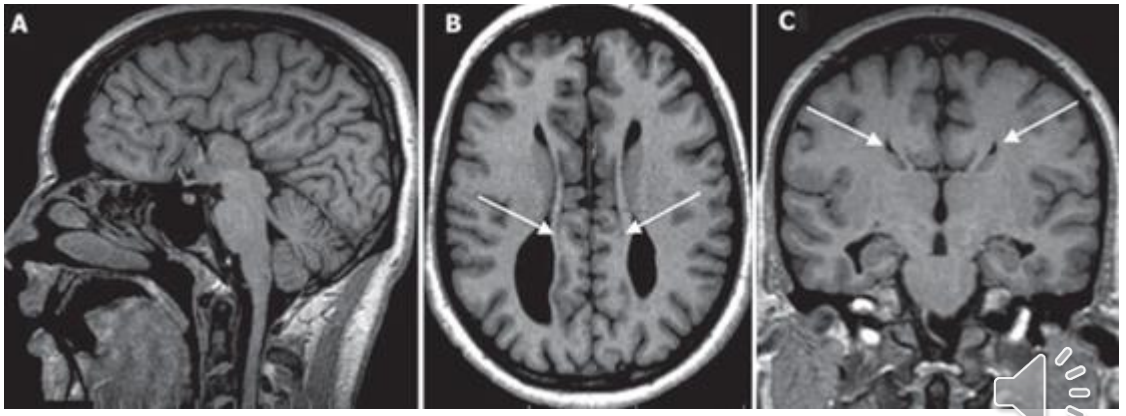
Microgyria

REMEMBER?





Periventricular heterotopia



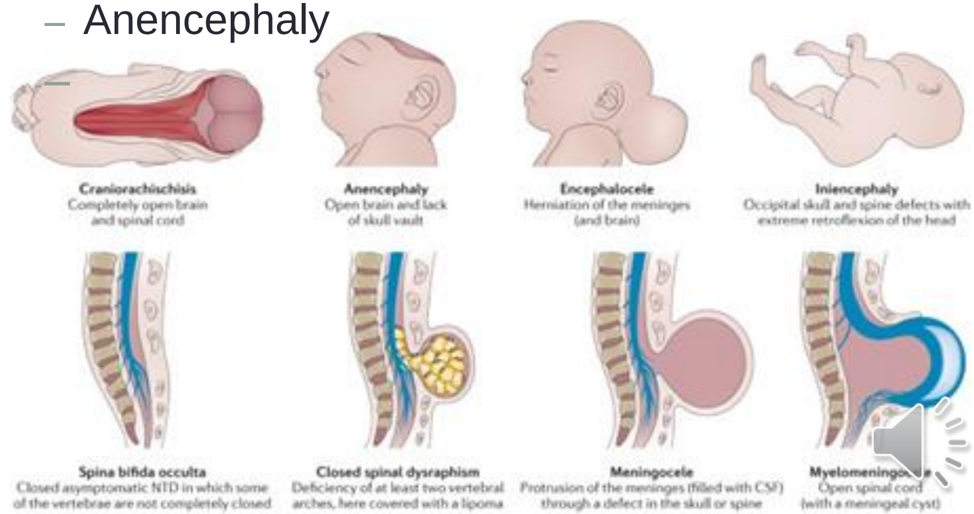
Agenesis corpus callosum

Most important dysraphisms

Dismesenquimosis

- I. Craneal (no sindrómica)
  - A. Craneosinostosis:
    1. Escafocefalia (sutura sagital).
    2. Braquicefalia (sutura coronal).
    3. Trigonocefalia (Sutura metópica).
    4. Plagiocefalia (craneosinostosis unilateral):
       - a) Anterior (hemicoronal).
       - b) Posterior (hemilandoidea).
    5. Sus combinaciones.
  - B. Craneosquisis:
    1. Meningocele
    2. Encefalocele
    3. Anencefalia
- II. Facial
  1. Fisuras de Tessier 0 a 3 (Fisura labio-alveolo-palatina).
  2. Fisuras de Tessier 4 a 6 (Fisura Facial Media).
  3. Fisuras de Tessier 7 (Microsomía Hemifacial).
  4. Fisuras de Tessier 6 a 8 (Treacher Collins).
  5. Sus combinaciones.
- III. Cráneofacial
  1. Fisuras de Tessier de la 9 a la 14 (no sindrómicas), con o sin craneosquisis
  2. Grandes Síndromes craneofaciales

- Craniosynostoses
  - Scaphocephaly (sagittal suture)
  - Trigonocephaly (metopic suture)
  - Plagiocephaly (unilateral coronal or lambdoid suture)
  - Brachycephaly (bilateral coronal suture)
  - Combinations
- Cranioschises
  - Meningocele
  - Encephalocele
  - Anencephaly



Most important dysraphisms

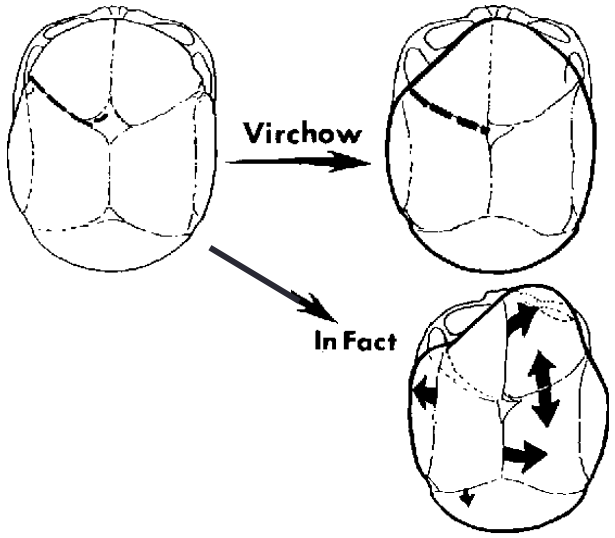
- **Skull**
  - Craniosynostoses = *premature suture closure*
  - Cranioschises = *suture closure failure*
  - Myeloencephalocele = *neural tube closure failure*
- **Cranio-cervical junction abnormalities**
  - Arnold-Chiari malformation = *failure posterior fossa development*
- *Spine*
  - *Myelomeningocele*

OTHER CONGENITAL PATHOLOGIES:

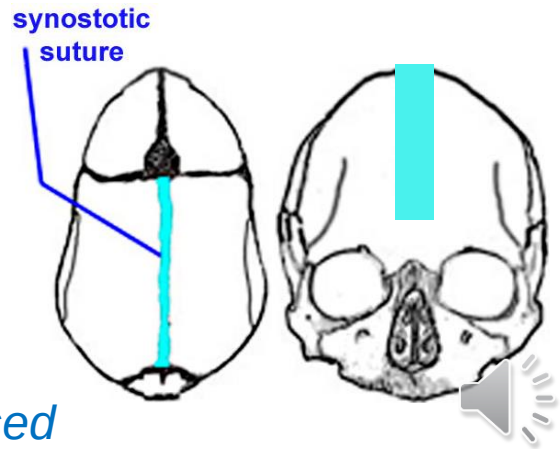
- **Skull size:** Macro and microcephaly
- **Brain:** Anencephaly, Dandy-Walker (fourth ventricle cyst), congenital hydrocephalus
- **Vascular malformations**
- **Calcifications**
- **Tumors** (neurofibromatosis)
- **Occult spinal dysraphism**: dermal sinus, tethered spinal cord, lumbosacral lipoma, diastematomyelia…

1. CRANIOSYNOSTOSES

- Premature closure of one or more cranial sutures → abnormal skull growth + development
  - Primary mesenchymal alteration
  - Sometimes hereditary
    - Usually a genetic mutation
    - Incidence 3 - 5 / 10,000 births



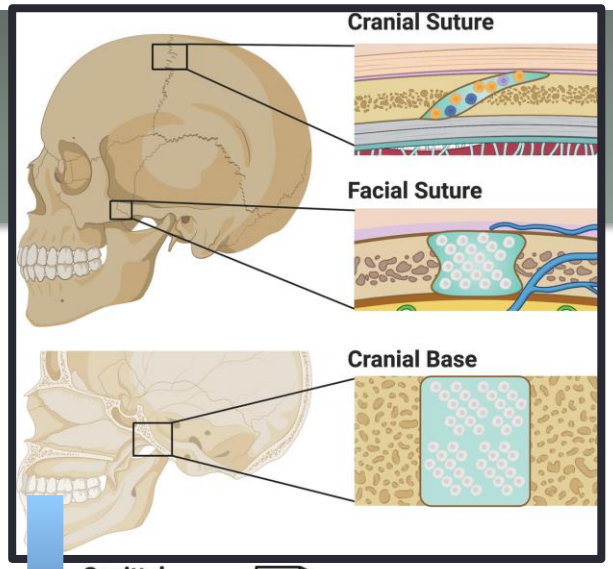
- History
  - Known since ancient times
  - Virchow (1851)
    - *Synostosis → No growth perpendicular to the involved suture + compensatory growth in other directions*
  - Surgery: Lannelongue (1890)
    - *Linear craniectomy removing the prematurely closed sutures*

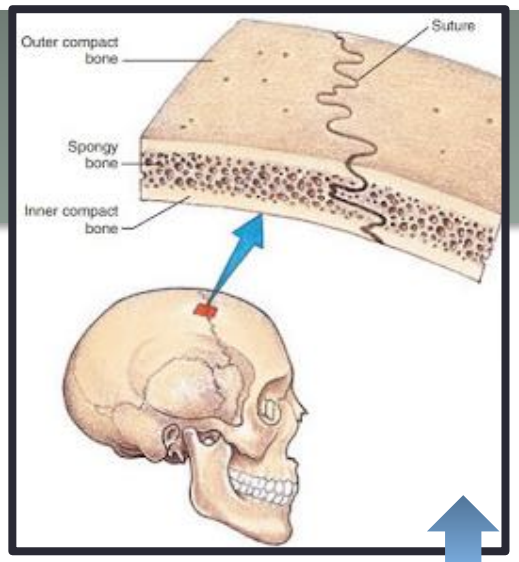


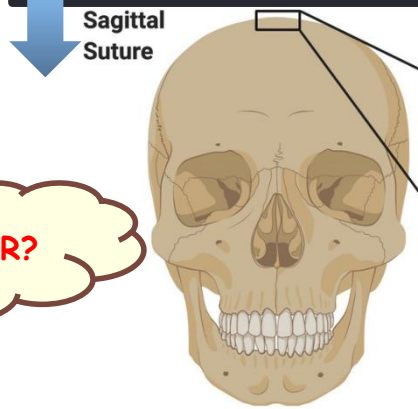
Lannelongue

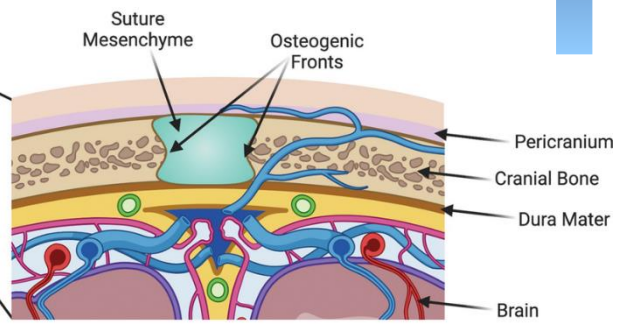
Sutures (1)

- Connective tissue layer between bones
  - Syndesmosis (cranial vault)
  - Synchondrosis (skull base)
- Closure at 6 – 12 months age
  - 12 – 13 years: fibrous tissue
  - 4th – 5th decade: ossification

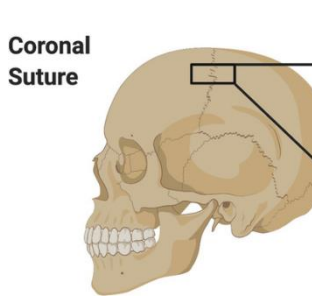


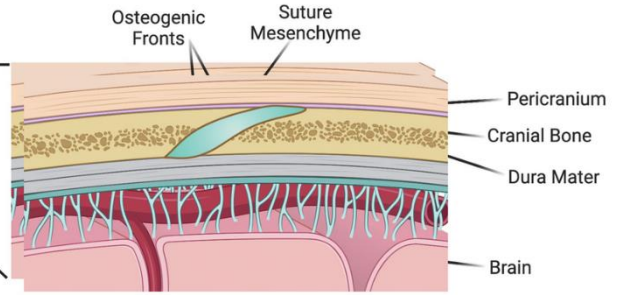






REMEMBER?

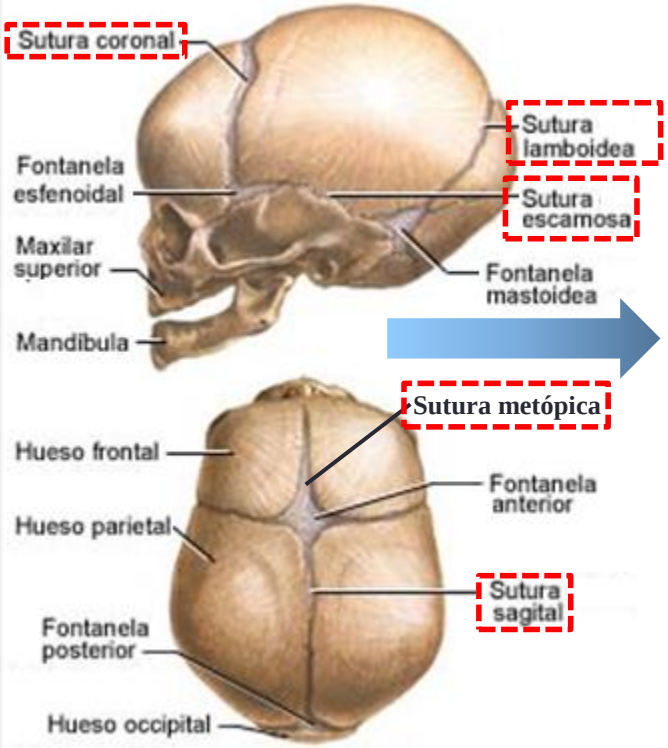




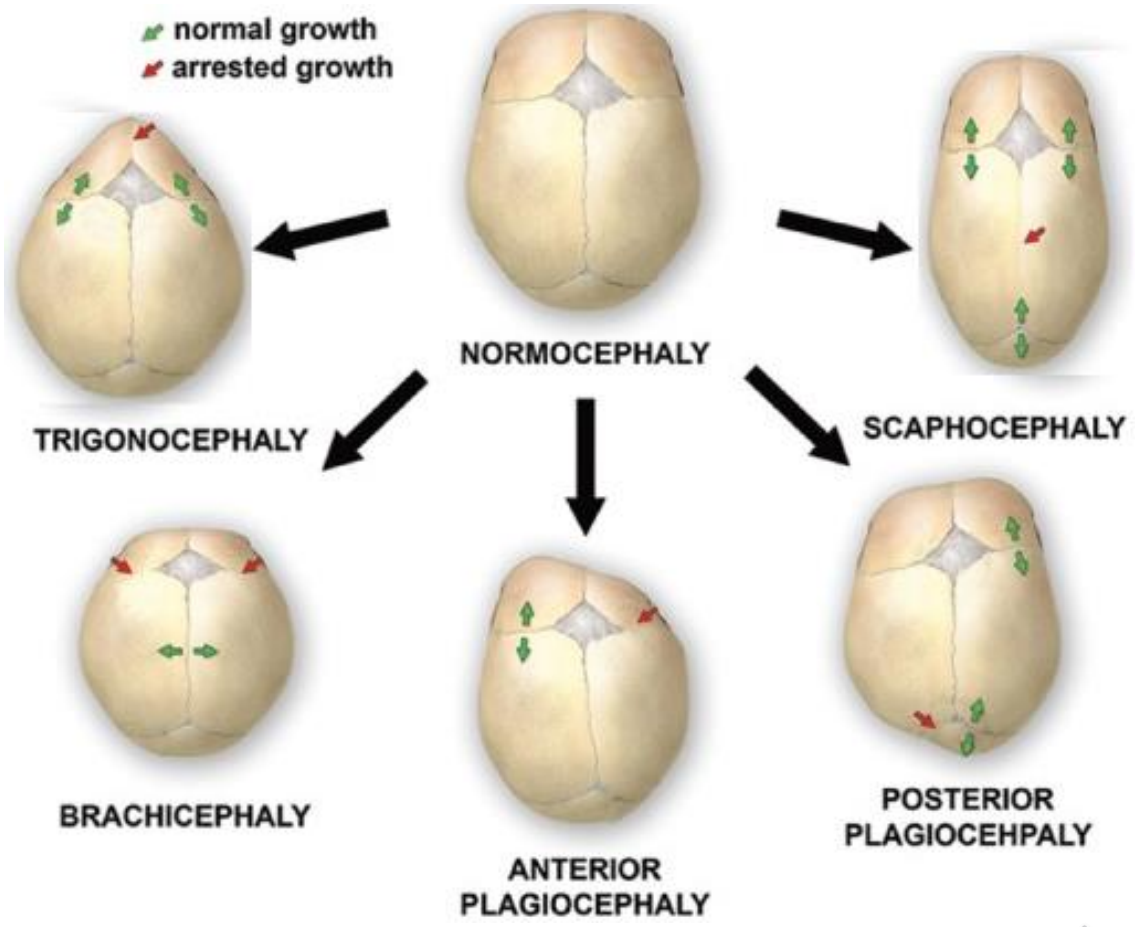
Early suture closure → Cranial malformation

Sutures and primary craniosynostoses

Side view



Top view



Craniosynostoses: diagnosis

- Cranial dysmorphia
  - First sign
  - Gives name to craniosynostosis
- **Content**: ↑ intracranial pressure
  - Intracranial hypertension
    - Headaches, exophthalmos, cranial nerve injury, psychomotor delay
  - Cranial nerve conduct stenosis: anosmia, blindness, deafness, ophthalmoplegia
- May be **part of a syndrome**
- **Associated malformations:** polydactyly, syndactyly, and anotia






Fontanelle palpation (fontanelle not always absent in craniosynostoses, and absence is not always diagnostic)
More reliable is high resolution multi-cut CT (3D reconstruction)

Polydactyly and syndactyly

- Sometimes isolated (familial?) malformation
- In syndromic cases = more severe

Isolated polydactyly

Isolated syndactyly

Syndactyly of syndromic cases

Pinna malformations / microtia / anotia

- Indicate severe syndromic case
- Uncommon

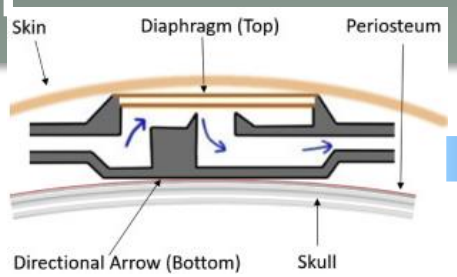
Pinna malformation

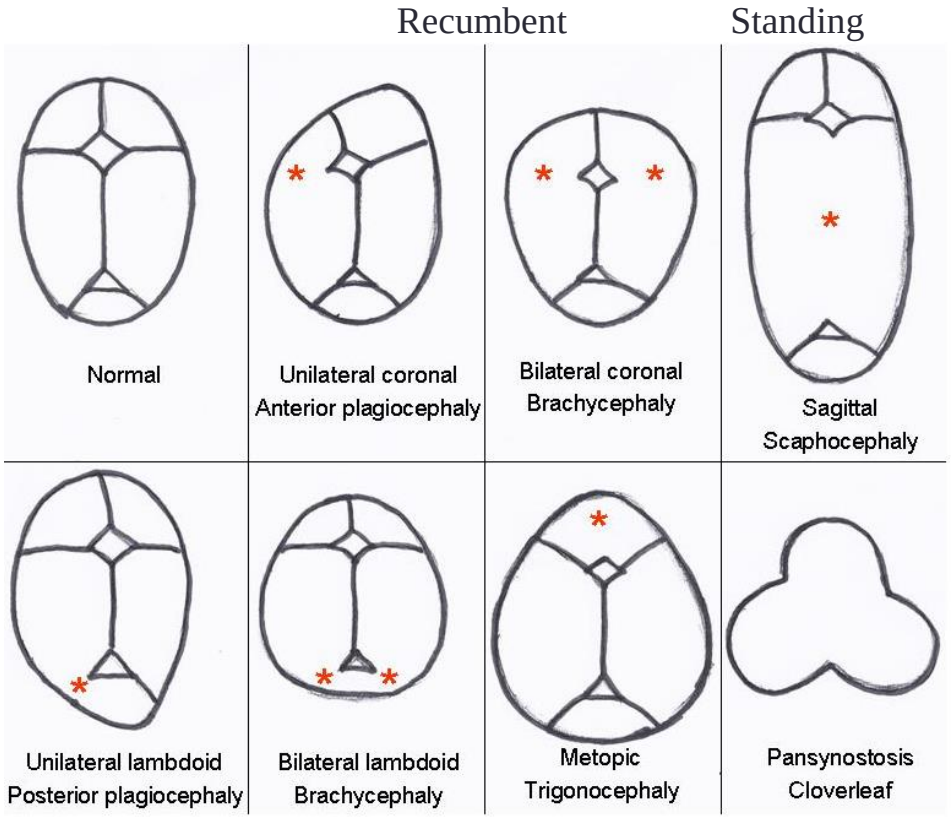
Anotia

Craniosynostoses: types

- **Primary**
  - **Simple**
    - Scaphocephaly
    - Trigonocephaly
    - Plagiocephaly
    - Oxycephaly
    - Turricephaly
  - **Complex (syndromes)**
    - Crouzon, Apert, Carpenter
    - Pfeiffer, Saethre-Chotzen, etc.
- **Secondary**
  - Metabolic disorders
    - Hyperthyroidism, polycythemia, thalassemia…
  - Meningitis
  - Osteoclastic diseases
  - Hydrocephalus shunted with low pressure valves or without anti-siphon device

Anti-siphon device →

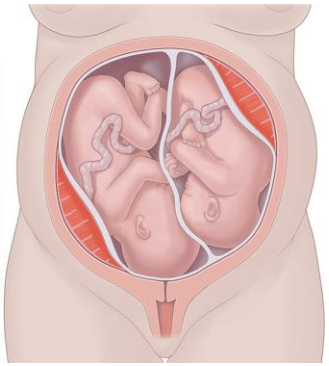


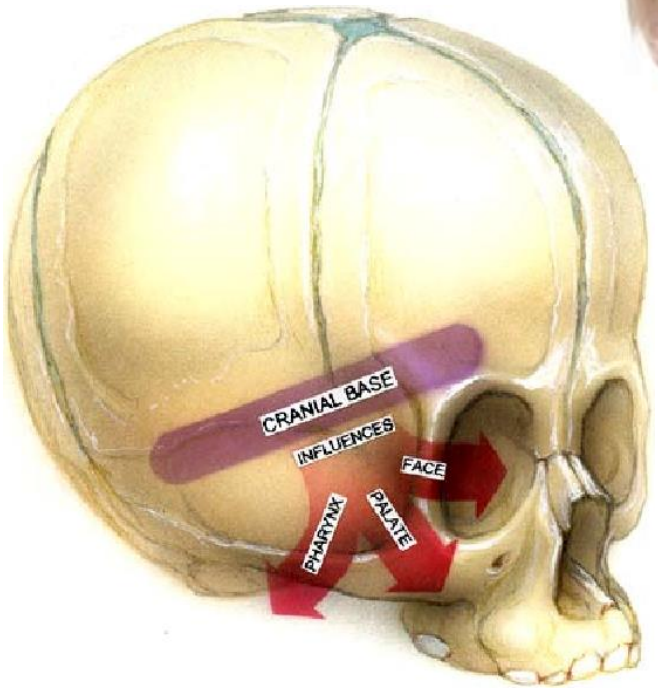


David, J.; Poswillo, D.; Simpson, D.: The craniosynostosis. Causes natural history and management. Springer-Verlag 3: 35-42, 1982

Craniosynostoses: production mechanism

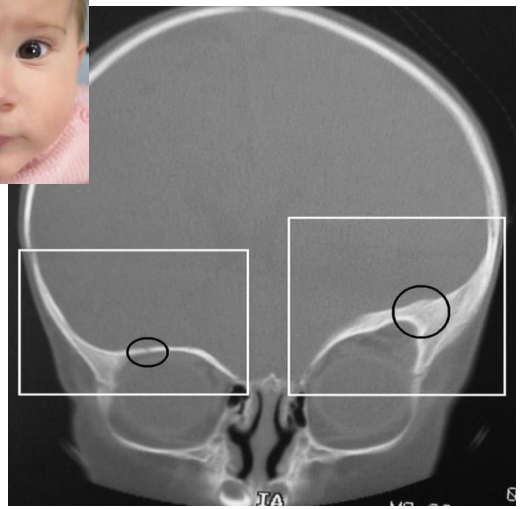
- May be due to intrauterine position or twin pregnancy
- **Not only affects cranial vault sutures**
  - Skull base suture involvement = reason why simple cranial vault synostectomies fail to correct problem
  - Facial involvement = deformities in eye / nasal pyramid / face
    - Need repair BEFORE unaesthetic deformity happens



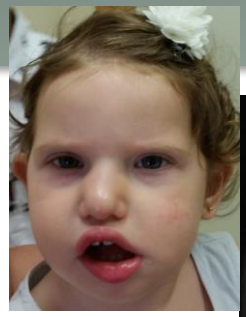


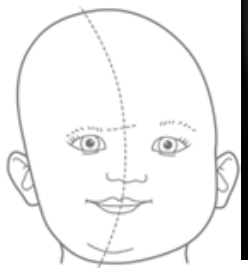
Craniosynostoses: facial consequences

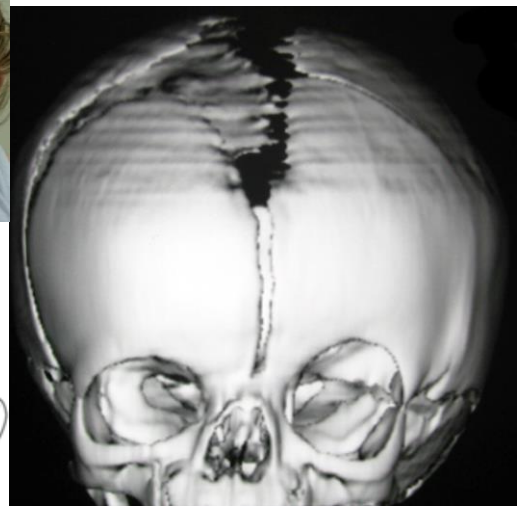
- Abnormal position orbits
- Nasal pyramid deviation
- Upper maxillary hypoplasia



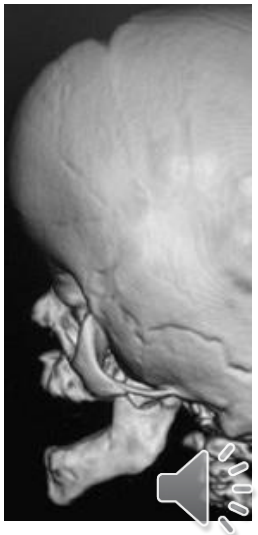
Orbit asymmetry





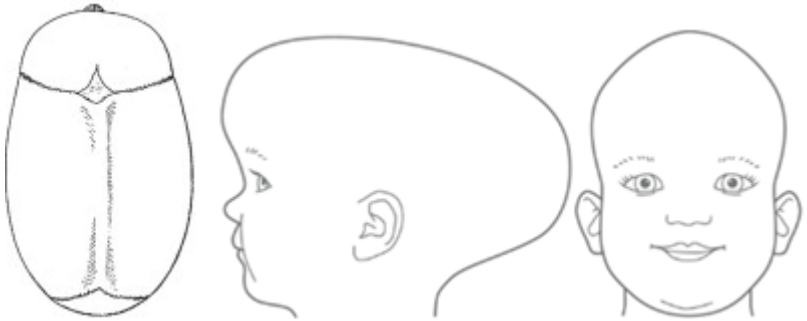


Nasal pyramid deviation

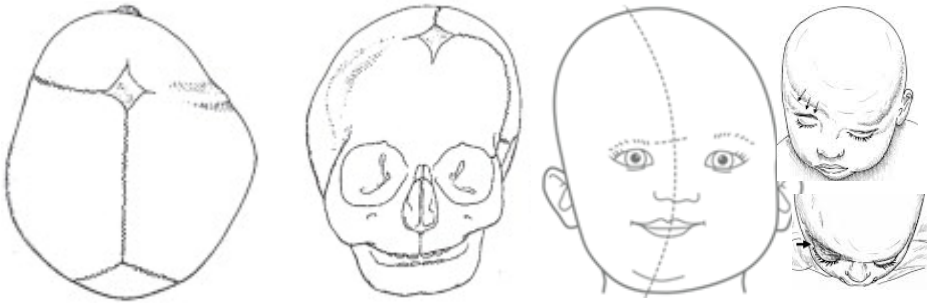


Upper maxillary hypoplasia

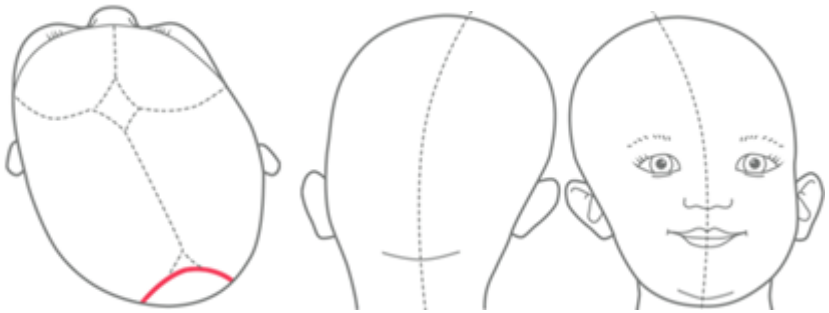
Simple primary craniosynostoses



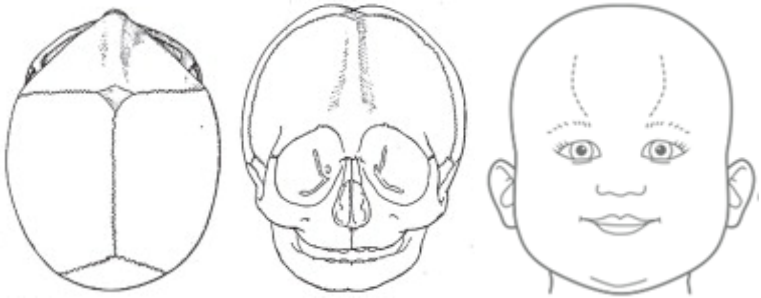
Scaphocephaly



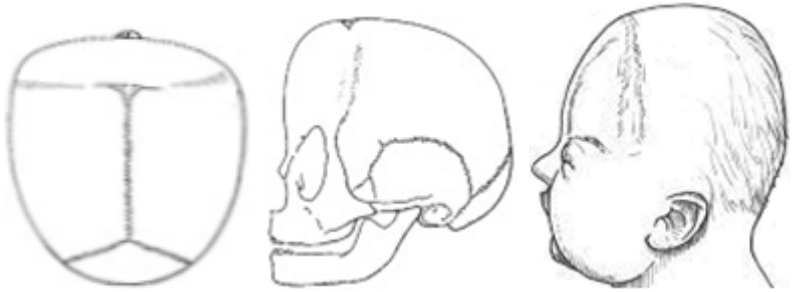
Frontal plagiocephaly



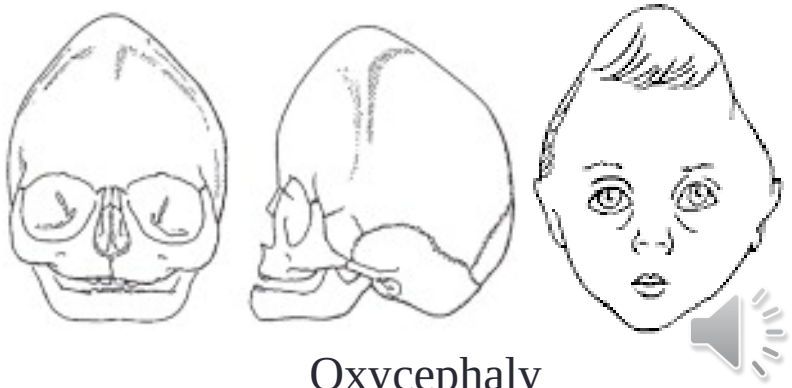
Occipital plagiocephaly



Trigonocephaly



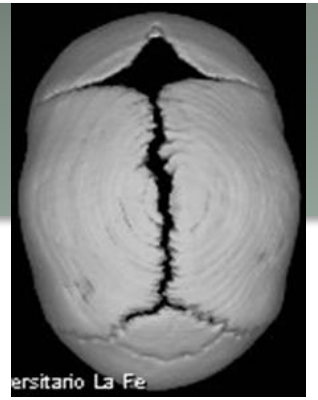
Brachycephaly

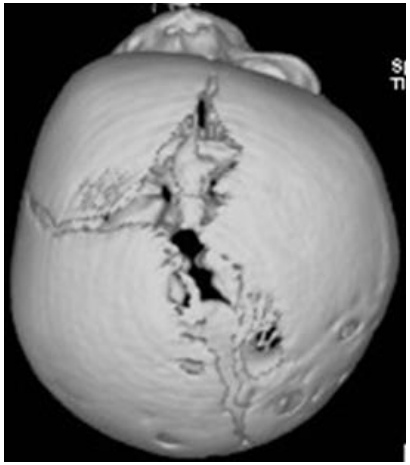


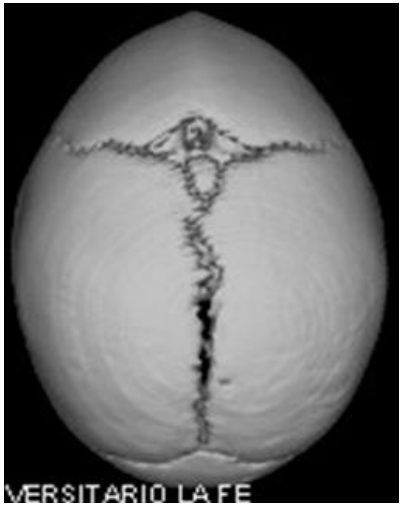
Oxycephaly

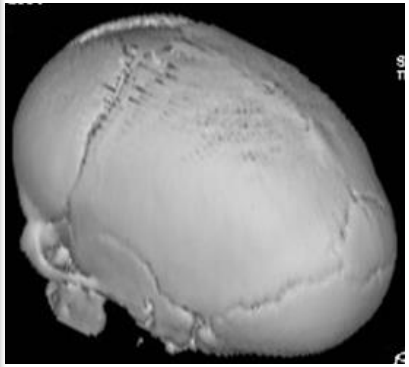
Craniosynostoses: diagnosis

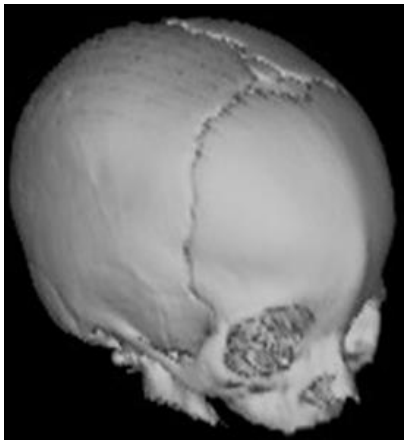
- Skull-face deformity, orbit-ear position
- CT-scan with 3D reconstruction





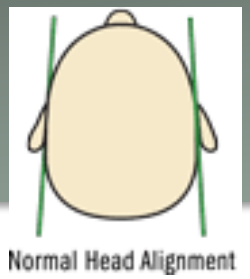


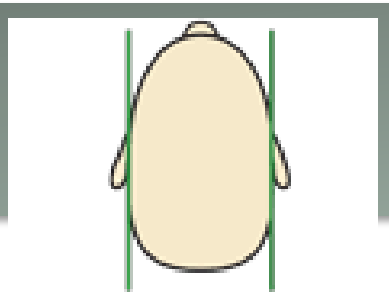


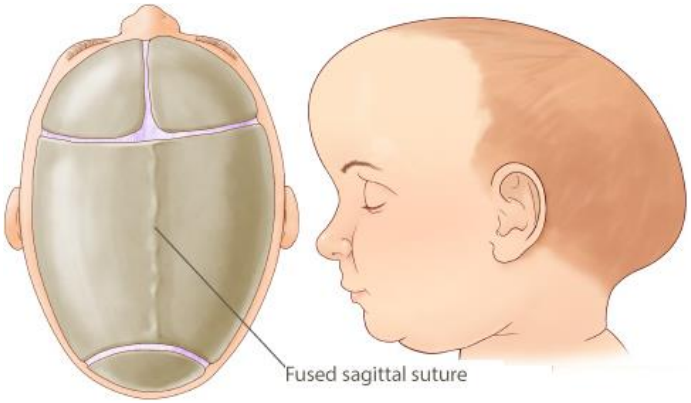


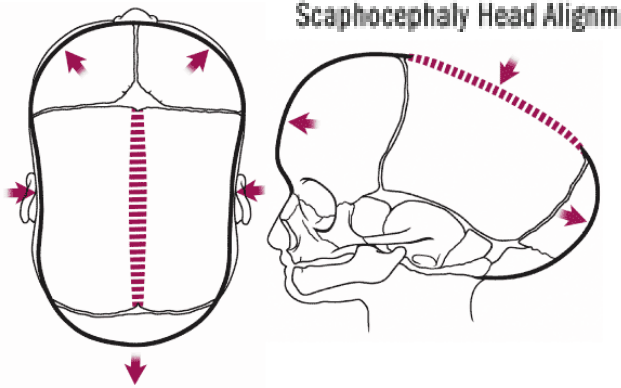
Simple primary CS: scaphocephaly (1)

