



# Neuropathology

## Congenital malformations

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# Congenital malformations

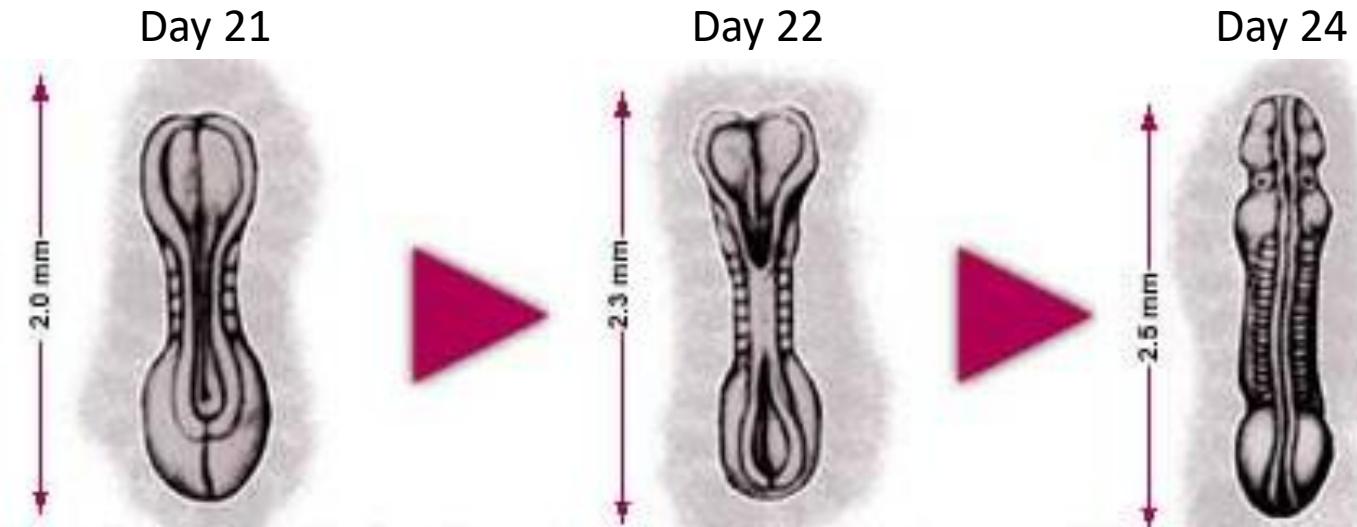
- Reflect interruptions in the completion of critical developmental processes
- Genetic abnormalities
- External insults
  - Chemicals
  - Infections

# Congenital malformations

- Neural tube defects
- Forebrain malformations
- Posterior fossa anomalies

# 1. Neural tube defects

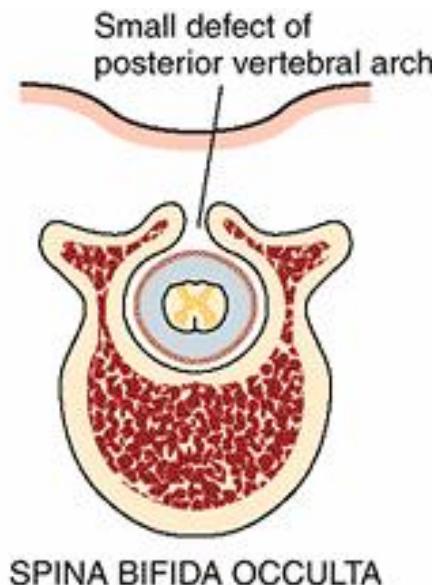
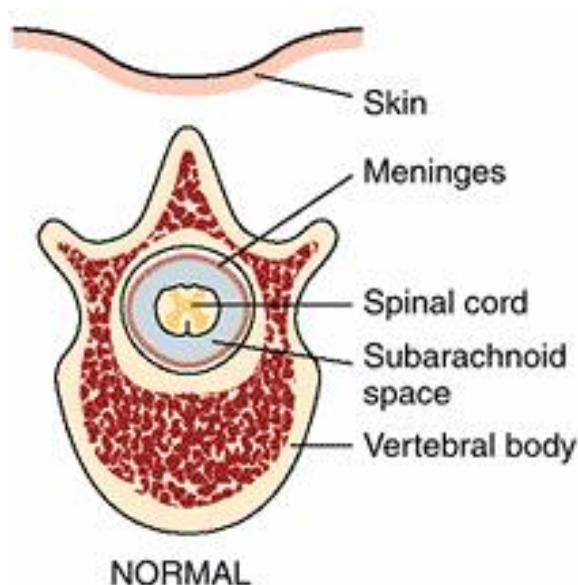
- Most frequent type of CNS malformations
- Impaired closure of the dorsal aspect of the vertebral column
- Genetic component
- Folate deficiency (initial weeks)



