Spinal dysraphism (spina bifida)

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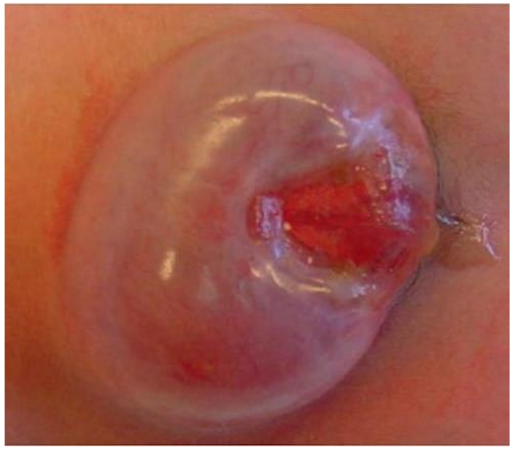
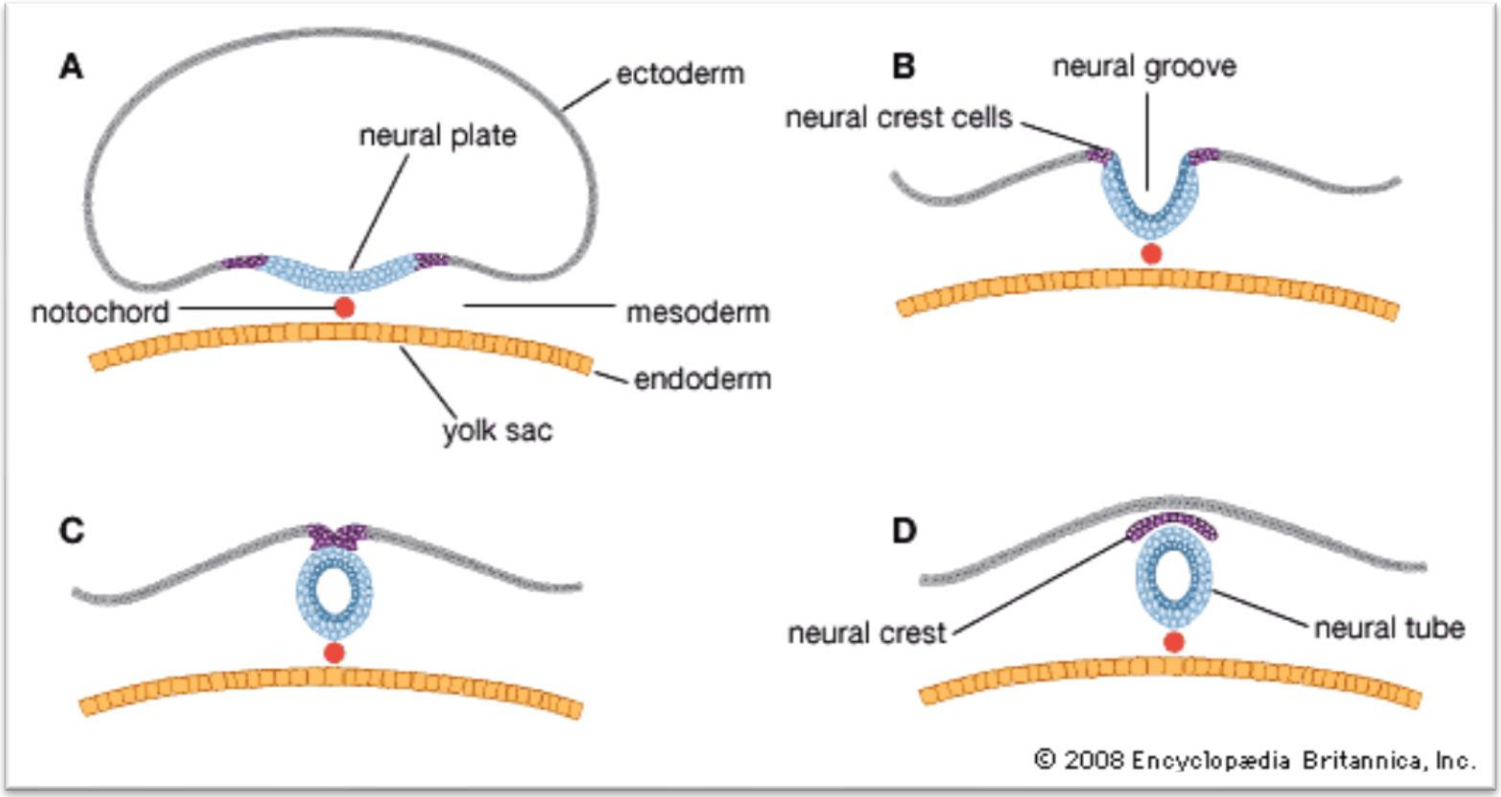
Definition



- A major birth defect and a type of neural tube defect that involves an opening in the vertebral column caused by the failure of the neural tube to close properly during embryonic development

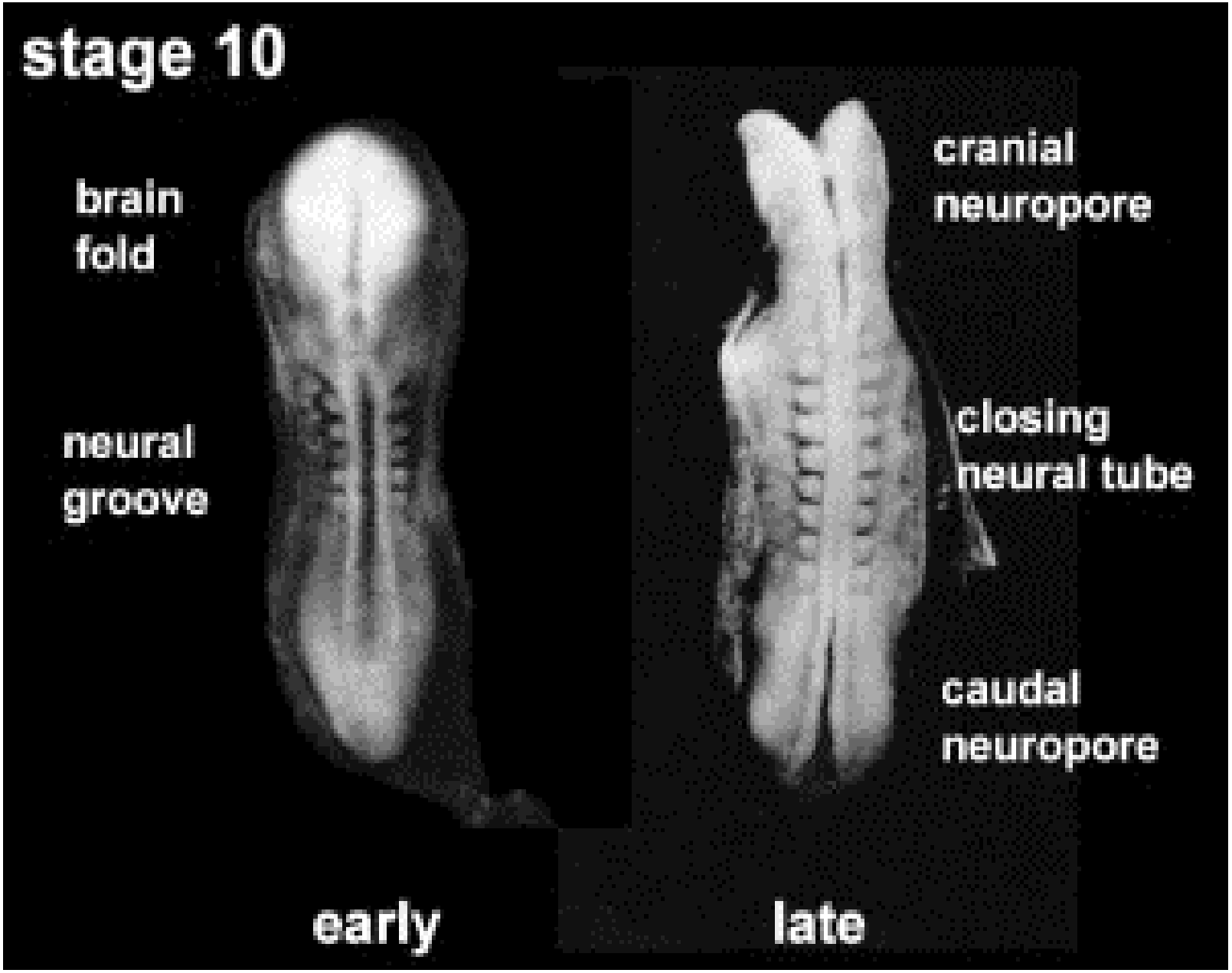
Embryology

- The process of primary neurulation occurs in the first month of gestation.



- The early central nervous system begins as a simple neural plate that folds to form a neural groove and then neural tube. This early neural is initially open initially at each end forming the neuropores(cranial and caudal).
- Failure of these opening to close contributes a major class of neural abnormalities (neural tube defects)
- Neural development begins quite early, neural plate and groove appear at 18 days and complete closure of neural tube by the end of the 4^{th} week.