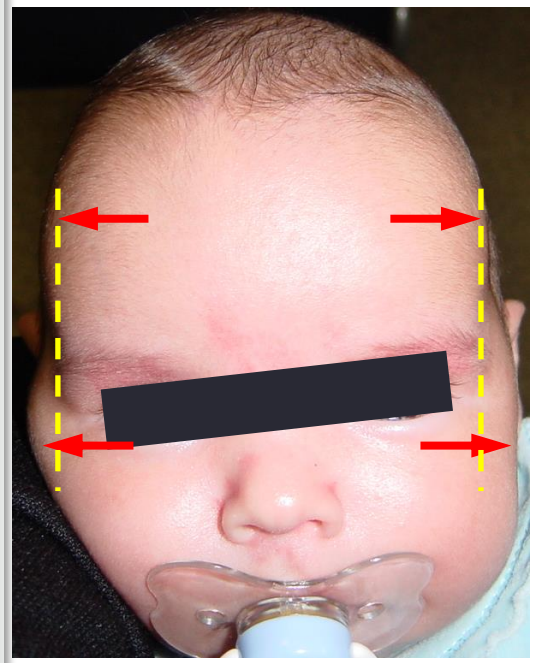
- ⇑ frequent CS
  - 1/4,000 births
- Sagittal suture closure → skull grows in AP direction

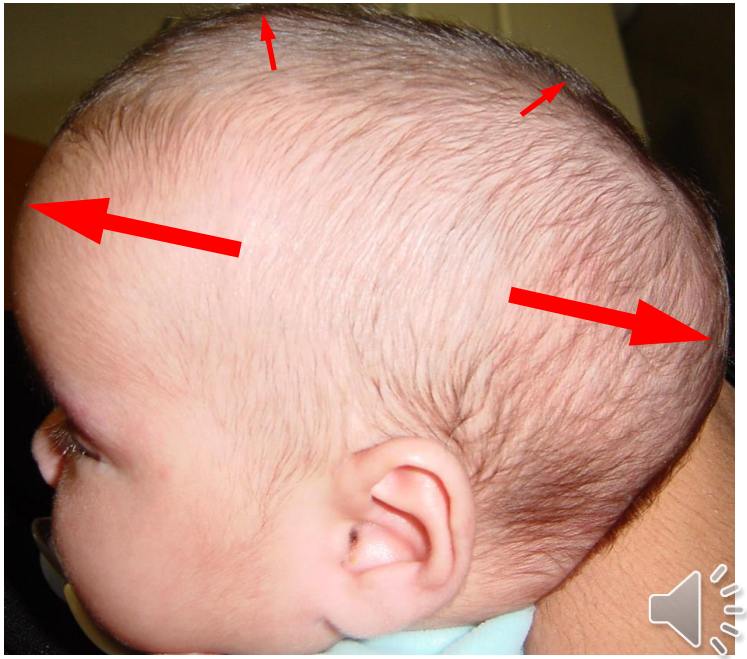




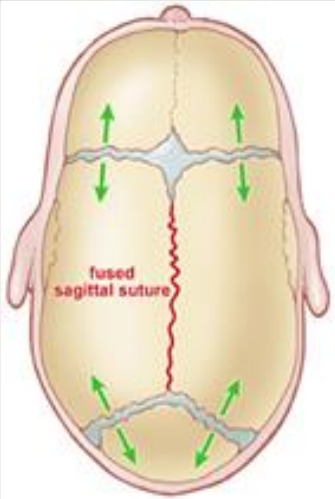


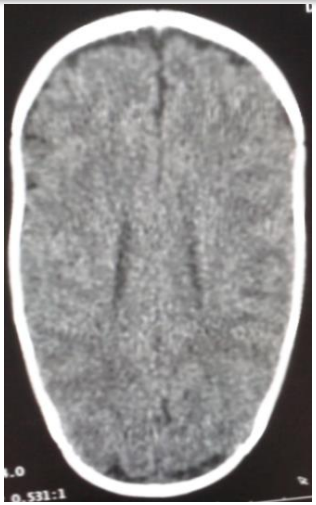


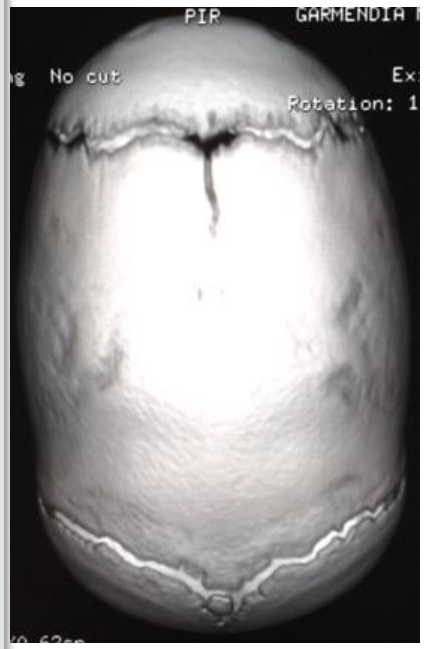


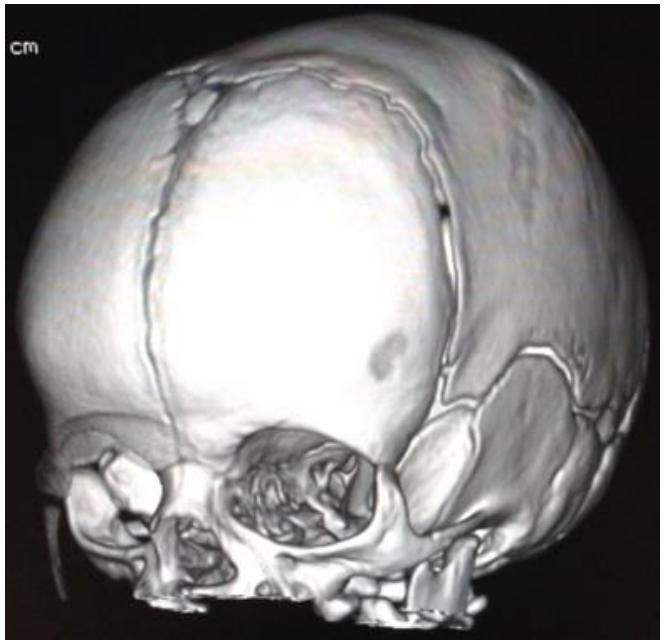


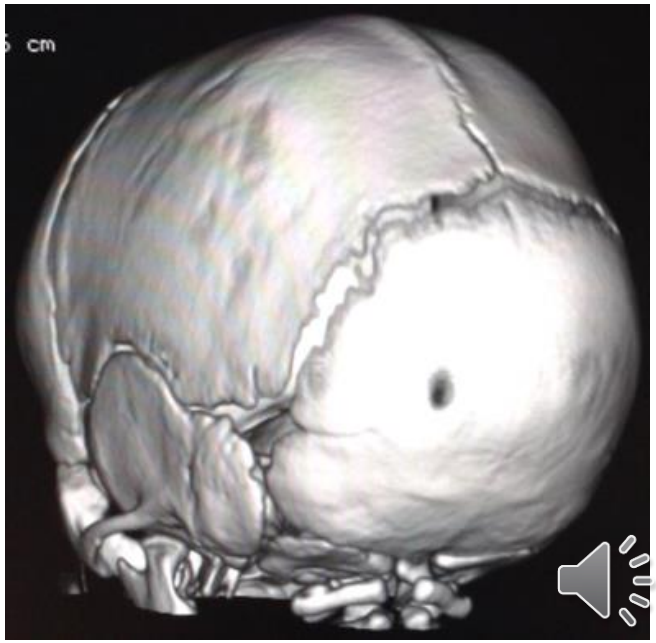
Scaphocephaly: CT-scan





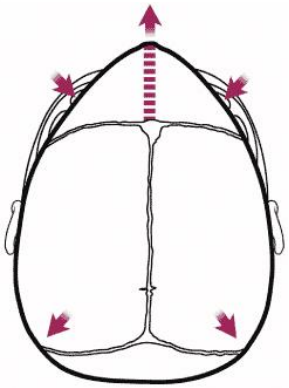


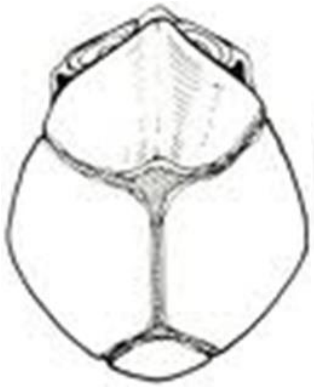


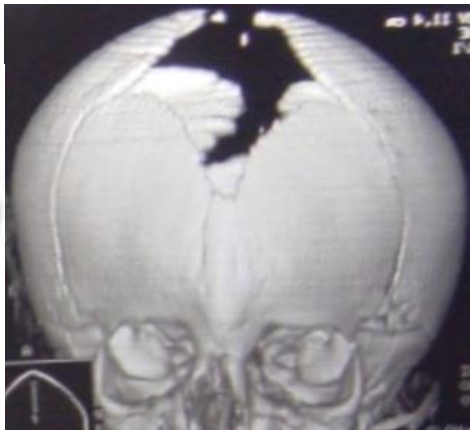


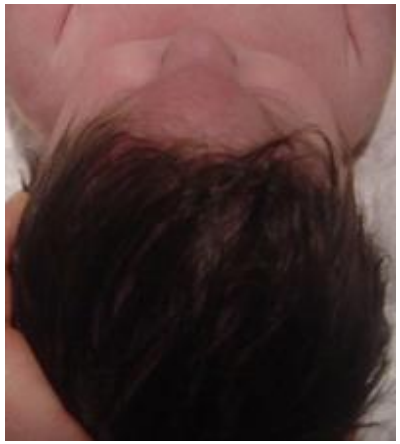
Simple primary CS: trigonocephaly

- Premature closure metopic suture → narrow triangular forehead, palpable midfrontal ridge, hypotelorism

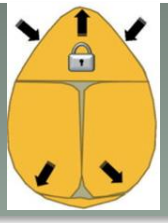




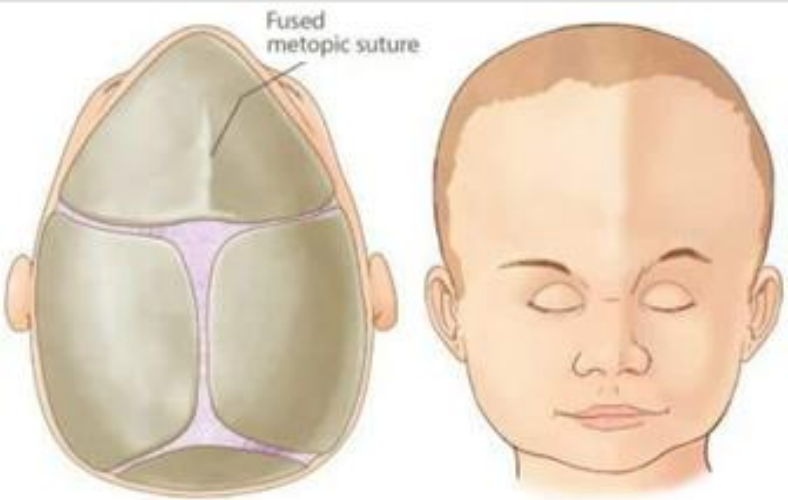


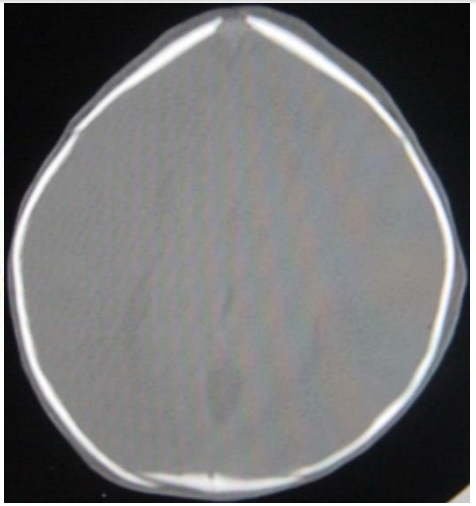


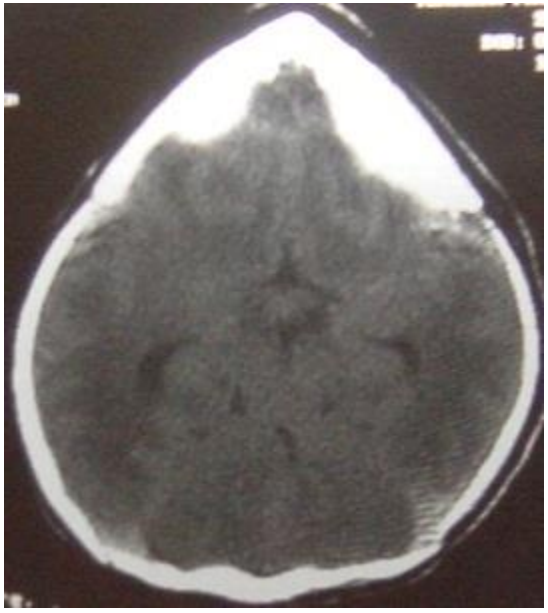
Trigonocephaly: CT-scan

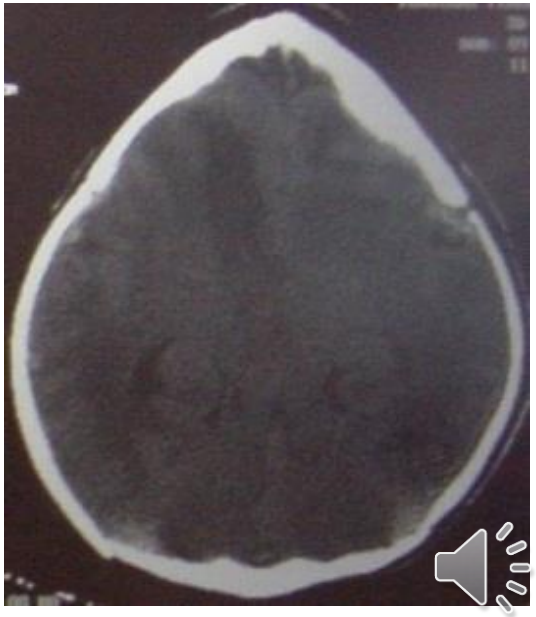


- Depicts metopic suture fusion + cranial deformity + hypotelorism = helps in surgical planning



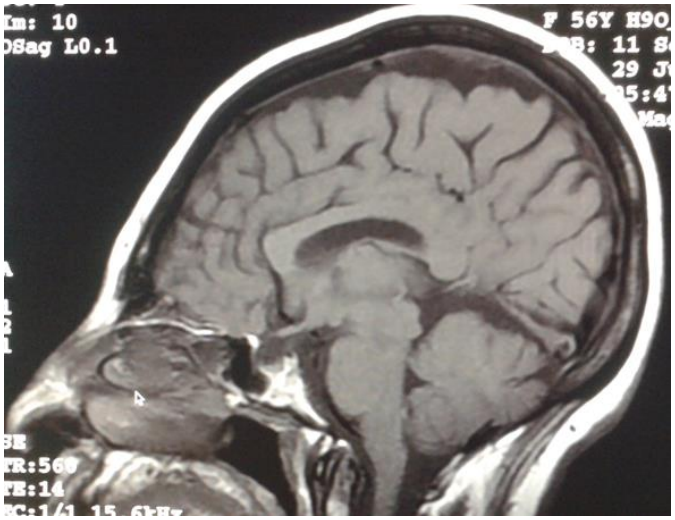


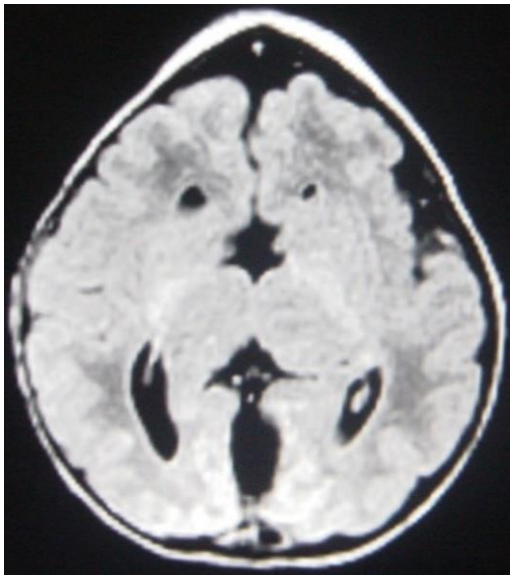


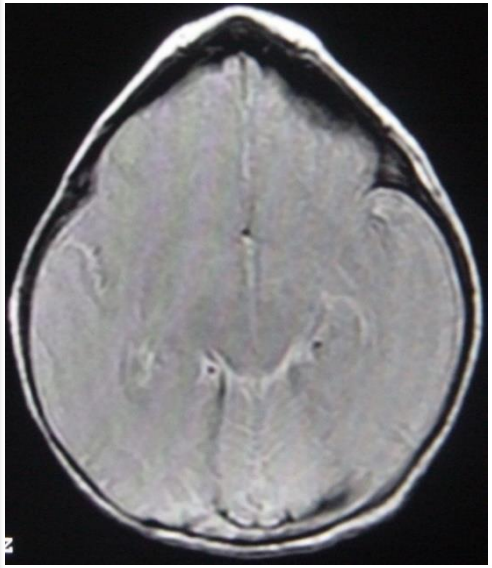


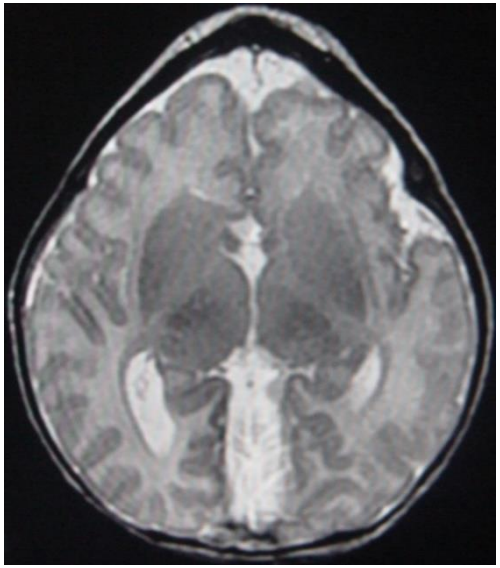
Trigonocephaly: MRI

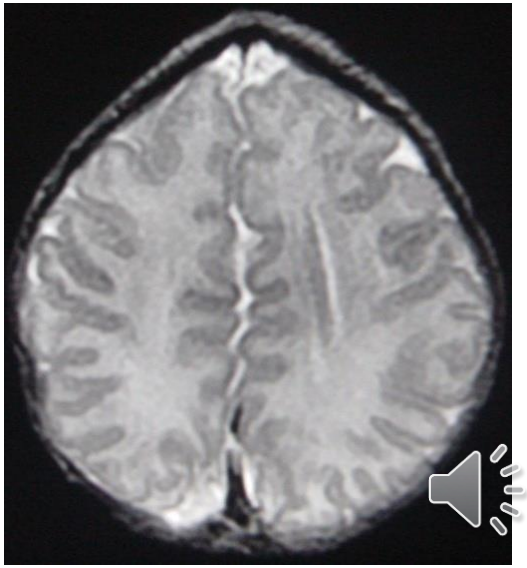
- Rules out brain malformations





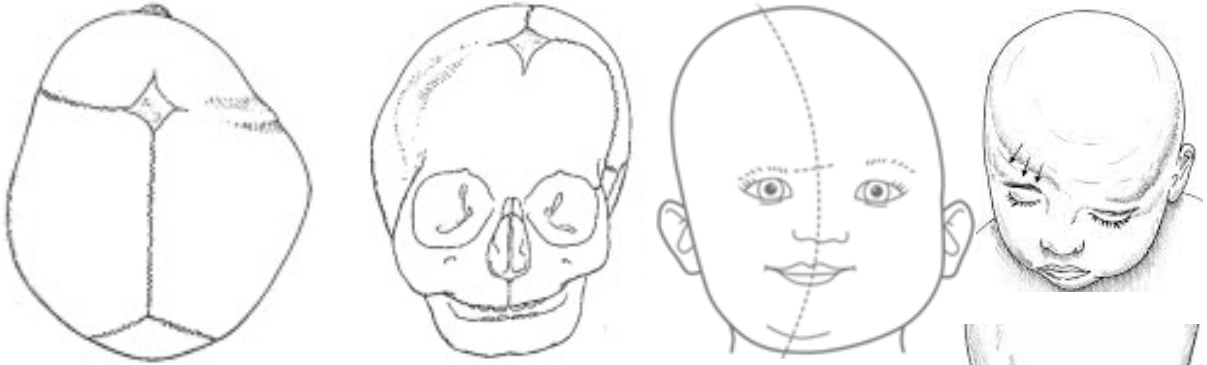






Simple primary CS: plagiocephaly

- **Frontal** = premature coronal hemi-suture closure → oblique skull shape, frontal bulging on healthy side, facial asymmetry



Frontal plagiocephaly

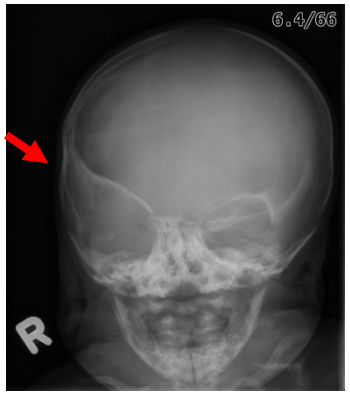
- ***Occipital*** = premature *lambdoid hemi-suture → oblique skull shape, occipital bulging*

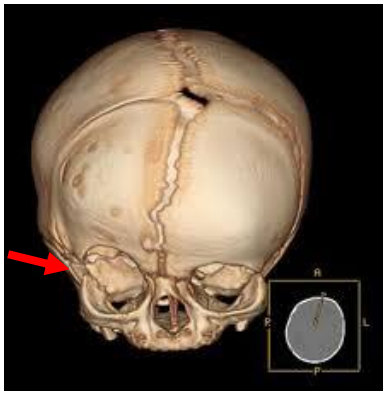
Occipital plagiocephaly

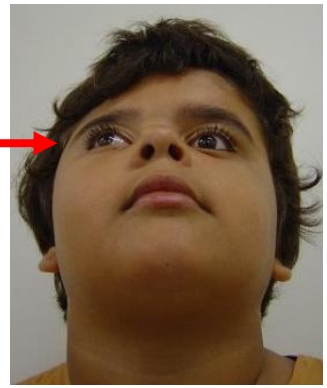
Simple CS: frontal plagiocephaly

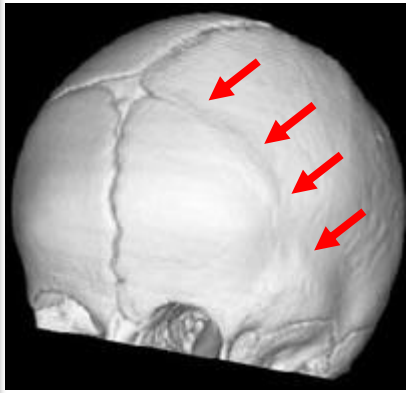
- Premature unilateral coronal suture closure → oblique skull shape, frontal bulging healthy side, facial asymmetry, Harlequin eye

Harlequin





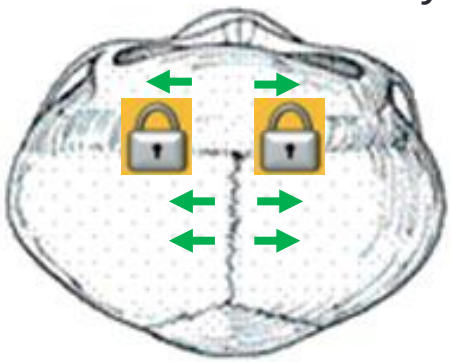




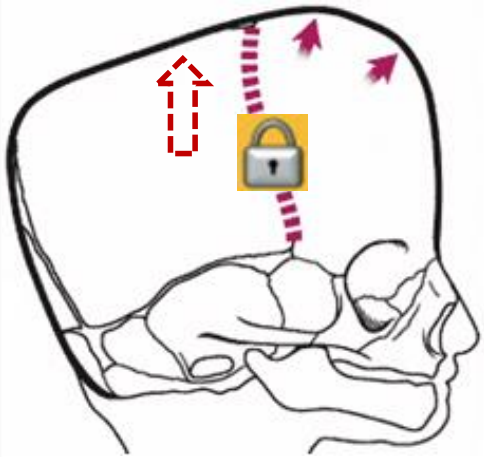
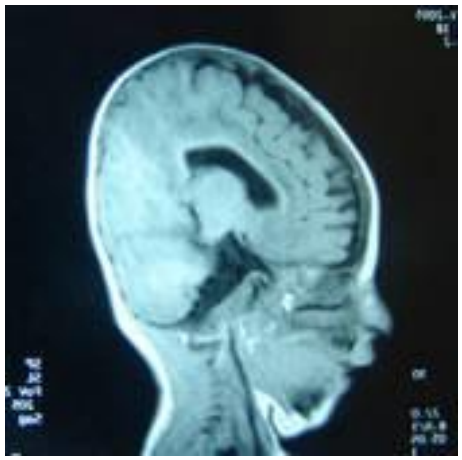
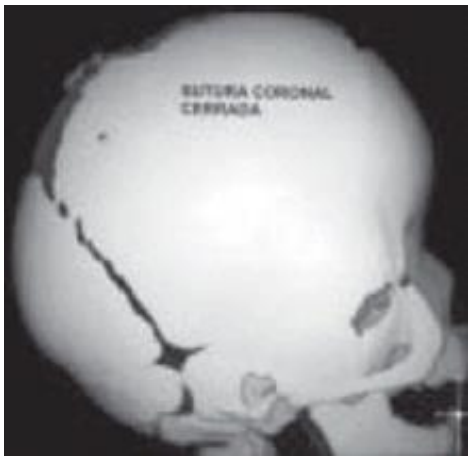
Brachycephaly

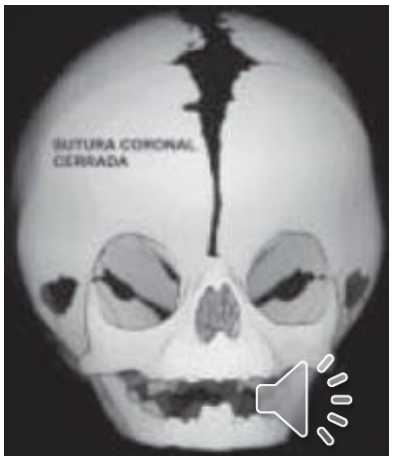
- Premature bilateral coronal suture closure (skull does not grow antero-posteriorly) → grows upwards
- Associated with syndromes



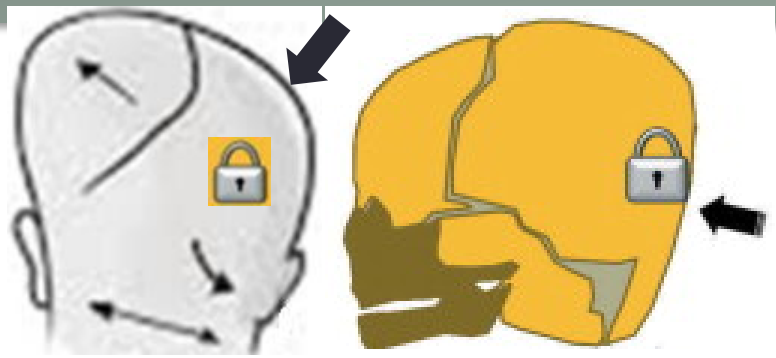


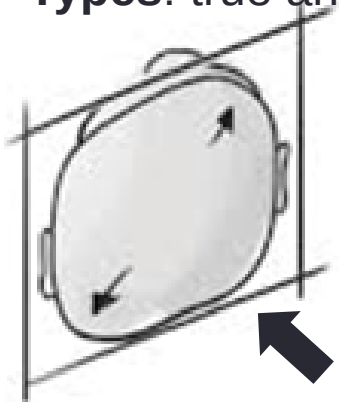




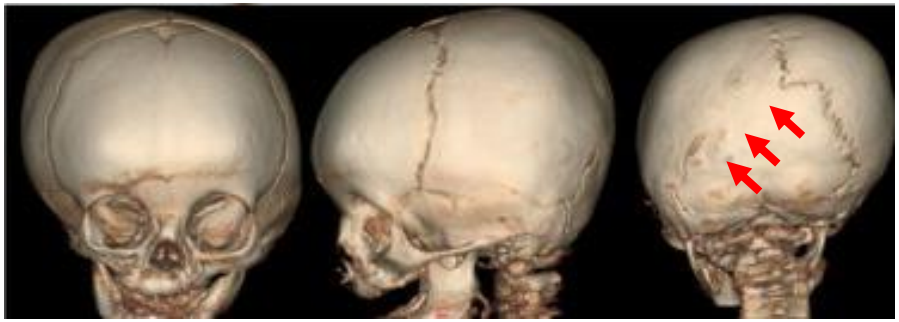


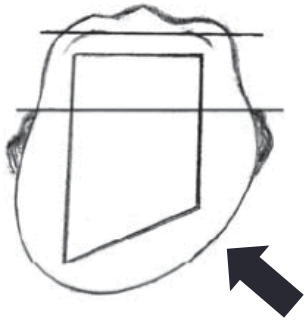
Occipital plagiocephaly

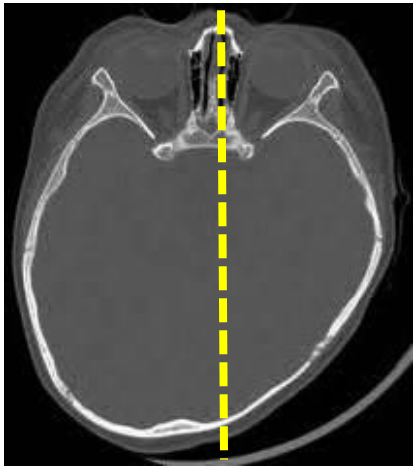
- Premature unilateral lambdoid suture closure → oblique skull shape, contralateral occipital bulging
- **Types**: true and positional



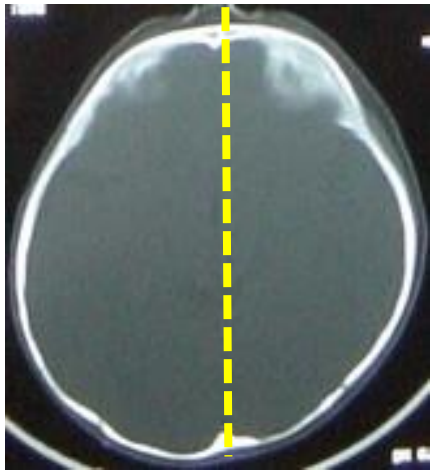


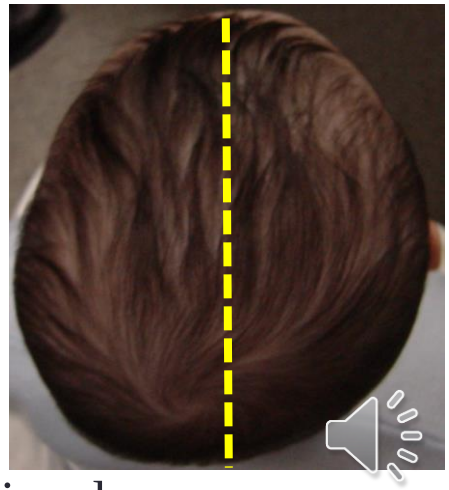






True

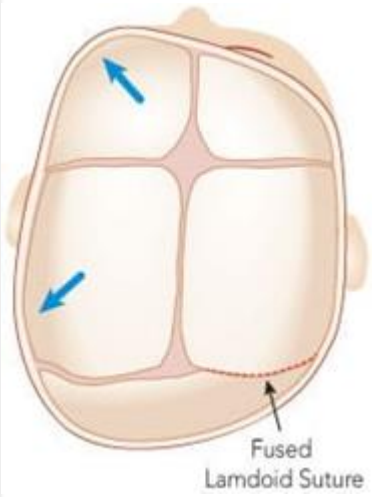


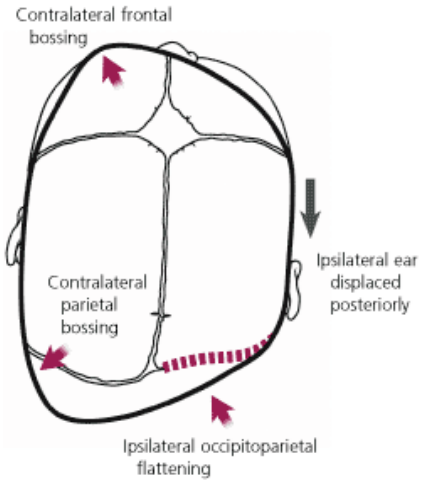


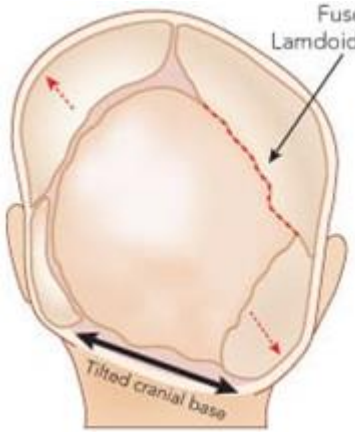
Positional

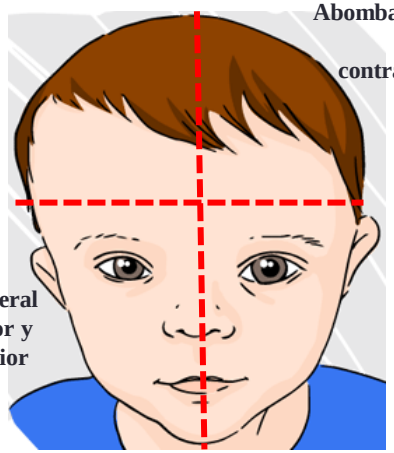
True versus positional occipital plagiocephaly (2)

- Importance: true requires surgery, positional requires molding helmet

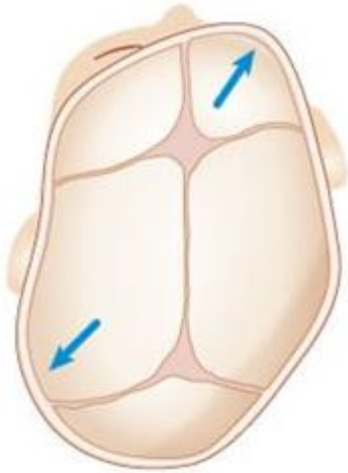


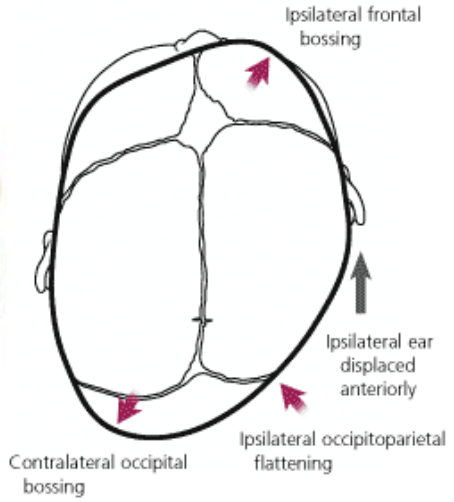


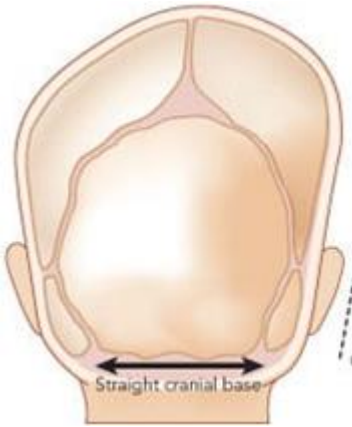


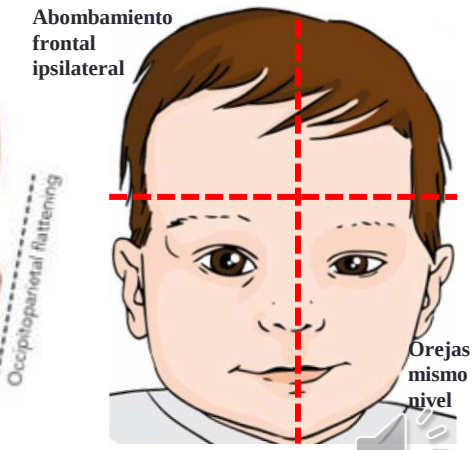


True





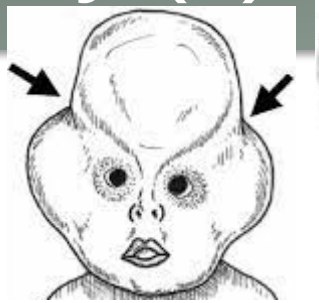




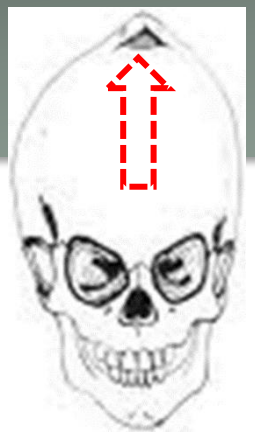
Positional

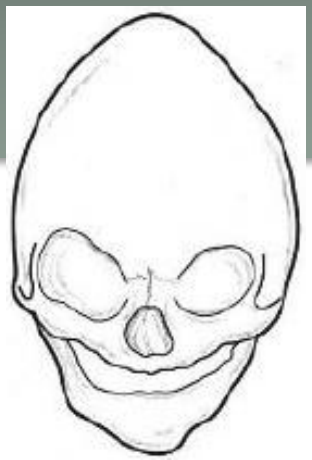
Oxycephaly (1)

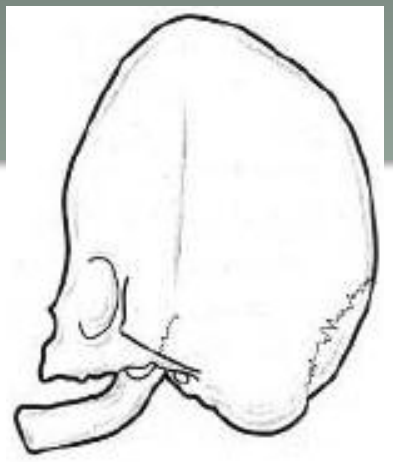
- Closure of ‘all’ sutures
  - At least coronal & sagittal
- Maybe
  - Harmonic = all sutures close at once → Microcephaly
  - Disharmonic