# Neural tube defects – posterior end

## Spina bifida occulta

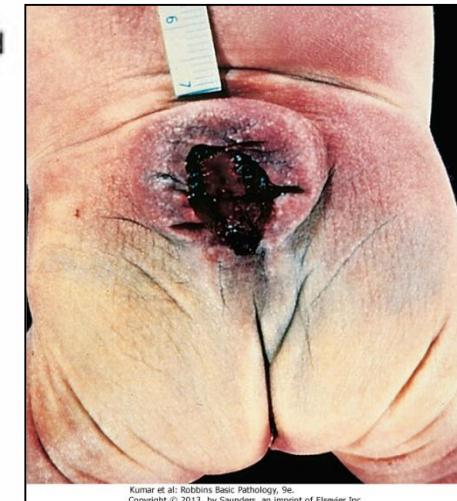
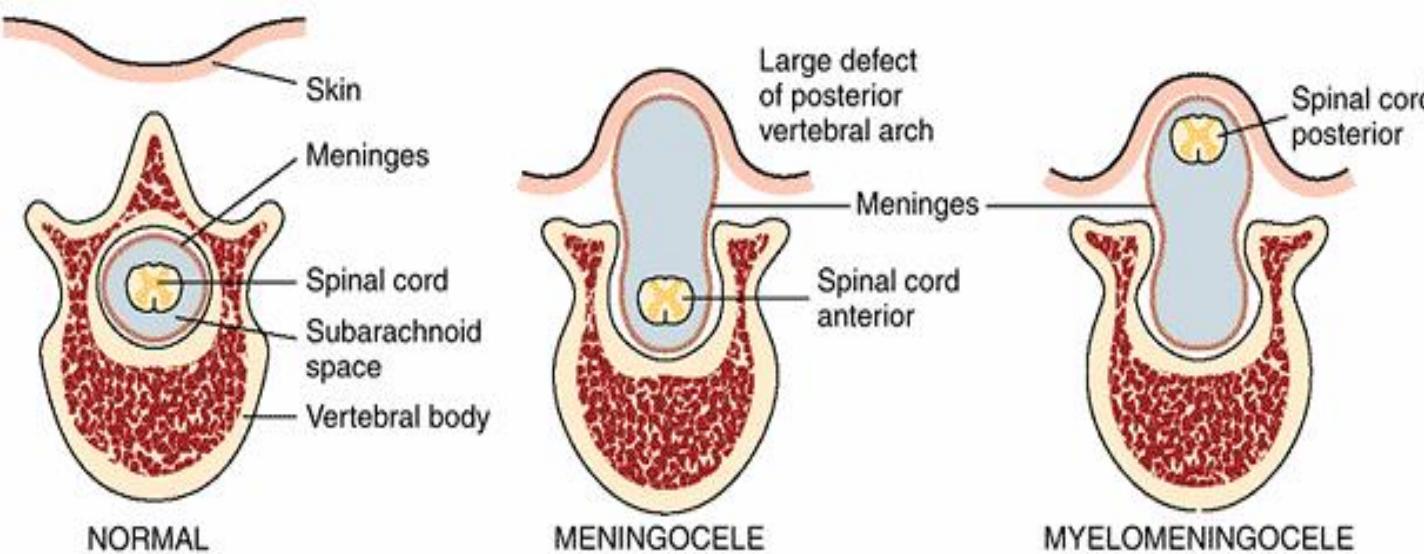
- asymptomatic bony defect
- hair tuft or dimple