- Within the neural tube stem cells generate the 2 major classes of cells that make the majority of the nervous system : neurons and glia. Both these classes of cells differentiate into many different types generated with highly specialized functions and shapes.


stage 10
brain fold
neural groove
cranial neuropore
closing neural tube
caudal neuropore
early
late

Types of spina bifida

- **spina bifida occulta**

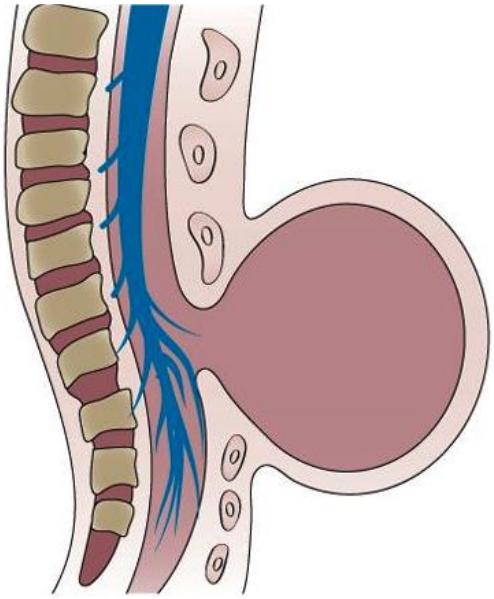
Congenital absence of a spinous process and variable amounts of lamina. No visible exposure of meninges or neural tissue .

- **spina bifida aperta** (*aperta* from the Latin for “open”) or **spina bifida cystica**.

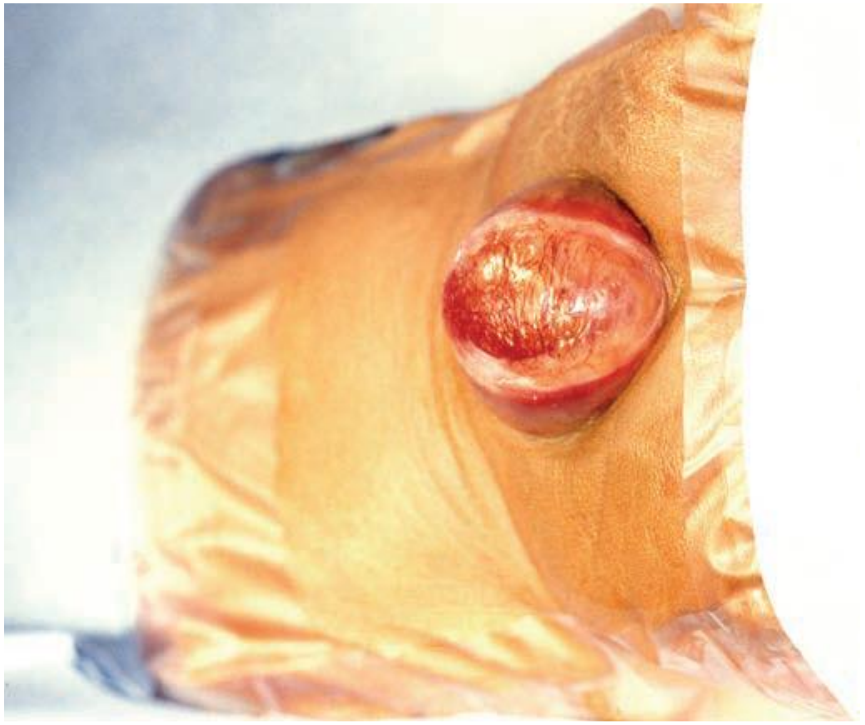
Spina bifida cystica

- ✓ Meningocele :Congenital defect in vertebral arches with cystic distension of meninges, but no abnormality of neural tissue. One third have some neurologic deficit.
- ✓ Myelomeningocele :Congenital defect in vertebral arches with cystic dilatation of meninges and structural or functional abnormality of spinal cord or cauda equina .

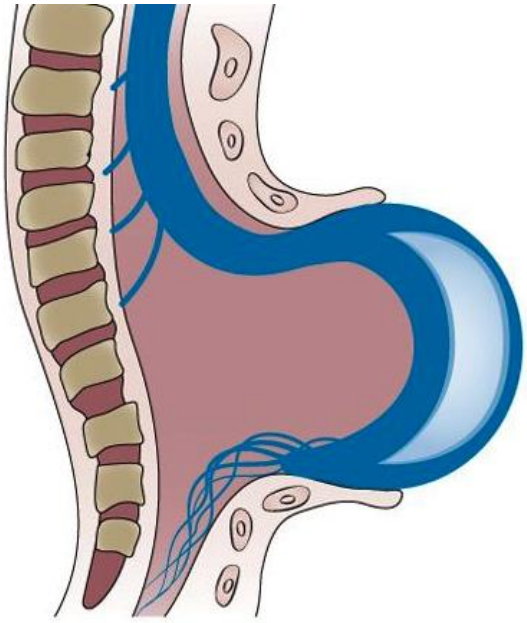
Meningocele



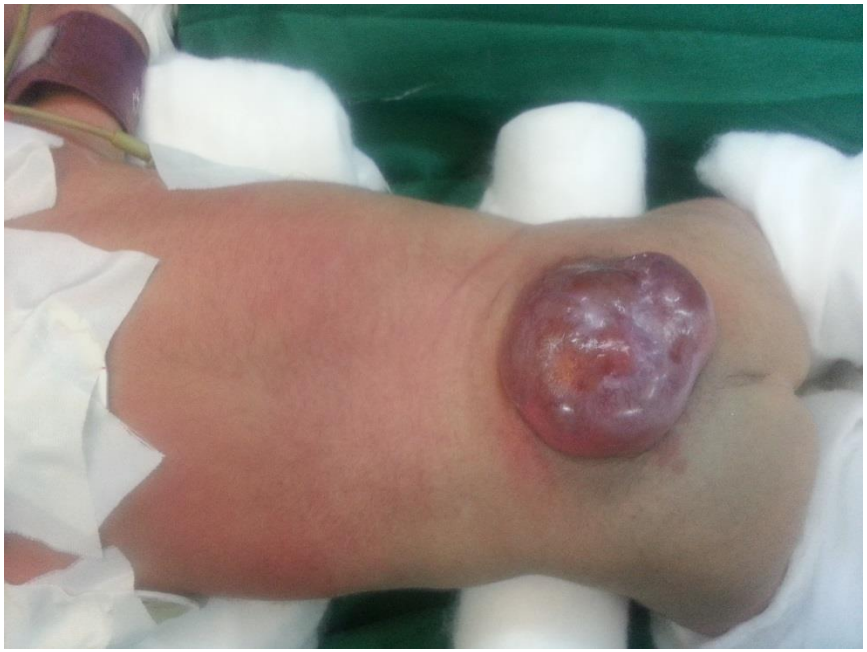
Meningocele
Protrusion of the meninges (filled with CSF) through a defect in the skull or spine



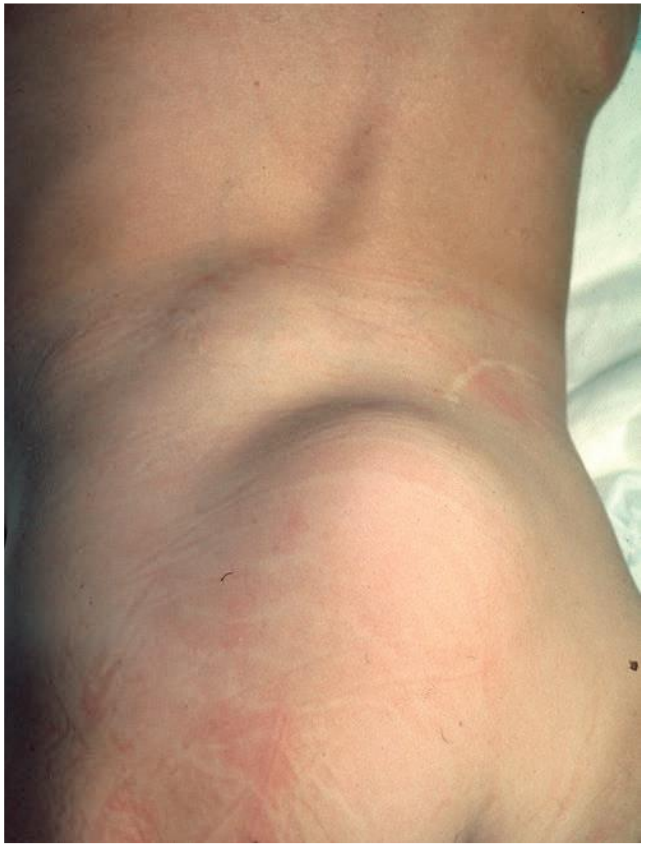
Myelomeningocele

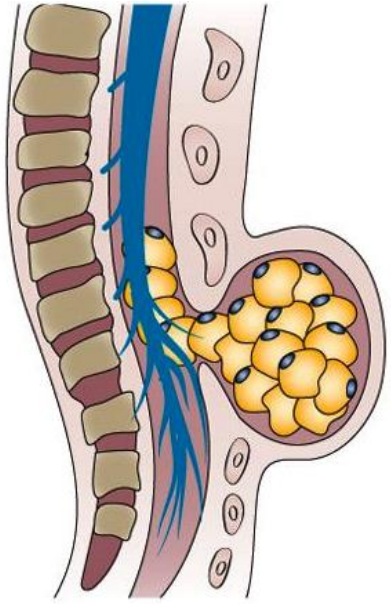


Myelomeningocele
Open spinal cord
(with a meningeal cyst)



Lipomyelomeningoceles





Closed spinal dysraphism
Deficiency of at least two vertebral arches, here covered with a lipoma

Etiology

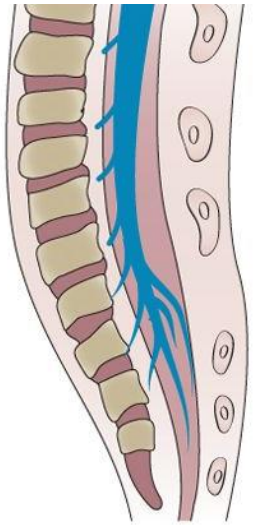
- Unknown cause but some predisposing factors are:
  - Nutritional deficiency of Folic Acid .
  - Genetic factors: Family history —> if a woman bears child with Spinal Bifida, there is a chance that of another child having spinal bifida
  - More common in females than males
  - Medications like. Anticonvulsants(valproate)
  - Conditions like:. Diabetes, obesity and fever also increases the chances of delivering of a baby with a spina bifida

SPINA BIFIDA OCCULTA (SBO)

Reported prevalence range of SBO: 5-30% of North Americans (5-10% is probably more realistic).