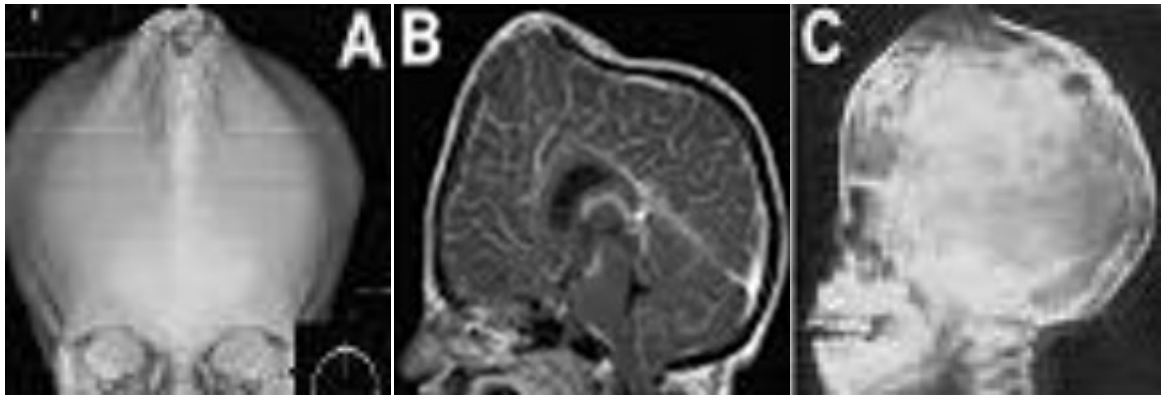


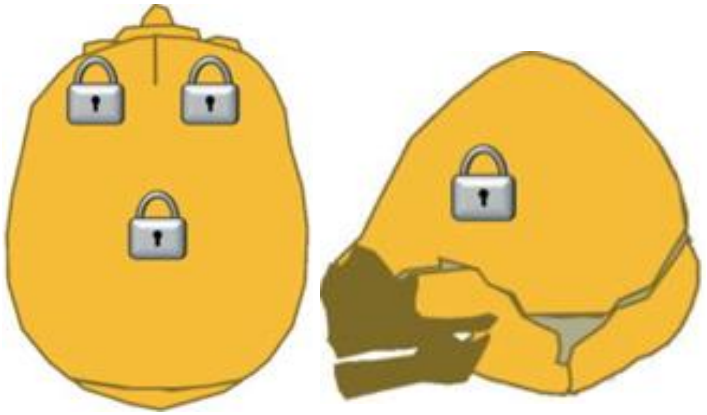
Cloverleaf skull





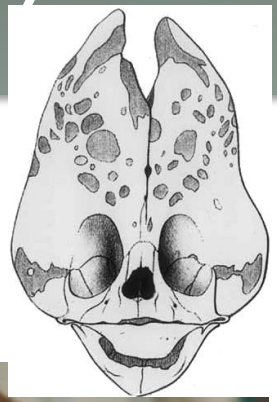


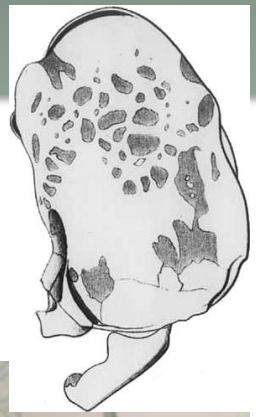




Oxycephaly (2)

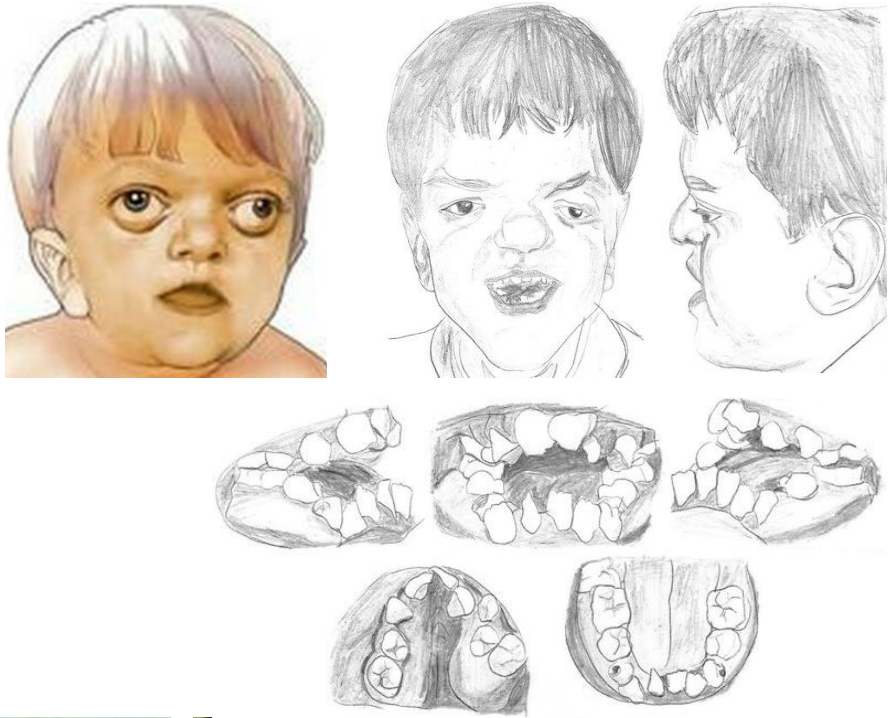
- In extreme cases = intracranial hypertension = blindness
- Brain growth impairment = severe psychomotor retardation





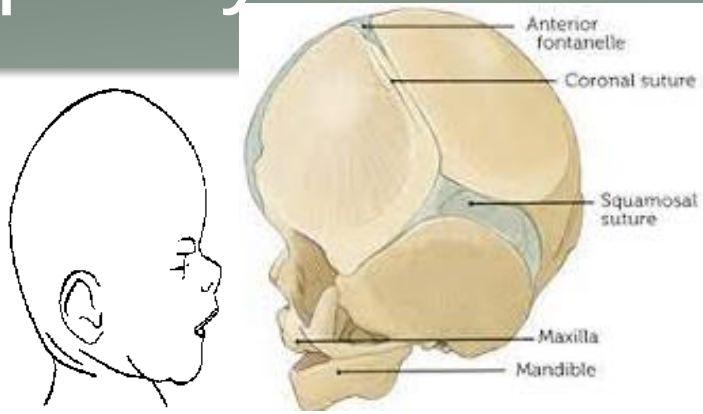
Complex primary CS: Crouzon syndrome

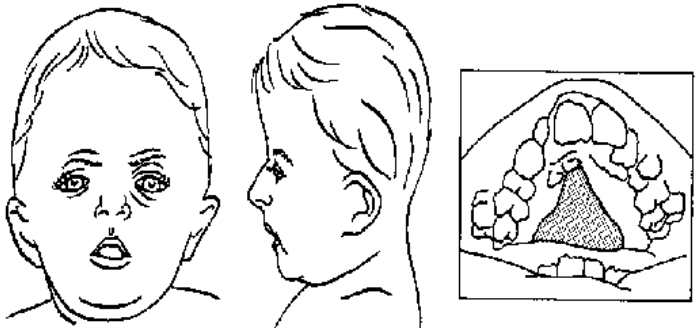
- Craniofacial synostosis
- Hereditary: dominant or spontaneous (elderly father)
- Brachycephaly
- Limited center face growth: **hypoplastic maxilla**, short and protruding upper lip, **peaked nose**, hypertelorism, **exophthalmos** and divergent strabismus



Complex primary CS: Apert syndrome

- Acrocephalosyndactyly type I
- Possible autosomal dominant
- Acrocephaly, **syndactyly (hands and feet)**
- First gill arch defects: choanal atresia, megalo-cornea, strabismus, orbital hypoplasia and other malformations



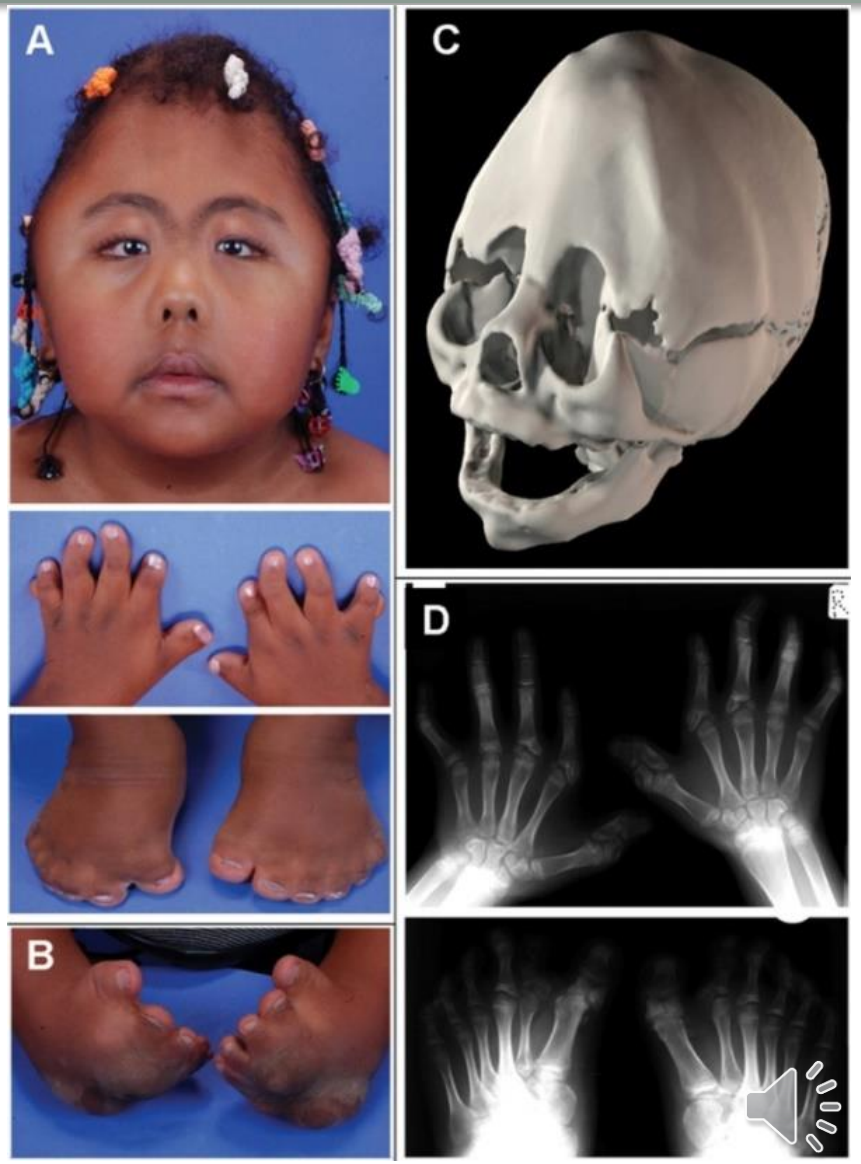


Turribraquicefalia con huesos frontales altos, inclinados y planos, nariz pequeña y contraída, estrabismo, proptosis ocular, inclinación antimongoloide de las hendiduras palpebrales, aplanamiento de la parte media de la cara; paladar ojival, con maloclusión dentaria

Diversos grados de sindactilia de los dedos de las manos y de los pies

Complex primary CS: Carpenter syndrome

- Acro cephalopolysyndactyly type II
- Possible autosomal recessive
- Chromosome 6
- Acrocephaly, brachysyndactyly hands, preaxial polydactyly and **foot syndactyly**
- Sometimes obesity, hypogenitalism, and mental retardation



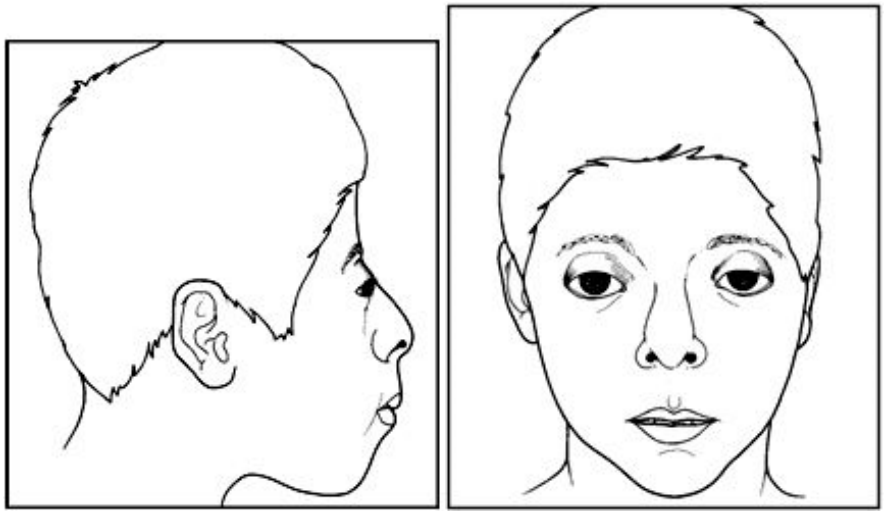
Complex primary CS: Pfeiffer syndrome

- Acrocephalosyndactyly type V
- Autosomal dominant (mutation)
- Varies from brachycephaly to **cloverleaf skull**
- Partial **syndactyly**, broad thumbs, large toes, normal intellect

Type I Type II Type III

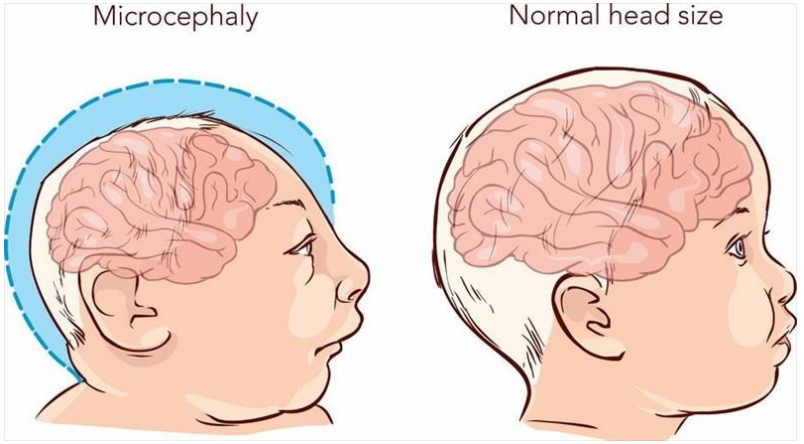
Complex primary CS: Saethre-Chotzen syndrome

- Acrocephalosyndactyly type III
- Gene mutation chromosome 7
- Hypertelorism, **palpebral ptosis**, mild mental retardation

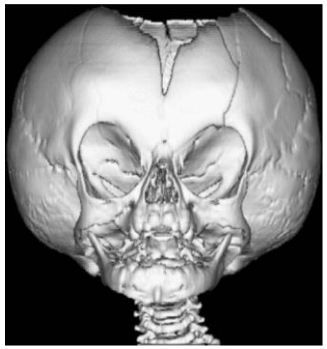


Craniosynostoses: differential diagnosis

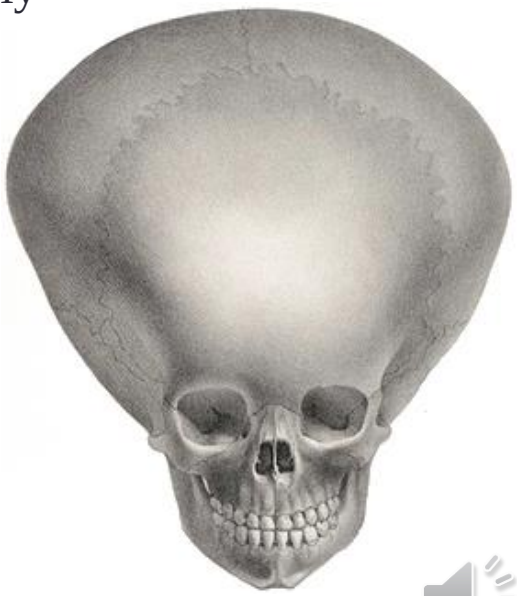
- **Microcephaly vera**
  - *Brain is normal but has not grown*
  - *Small cranial perimeter, sutures present*
- **Postural defects**
  - *Infants with psychomotor retardation, rickets, osteogenesis imperfecta*
- **Hydrocephalus**
  - *Fontanelle present and bulging*
- Others
  - *Arachnoid cysts, hemorrhages, tumors, intra-uterine infections…*



Microcephaly



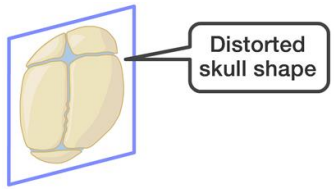
Osteogenesis imperfecta

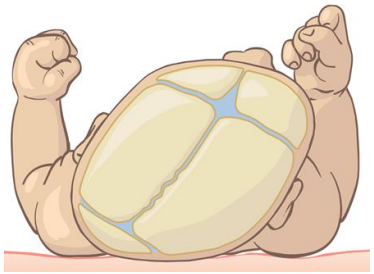


Hydrocephalus

Craniosynostoses: treatment

- **Conservative**
  - Cranial orthosis (helmet)
- **Surgical (3 – 6 months)**
  - Indications
    - Intracranial hypertension
    - Optic nerve atrophy
    - Avoid psychomotor retardation
    - Aesthetics
      - Allow correct brain growth
      - Avoid/correct facial deformities
  - Surgical techniques
    - Closed suture opening (synostectomy)
    - Multiple cranial fragmentation (morcellation)
    - Cranial decompressions or flaps
    - Craniofacial advancements

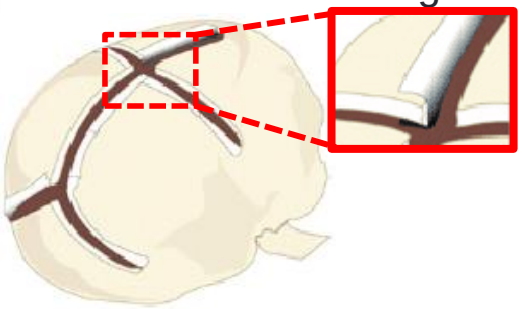




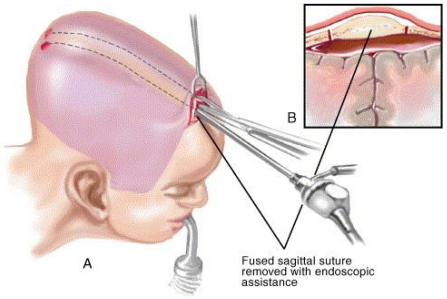
Positional occipital plagiocephaly

Craniosynostoses: surgical treatment (1)

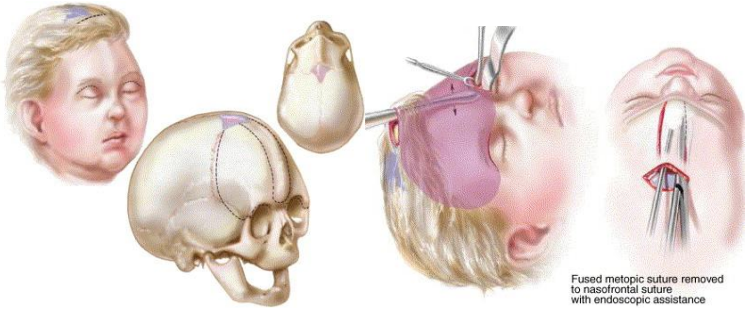
- Surgical procedure indicated according to patient age, degree of cranial / facial deformity, simple craniosynostosis, or associated with a syndrome
  - Skull growth 60% 1st year, 80% 2nd year, and 90% 3rd year
    - Corrections between 3-6 months = better results with less aggressiveness
      - Possibility of endoscopic synostectomy = minimal surgical aggression, but need for molding skull helmet for 1-2 years



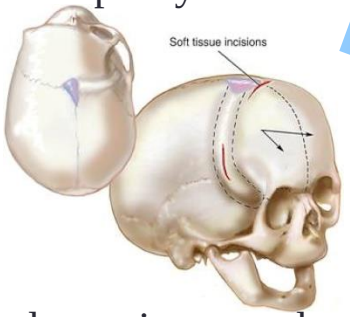
Simple synostectomy



Endoscopic sagittal synostectomy



Endoscopic metopic synostectomy

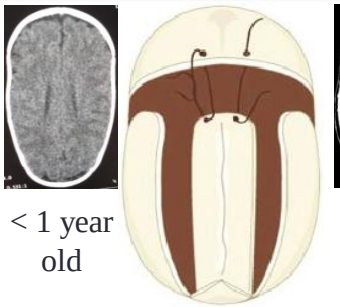


Endoscopic coronal synostectomy

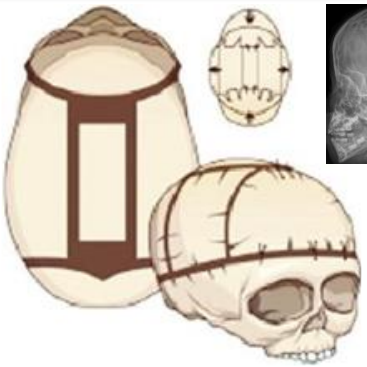
Molding helmet

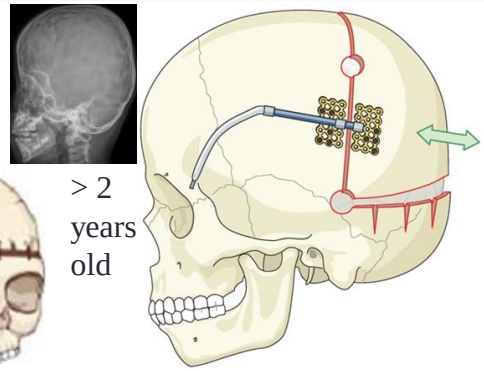
Craniosynostoses: surgical treatment (2)

- **Cranial deformity**
  - Craniectomy
  - Cranial remodeling
  - Skull expansion
- **Facial deformity**
  - Hyper/hypotelorism correction
  - Craniofacial advancement

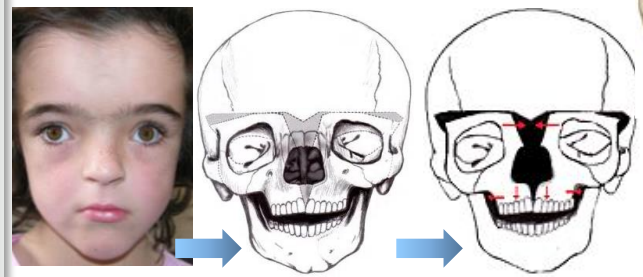


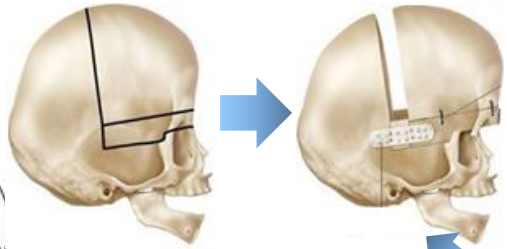
Craniectomy


Cranial remodeling

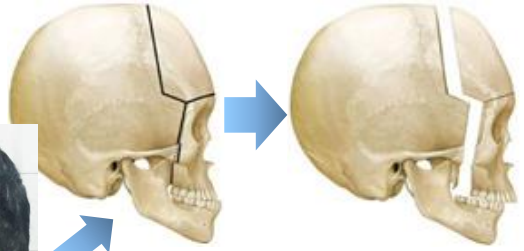


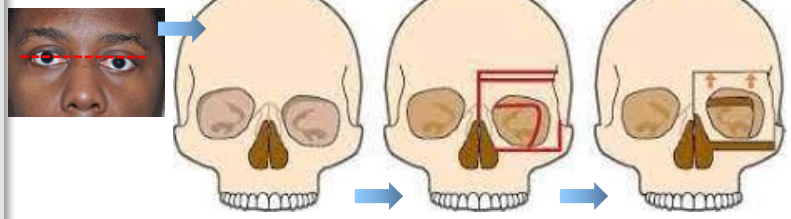
Skull expansion

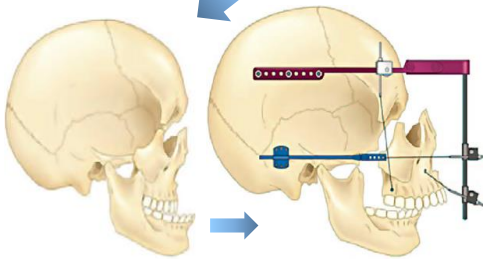

Hypertelorism correction

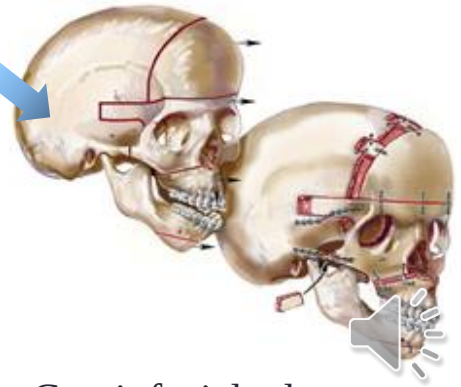

Frontal advancement


CS síndromes

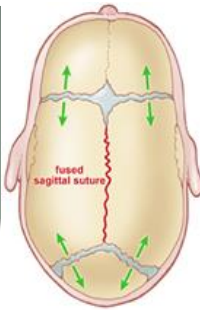

Craniofacial advancement


Orbital situation correction

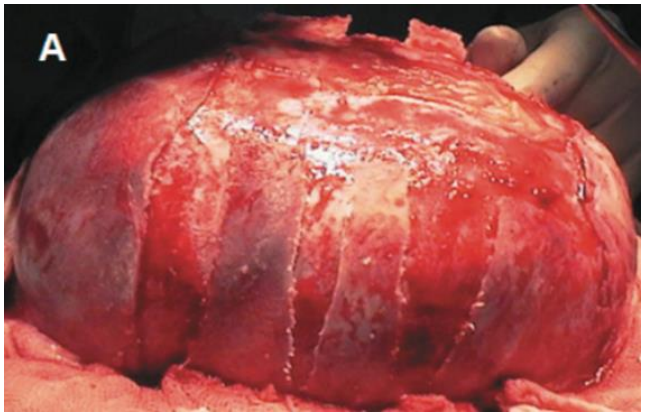

Facial advancement

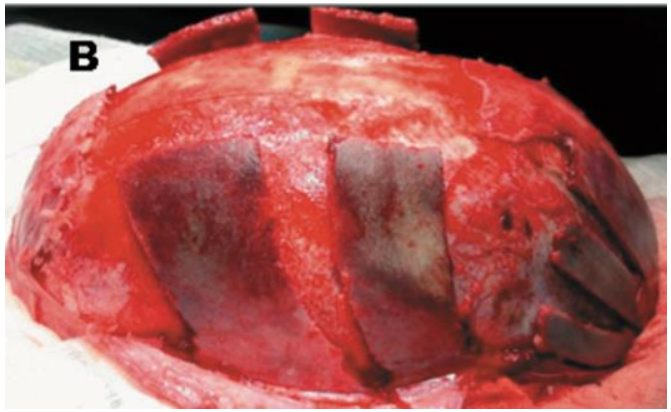

Craniofacial advancement

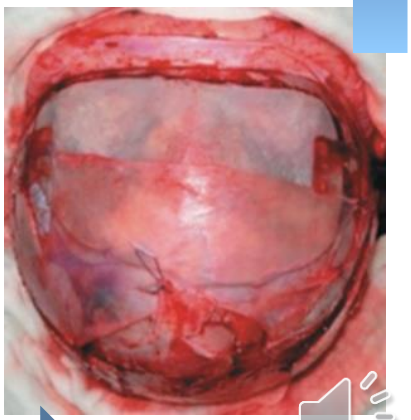
Craniosynostoses surgical treatment (3)



- Avoid delays = ↓ aggressiveness

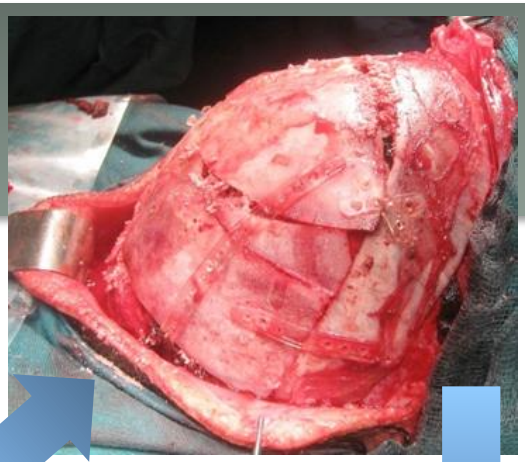






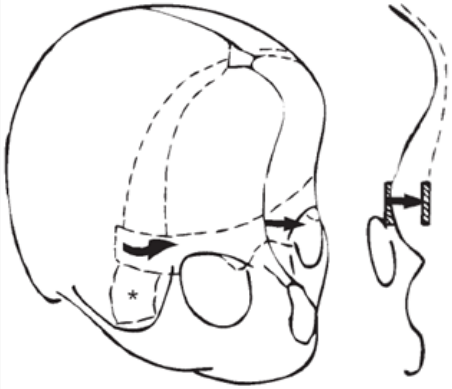
True occipital plagiocephaly

- Few cases

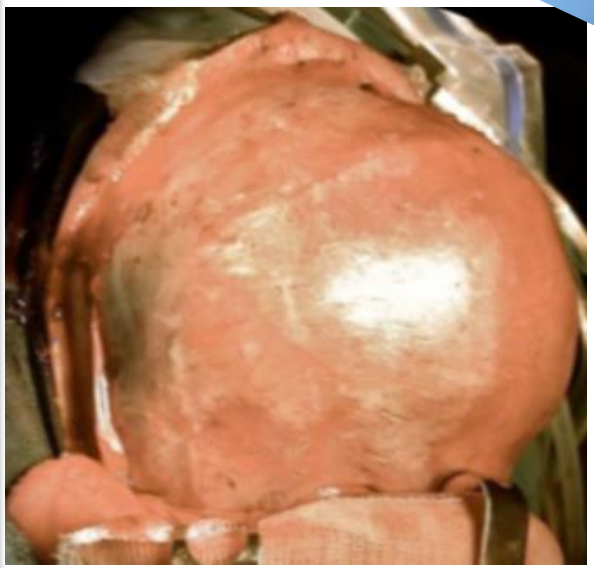
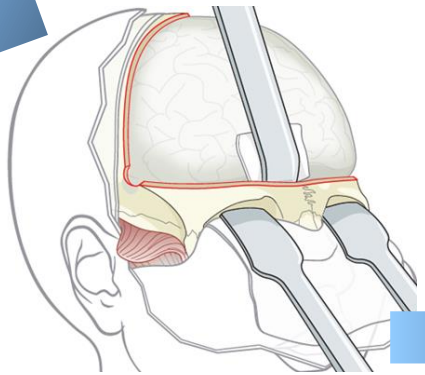
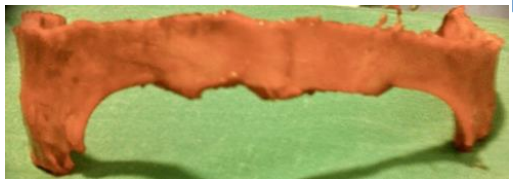
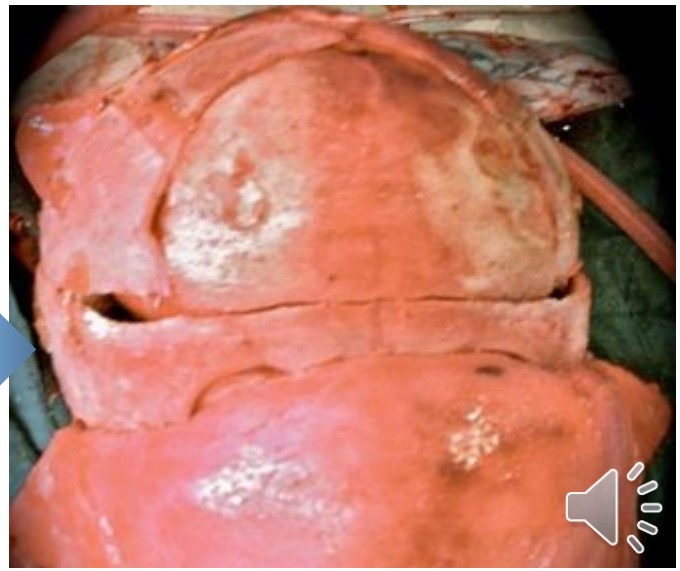


Craniofacial advancement

- Typical in craniosynostosis associated with syndromes

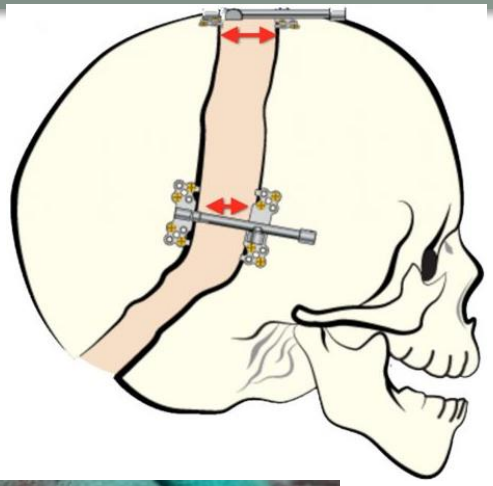


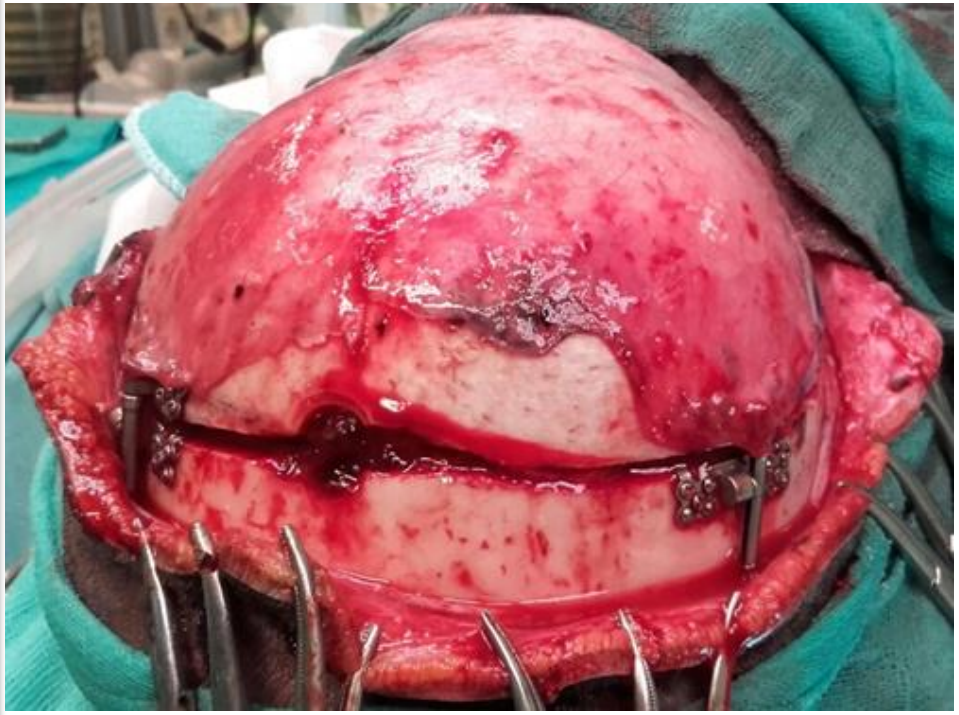


Craniosynostoses: percutaneous cranial distraction

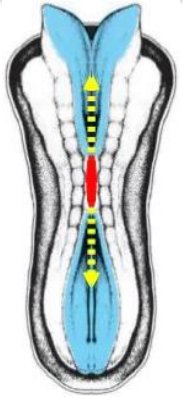
- Progressive cranial distraction
- Risk of infection

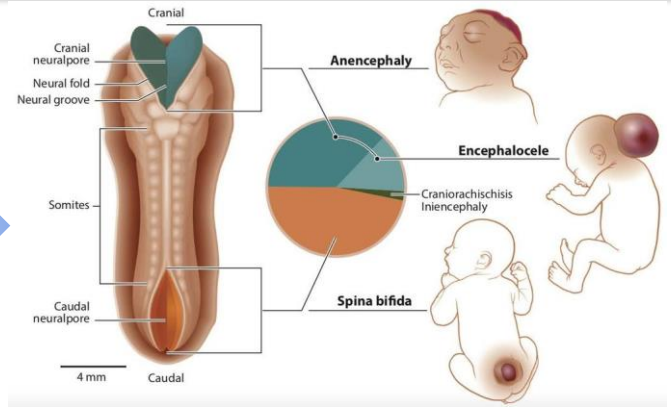


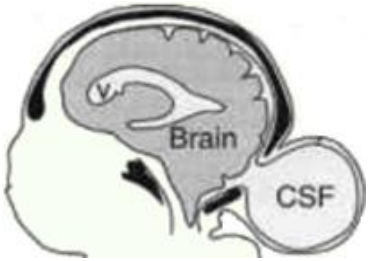


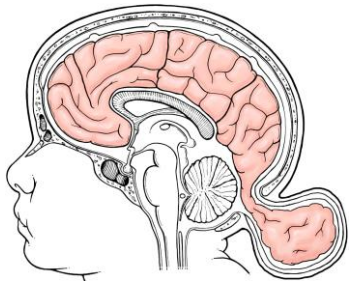
2. CRANIOSCHISIS

- *'Cranioschisis' = 'split skull'*
- **Cranial neuropore lack of closure**
  - Skull ± neural tube defects
- **Severity**
  - Cranial meningocele
    - Meninges only
  - Meningoencephalocele
    - Meninges & brain
  - Anencephaly
    - Meninges & brain, open to amniotic fluid
    - Non-closure of the neural tube
    - Brain formation lacking, but with basal ganglia & brainstem preserved