# Neural tube defects – posterior end

## Meningocele, myelomeningocele

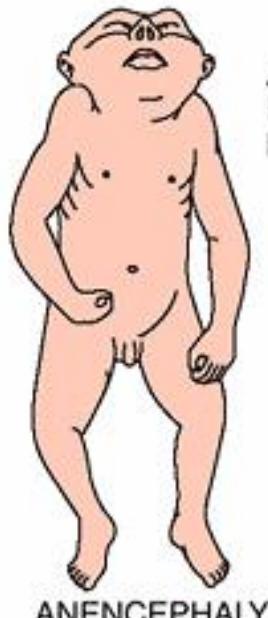
- protrusion of meninges and nerve roots
- motor and sensory deficits, infections



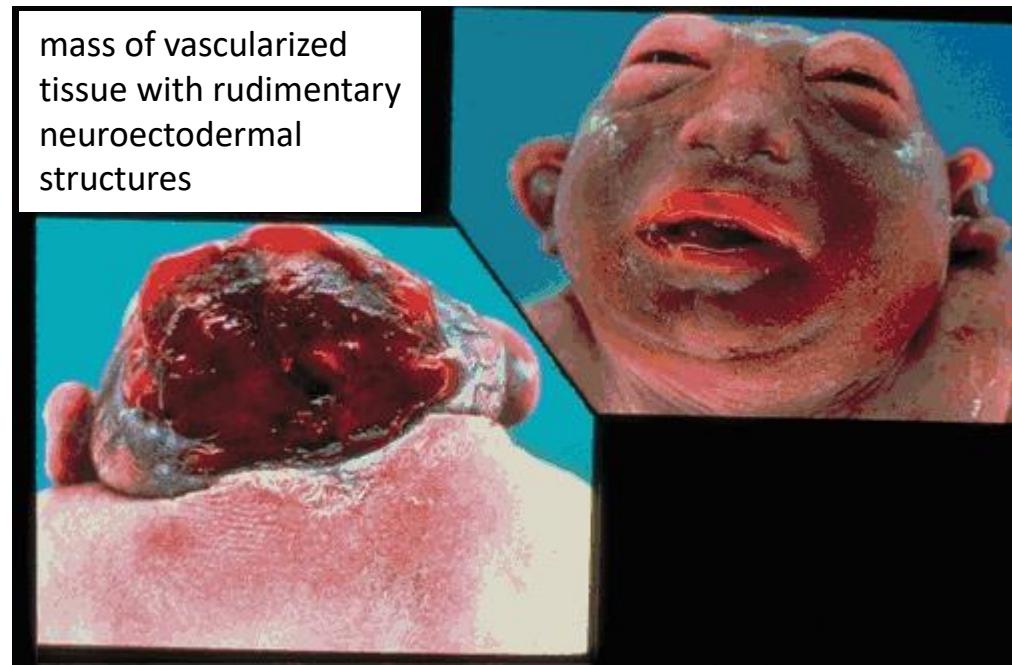
# Neural tube defects – anterior end

## Anencephaly

- absence of the brain and the top of skull
- normally formed eyes, brain stem
- stillborn or die within the first few days of life