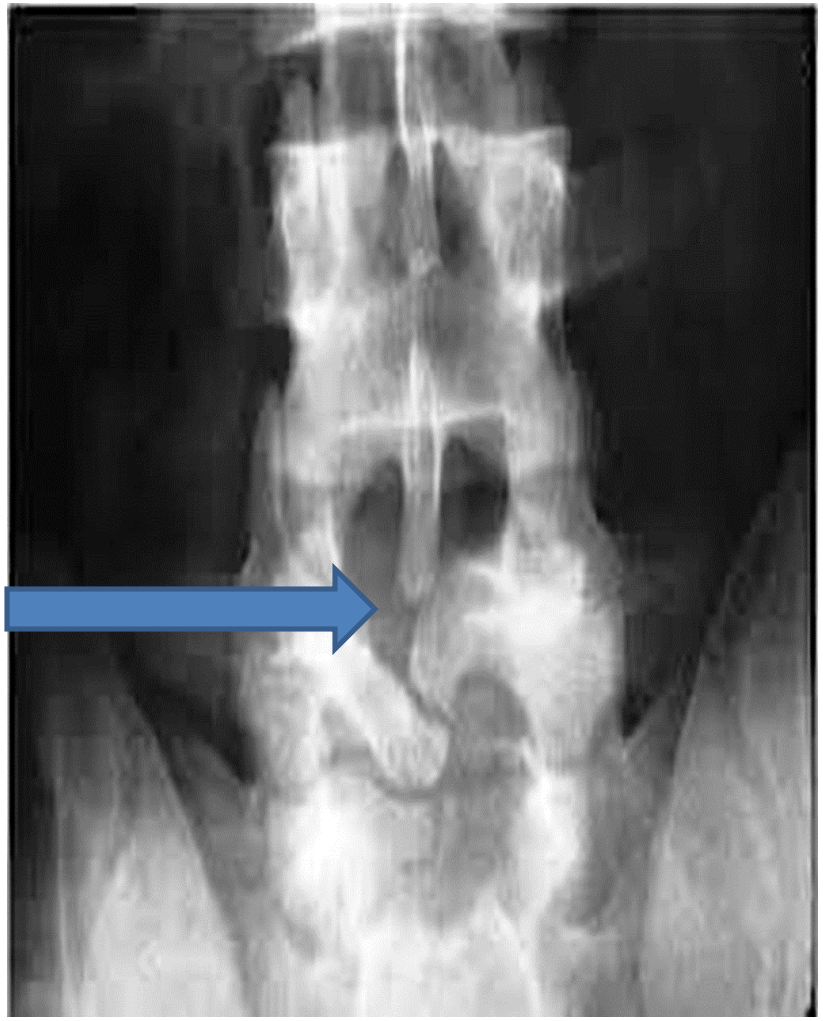
The defect may be palpable, and there may be overlying cutaneous manifestations

Often an incidental finding, usually of no clinical importance when it occurs alone.



Spina bifida occulta
Closed asymptomatic NTD in which some of the vertebrae are not completely closed

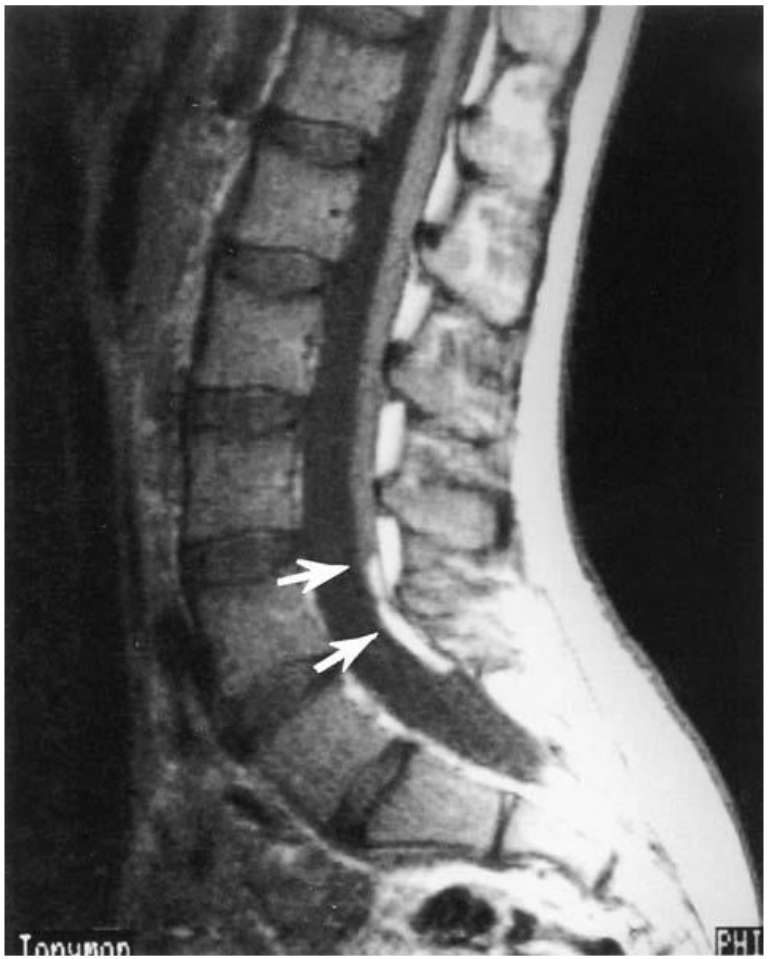
- ✓ Numerous reviews have shown no statistical association of SBO with nonspecific Low back pain. An increased incidence of disc herniation was shown in one study.
- ✓ SBO may occasionally be associated with diastematomyelia, tethered cord, lipoma, or dermoid tumor.
- ✓ When symptomatic from one of these associated conditions, the presentation is usually that of tethered cord (gait disturbance, leg weakness and atrophy, urinary disturbance, foot deformities…,).