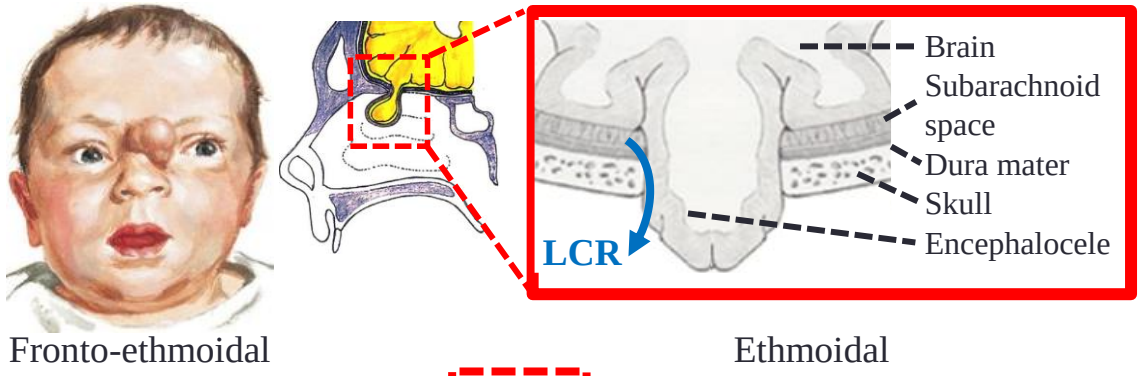
Meningocele

Meningoencephalocele

Anencephaly

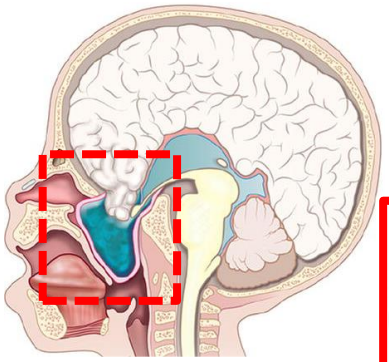
Meningocele & meningoencephalocele

- **Location**: anywhere in the skull
  - ↑frequent occipital or frontal-basal areas
- **Size**: variable
- Coverage and content: variable
- **Prognosis**: depends on
  - Size, location, brain structures, presence of microcephaly, association with syndromes
  - **Mortality**
    - ***Meningocele 11%***
      - *No brain tissue → good result after surgical repair*
    - ***Encephalocele > 70%***

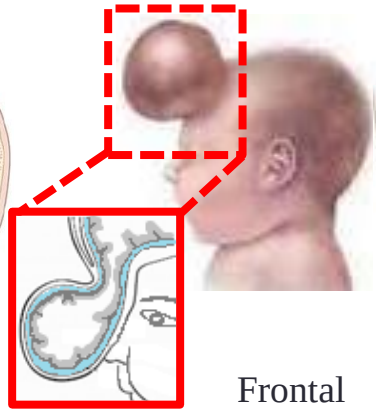


Fronto-ethmoidal

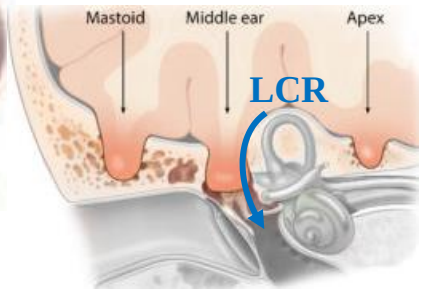
Ethmoidal



Sphenoidal

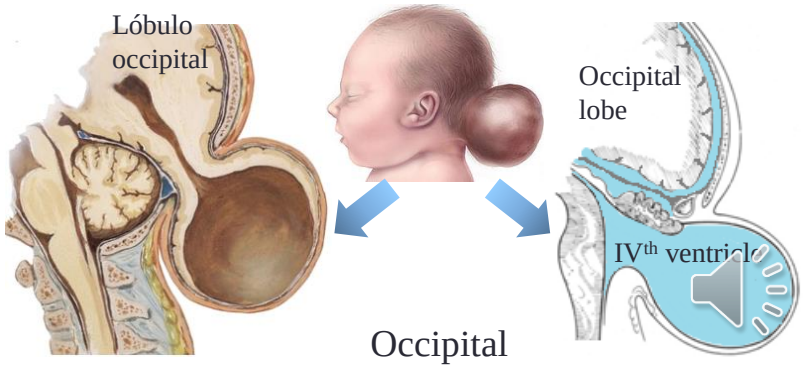


Frontal



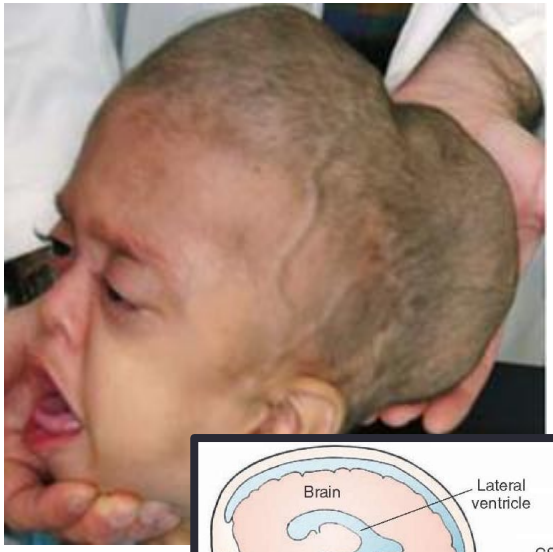
Middle fossa

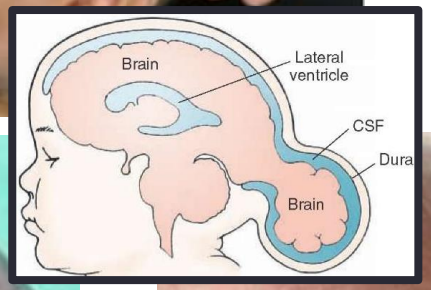
Parietal



Occipital

Meningocele & meningoencephalocele

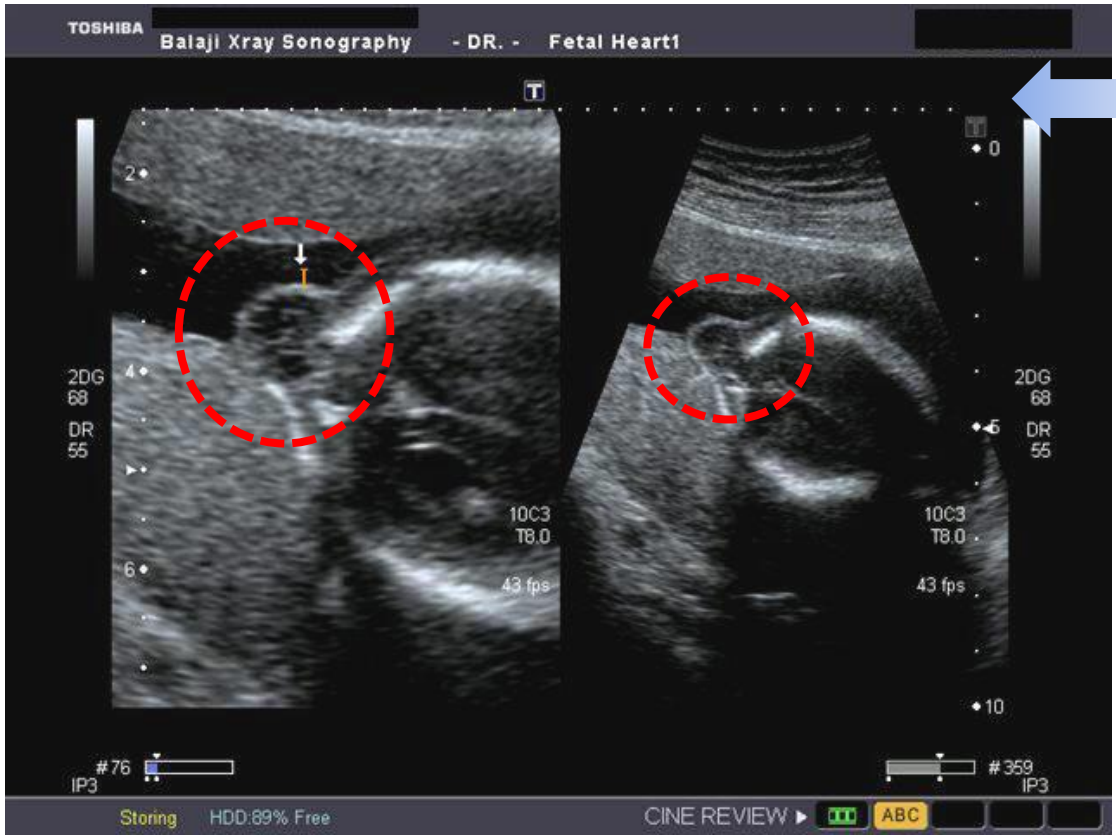


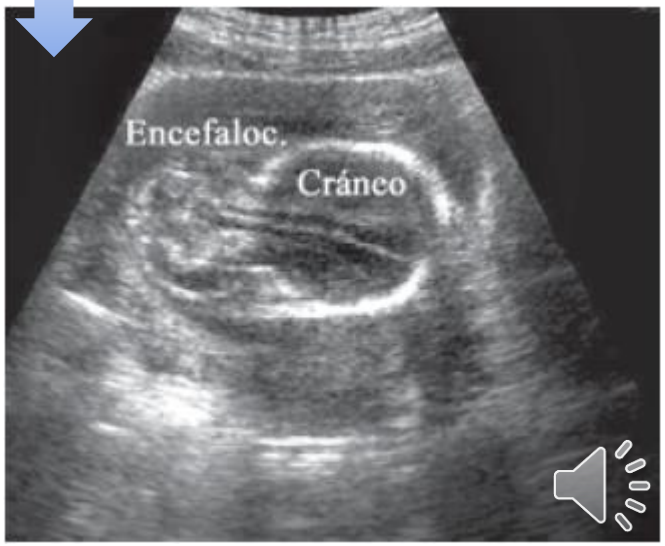


Meningocele & meningoencephalocele

- Intrauterine diagnosis
  - Prenatal ultrasound → Abortion?
  - Prenatal MRI

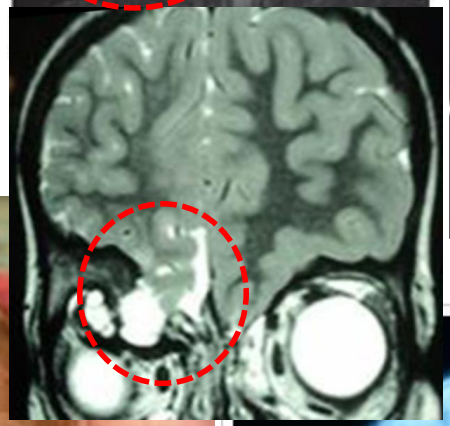
MRI

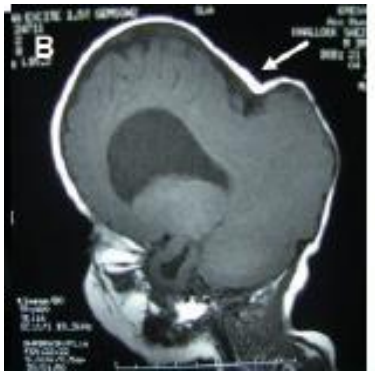




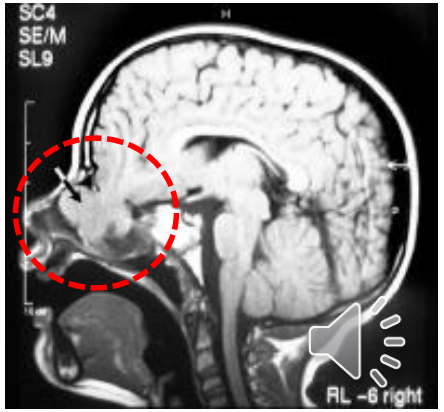
Meningocele & meningoencephalocele

- Diagnosis at birth
  - Clinical features
  - Ultrasound
  - MRI



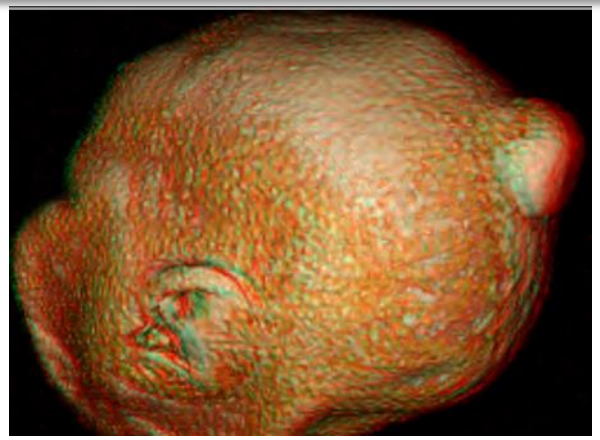


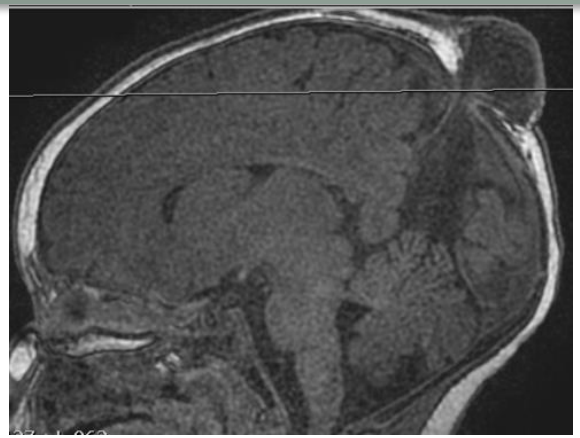
Atresic parietal meningo-encephalocele

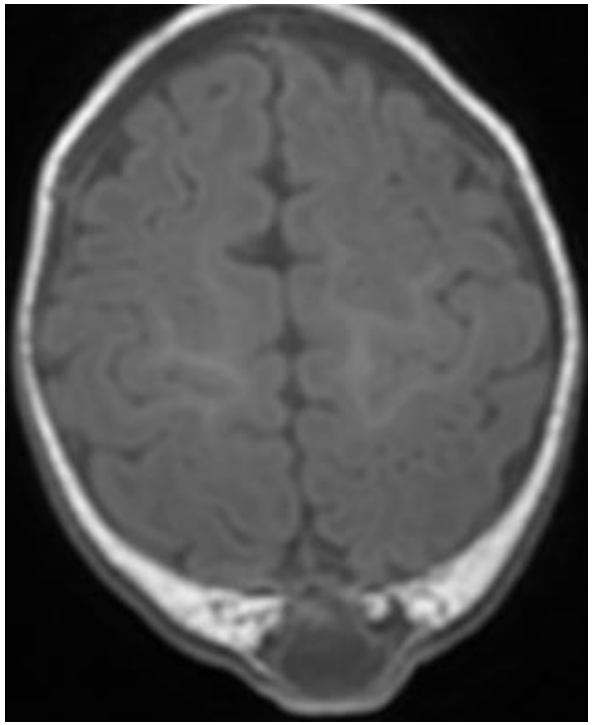


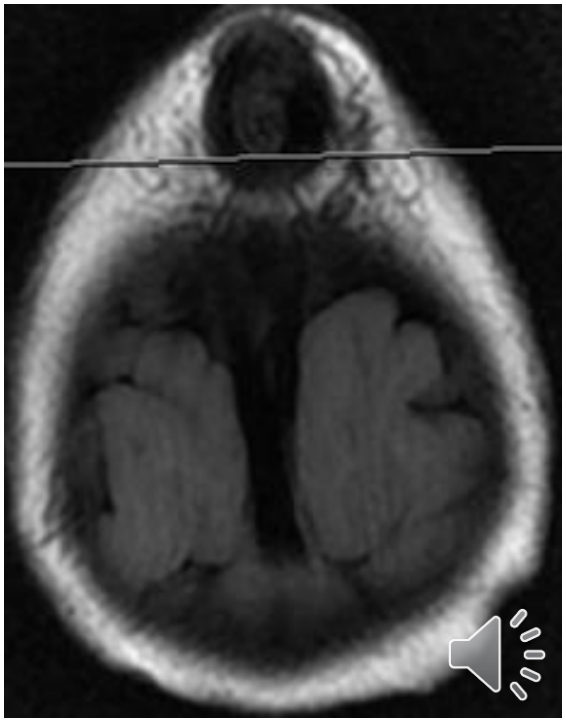
Cranial meningocele: intrauterine diagnosis

- Presence may be suspected with ultrasound
- Confirmation by magnetic resonance imaging



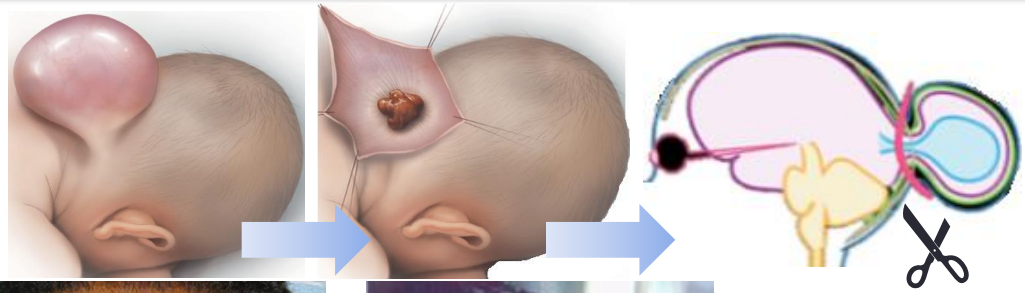


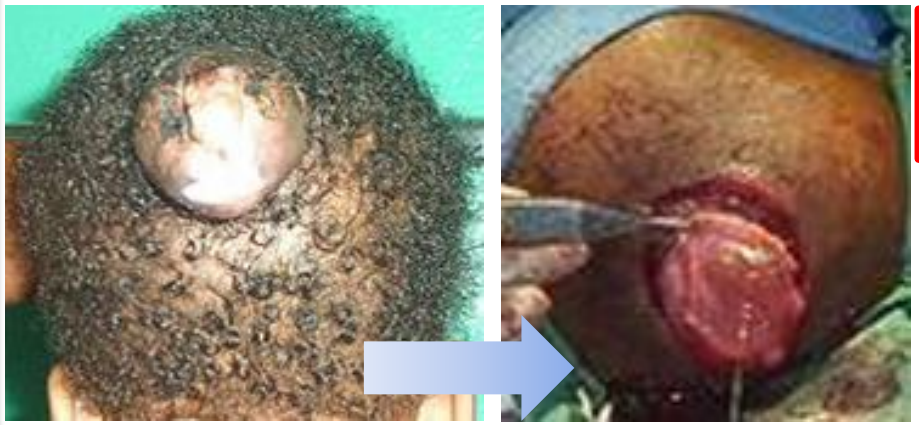


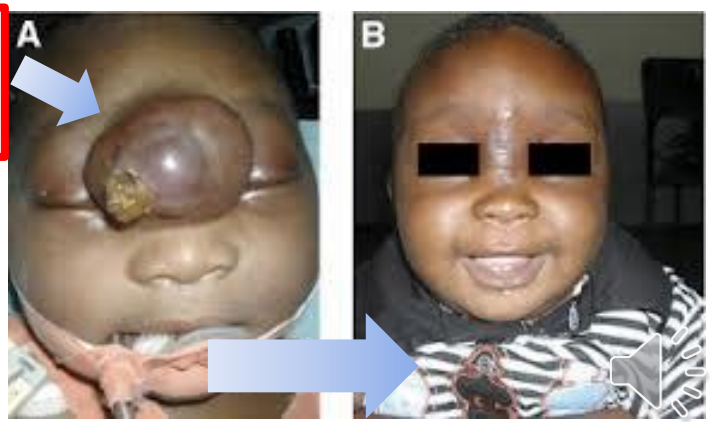


Meningocele & meningoencephalocele

- Treatment= surgical = **defect repair & closure**
  - Goal: To preserve viable brain tissue
  - Highly variable results, high mortality







Occipital meningoencephalocele: venous sinuses

- Abnormal venous sinuses = inability to expand skull to introduce herniated nerve tissue intracranially = need to remove that tissue = severe neurological deficits

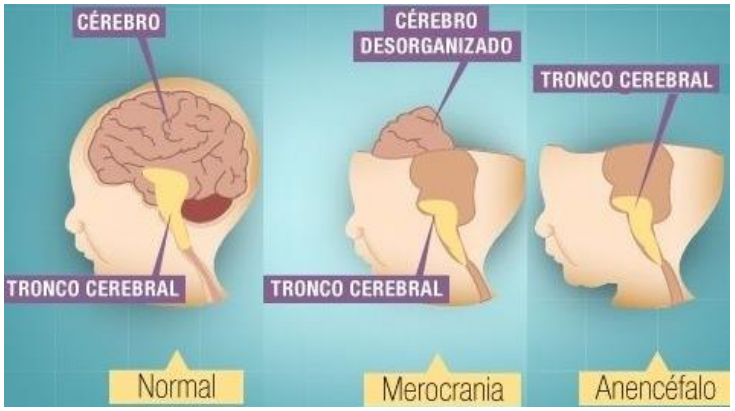
Normal

Occipital meningoencephalocele

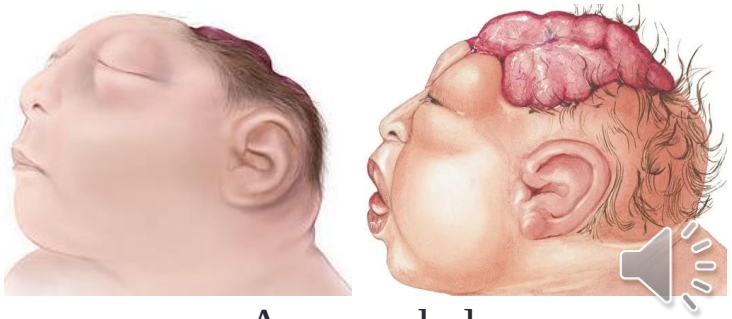
Anencephaly (1)

- Definitions
  - **Acrania** = absence cranial vault
    - Usually associated with spine defects
  - **Meroanencephaly** = no closure of neural tube cranial end (4th week) → no cranial vault closure
  - **Anencephaly** = partial brain absence
- Incidence: 1 / 1000 births
- **Incompatible with life**
  - Survival time depends on the amount of existing brain and its degree of organisation

Acrania



Meroanencephaly



Anencephaly

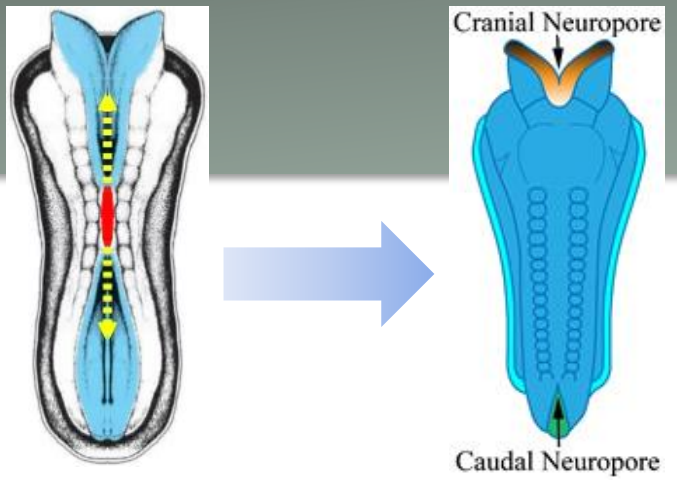
Anencephaly (2)

- Intrauterine diagnosis
  - Ultrasound (14 weeks) → Abortion?
- **Prognosis**
  - Death at birth or soon after
  - Rare survivals > 1 year

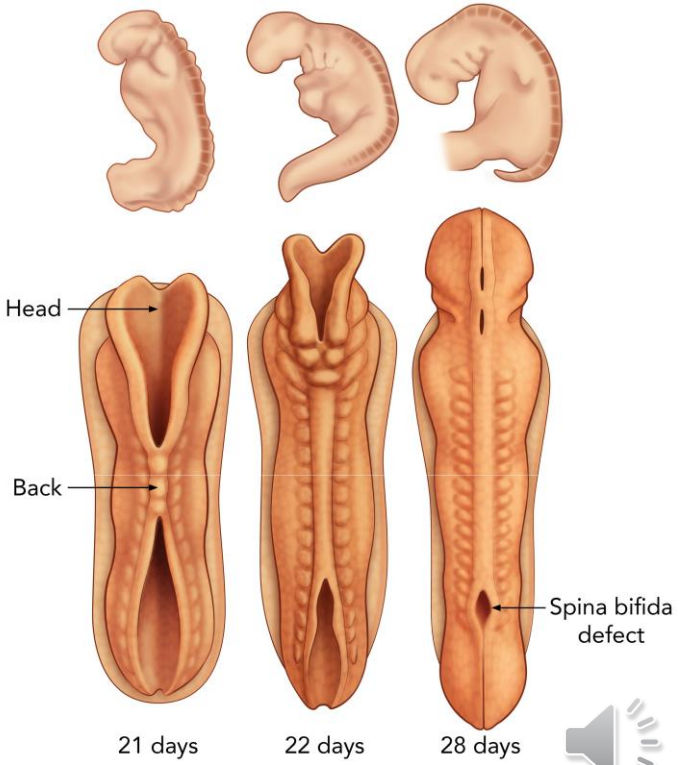
Vitória do Cristo,
2 ½ years
†July 2012

3. SPINA BIFIDA

- Spinal cord neural tube closure defect
- May involve
  - Meninges = meningocele
  - Spinal cord = myelomeningocele
  - Fatty tissue = lipomyelomeningocele
  - Bone = diastematomyelia
- Diagnosis = clinical features, CT/MRI
- Treatment = surgical
  - Meningocele = dural + soft tissue repair
  - Myelomeningocele = spinal cord, meninges + soft tissues repair
  - Lipomyelomeningocele = untether spinal cord followed by meninges + soft tissue repair
  - Diastematomyelia = bone spicule removal

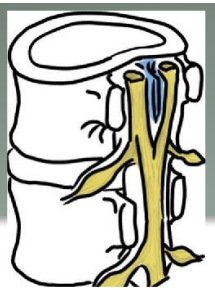


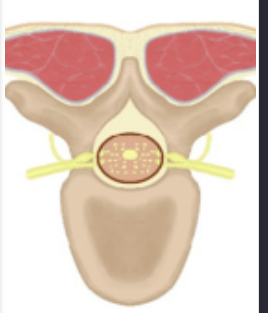
Normal neural tube closure

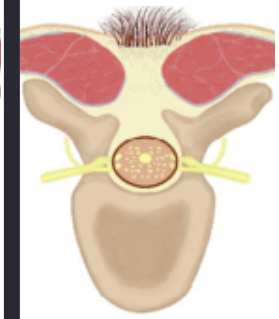


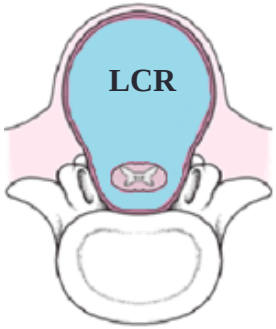
Spina bifida

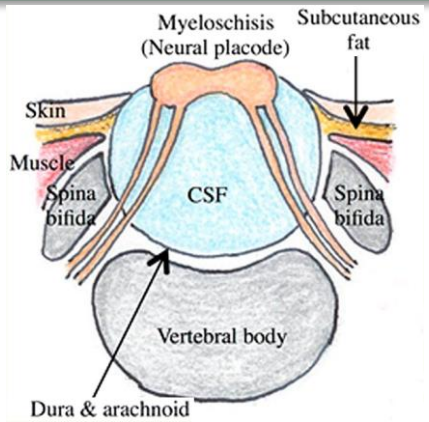
Spinal dysraphisms (1)

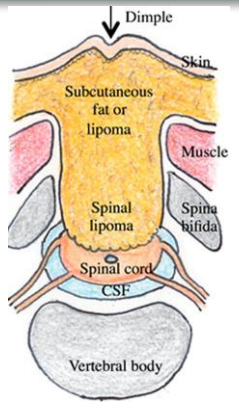


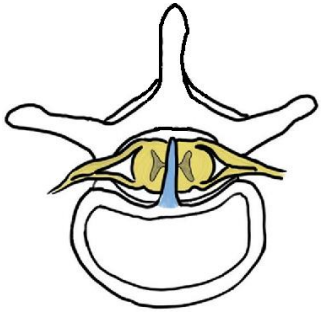


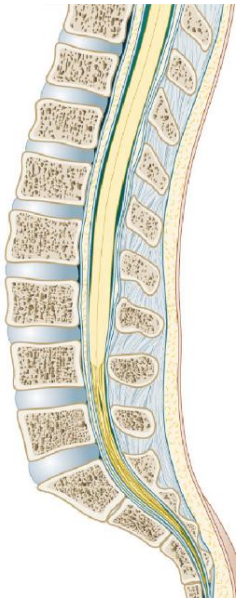


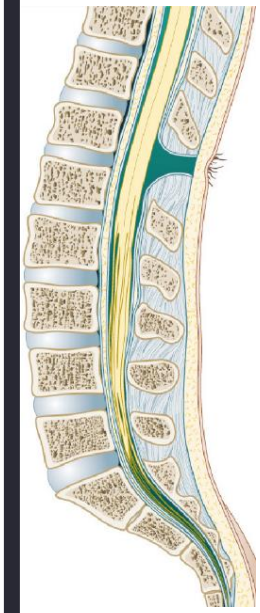


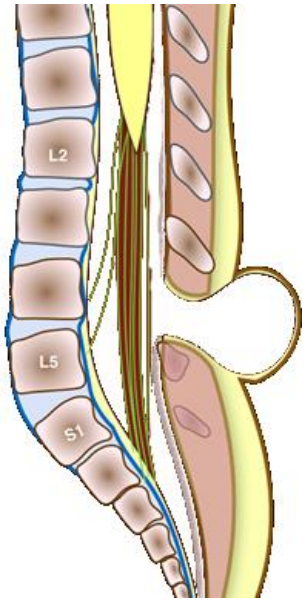


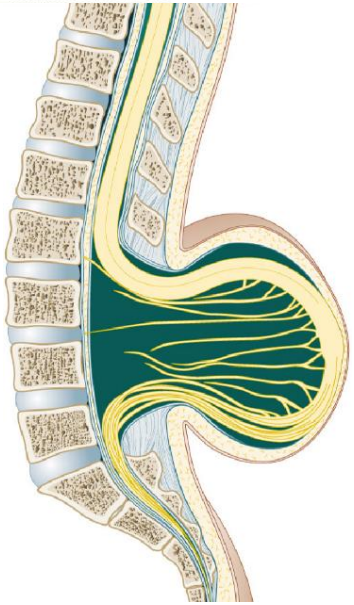


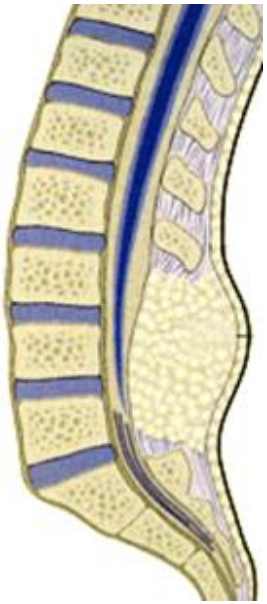


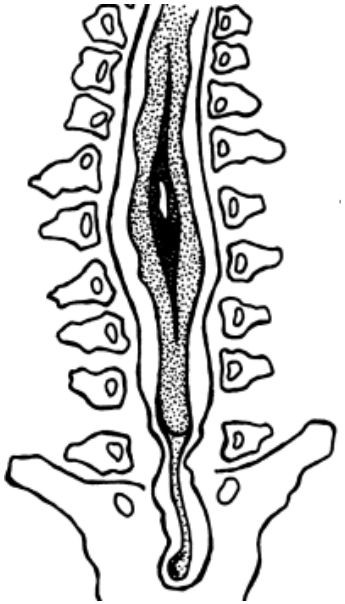












Spina bifida occulta

Meningocele

Myelomeningocele

Lipomyelo-meningocele

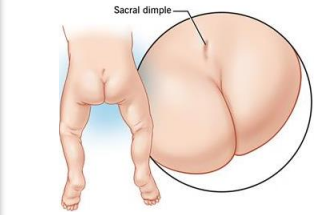
Diastema-tomyelia

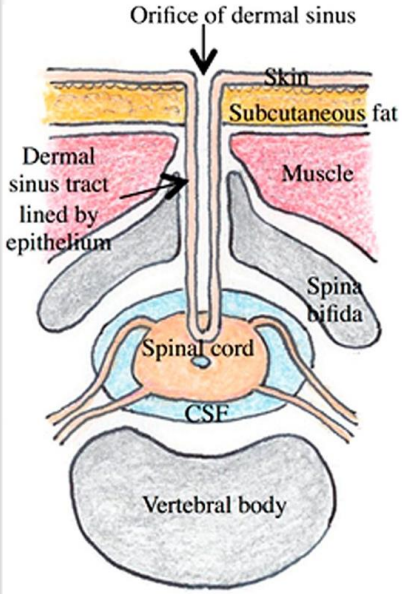
Normal

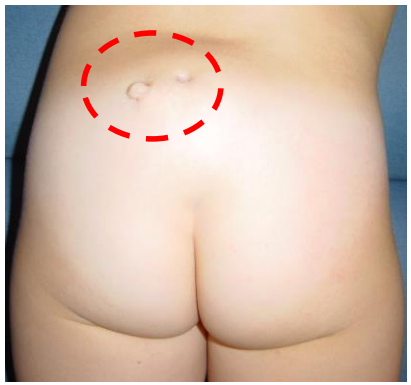
Dysraphisms

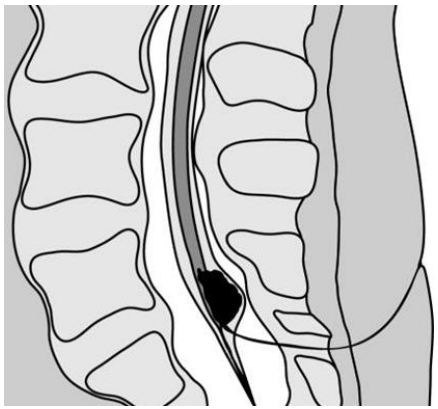
Spinal dysraphisms (2)

- **Dermal sinus** = cutaneous connection between intradural compartment and outside world = bacterial meningitis risk
- **Thickened filum terminale** = tethered spinal cord = with growth traction on thoracic spinal cord = spinal cord impairment = lower limb paraparesis

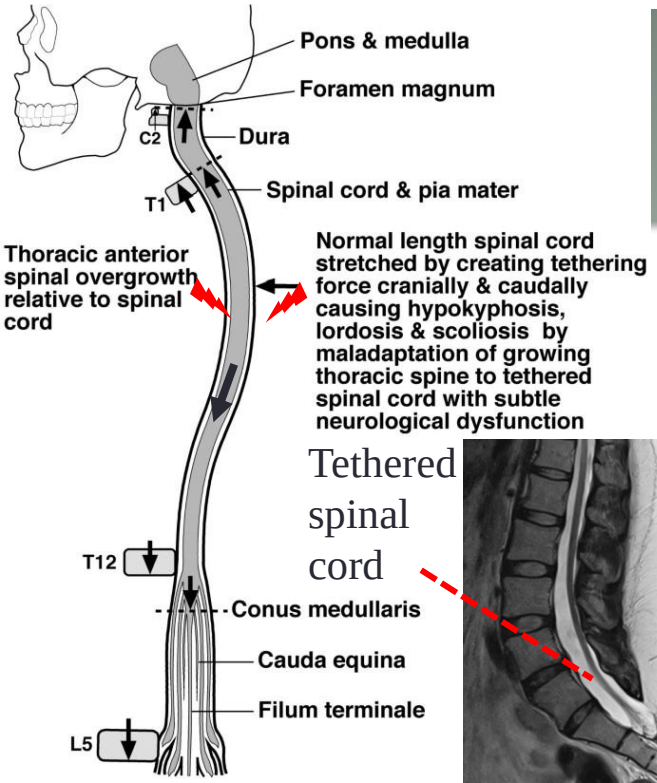




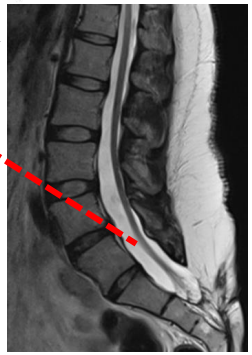


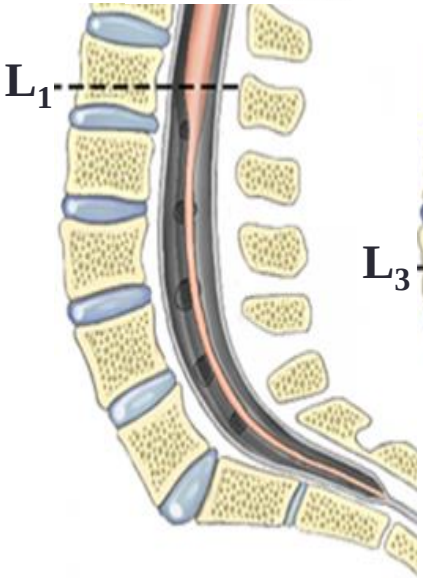


Dermal sinus

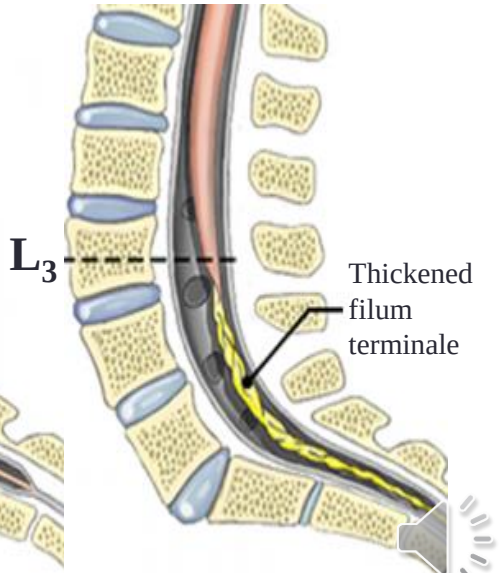


Tethered spinal cord





Normal

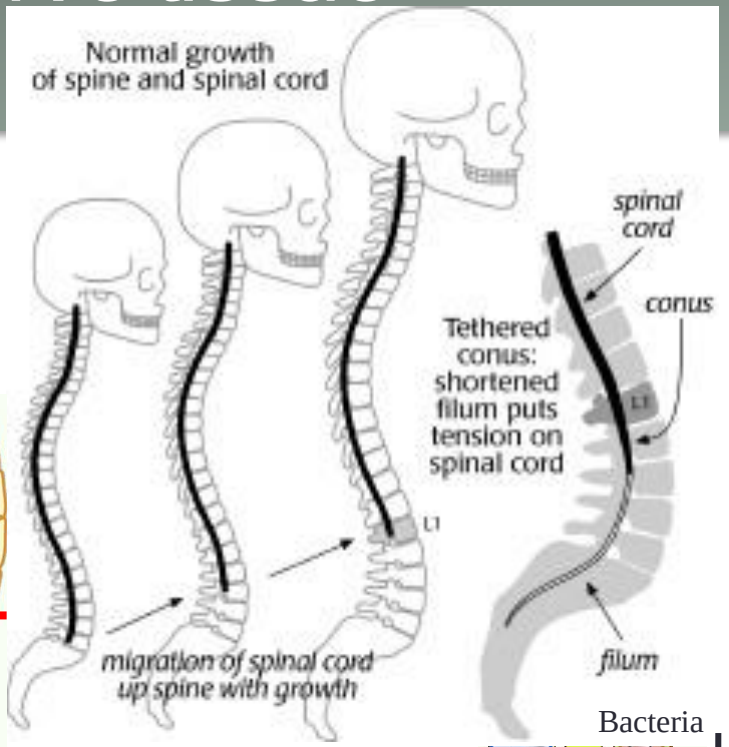


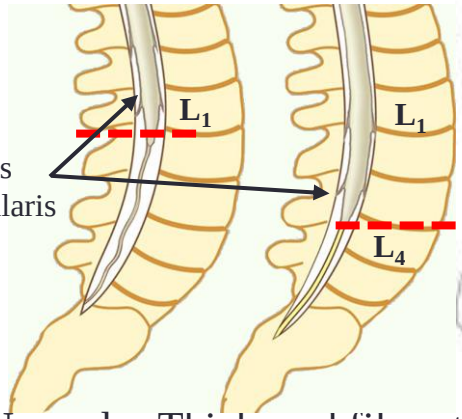
Tethered spinal cord

Spinal dysraphisms: nerve tissue injury mechanisms

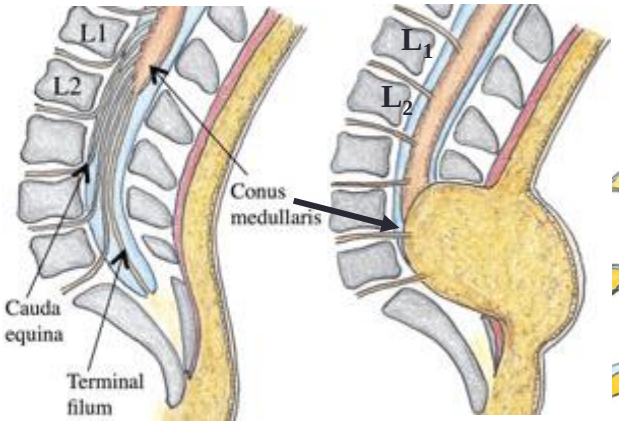
- **Outside world nerve tissue exposure**
  - Myelomeningocele
- **Spinal cord tethering**
  - Myelomeningocele Lipomyelomeningocele
  - Diastematomyelia
  - Thickened filum terminale
- **Bacterial meningitis**
  - Non-repaired myelomeningocele
  - Dermal sinus