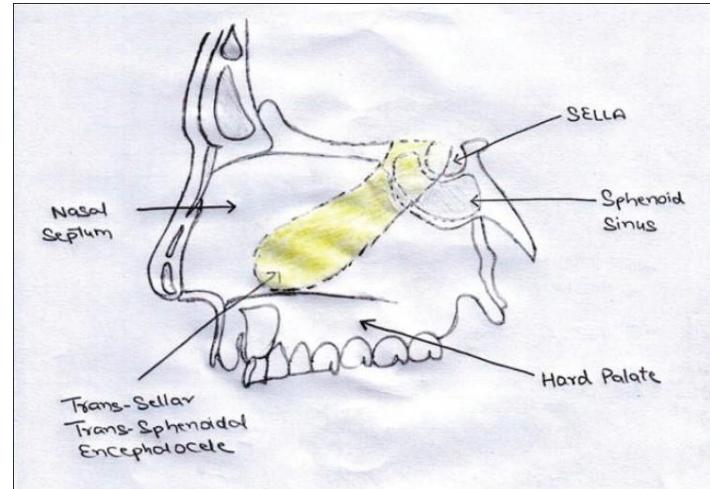
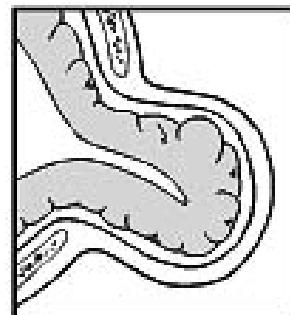
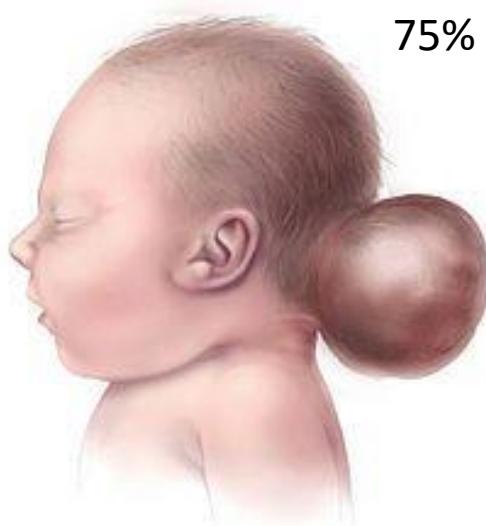
Acrania  
Protruding eyes  
Long arms



# Neural tube defects – anterior end

## Encephalocele

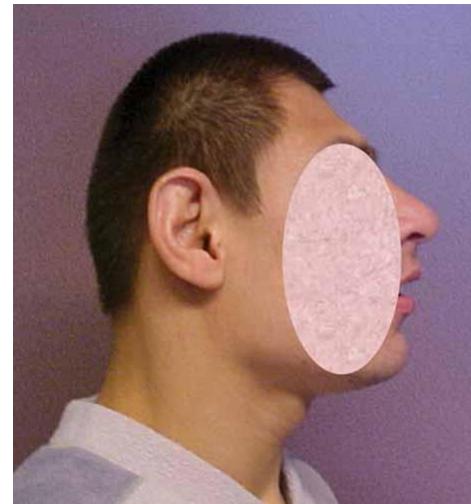
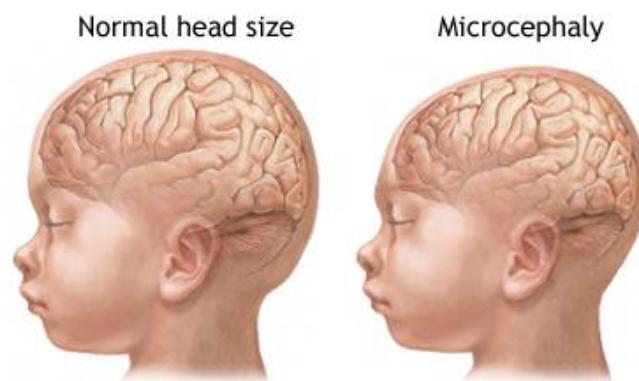
- sac comprising brain and membranes
- a number of forms based on its location
- +/- other structural aberrations



## 2. Forebrain malformations

### Micro(en)cephaly

- The volume of the brain is abnormally small (decreased generation of cortical neurons) → subnormal intelligence
- chromosomal abnormalities  
fetal alcohol syndrome  
in utero HIV or Zika virus infection



# Forebrain malformations

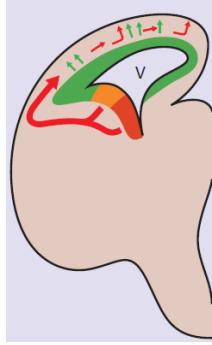
## Megalencephaly (hemimegalencephaly)