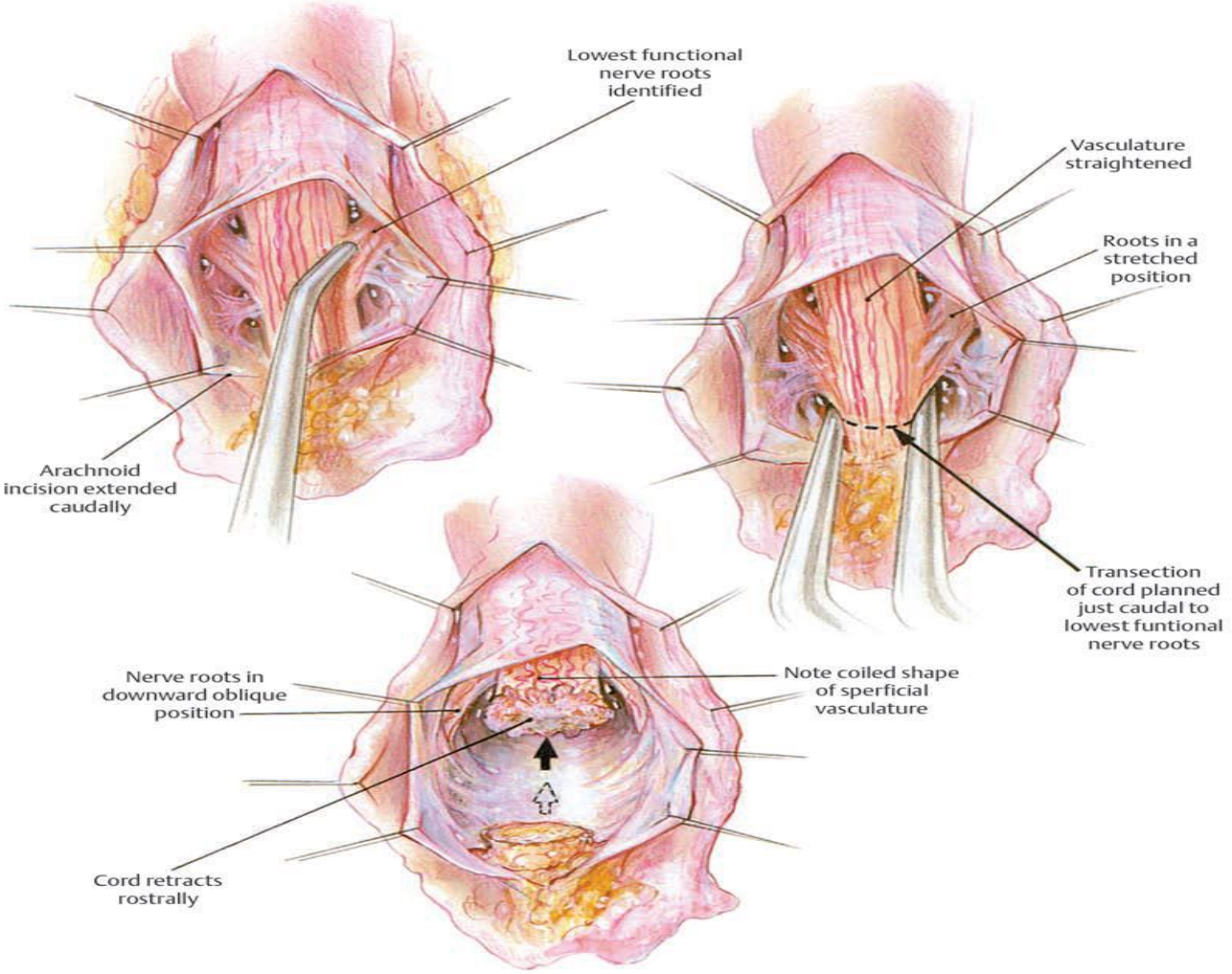
Tethered cord syndrome

Abnormally low conus medullaris. Usually associated with a short, thickened filum terminale, or with an intradural lipoma (other lesions, e.g. lipoma extending through dura, or diastematomyelia are considered as separate entities). Most common in myelomeningocele **(MM)**.

A sagittal T1-weighted magnetic resonance image through the lumbar region demonstrates a thickened filum terminale. The hyperintensity (*arrows*) indicates fatty infiltration of the filum



Detethering(release of the tethered cord)



Myelomeningocele

EMBRYOLOGY

The anterior neuropore closes at gestation day 25. The caudal neuropore closes at day 28.

EPIDEMIOLOGY/GENETICS

- Incidence of spina bifida with meningocele or myelomeningocele **(MM)** is 1-2/1000 live births (0.1-0.2%).
- Risk increases to 2-3% if there is one previous birth with MM, and 6-8% after two affected children.
- The risk is also increased in families where close relatives (e.g. siblings) have given birth to MM children, especially when on the mother's side of the family.
- Incidence may increase in times of war, famine or economic disasters, but it may be gradually declining overall.

Transmission follows non-Mendelian genetics, and is probably multifactorial.

PRENATAL DETECTION OF NEURAL TUBE DEFECTS

- **Serum alpha-fetoprotein (AFP)**

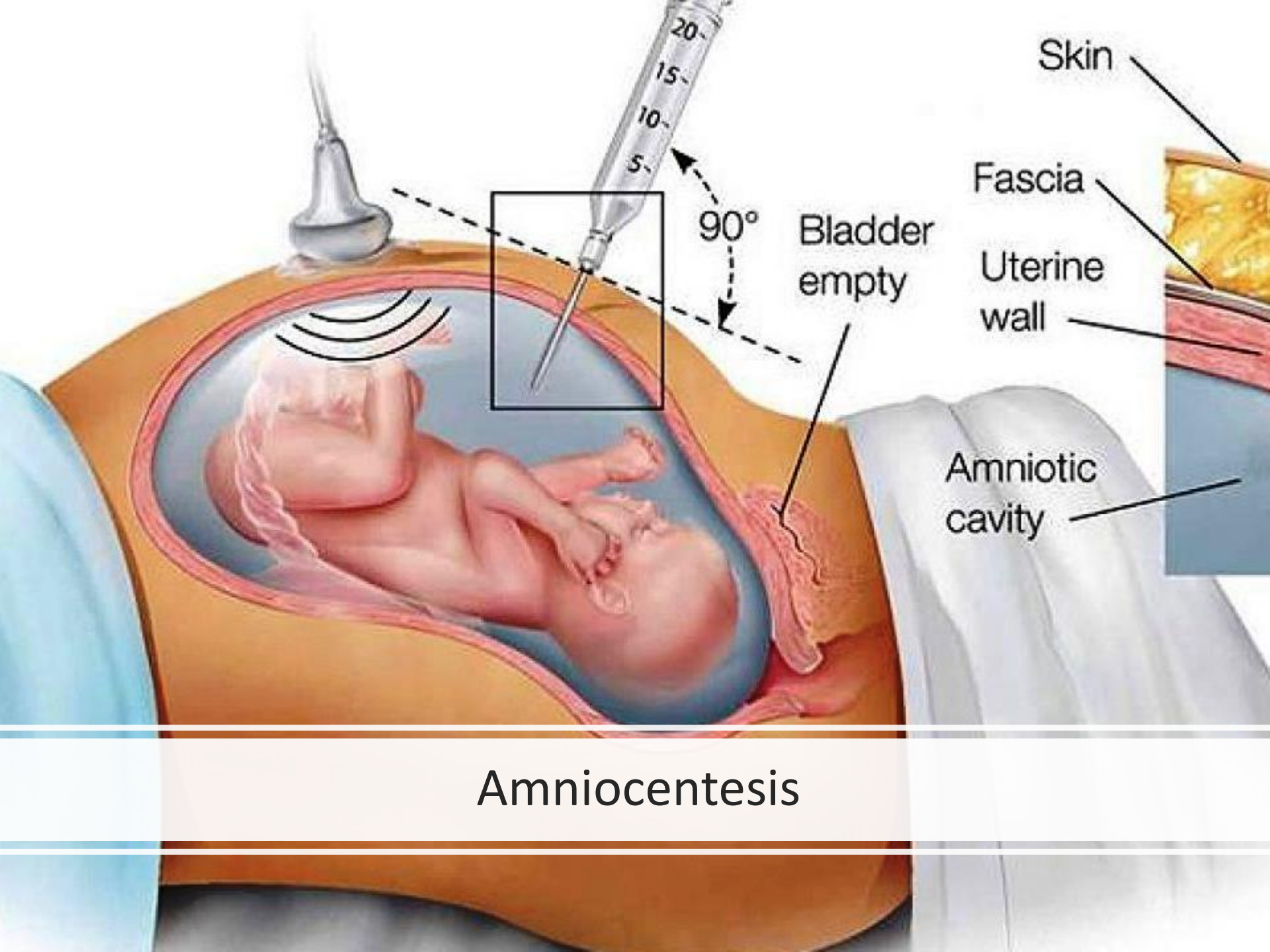
A high maternal serum AFP (≥ 2 multiples of the median for the appropriate week of gestation) between 15-20 weeks gestation carries a relative risk of 224 for neural tube defects.

- **Ultrasound**

Prenatal ultrasound will detect 90-95% of cases of spina bifida, and thus in cases of elevated AFP, it can help differentiate NTDs from non-neurologic causes of elevated AFP (e.g. omphalocele),and can help to more accurately estimate gestational age.

- **Amniocentesis**

For pregnancies subsequent to a MM, if prenatal ultrasound does not show spinal dysraphism, then amniocentesis is recommended (even if abortion is not considered, it may allow for optimal post-partum care if MM is diagnosed). Amniocentesis also carries a ≈ 6% risk of fetal loss in this population.



Amniocentesis

Clinical presentation

Neurological assessment :

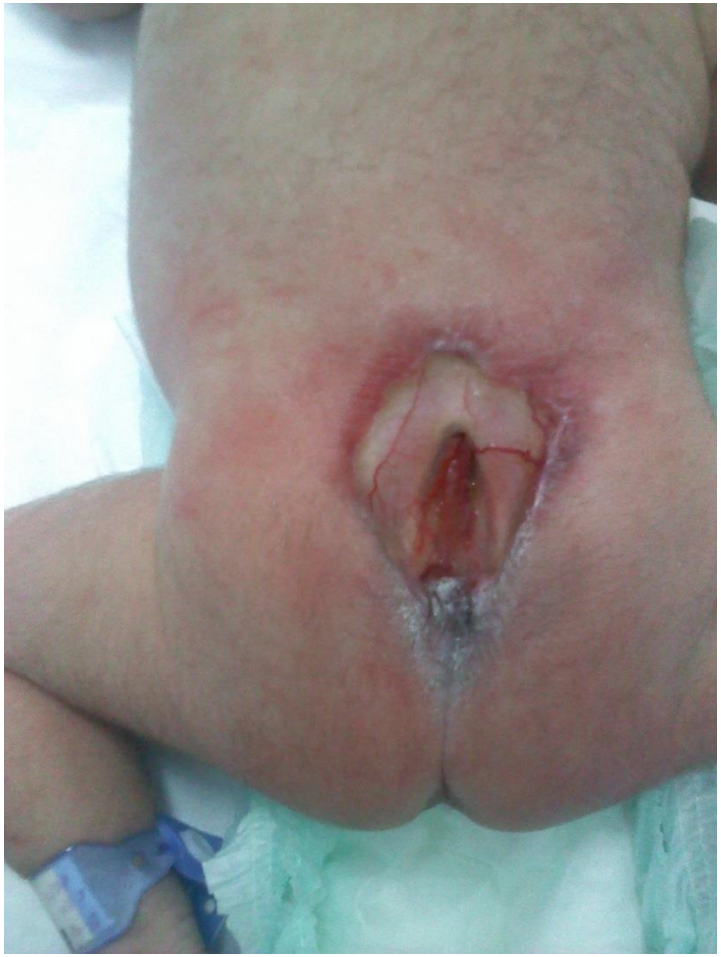
- Items related to spinal lesion

1. watch for spontaneous movement of the LLs (good spontaneous movement correlates with better later functional outcome)
2. assess lowest level of neurologic function by checking response of LLs to painful stimulus:.