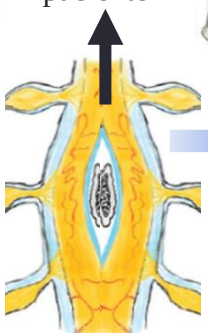
Myelomeningocele

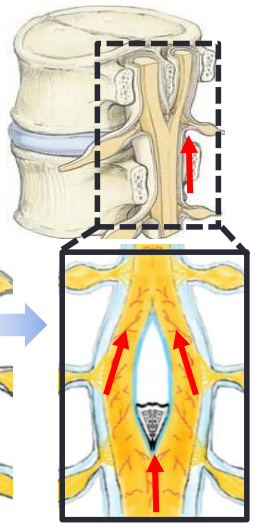


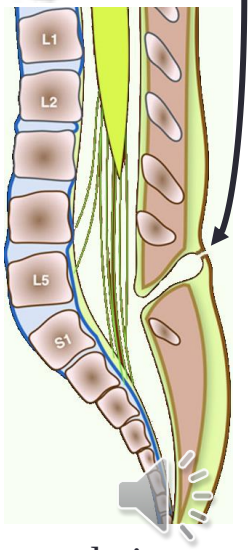


Normal Thickened filum terminale





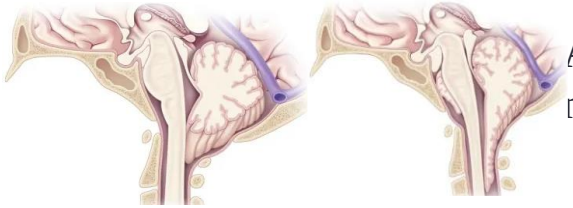




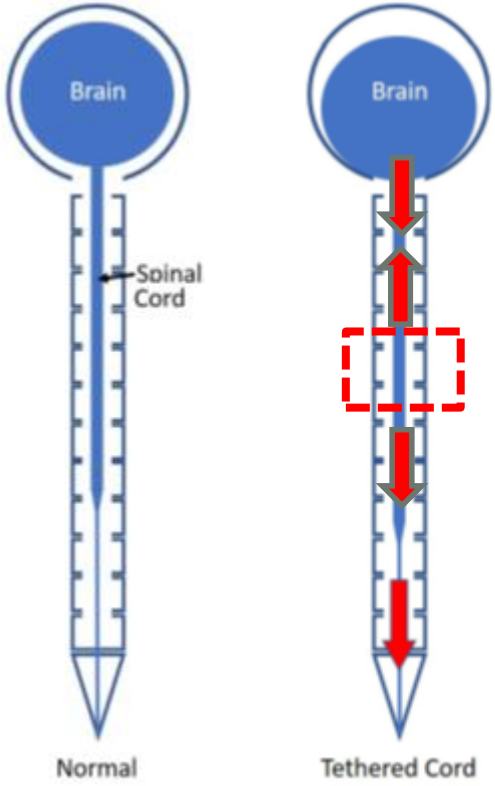
Normal Lipomyelomeningocele Diastematomyelia Dermal sinus

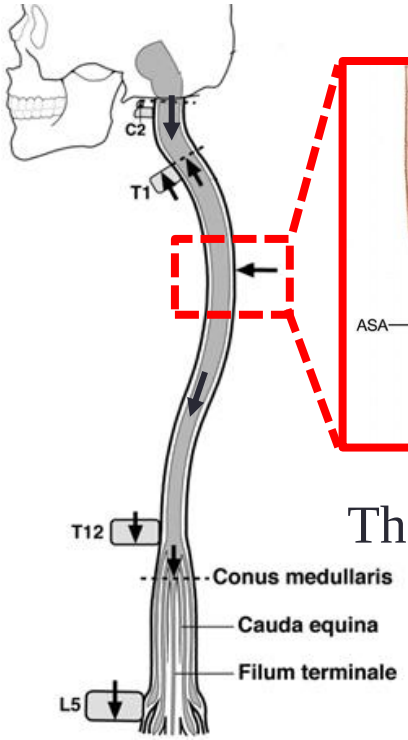
Spinal cord tethering & nerve tissue injury

- Arnold-Chiari type II malformation
- Thoracic spinal cord myelopathy

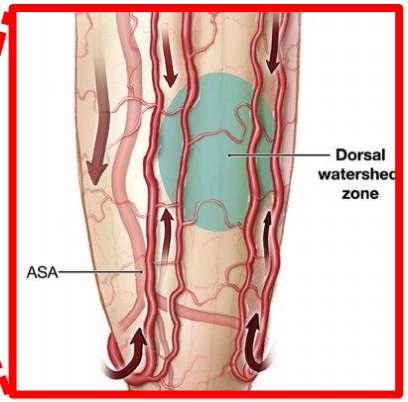


Arnold-Chiari type II malformation





Tethered cord

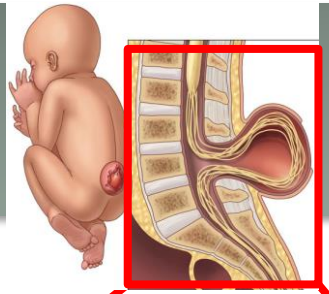


Thoracic myelopathy

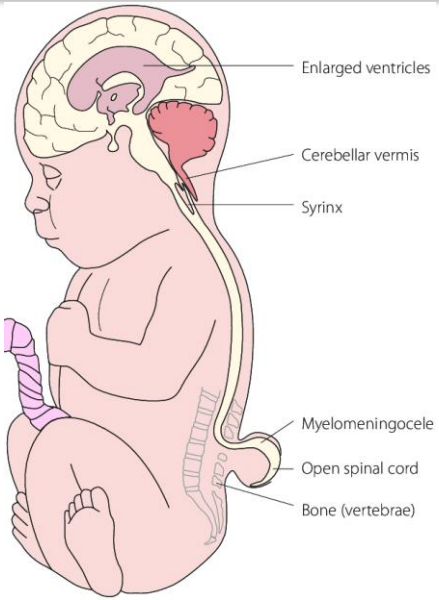
Pareto-spastic gait

Lower limb muscle atrophy

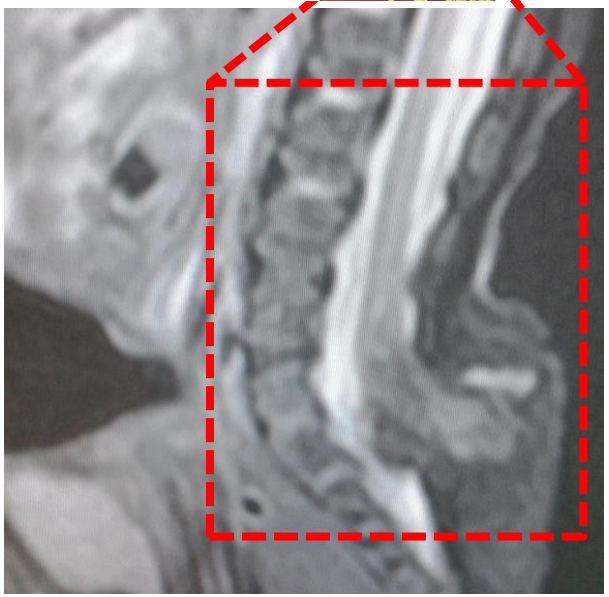
Myelomeningocele

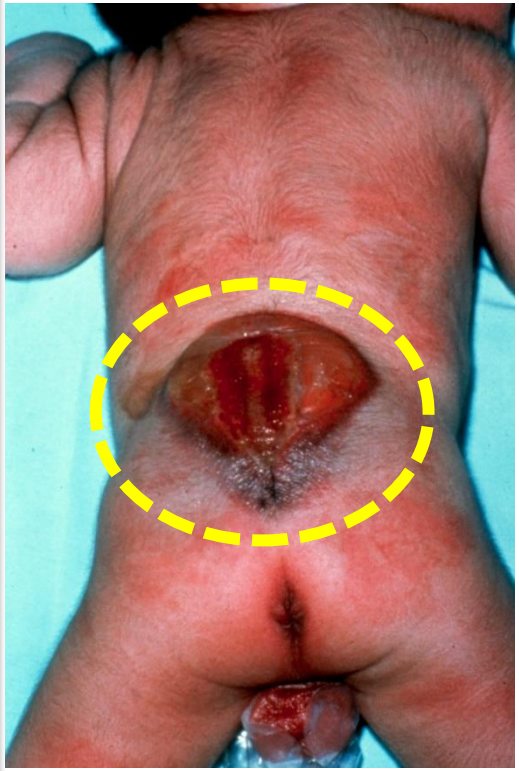


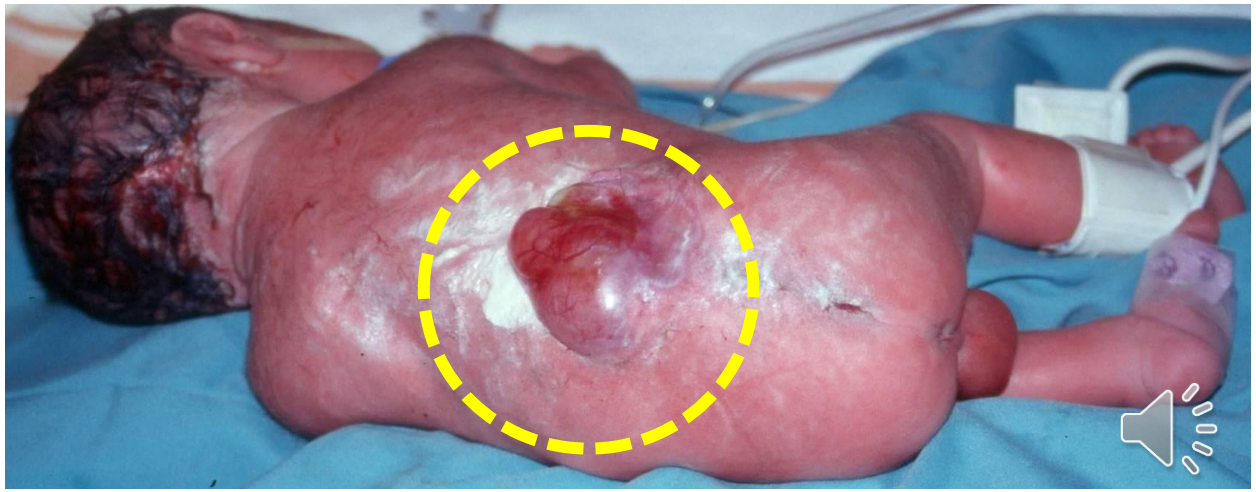
- **Spinal cord damage since inception**
  - Severity depends on the spinal level
- The scar tethers spinal cord after surgical repair
  - Arnold-Chiari type II malformation



Arnold-Chiari type II malformation





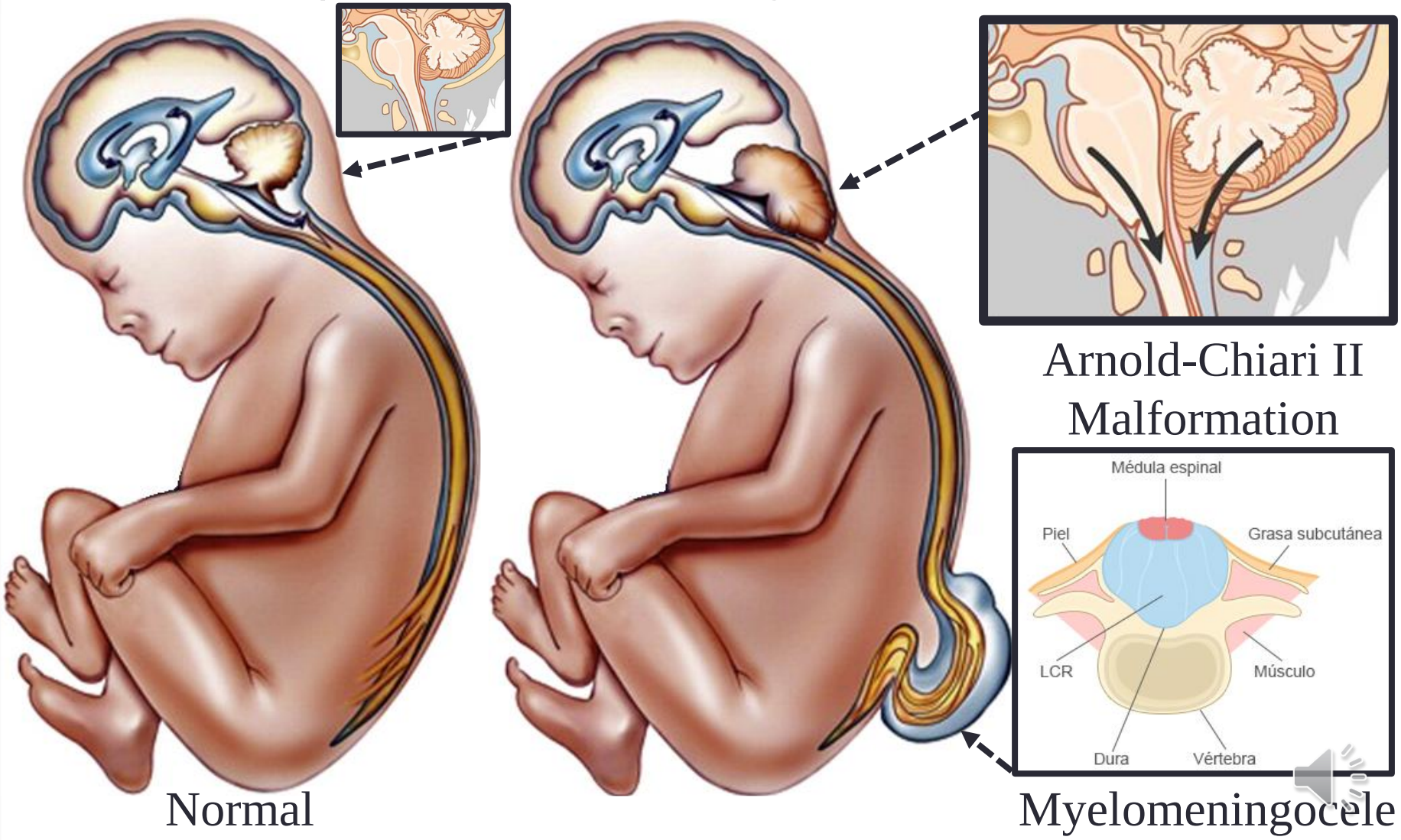


Myelomeningocele & Arnold-Chiari type II malformation: pathophysiology

- Due to spinal cord tethering by myelomeningocele
- Downward spinal cord traction = herniation cerebellar tonsils
- Hydrocephalus
  - Partly due to CSF leakage into the amniotic fluid = poor development of CSF drainage system at venous sinuses

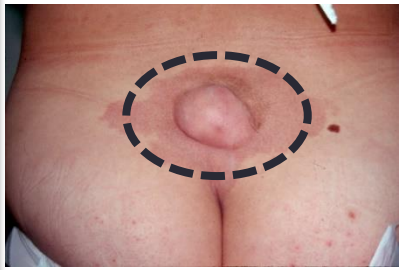
Myelomeningocele & Arnold-Chiari type II malformation: treatment

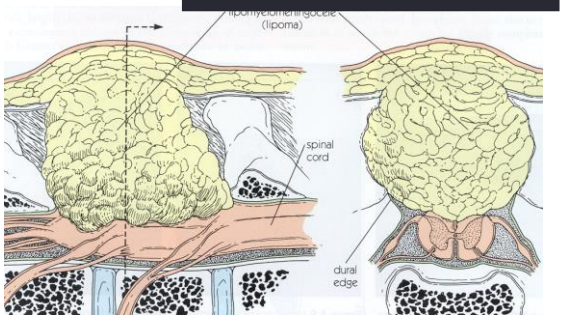
- Treatment: spinal cord untethering + CSF shunt

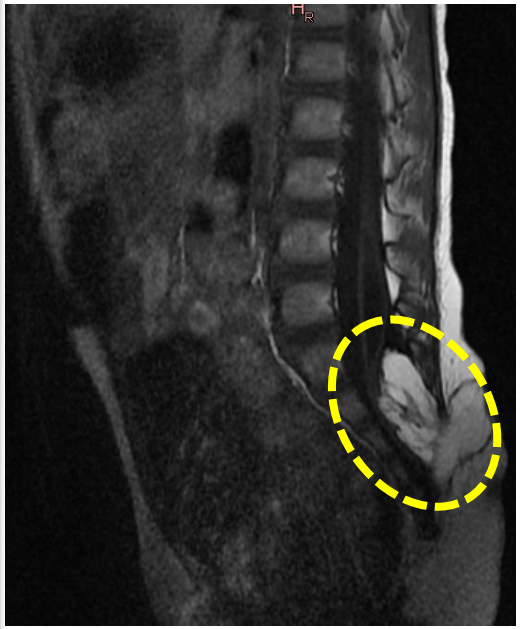


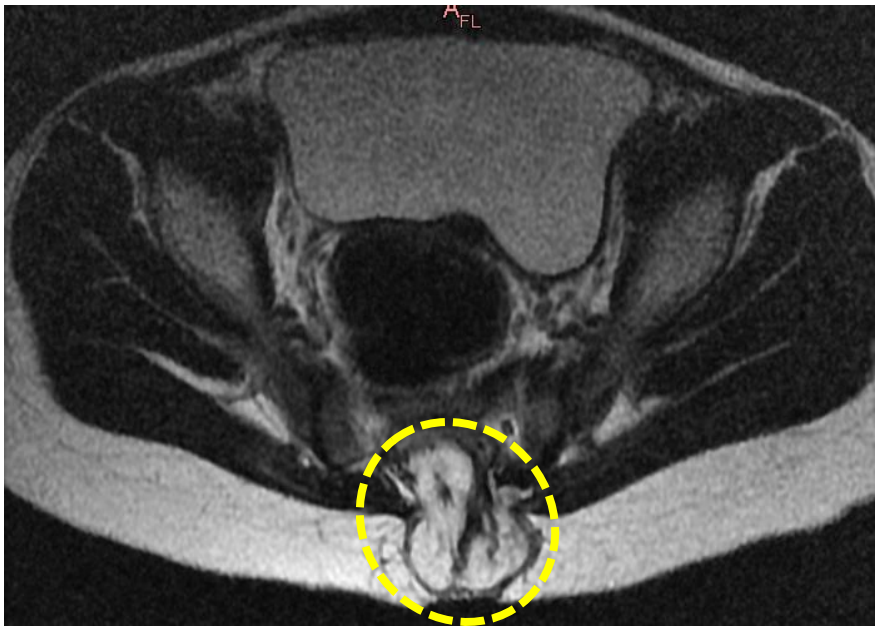
Lipomyelomeningocele

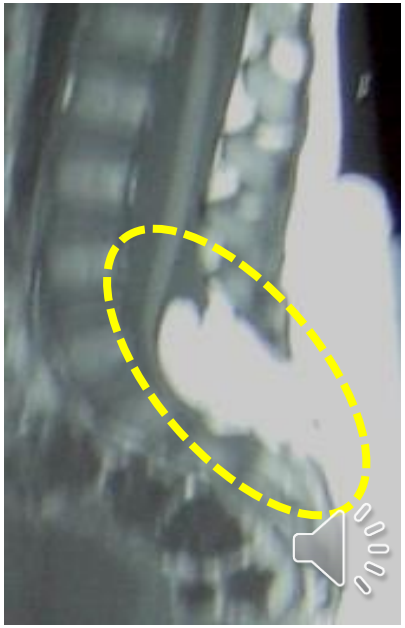
- NORMAL lipomatous tissue tethering the spinal cord
  - The problem is the tethering





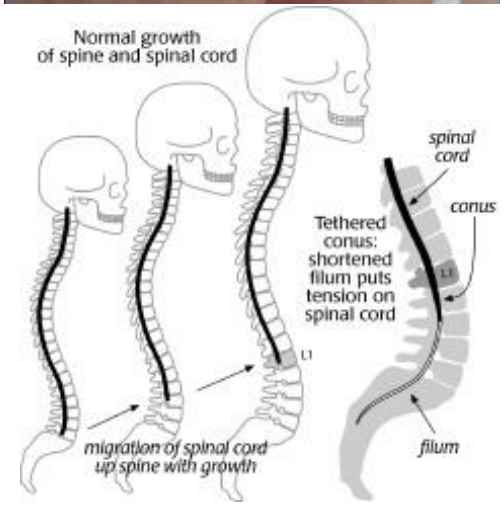






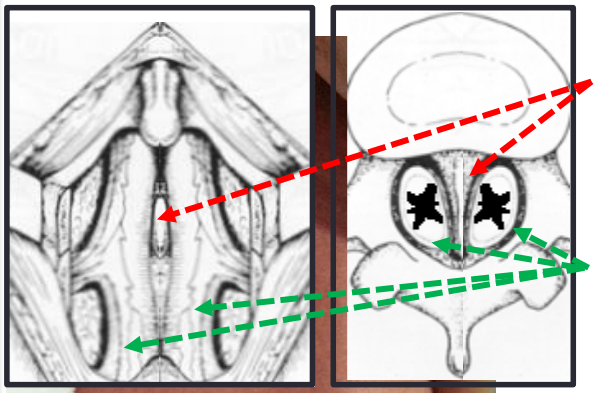
Lipomyelomeningocele

- Good skin coverage
  - No risk of meningitis
  - Surgical repair can be delayed
- Spinal cord medium and long-term impairment due to tethering
  - Thoracic spinal cord undergoes stretching and ischemia as the child growths

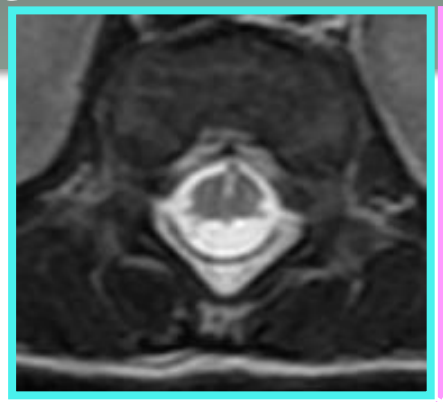


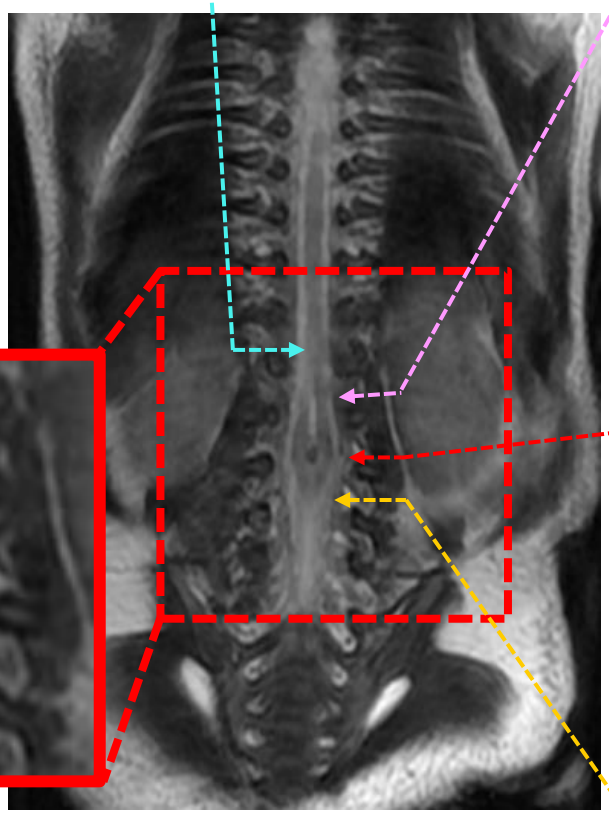
Diastematomyelia

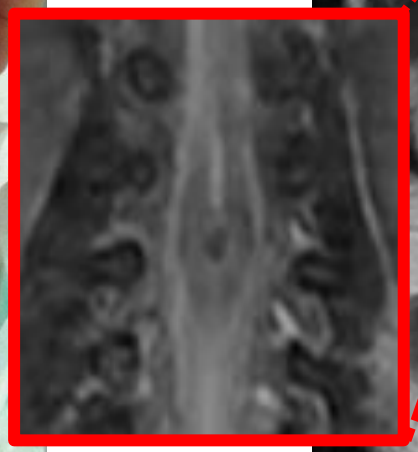
- Bone spicule invading spinal canal and splitting spinal cord in two
  - Skin sign = lock of hair = Faun's beards
  - Problem = spinal cord tethering

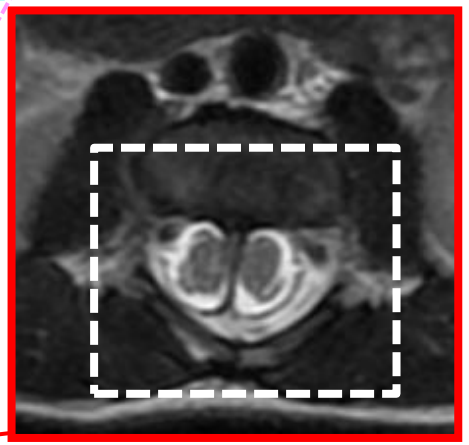


Fauns beard







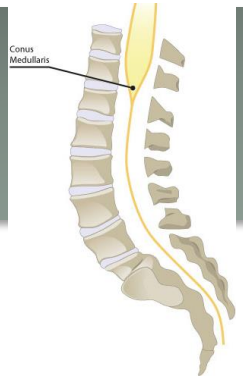


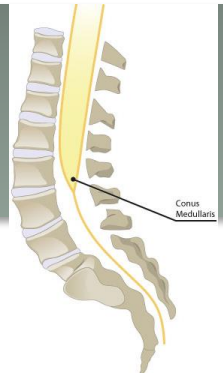
Dermal sinus

- Frequent consultation
- Rarely symptomatic
- Requires lumbar MRI
- Risk of bacterial meningitis

Spinal cord tethered by thickened filum terminale (1)

- Very rare
- Possible association with scoliosis
- No relationship with syringomyelia
-

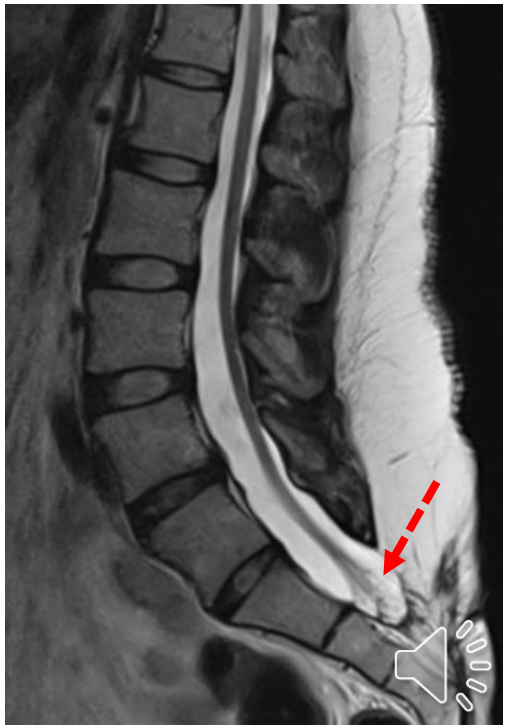




Normal **Tethered spinal cord**

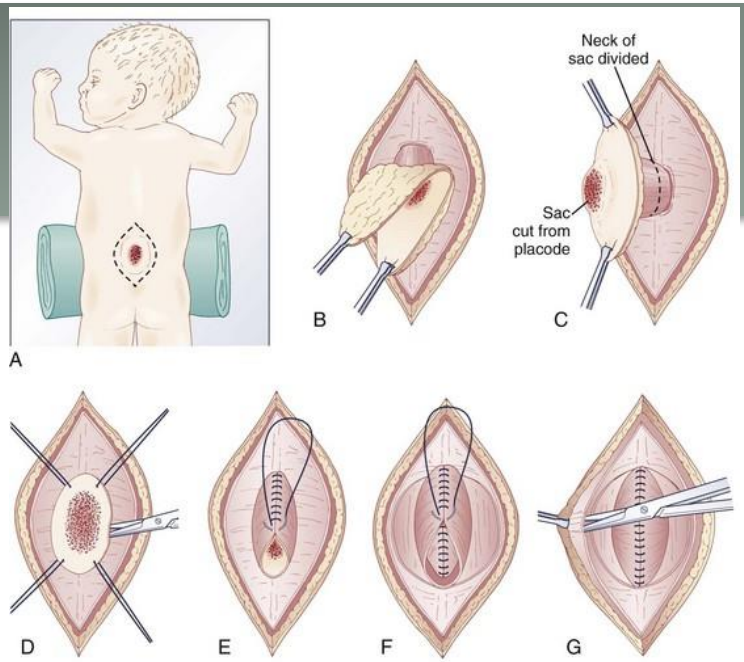
Spinal cord tethered by thickened filum terminale(2)

- Lower back and lower limb pain that worsens in the lotus position

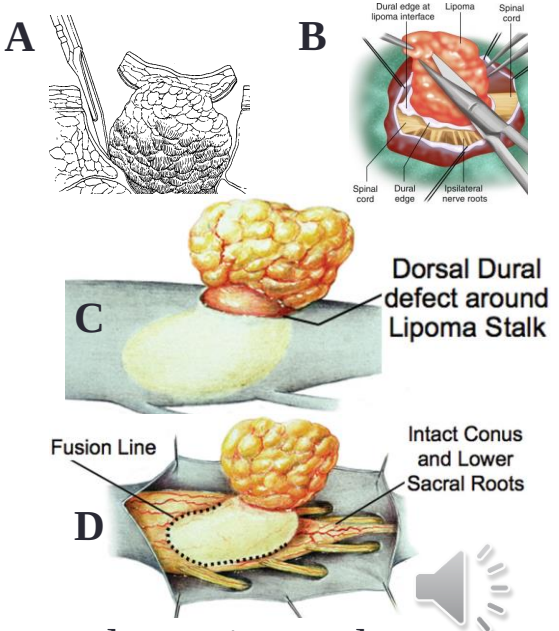


Spinal dysraphisms: surgical treatment (1)

- **Objectives**
  - Untethering spinal cord
  - Achieve skin coverage
  - Avoid spinal retethering through scar
- Procedure: microsurgical technique
- **Results**
  - Myelomeningocele: achieve skin cover = scar retethers spinal cord = Arnold-Chiari type II malformation
  - Lipomyelomeningocele: lipoma remnants attached to spinal cord = retethering= need for reintervention in adolescence
  - Diastematomyelia: laminectomy = kyphosis
  - Dermal sinus & spinal cord tethered by thickened filum terminale = no recurrences



Myelomeningocele repair

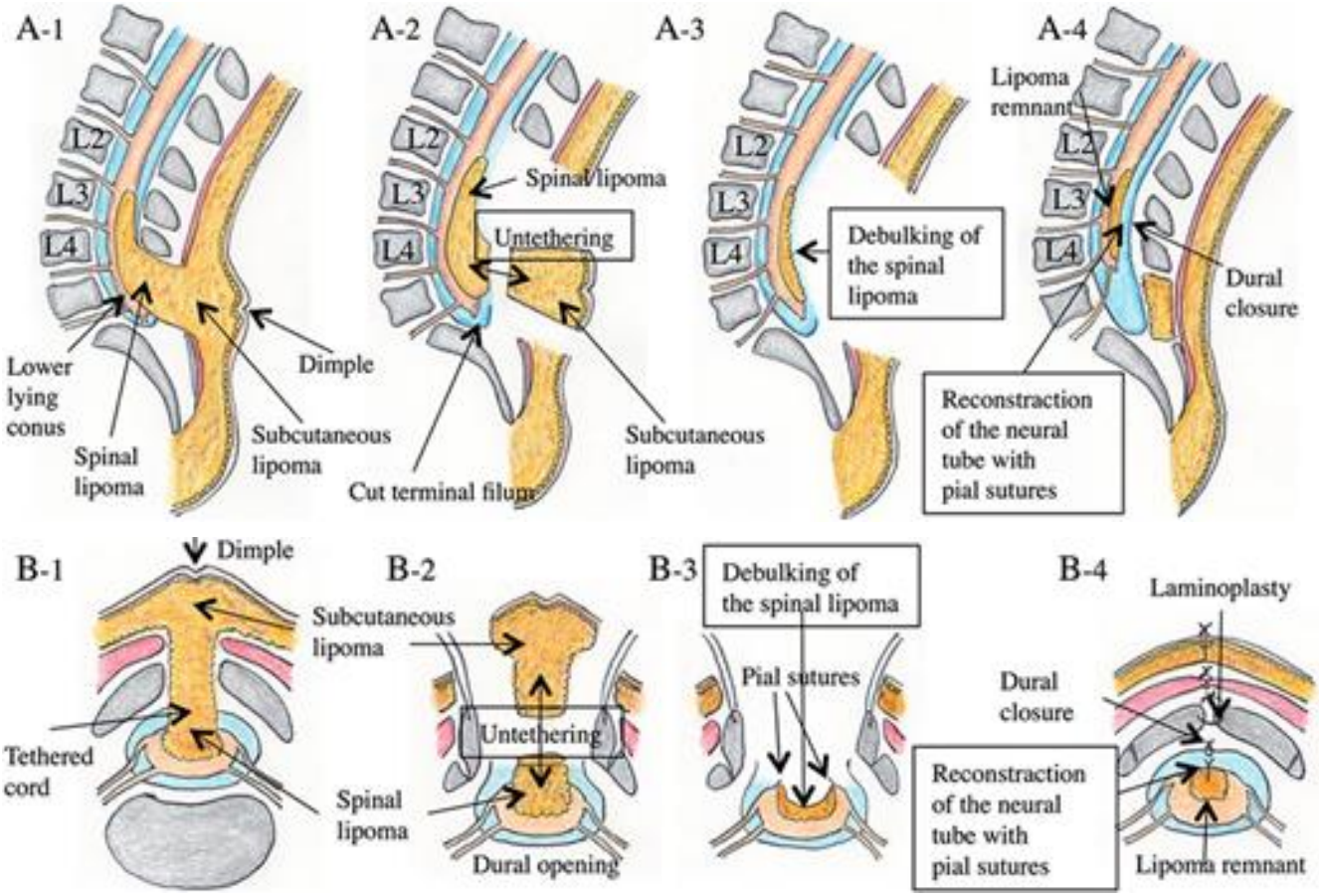


Lipomyelomeningocele repair

Lipomyelomeningocele: surgery

- Problem: **spinal cord tethering**
  - Spinal cord retethers again after surgery
  - Reinterventions common
  - Possibility of thoracic myelopathy

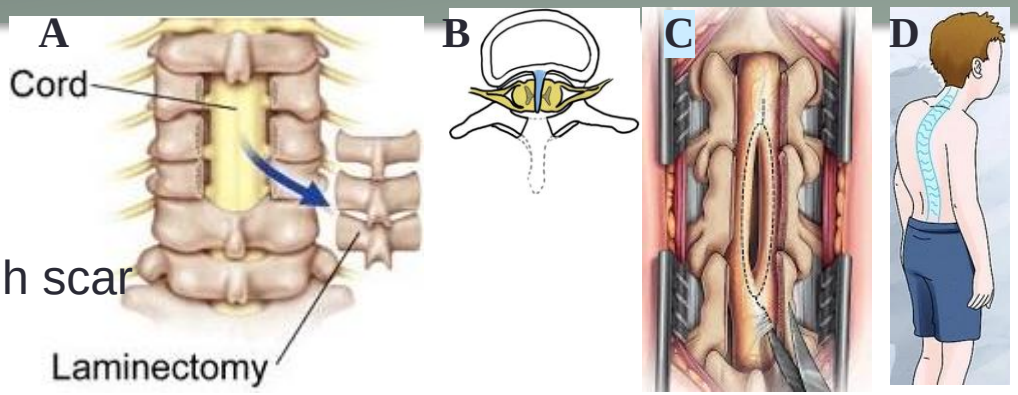
Lumbosacral lipoma remnants after surgery