-differentiating reflex movement from voluntary may be difficult. In general, voluntary movement is not stereotyped with repetitive stimulus .

- Items related to the commonly associated Chiari type 2 malformation:

1. measure OFC(occipitofrontal circumference): risk of developing hydrocephalus.
2. head U/S within ≈ 24 hrs
3. check for inspiratory stridor, apneic episodes



Ancillary assessment and management:

- Evaluation by neonatologist to assess for other abnormalities, especially those that may preclude surgery (e.g. pulmonary immaturity). There is an average incidence of 2-2.5 additional anomalies in MM patients
- Bladder: start patient on regular urinary catheterizations, obtain urological consultation(non-emergent)
- AP & lat spine films: assess scoliosis (baseline)
- Orthopedic consultation for severe kyphotic or scoliotic spine deformities and for hip or knee deformities

Management

ADMISSION

- assessment and management of lesion:
  - measure size of defect
  - assess whether lesion is ruptured or unruptured

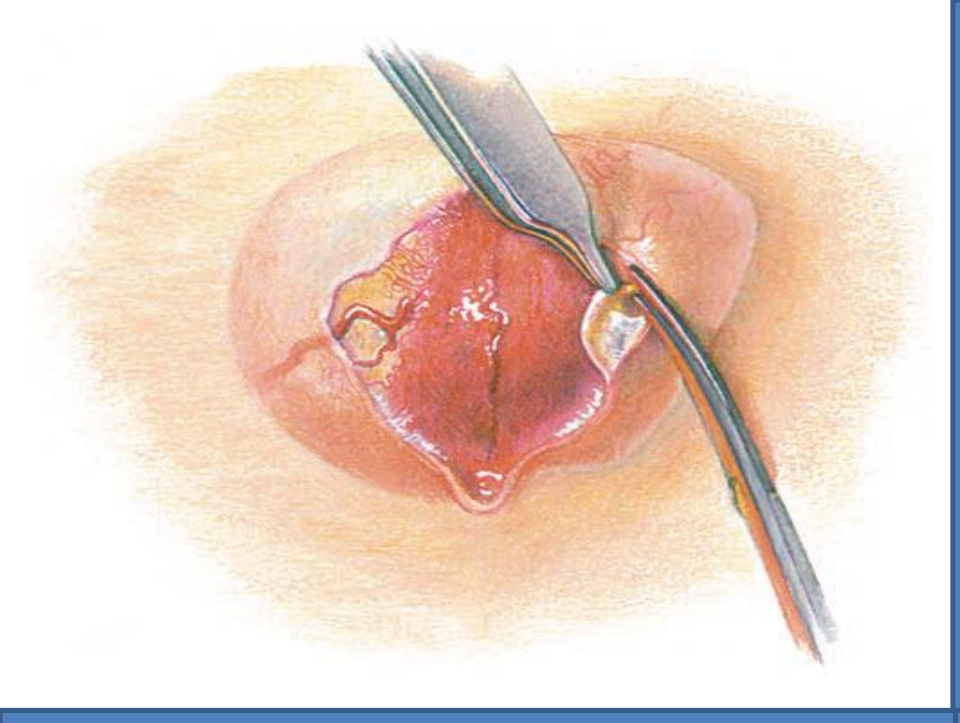
1. ruptured: start antibiotics
2. unruptured: no antibiotics necessary

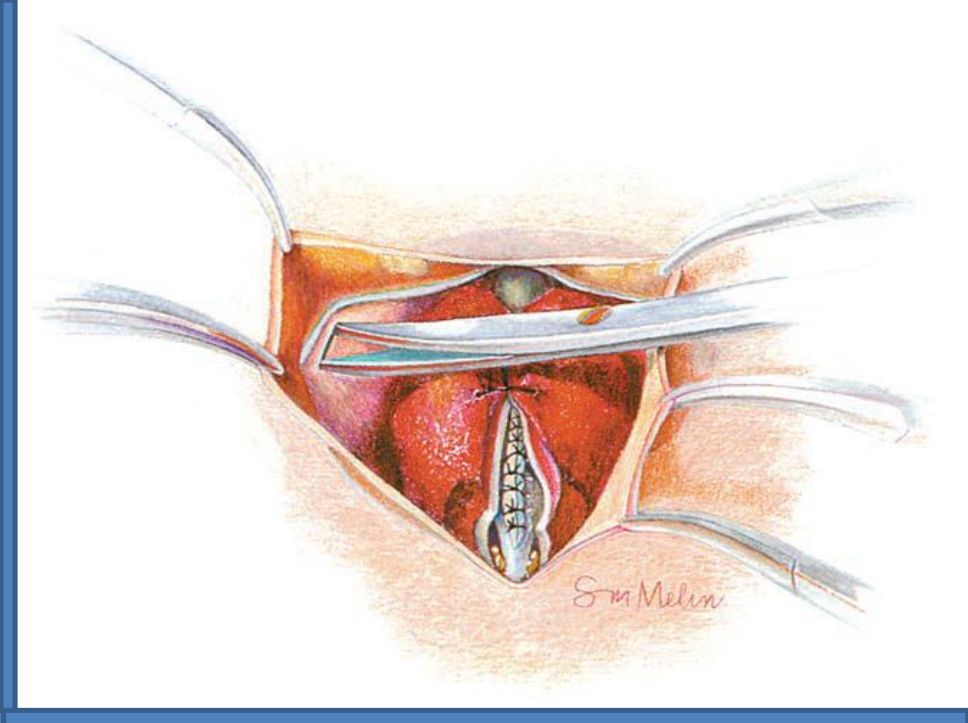
  - cover lesion with telfa or wet dressing , to prevent desiccation
  - Trendelenburg position.
  - perform surgical closure within 36 hrs unless there is a contraindication to surgery

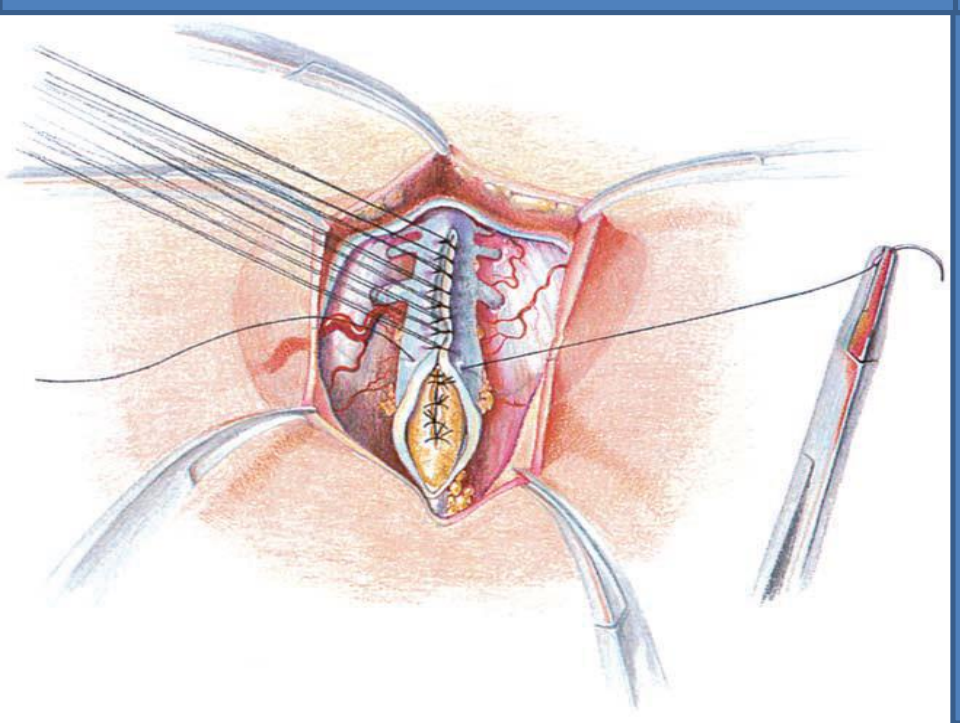
SURGICAL REPAIR

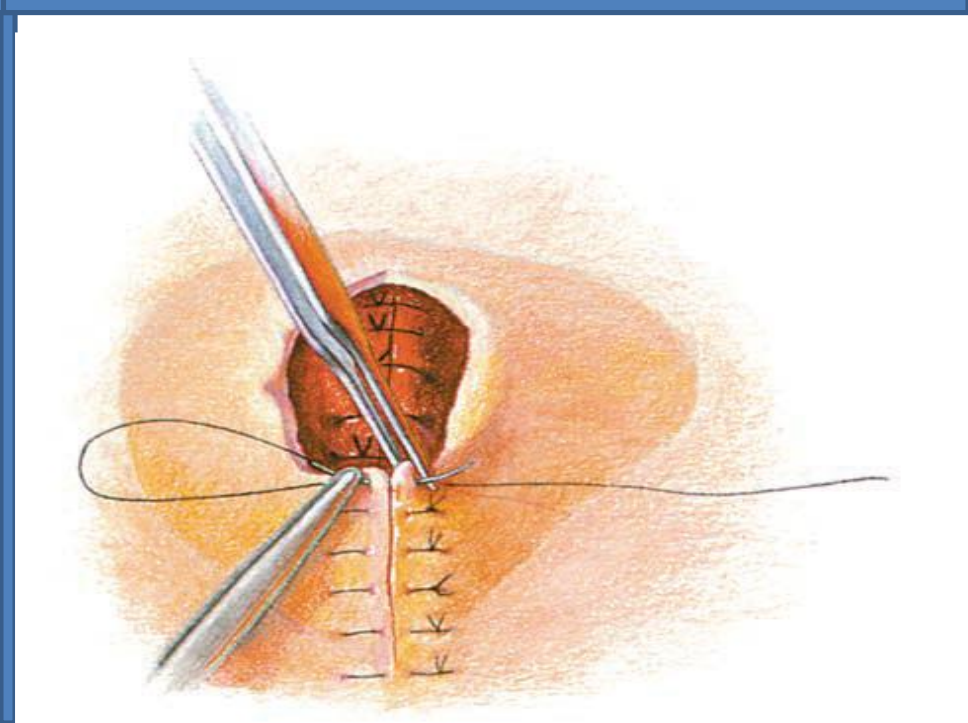
- *TIMING OF CLOSURE:*

Early closure of MM defect is not associated with improvement of neurologic function, but evidence supports lower infection rate with early closure. MM should be closed within 24 hrs whether or not membrane is intact (after ≈ 36 hrs the back lesion is colonized and there is increased risk of postoperative infection).







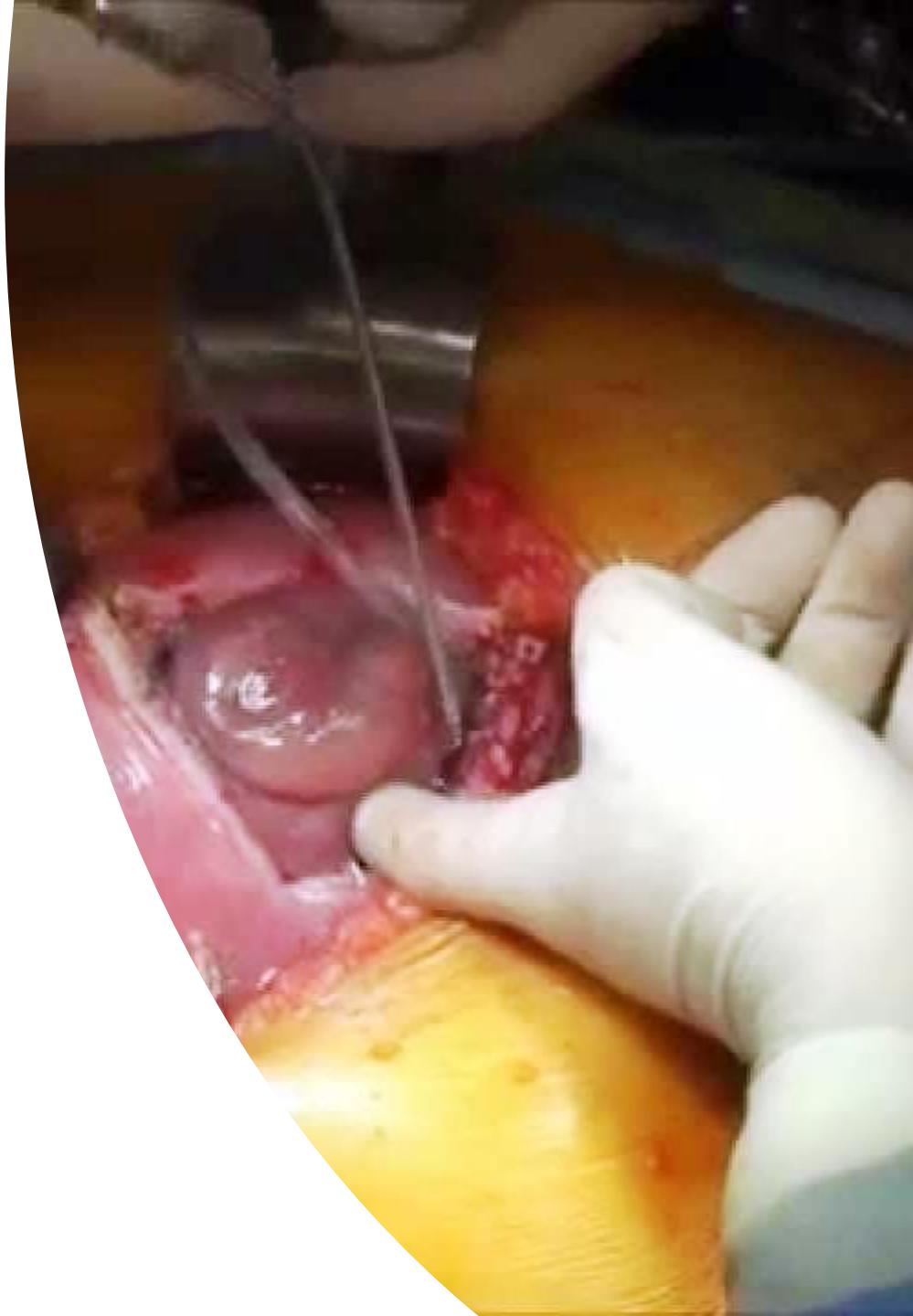


Intrauterine closure of MM defect

- Controversial.
- Does reduce incidence of Chiari II defect, but it has not been determined if this is

clinically significant.

- Argued whether this reduces incidence of hydrocephalus.
- Does not improve distal neurologic function.



MMC and hydrocephalus

- Incidence of hydrocephalus is 85-90% of all cases of myelomeningocele.
- Usually it is a part of type 2 (arnold)-chiari malformation.
- May not be apparent after birth immediately, may be seen as far as 2 years ,but most cases will need shunt by the age of 6 months.

Simultaneous MM defect closure and VP shunting

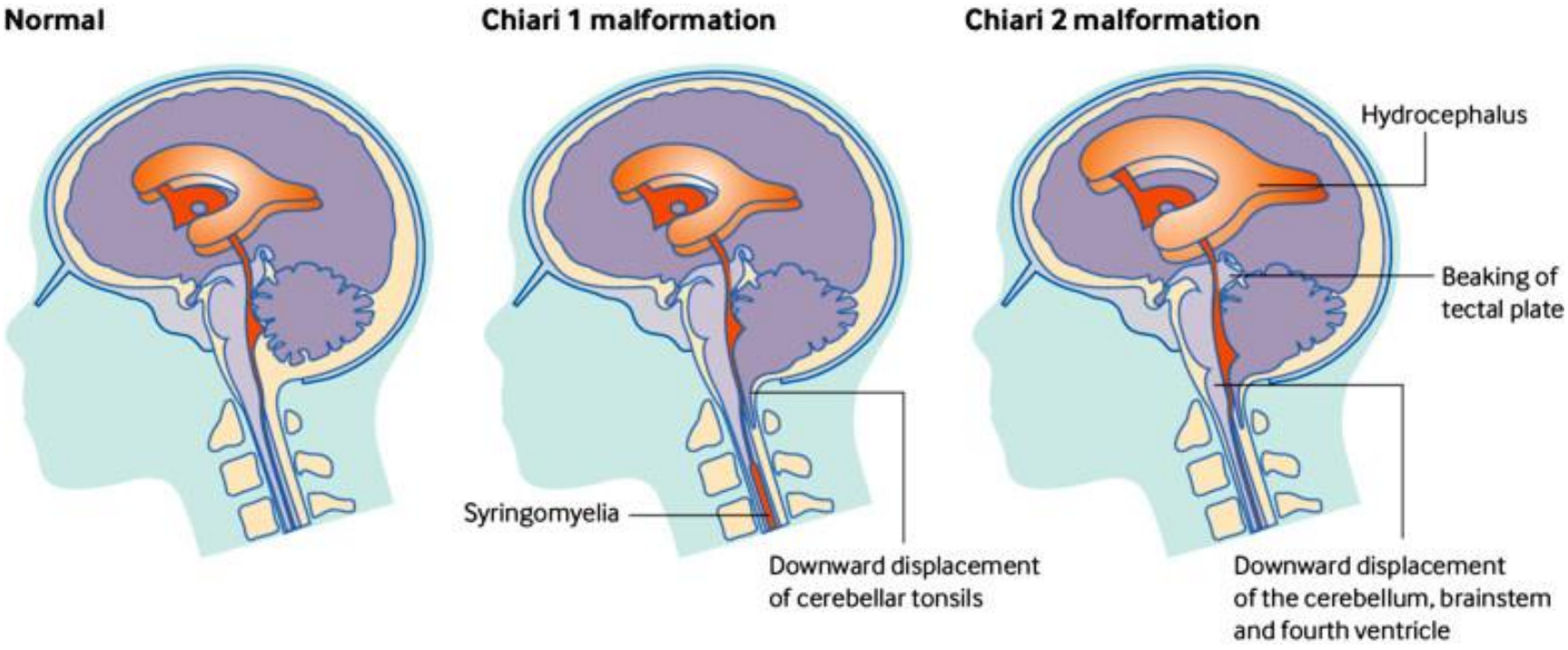
- In patients without hydrocephalus, most surgeons wait at least ≈ 3 days after MM repair before shunting.
- In MM patients with clinically overt HCP at birth (ventriculomegaly with enlarged OFC and/or symptoms), MM repair and shunting may be performed in the same sitting without increased incidence of infection, and with shorter hospitalization.