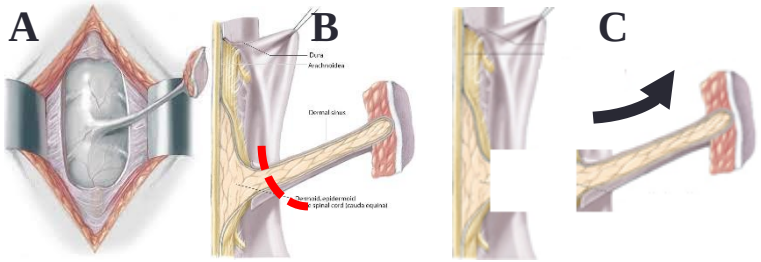
Lower limbs with thoracic myelopathy after lumbosacral lipoma removed in childhood

Spinal dysraphisms: surgical treatment (2)

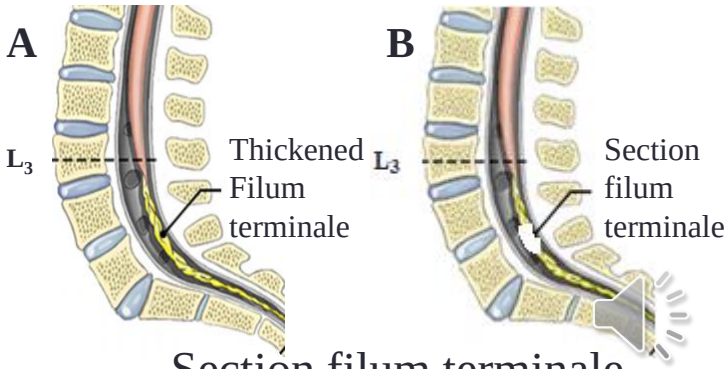
- **Objectives**
  - Untethering spinal cord
  - Achieve skin coverage
  - Avoid spinal reanchoring through scar
- Microsurgical technique
- **Results**
  - Myelomeningocele: achieve skin cover = scar retethers spinal cord = Arnold-Chiari type-II malformation
  - Lipomyelomeningocele: lipoma remnants attached to spinal cord = retethering= need for reintervention in adolescence
  - Diastematomyelia: laminectomy = kyphosis
  - Dermal sinus & spinal cord tethered by thickened filum terminale = no recurrences



Diastematomyelia repair



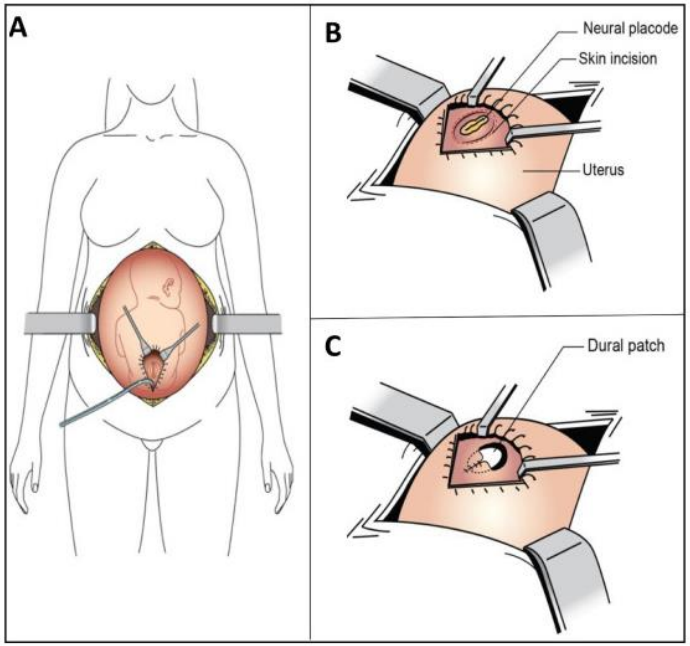
Dermal sinus repair

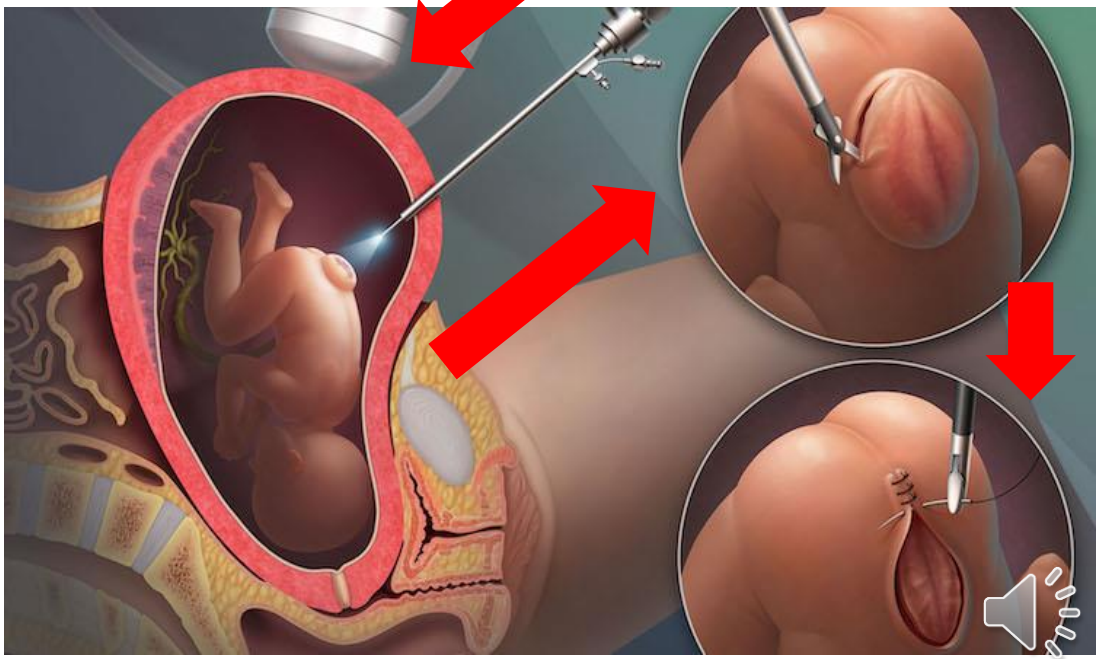


Section filum terminale

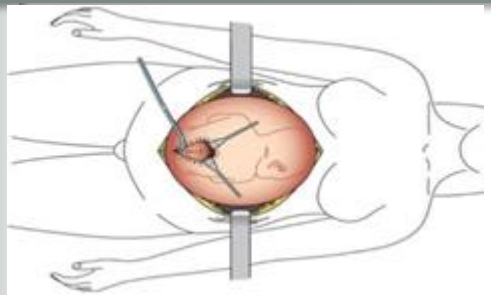
Myelomeningocele: intrauterine repair (1)

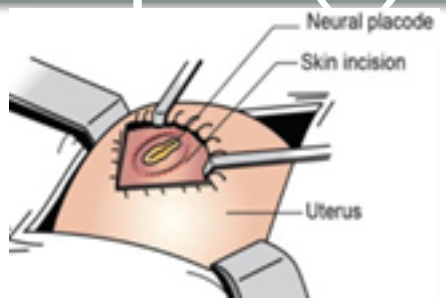
- Possible but technically ↑ complex
  - Opening uterus = avoid uterine contractions & avoid abortion
- **Results in evaluation**
  - ↓ nerve tissue damage?
  - ↓ hydrocephalus incidence?
  - ↓ spinal cord retethering?



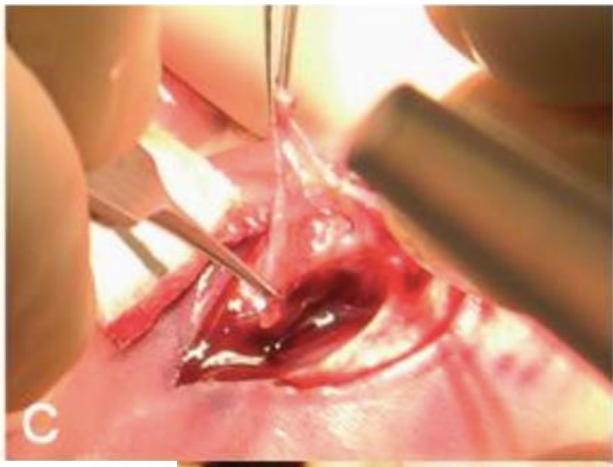


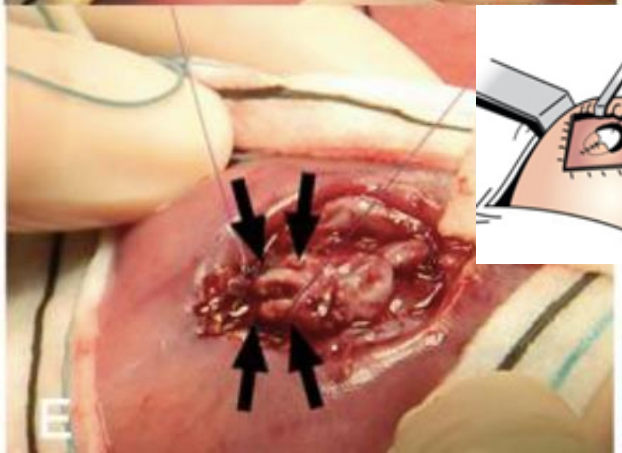
Myelomeningocele: intrauterine repair (1)

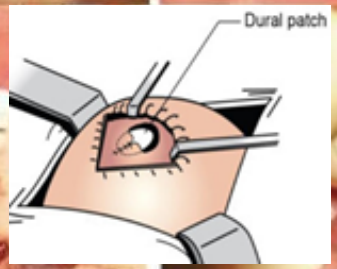


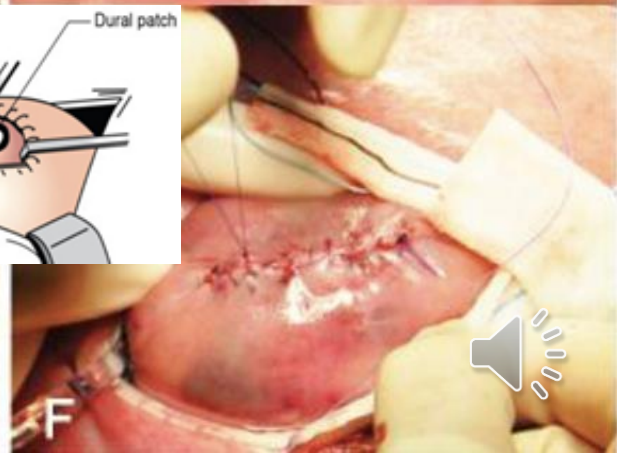


Pregnancy ultrasound



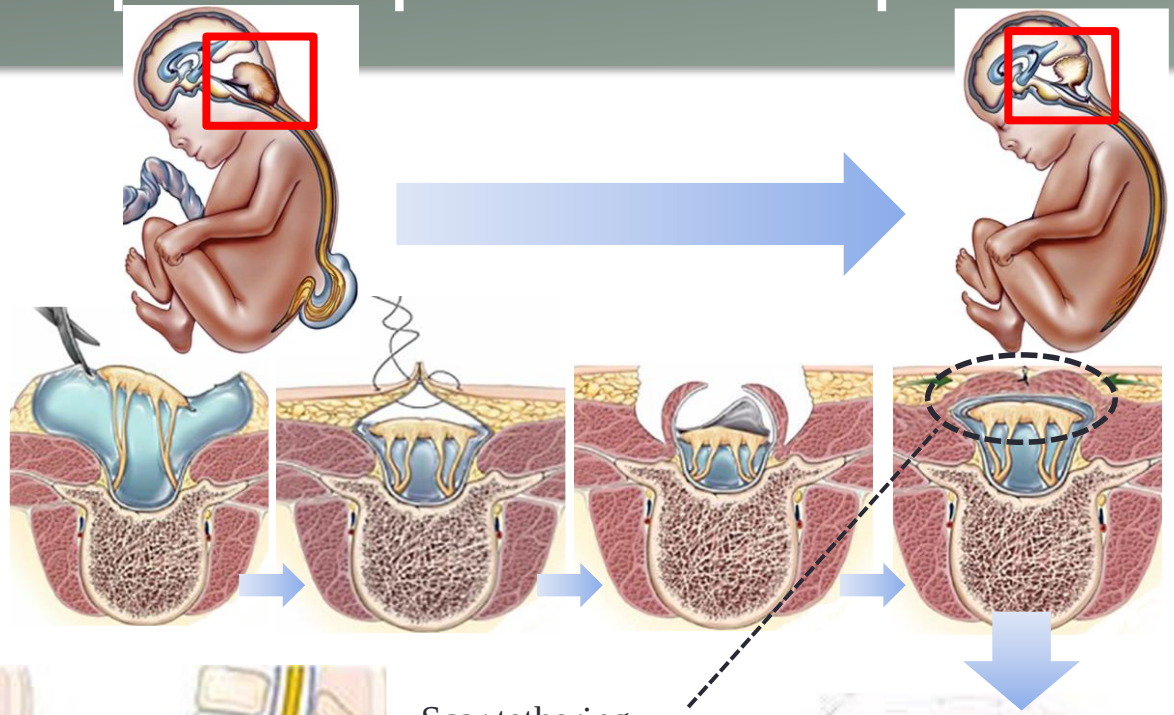


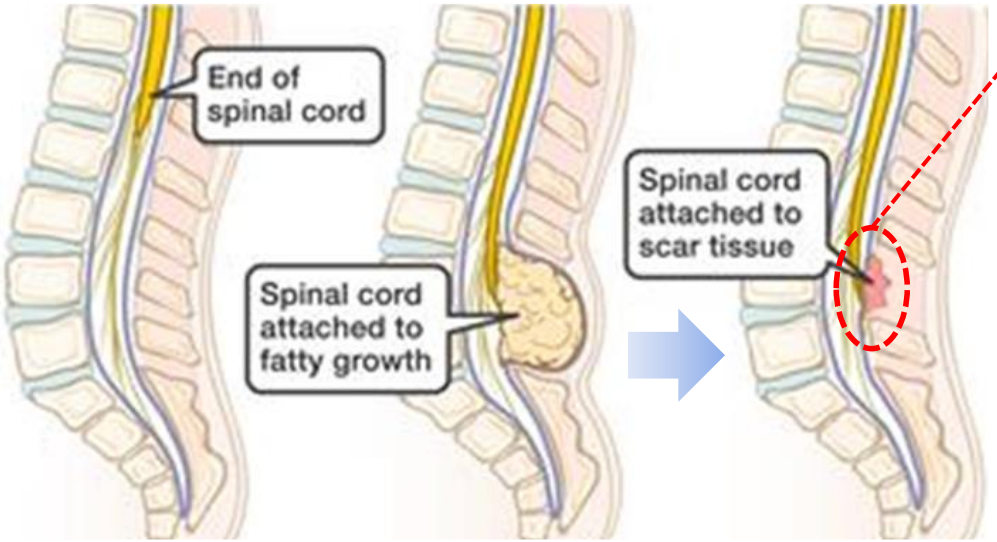




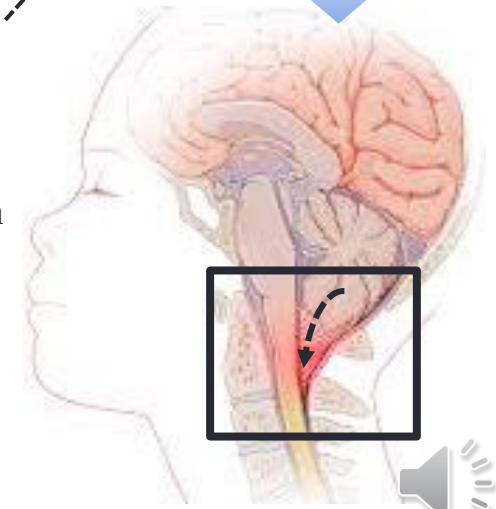
Dysraphisms: post-repair follow-up

- Scarring surgical field = spinal cord retethering = need reintervention
  - Particularly in the adolescence growth spurt





Scar tethering spinal cord and preventing its cranial migration with child growth in height



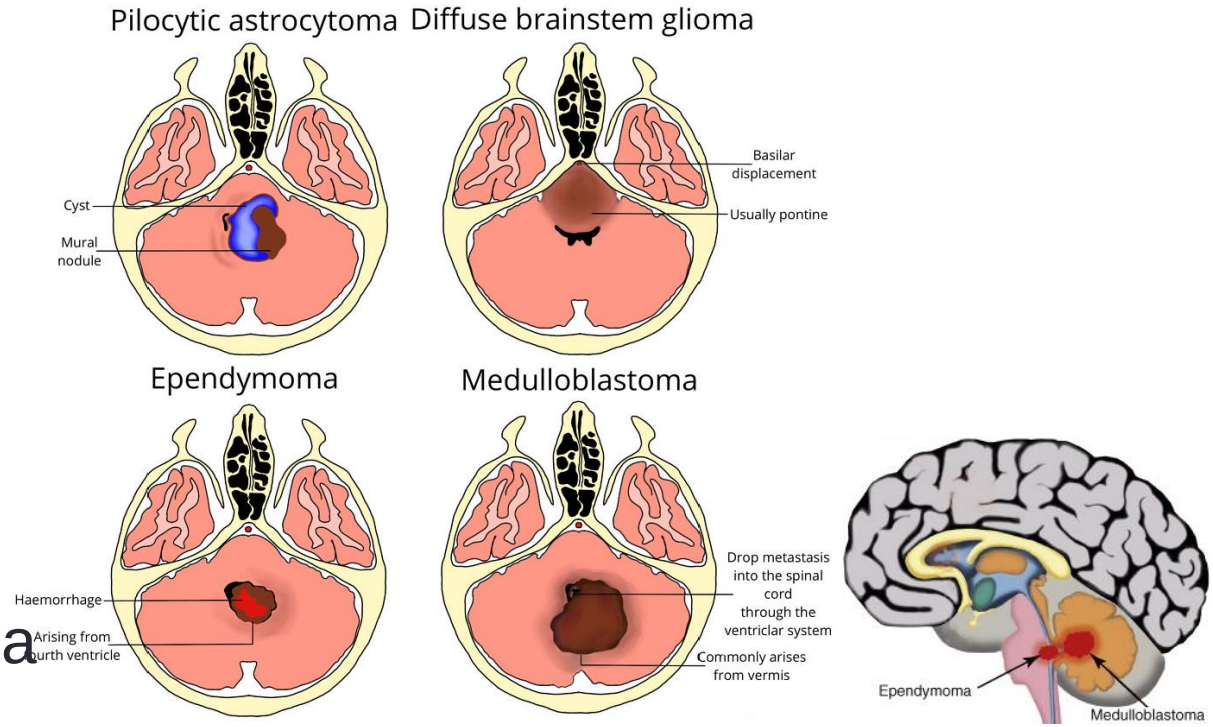
Normal

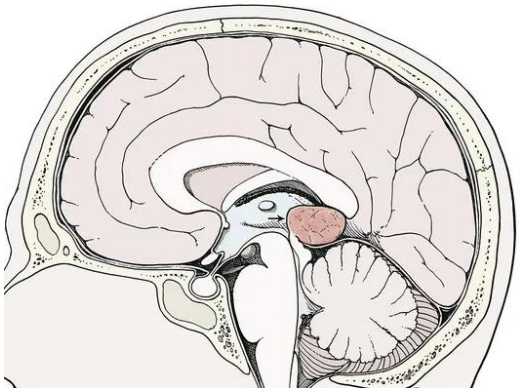
Evolution after lipomyelomeningocele surgery

Evolution after myelomeningocele surgery

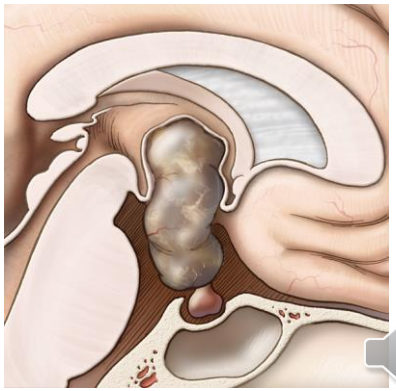
PEDIATRIC BRAIN TUMORS

- Medulloblastoma
- Ependymoma
- Cystic cerebellar astrocytoma
- Infiltrating brain stem glioma
- Craniopharyngioma
- Pineal region tumors



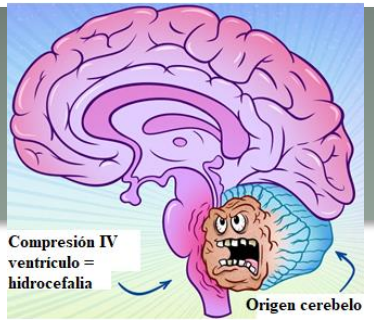


Pineal region tumor

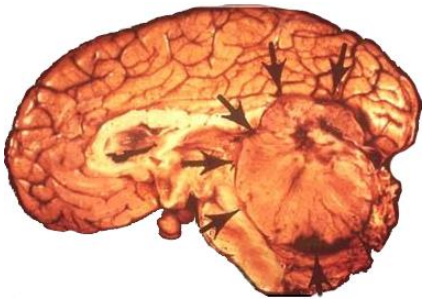


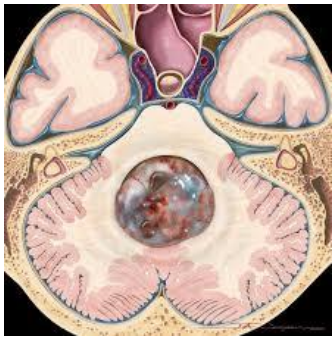
Craniopharyngioma

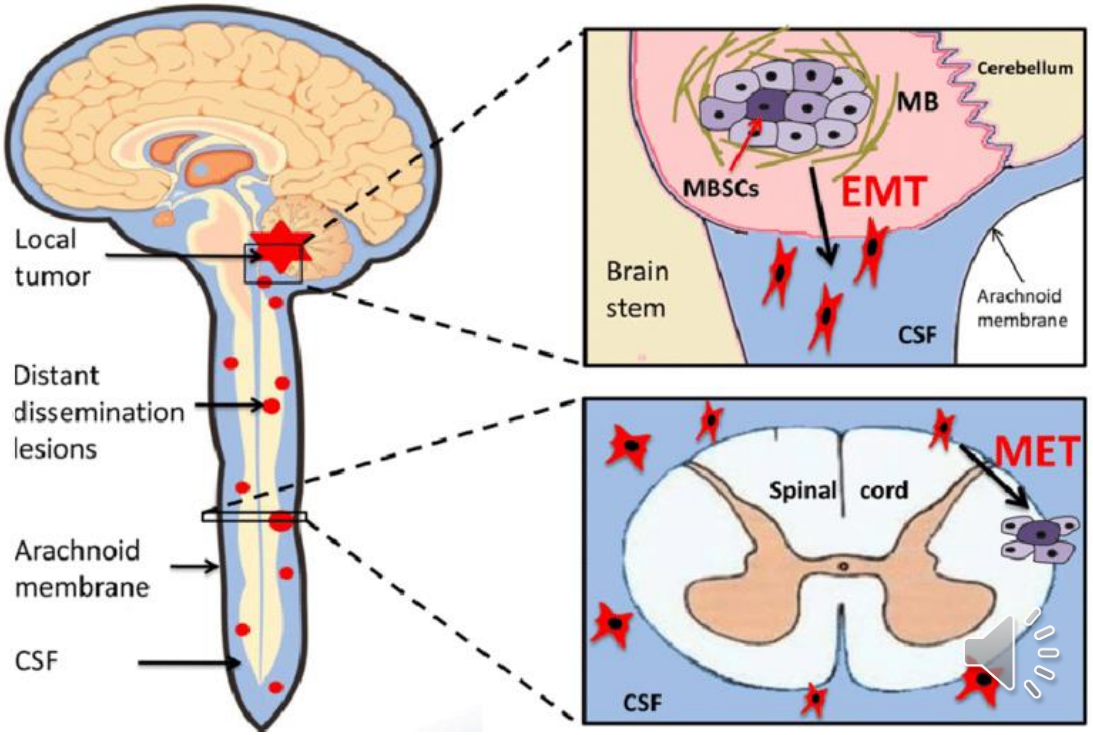
Medulloblastoma in children



- 20-25% childhood brain tumors
- Age 5-7 years
- Origin: roof fourth ventricle = does not invade cranial nerve nuclei
- Symptoms = hydrocephalus + compression brain stem & cerebellum
  - Headache
  - Dizziness, nausea & vomiting
  - Visual disturbances
  - Truncal ataxia
  - Sensory deficits
- Diagnosis: MRI
  - Rule out leptomeningeal spread
- Treatment: surgery + chemo + radiotherapy (only if child > 5 years of age)

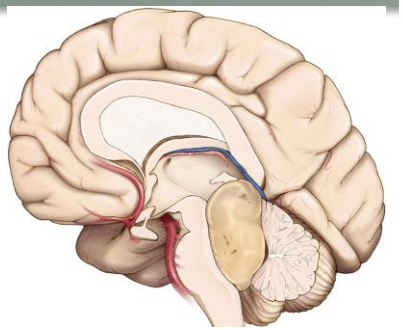


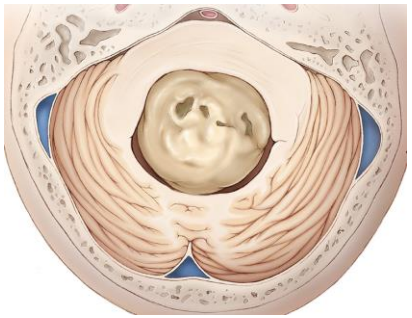


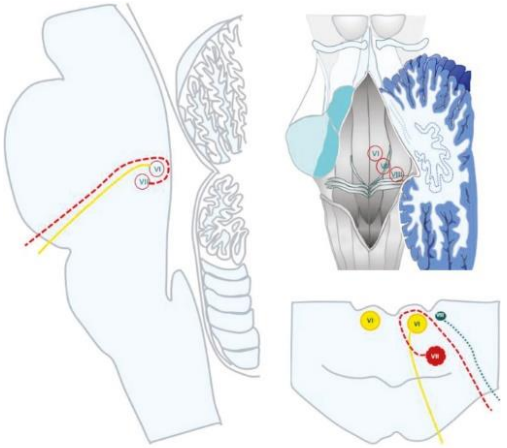


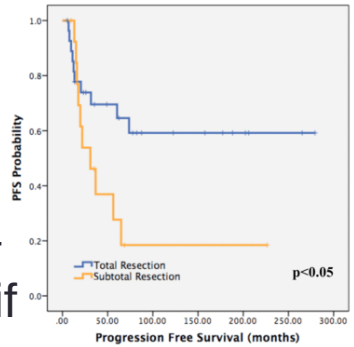
Ependymoma in children

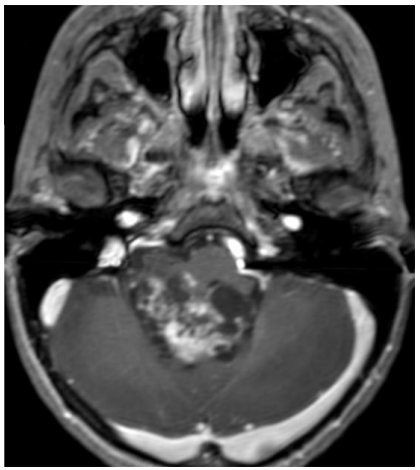
- 90% are intracranial
  - ↑posterior fossa
- Origin: floor fourth ventricle = cranial nerve deficits
- Diagnosis: MRI
  - Rule out leptomeningeal spread
- Treatment:
  - Surgery + chemo + radiotherapy (only if > 5 years age)





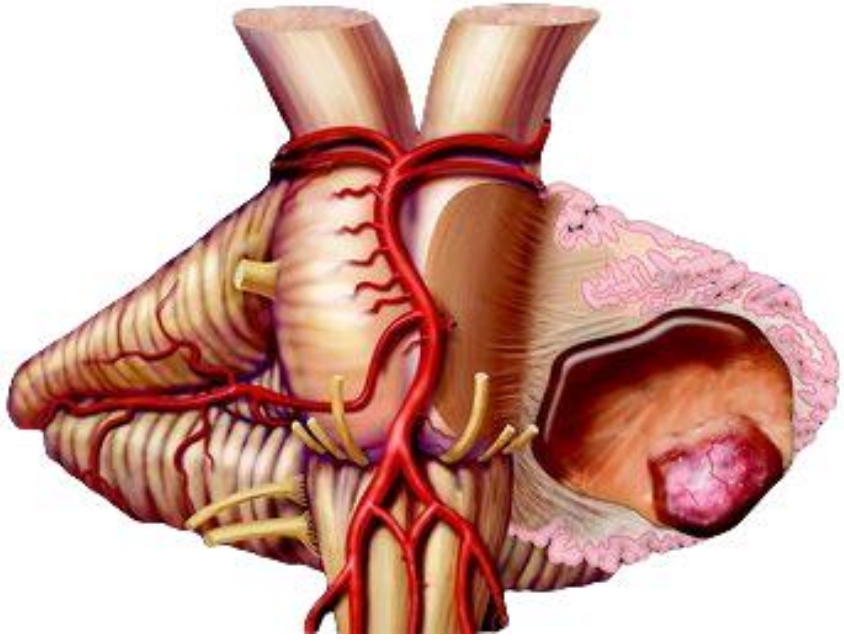


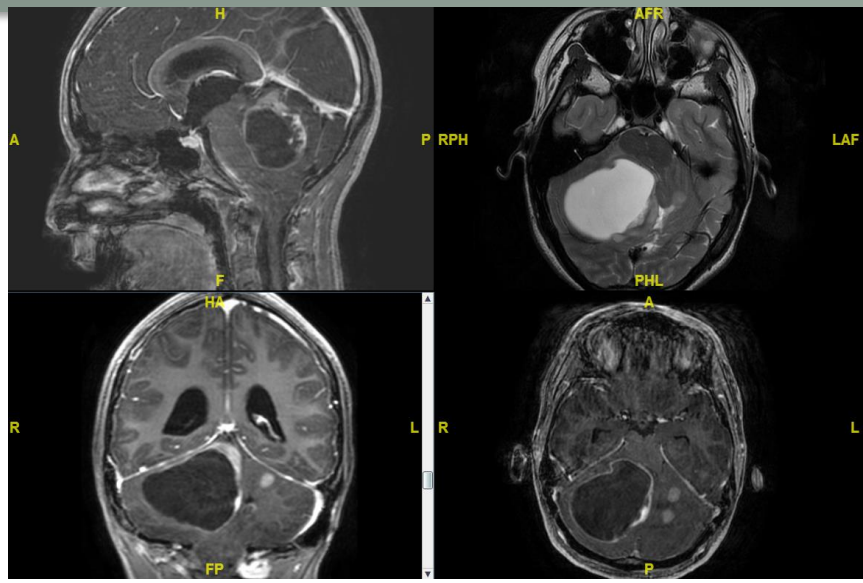


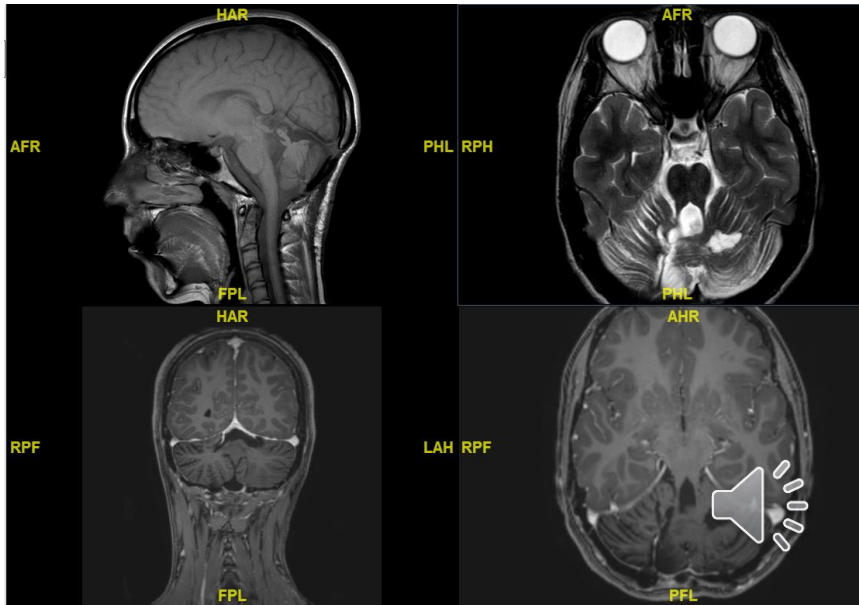


Cystic cerebellar astrocytoma

- The most common glial tumor in children
- Benign
- Only the nodule is tumoral
  - Nodule excision cures the tumor

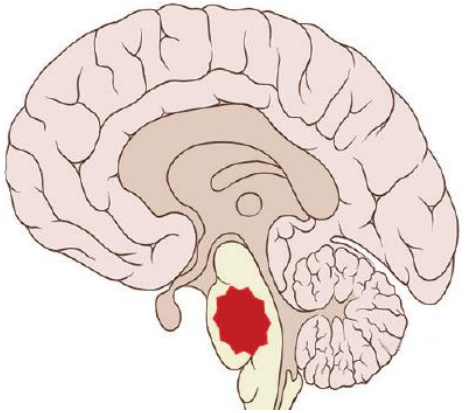






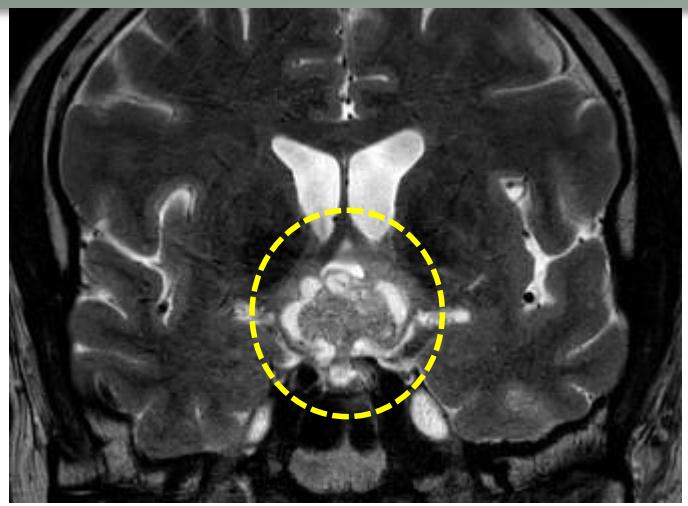
Infiltrating brain stem glioma

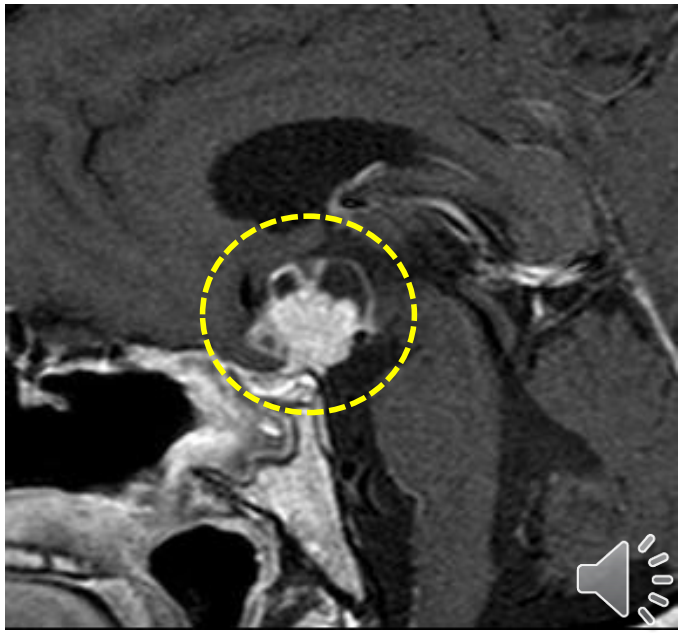
- 10% gliomas in children
- Infiltrating
- Symptoms
  - Cranial nerve deficits
- Treatment: palliative radiotherapy
- Prognosis: gloomy



Craniopharyngioma

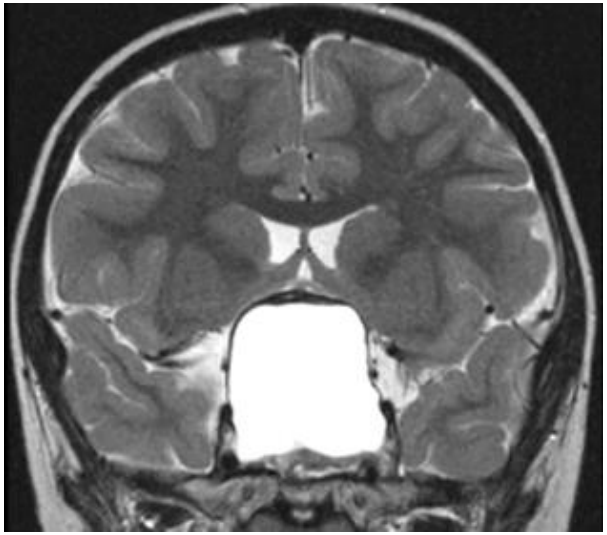
- 2.5 - 4 % brain tumors
- 60 % patients < 16 years old
- ↑ frequent in ♂
- Localization: usually sellar/parasellar
  - 75 % suprasellar
- Clinical features
  - Visual disturbances
  - Hypothalamic-pituitary axis dysfunction
  - Intracranial hypertension
  - Hydrocephalus
  - Cognitive impairment