Chiari malformations

- Basically they are hindbrain malformations, with associated anomalies that may involve the rest of the CNS.
- The term “Chiari malformation” (after pathologist, Hans Chiari) is preferred for type 1 malformations, with the commonly used term “Arnold-Chiari malformation” reserved for type 2 malformation.
- The Chiari malformations consists of four types of hindbrain abnormalities, probably unrelated to

each other.

- The majority are types 1 or 2), a very limited number of cases comprise the remaining types.


Normal
Chiari 1 malformation
Chiari 2 malformation
Hydrocephalus
Beaking of tectal plate
Syringomyelia
Downward displacement of cerebellar tonsils
Downward displacement of the cerebellum, brainstem and fourth ventricle

Finding	Chiari type 1 (*see below*)	Chiari type 2 (*see page 238*)
caudal dislocation of medulla	unusual	yes
caudal dislocation into cervical canal	tonsils	inferior vermis, medulla, 4th ventricle
spina bifida (myelomeningocele)	may be present	rarely absent
hydrocephalus	may be absent	rarely absent
medullary "kink"	absent	present in 55%
course of upper cervical nerves	usually normal	usually cephalad
usual age of presentation	young adult	infancy
usual presentation	cervical pain, suboccipital H/A	progressive hydrocephalus, respiratory distress

LATE PROBLEMS/ISSUES

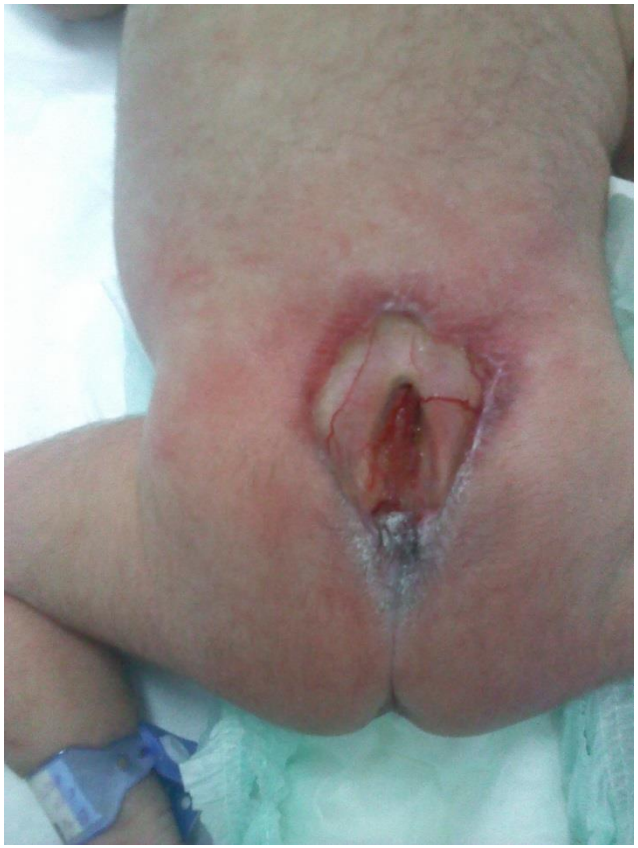
Include:

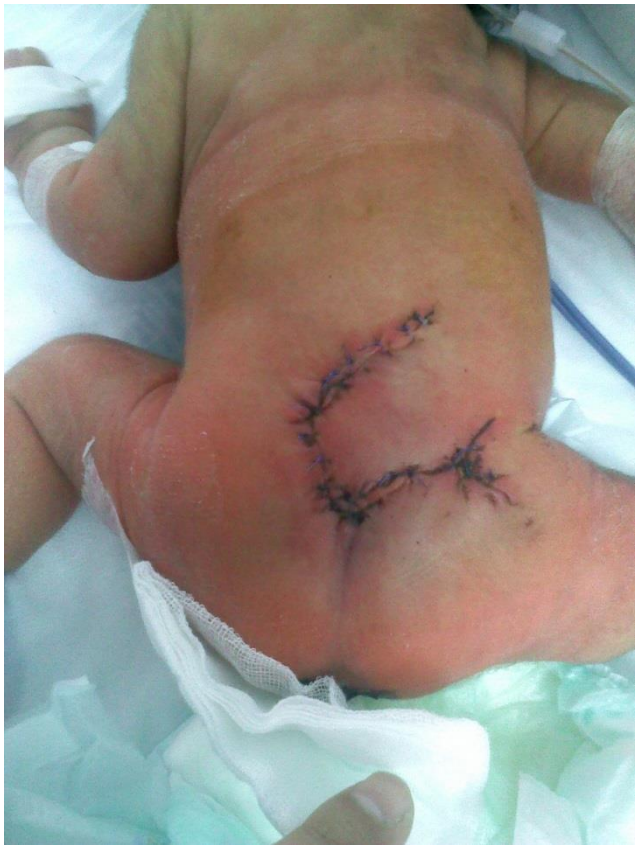
- 1. Hydrocephalus: may mimic ≈ anything listed below. always rule out shunt malfunction when a MM patient deteriorates
- 2. Syringomyelia (and/or syringobulbia):
- 3. Tethered cord as many as 70% of MM patients have a tethered cord radiographically (some quote 10-20%), but only a minority are symptomatic.
- 4. Dermoid tumor at the MM site131: incidence ≈ 16% .
- 5. Medullary compression at foramen magnum (symptomatic Chiari II malformation.
- 6. Use of growth hormone to increase stature is controversial

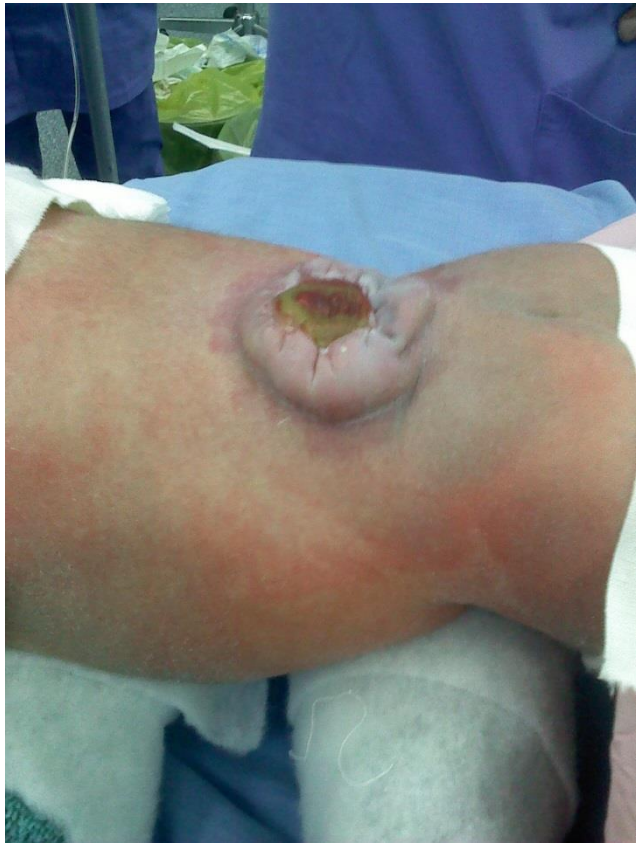
OUTCOME

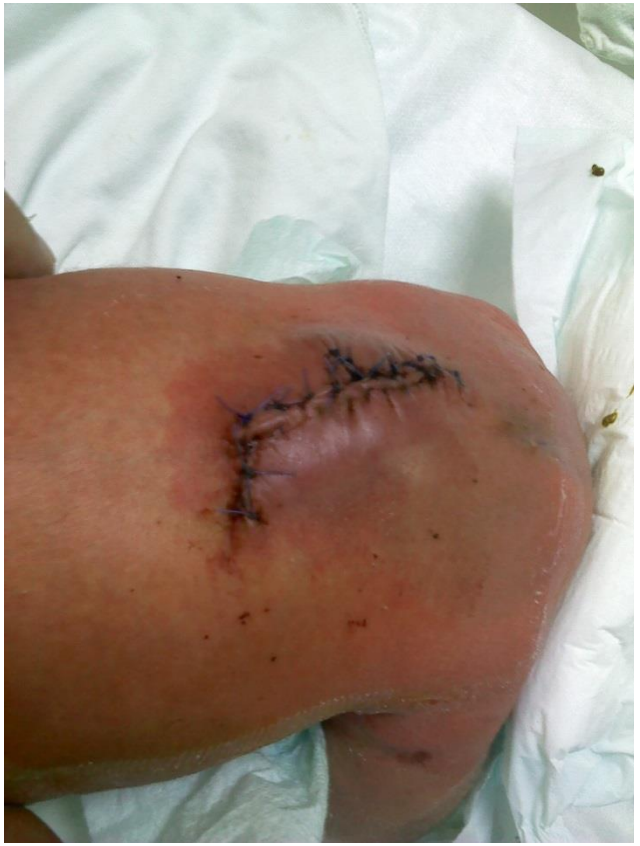
- Without any treatment, only 14-30% of MM infants survive infancy; these usually represent the least severely involved.
- With modern treatment, ≈ 85% of MM infants survive.
- The most common cause of early mortality are complications from the Chiari malformation (respiratory arrest, aspiration…), where late mortality is usually due to shunt malfunction.
- 80% will have normal IQ. Mental retardation is most closely linked to shunt infection. 40-85% are ambulatory with bracing, however, most choose to use wheelchairs for ease.
- 3-10% have normal urinary continence, but most may be able to remain dry with intermittent catheterization.