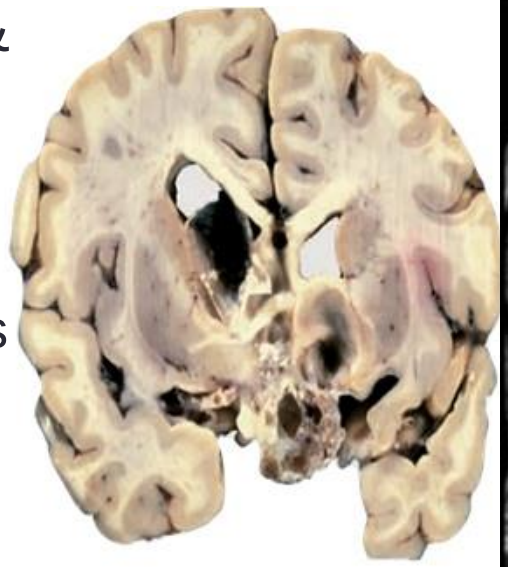


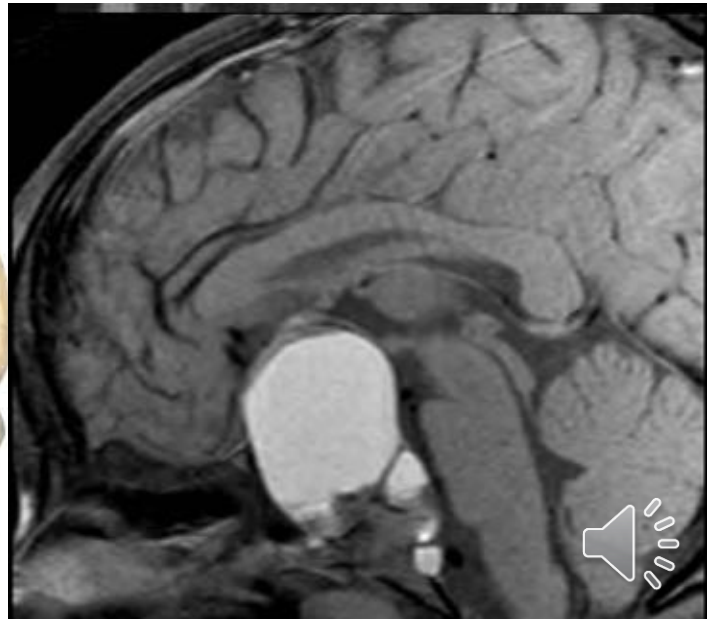


Craniopharyngioma

- Symptoms
  - Short stature
  - Hypopituitarism
  - Loss visual acuity/visual fields
- Cholesterol cysts
- Benign tumor but attached to chiasm & vessels & infiltrates hypothalamus
  - Incomplete resections = frequent recurrences = need for radiation therapy

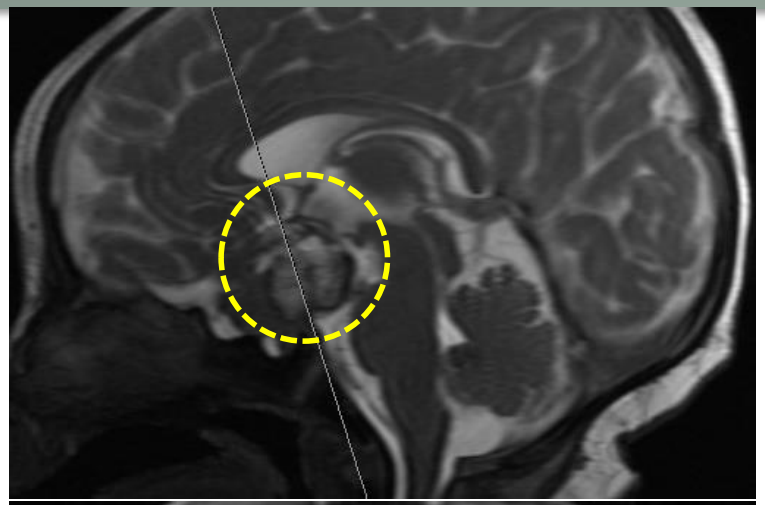


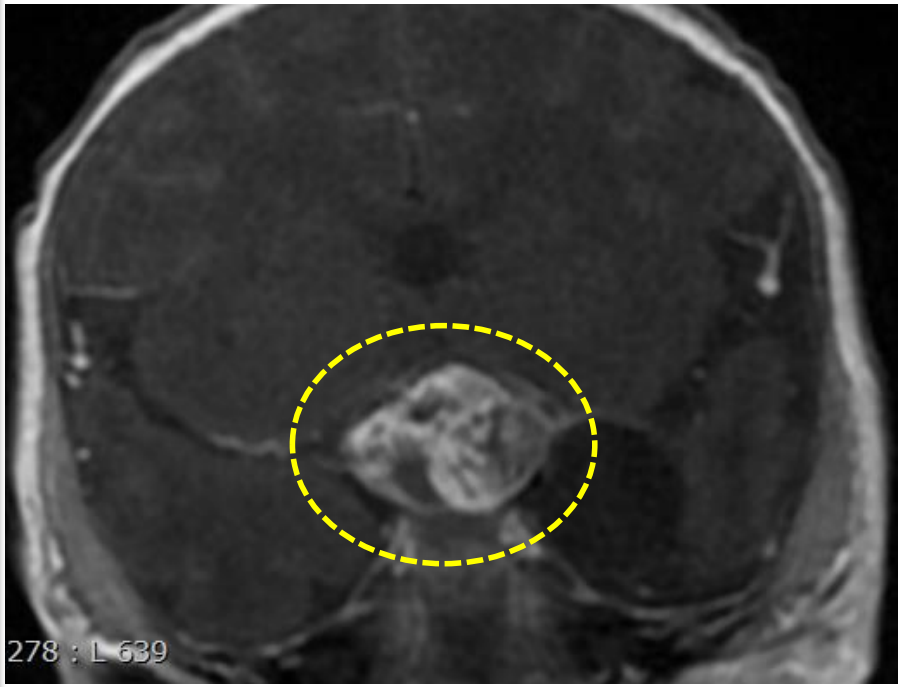


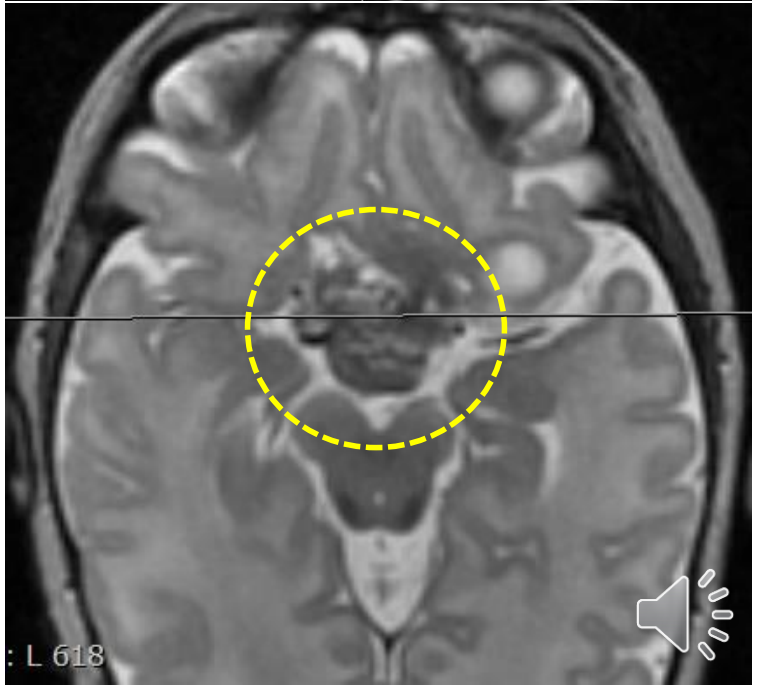


Suprasellar craniopharyngioma

- Newborn 39 weeks
- Visual disturbances
- Suprasellar cystic lesion

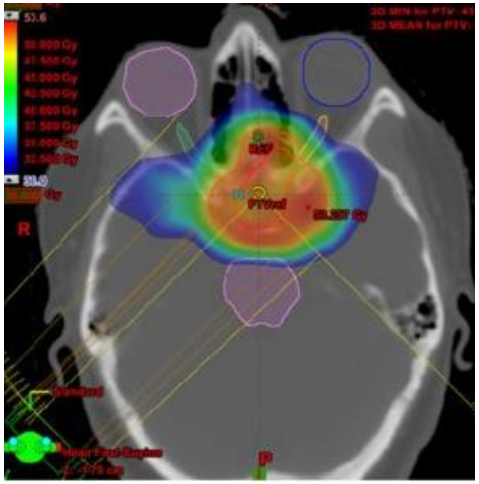


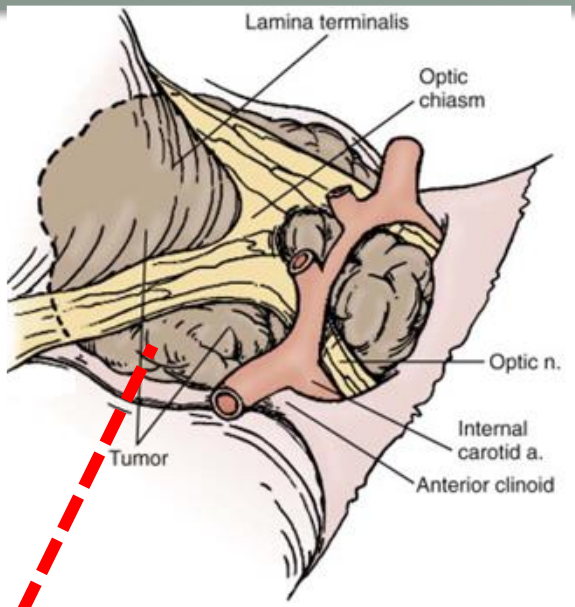


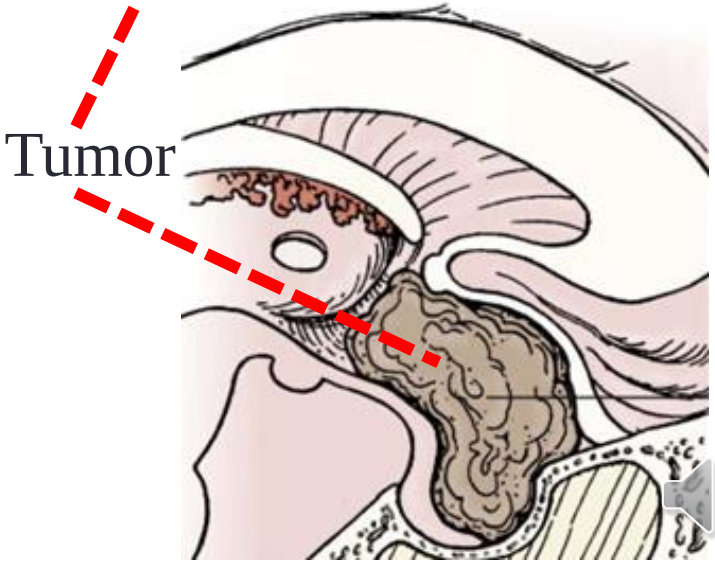


Craniopharyngioma

- Treatment
- Radical removal
- Radiotherapy?
  - Patient age > 5 years
  - Radiosurgery
- Intratumoral chemotherapy?

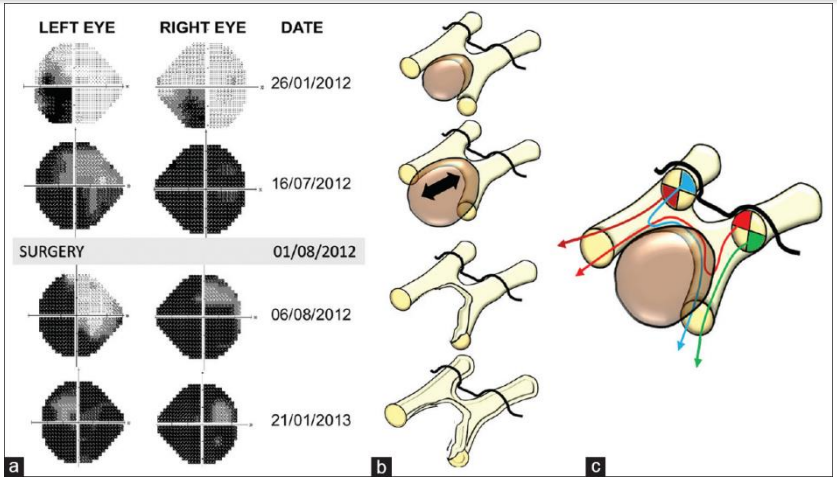






Craniopharyngioma: quality of life

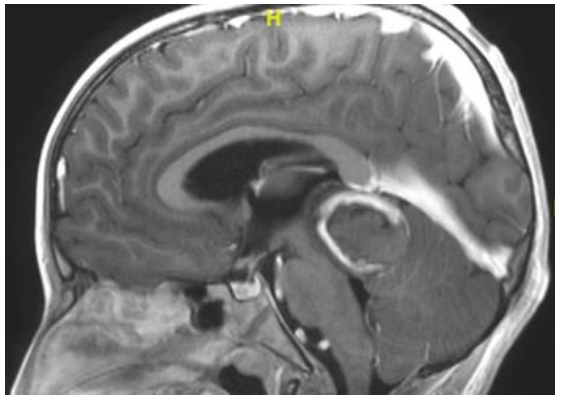
- Vision loss
- Endocrine deficits = short stature
- Neurocognitive deficits (memory loss)
- Motor deficits
- Epilepsy
- Result: good survival rate but long-term ↑ morbidity = ↑reduction in quality of life



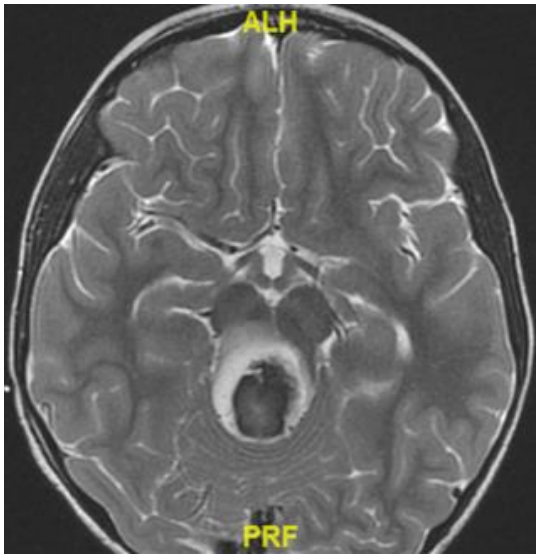
- Karavitaki N, Brufani C, Warner JT, et al: Craniopharyngiomas in children and adults: systematic analysis of 121 cases with long-term follow-up. Clin Endocrinol (Oxf) 2005; 62:397-409.
- Merchant TE: Craniopharyngioma radiotherapy: endocrine and cognitive effects. J Pediatr Endocrinol Metab 2006; 19(suppl 1):439-446. Dekkers OM, Biermasz NR, Smit JW, et al: Quality of life in treated adult craniopharyngioma patients. Eur J Endocrinol 2006; 154:483-489

Pineal germinomas(1)

- ♂ Age 7 years
- Precocious puberty 6 months evolution
- 126 cm & 30 kg (percentile 72)
- ↑Testosterone
- ↑β hCG blood & CSF
- Thoraco-abdomen-pelvic MRI NO findings



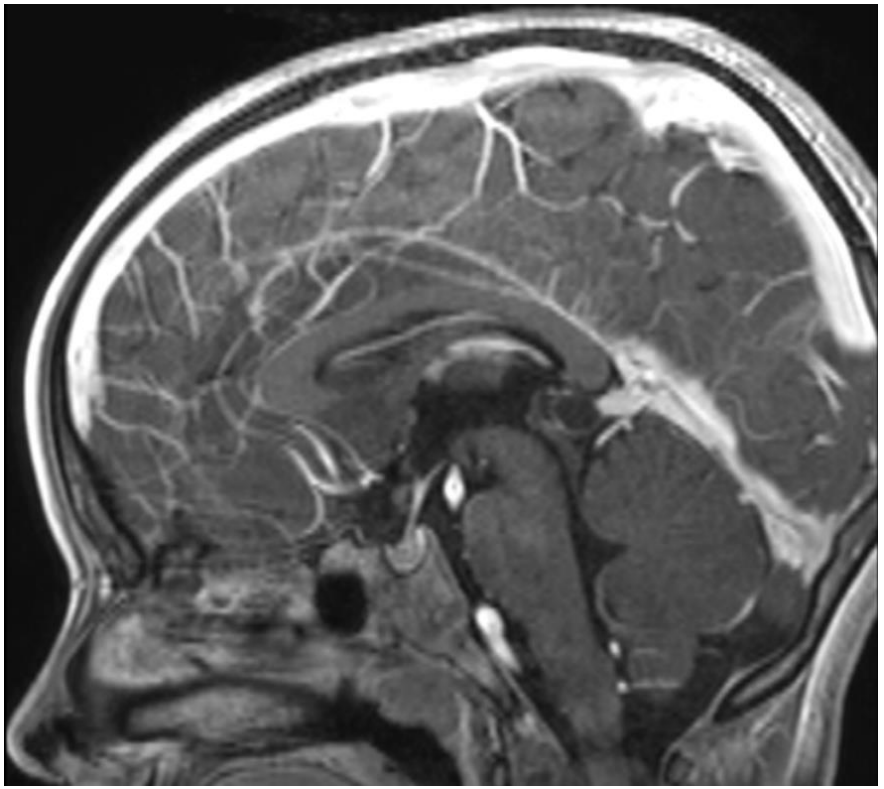
Setting sun eyes



Pre-op MRI

Germinoma pineal region (2)

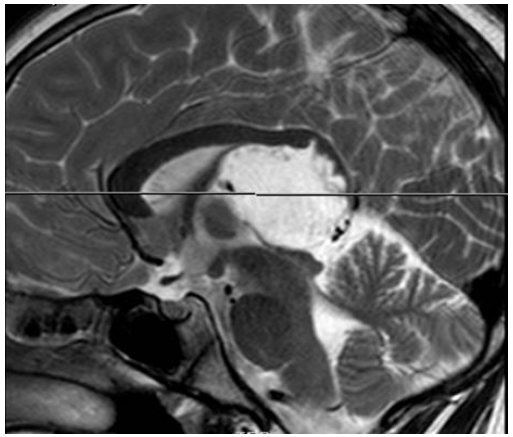
- Treatment
  - Ketoconazole to ↓testosterone
  - Cranial radiation therapy
  - Chemotherapy

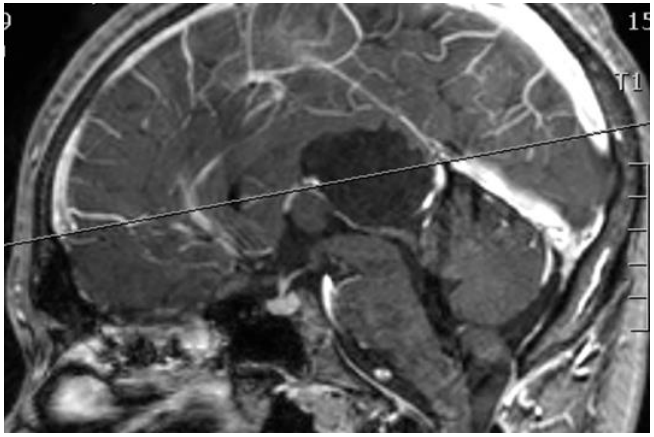


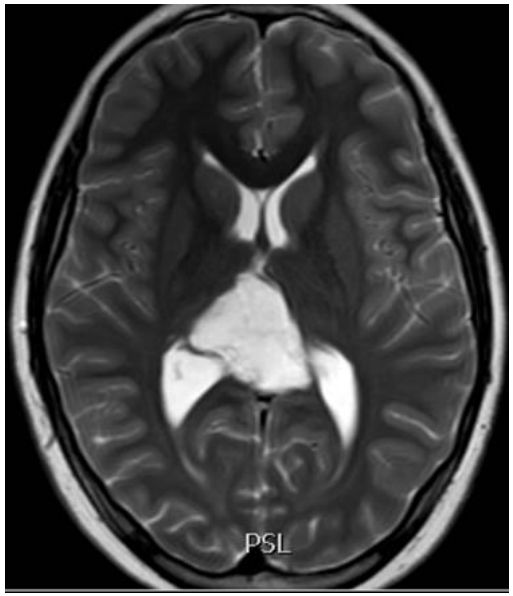
Post-treatment MRI

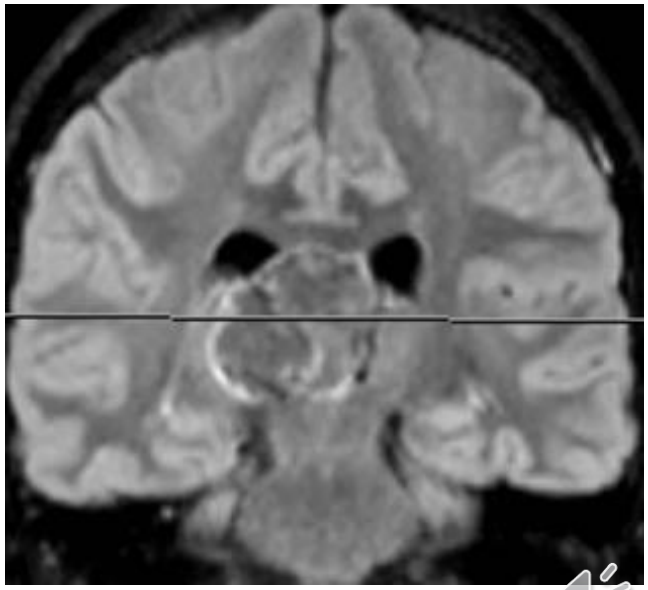
Non-germinomatous pineal region tumor

- Radioresistant
- Surgical removal
- 10 years ♀
- Symptoms
  - Precocious puberty + headache for 2 months



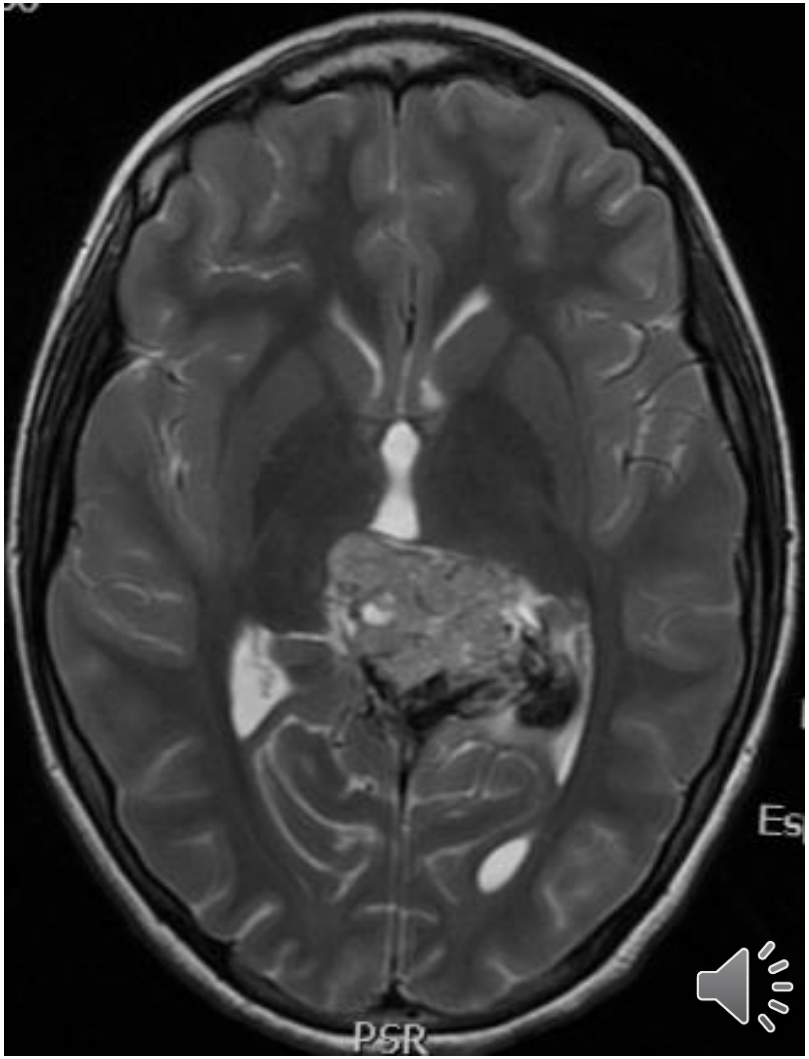






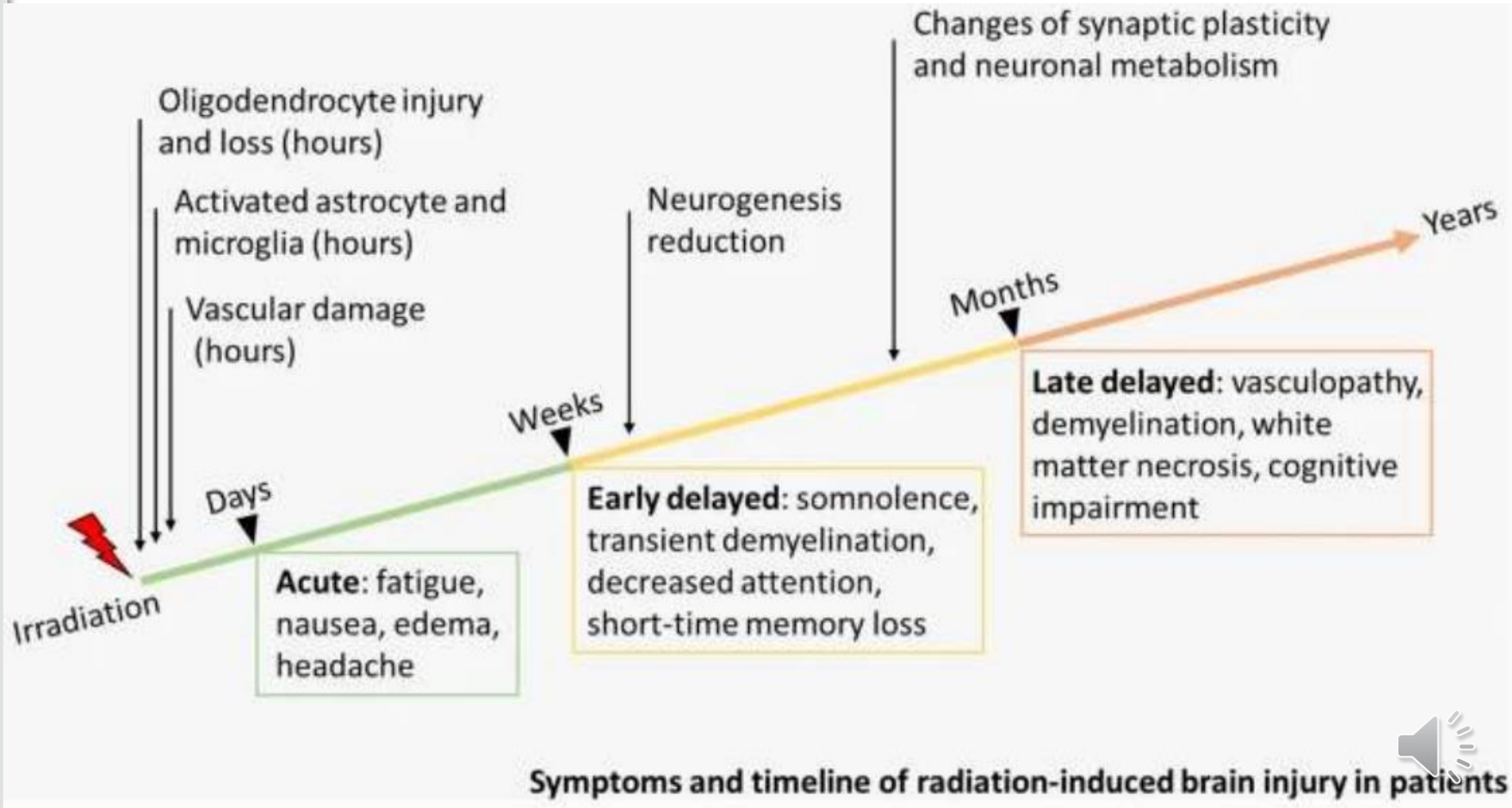
Non-germinomatous pineal region tumor

- Many histological types
- Challenging surgical removal
- 12 years ♂, 2 weeks headache + vomiting



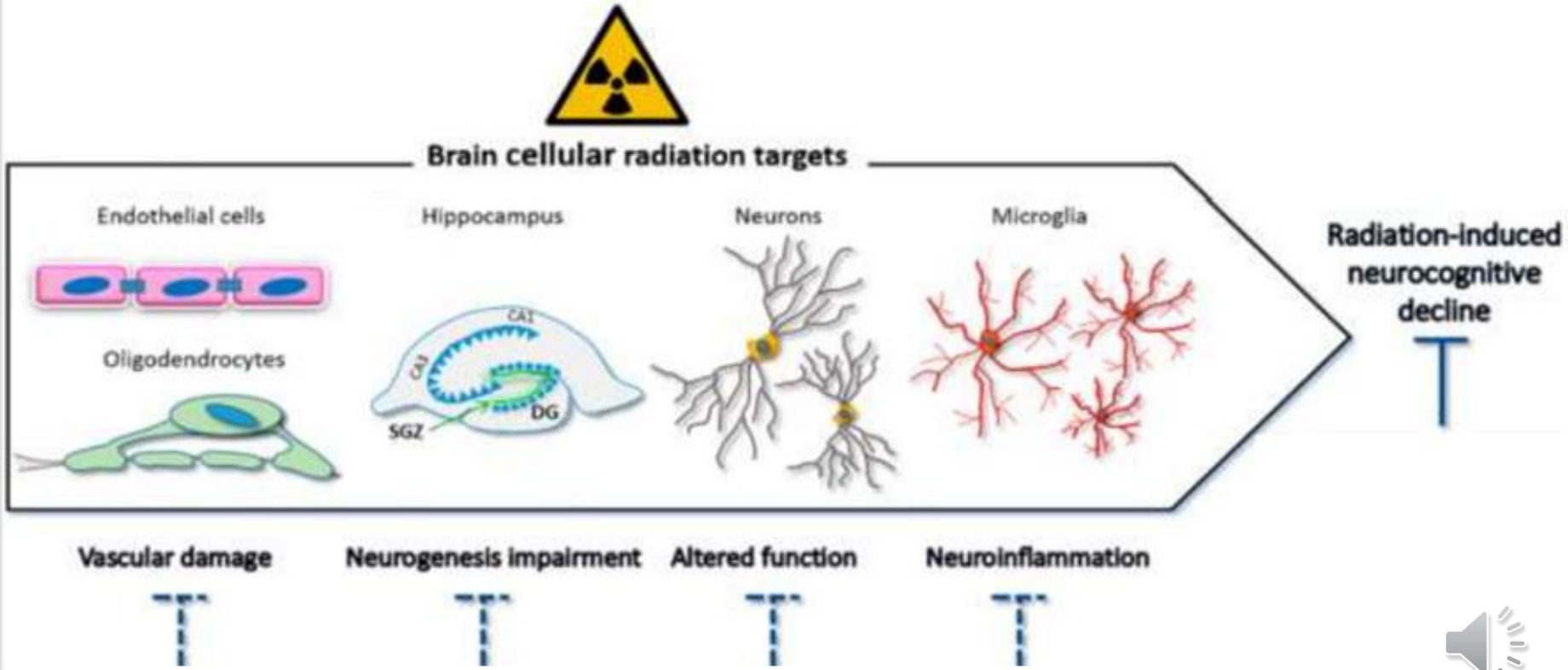
RADIOTHERAPY EFFECTS

- Progressive damage to different cell types = neurocognitive impairment in the intermediate & long term



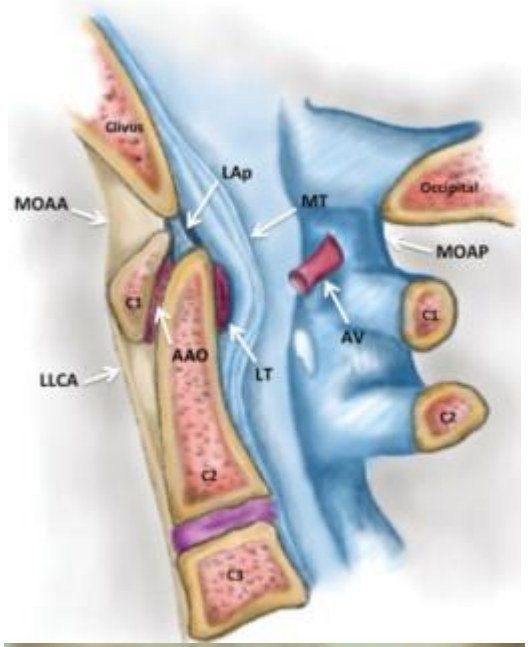
CRANIAL RADIOTHERAPY EFFECTS ON PEDIATRIC SURVIVORS

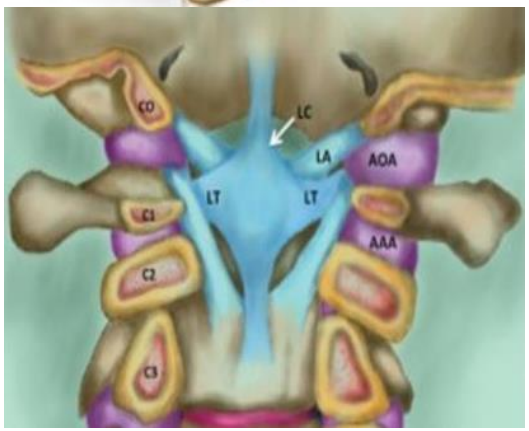
- The younger the age,↑↑progressive deterioration in reading, attention & behavior
  - Not in executive tasks (planning, organisation, and working memory)

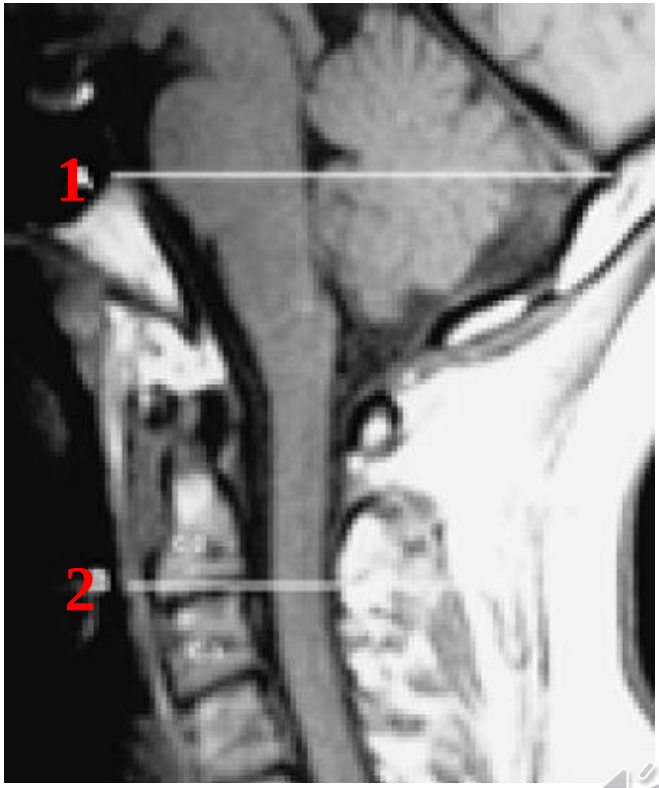


4. CRANIO-CERVICAL JUNCTION MALFORMATIONS

- Cranio-cervical junction
  - Area between (1) lower portion of occipital bone around foramen magnum and (2) first two cervical vertebrae
  - Funnel where the spinal cord attaches to the medulla and where the cerebellum rests





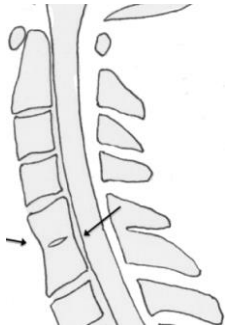


Cranio-cervical junction malformations

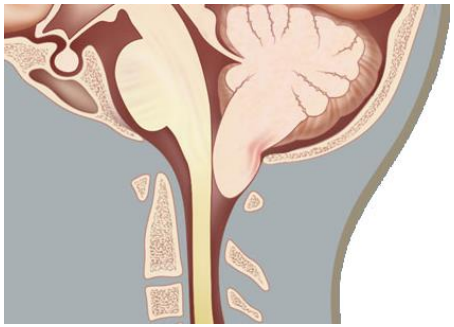
- **Bone malformations**
  - Platybasia
  - Basilar invagination
  - Craniocervical abnormalities
  - Klippel-Feil syndrome
- **Neurological malformations**
  - Arnold-Chiari malformations
  - Syringomyelia

Platybasia

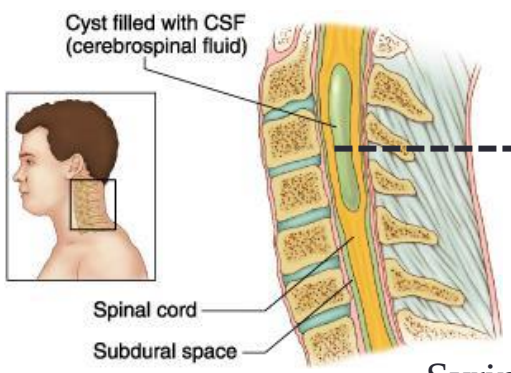
Basilar invagination

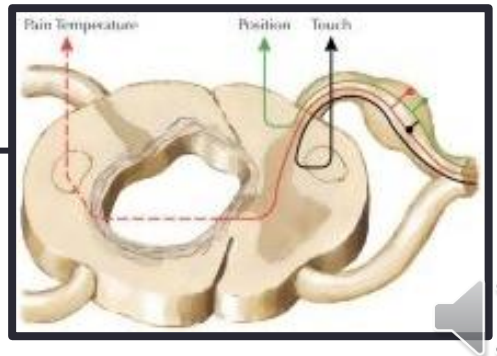


Klippel-Feil syndrome



Chiari I Malformation

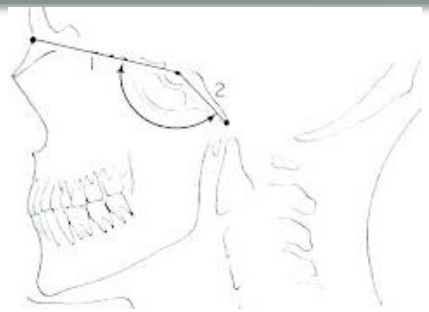




Syringomyelia

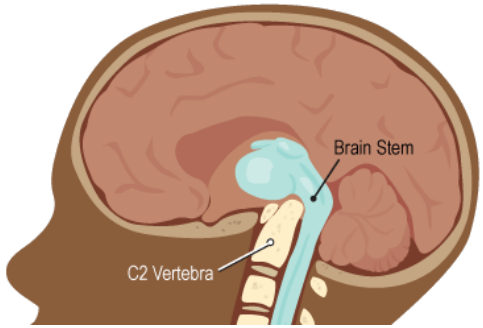
Bone malformations

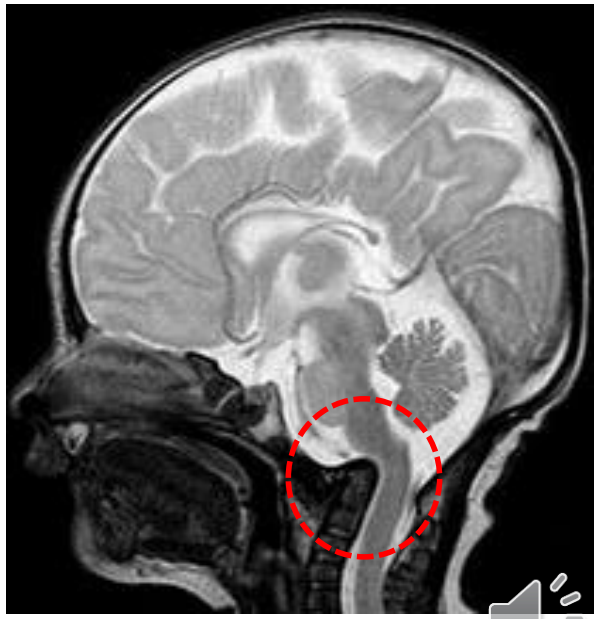
- **Platybasia**
  - Skull base flattening (basal or Welcher angle > 145º)
  - May be asymptomatic, but often associated with other abnormalities
- **Basilar invagination**
  - Skull base collapse over the cervical spine
  - Congenital or acquired (Paget's disease, tumors)
  - Symptoms
    - Occipital-cervical pain
    - Cervical spinal cord compression with tetraparesis



Platybasia

Normal

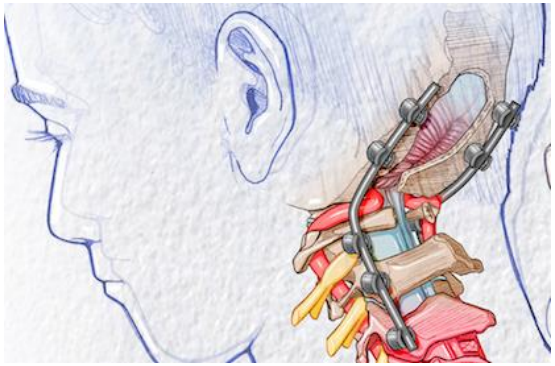




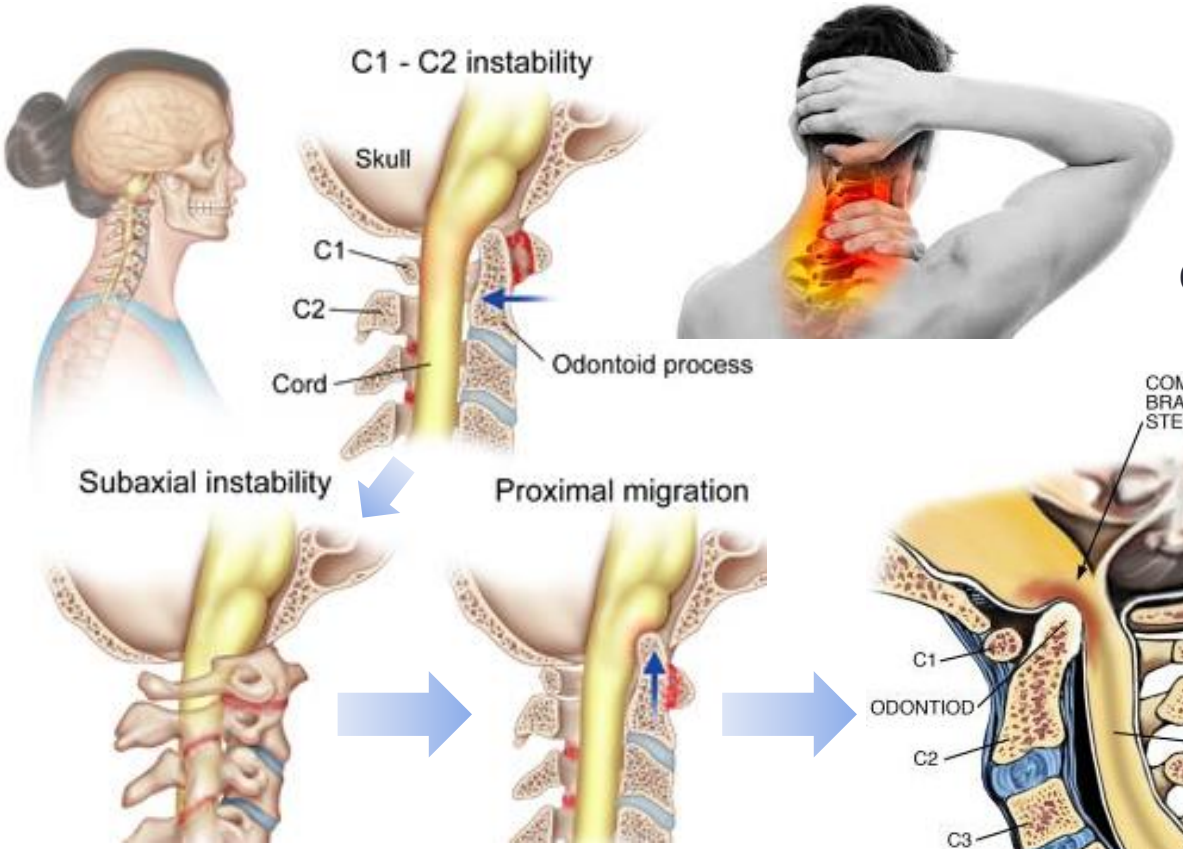
Basilar invagination

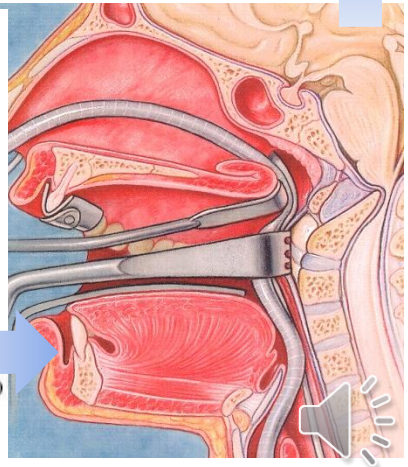
Bone malformations: basilar invagination

- Due to ligament instability and/or poor bone quality at the craniocervical junction
- Treatment surgical with double approach: transoral decompression + posterior arthrodesis



Craniocervical arthrodesis

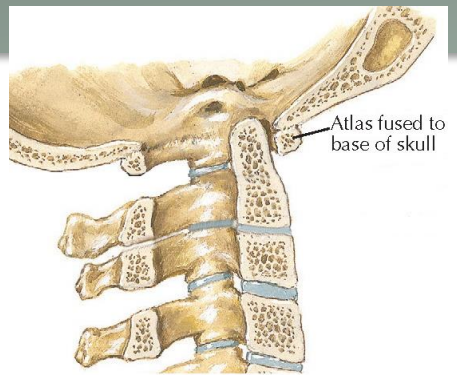




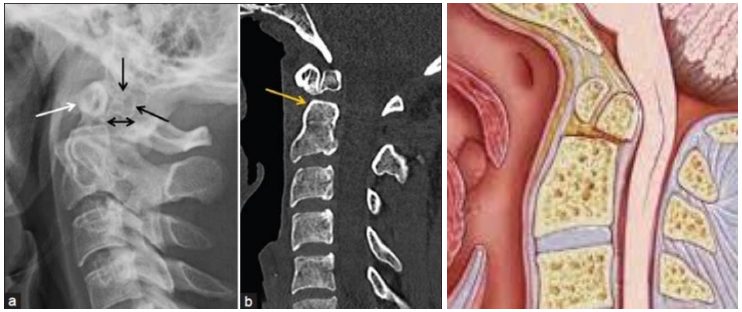
Transoral approach

Bone malformations: craniocervical junction abnormalities

- **Craniocervical junction abnormalities**
  - Atlas or odontoid malformations
  - Atlas-axis dislocation
  - Treatment: posterior occipital-cervical arthrodesis
- **Klippel-Feil syndrome**

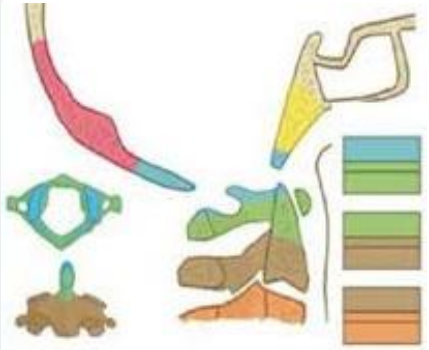


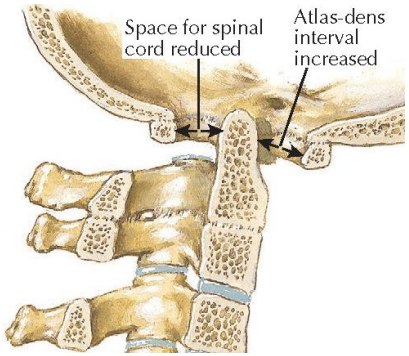
Atlas occipitalization



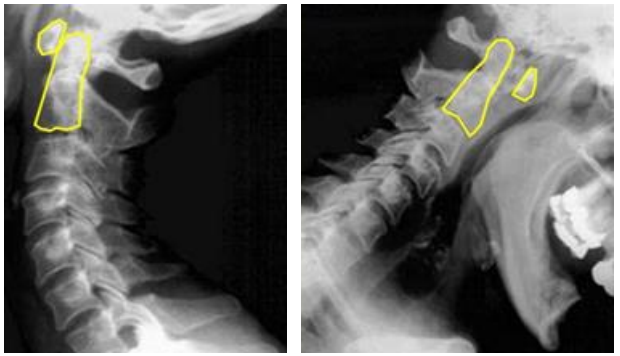
Odontoid malformation

C_1-C_2 arthrodesis

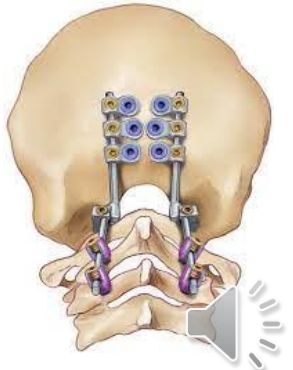




Craniocervical junction bone formation



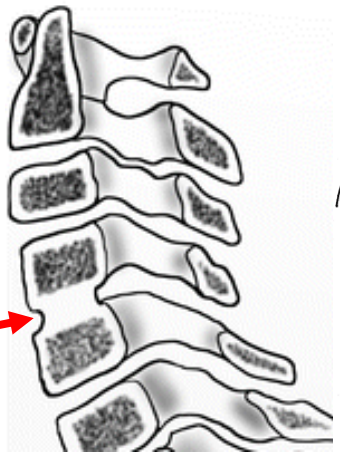
Atlas-axis dislocation

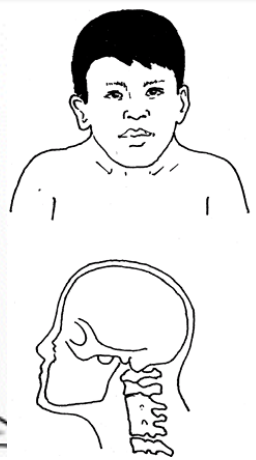


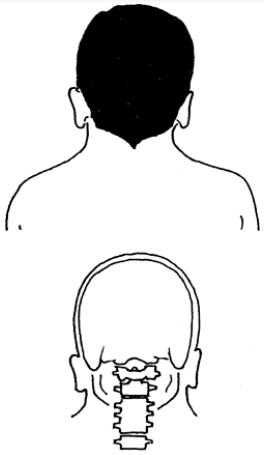
Arthrodesis occiput-C_1-C_2

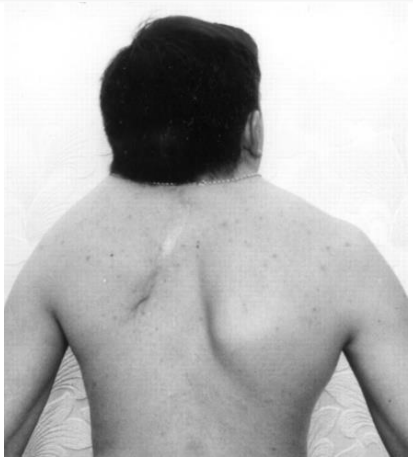
Bone malformations: Klippel-Feil syndrome

- **Craniocervical junction abnormalities**
- **Klippel-Feil syndrome**
  - Congenital fusion 2/+ cervical vertebrae (unsegmented)
  - Low hair implantation, short neck, cervical spine movement limitation
  - Associated with bone, kidney, cardiac abnormalities





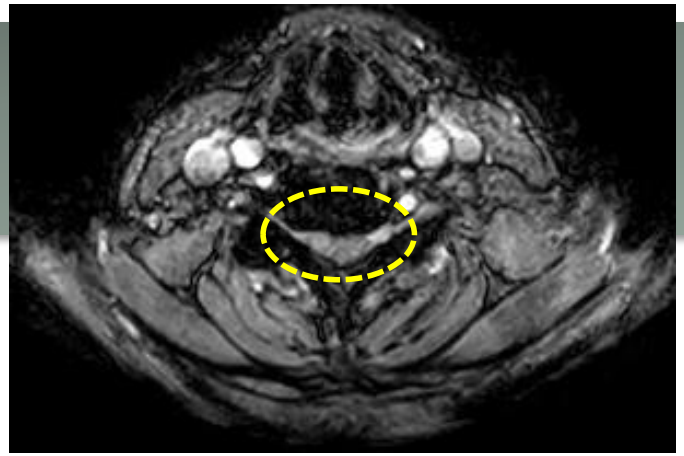


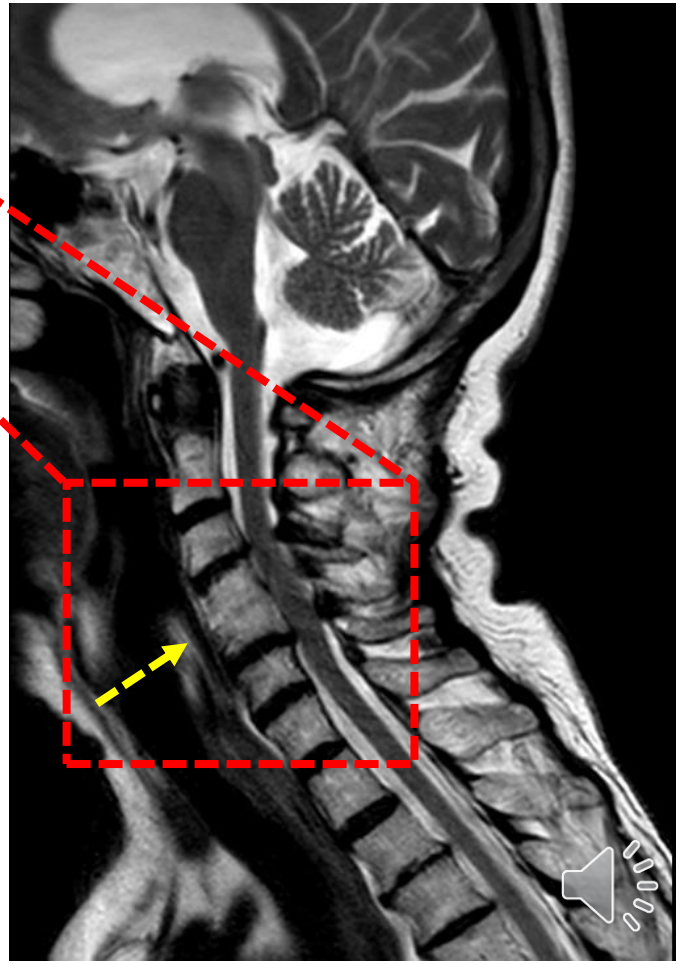


Klippel-Feil syndrome

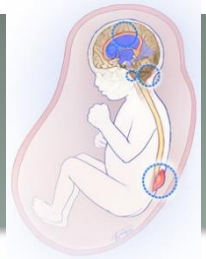
- Sometimes slight mental retardation
- Usually asymptomatic

♀53 years, cervical myelopathy

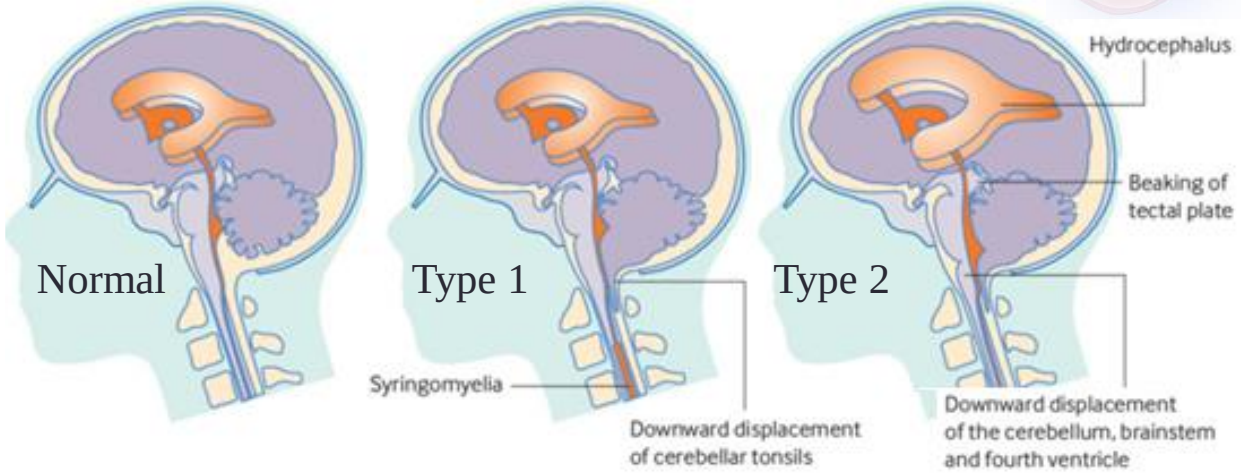


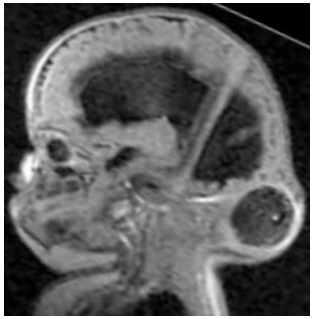


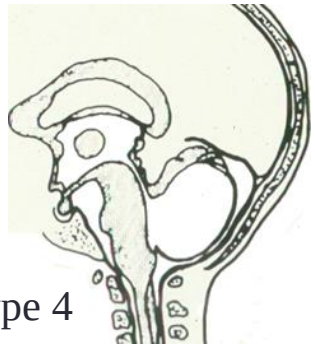
Neurological malformations

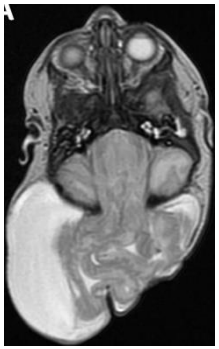


- **Arnold-Chiari malformations**
  - Cerebellar tonsil herniation
  - Types
    - Type I = adults
    - Type II = children, associated with spina bifida and hydrocephalus
    - Type III
    - Type IV
  - Associated with other skull base and neurological malformations (syringomyelia and syringobulbia)



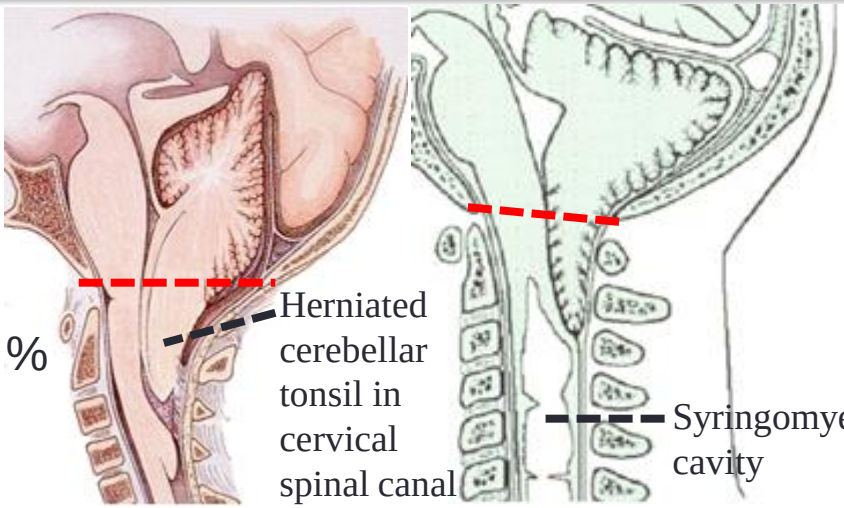


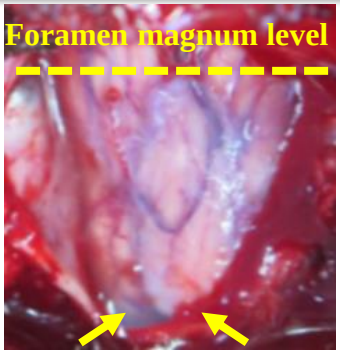




Neurological malformations: Chiari type 1 malformation

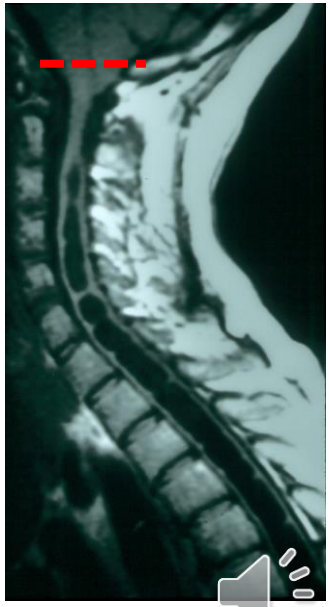
- Cerebellar tonsil herniation > 3 mm below foramen magnum
  - 5-10mm→symptoms 30 %
    - Association with syringomyelia > 20%
  - >12 mm ‘always’ symptoms
- Clinical features (1)
  - Vague symptoms due low cranial nerve compression
  - Nuchal headache that ↑cough, sneeze, Valsalva & sexual intercourse
  - Hydrocephalus
  - Syringomyelia





Herniated cerebellar tonsils

Chiari I malformation



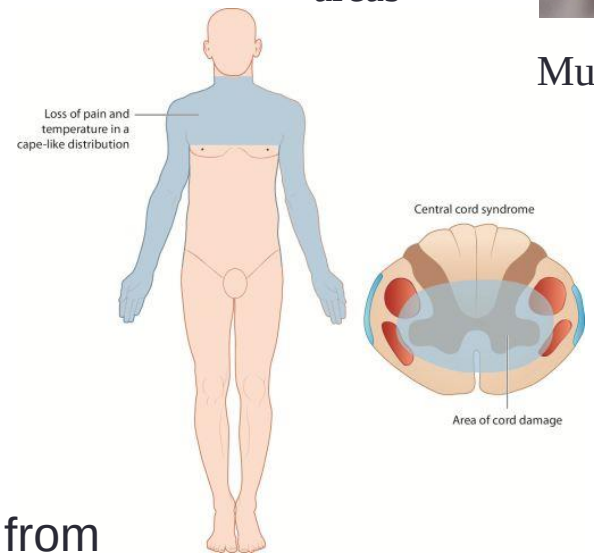
Chiari I malformation + syringomyelia

Chiari type 1 malformation: features

- Sleep apnea, ‘clumsiness’
- **Syringomyelia**
  - Paresthesias & thermoalgesic anesthesia in a cape-like distribution
  - Atrophy & motor deficits hands & arms
  - Myotatic reflex abolition
    - Due to 2nd motoneuron damage
  - Spinal tract involvement
    - Due to intramedullary cysts and/or cranio-cervical junction malformation
- Cranial nerve involvement
  - Diplopia, **dysphagia, dysphonia**
- Brainstem: **nystagmus**
- Cerebellum
  - Cerebellar syndrome (11%)
  - Truncal ataxia
- **Lhermitte's sign** = electric shock from nape to arms when flexing the neck

Burns in insensitive areas

Muscle atrophy in hands

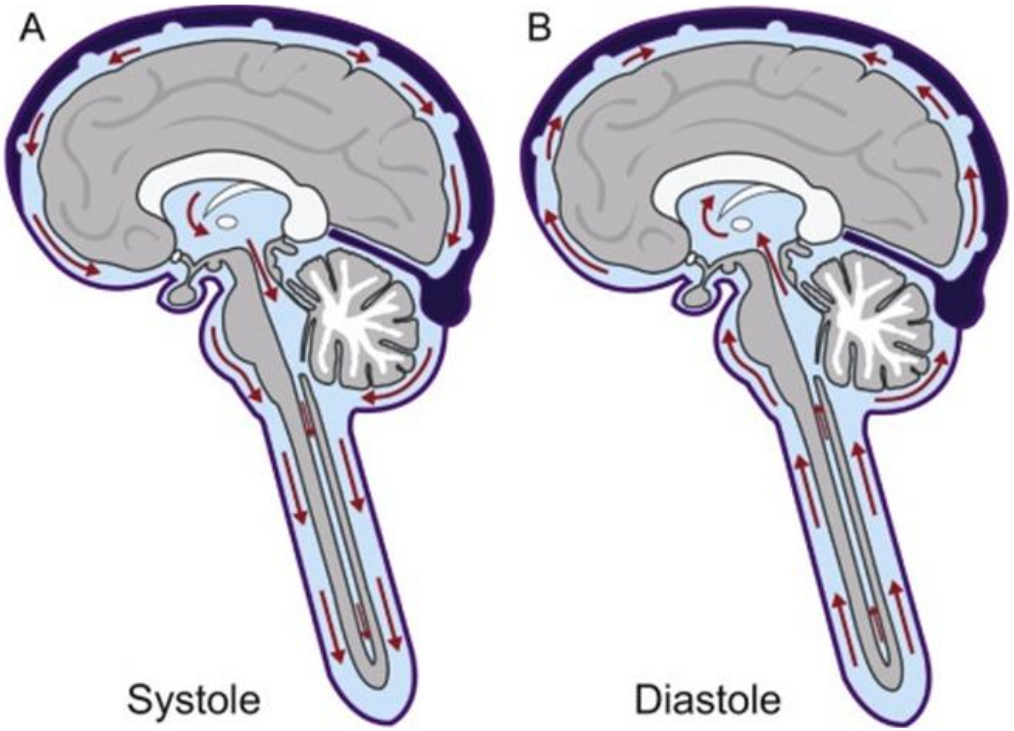


Cape-like distribution anesthesia

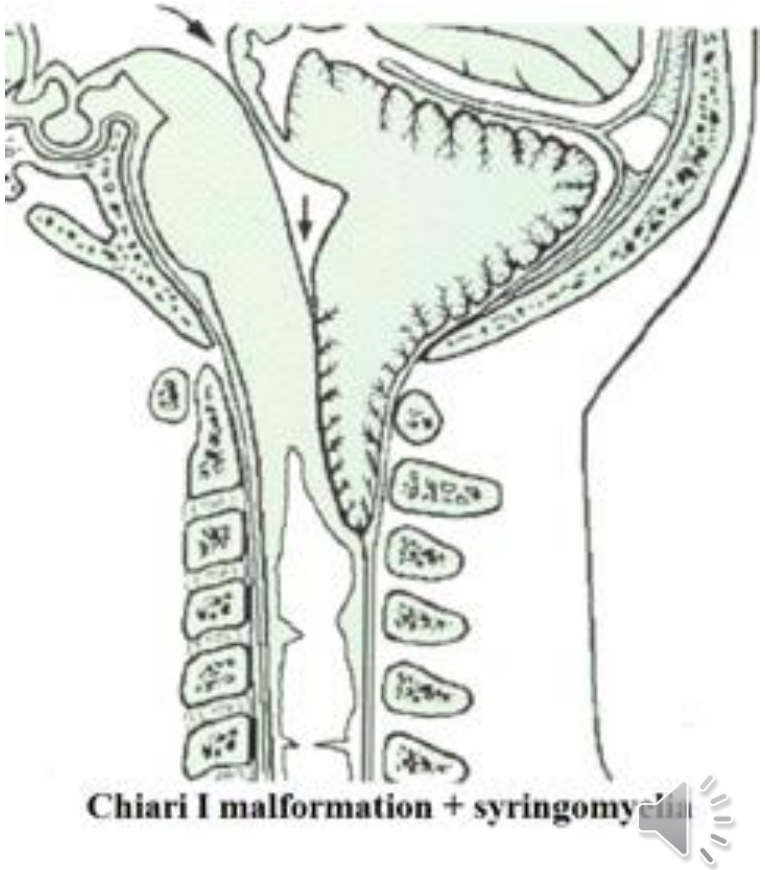
Lhermitte’s sign

Chiari type 1 malformation: Pathophysiology

- CSF cannot circulate properly because of the ‘plug’ created by the cerebellar tonsils



Normal

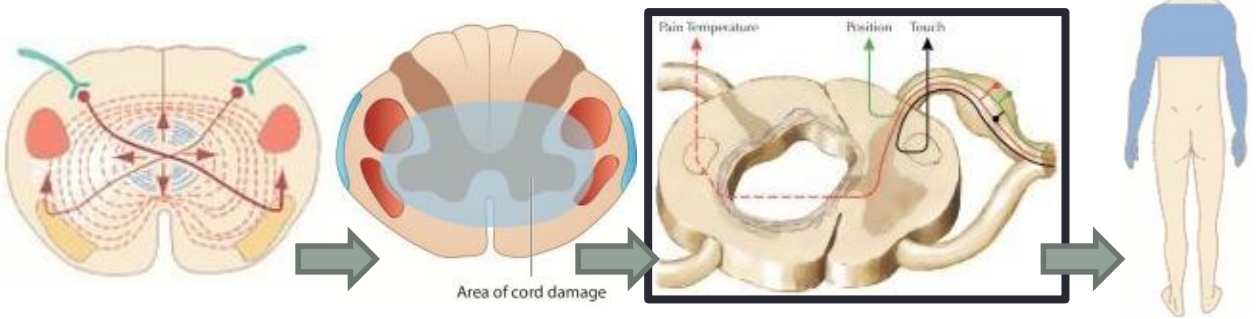


Chiari I malformation + syringomyelia

Syringomyelia

Syringomyelic intramedullary cyst

- Intramedullary cystic dilation
- Age presentation 30-40 years age
- ***Thermoalgesic dissociation*** in a cape-distribution
- ***Muscle atrophy in hands & upper limbs***
  - *Spastic paraparesis if long evolution*
- Localization
  - *Cervical-dorsal*
  - *Bulb: syringobulbia*
    - *Ninth to twelfth cranial nerve involvement*
- ***Progressive neurological deficits NEVER improve***



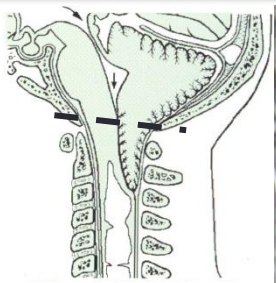
Thermoalgesic dissociation in cape-like distribution

Syringomyelic hands

Cervical-dorsal syringomyelia Syringobulbia

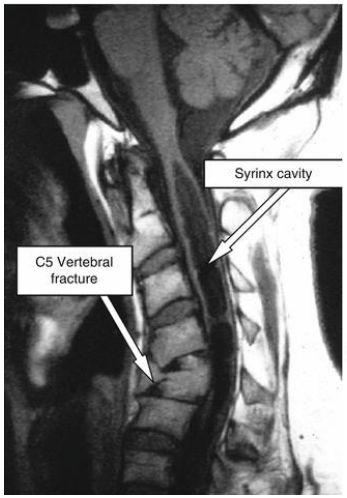
Syringomyelia: causes

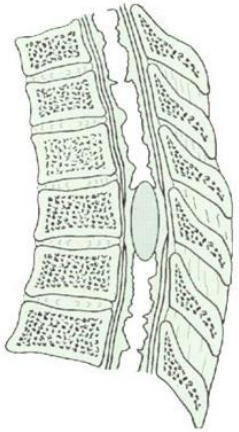
- **Cranio-cervical junction abnormalities**
  - Chiari type 1 malformation, basilar invagination or other cranio-cervical junction malformations (80%)
  - Foramen magnum tumors
  - Meningitis, hemorrhages
- **Spinal cord injuries**
  - Spinal cord trauma (5)%
  - Intramedullary hemorrhage
  - Tumors, vascular malformations
  - Spinal cord arachnoiditis
    - Tuberculous meningitis
- **Hydrocephalus** 1%



Chiari type 1 malformation

Basilar invagination





Spinal cord trauma

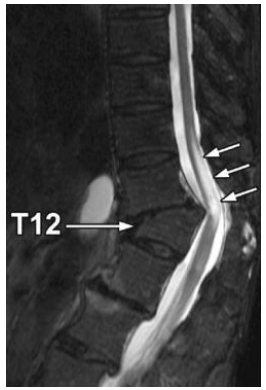
Spinal cord tumor

Syringomyelia: diagnosis

- **Symptoms**
  - The patient does not notice hot and cold water on upper limbs when showering
- **MRI**
  - Cranio-cervical junction malformations
    - Neurological (Chiari type I malformation)
    - Bony (basilar invagination),…
  - Tumors, spinal cord trauma
  - Helps to depict syringomyelia cavity extension

Chiari type 1 Malformation

Basilar invagination



Spinal cord trauma

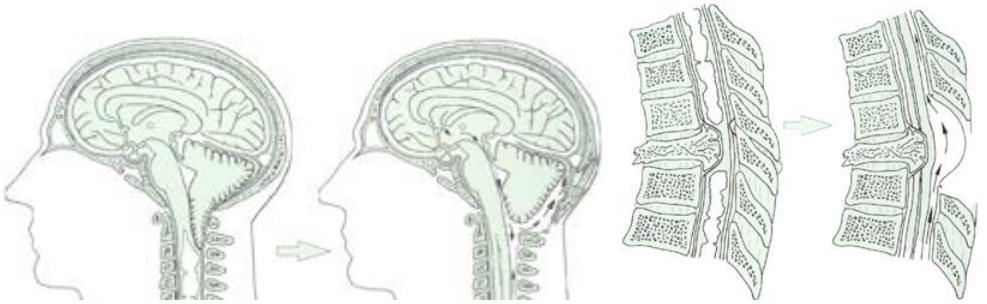
Hydrocephalus + Chiari 1 malf.

Intramedullary tumor

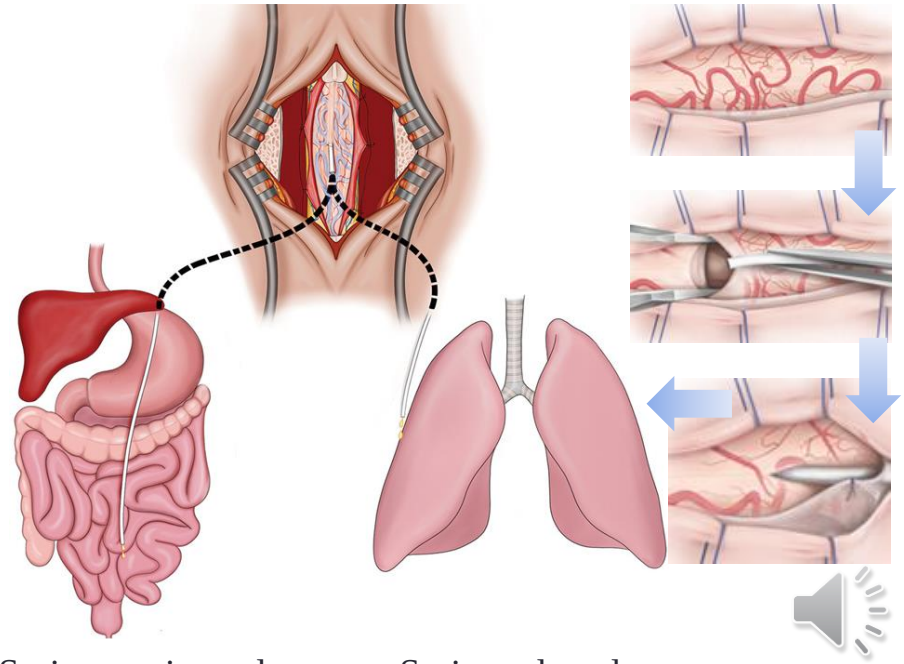
Cervical-dorsal extension

Syringomyelia: treatment

- **Treatment**
  - Chiari 1 malformation = posterior fossa decompression
  - Tumor = tumor removal
  - Post-traumatic = decompression + duraplasty
  - Arachnoiditis = syringo-pleural/peritoneal shunt
  - Hydrocephalus = ventricle-peritoneal shunt
- **Prognosis**
  - Stops disease but neurological deficits not reversed
  - Posterior fossa decompression = good results
  - Syringopleural/peritoneal shunt = catheter obstruction = frequent reoperations



Decompression Chiari 1 malformation Posttraumatic



Syringoperitoneal Syringopleural

SUMMARY CONCEPTS PEDIATRIC PATHOLOGY SEMINAR

- **Hydrocephalus**
  - Intracranial CSF increase
  - Risk: brain atrophy, psychomotor retardation & blindness
  - Valve selection appropriate to patient needs
- **Craniosynostosis**
  - Simple = aesthetic defect
  - Syndromic = other malformations
    - Complex treatment
- **Spina bifida**
  - Surgical repair needed
  - Prognosis according to nerve tissue involvement
  - Follow-up: due to post-repair scar, monitor spinal cord injury by retethering
- **Cranio-cervical junction malformation**
  - Symptomatic Chiari type 1 ⇨ cranio-cervical junction decompression
  - Syringomyelia ⇨ treatment cause

ANY QUESTIONS?

vivava@uv.es
pedro.roldan@uv.es