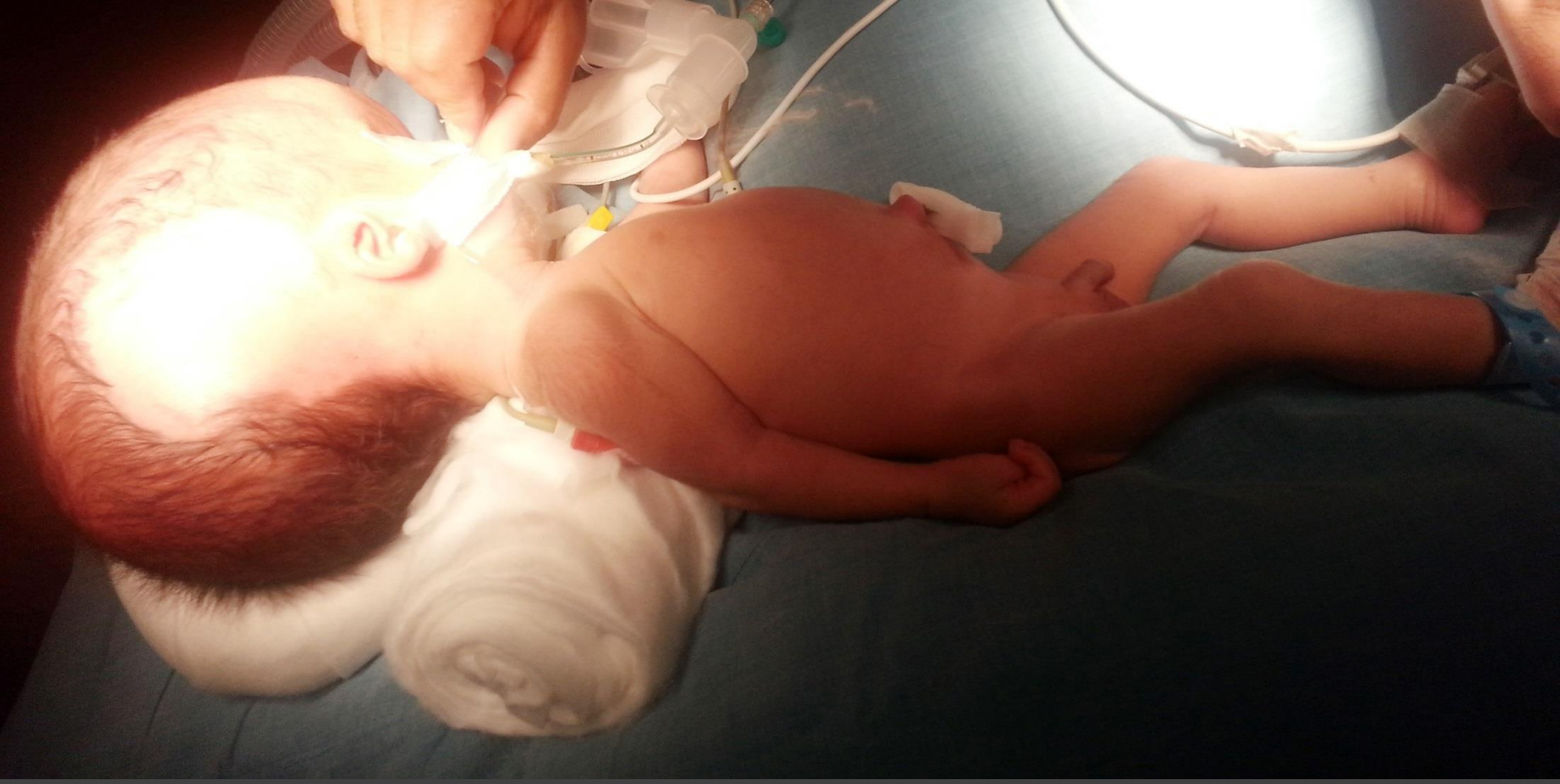
MMC repair-from our series







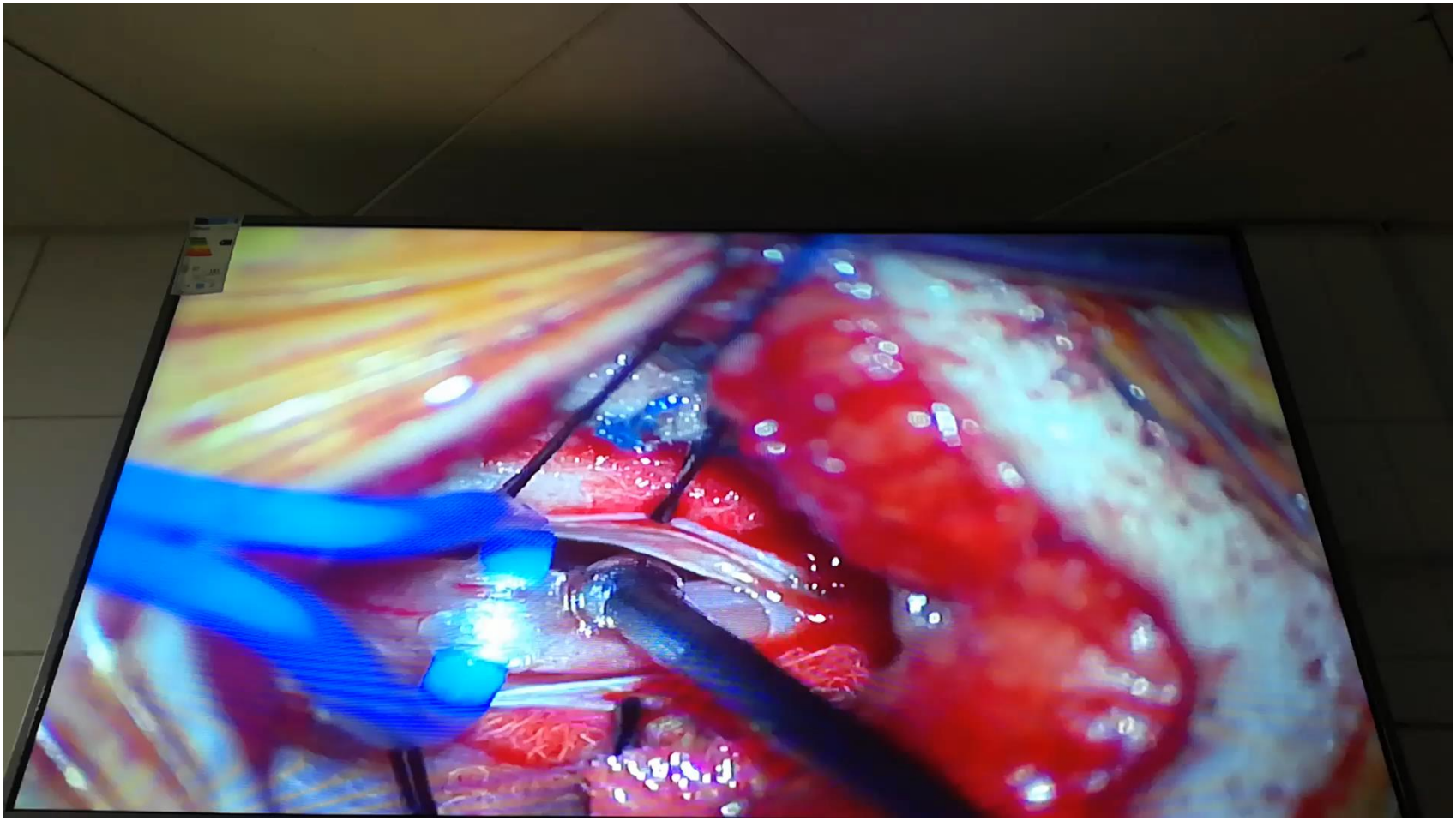




Patient with hydrocehalus,positioned and intubated in prepare for VP shunting

Huge occipital encephalocele

Detethering of cord-video



And here we come to the:

THE END