- The volume of the brain is abnormally large
- Associated with rare genetic disorders



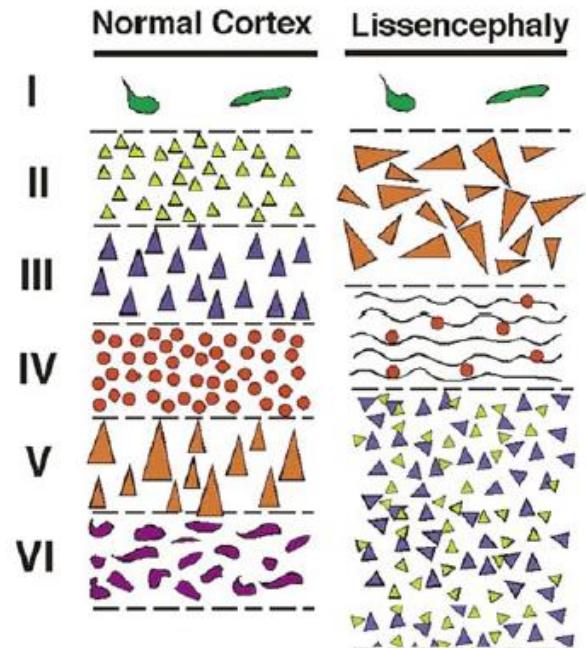
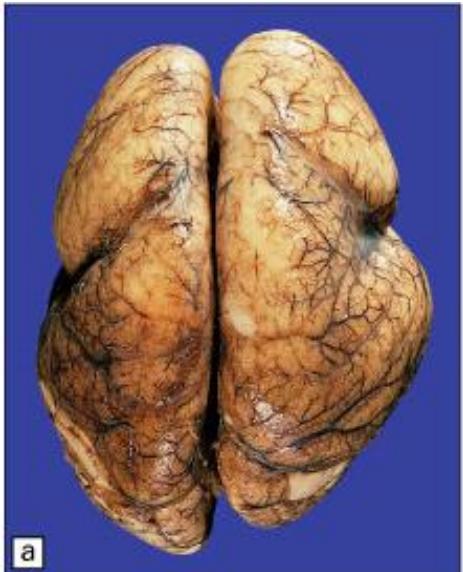
# Forebrain malformations

## Disrupted neuronal migration



### Lissencephaly (agyria)/Pachygryia (patchy)

- Smooth brain surface
- Thickened cortex with 4 layers

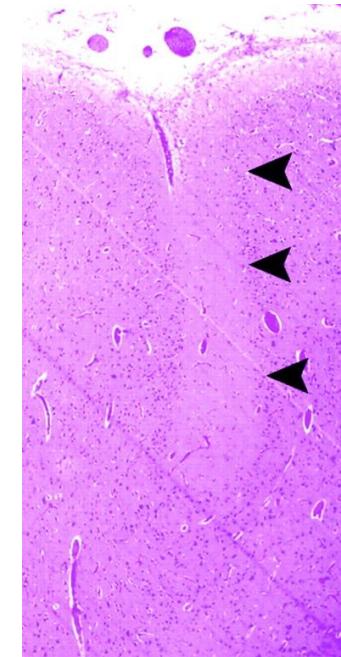
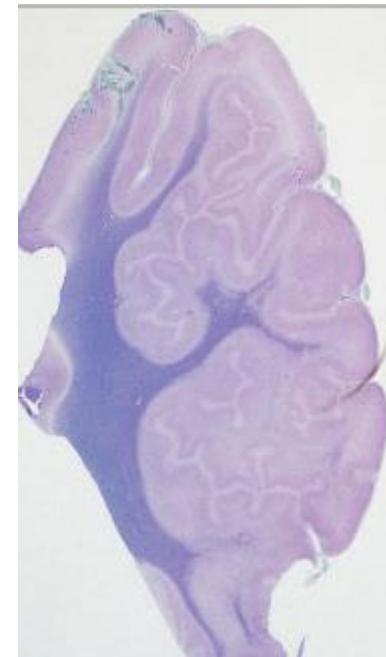
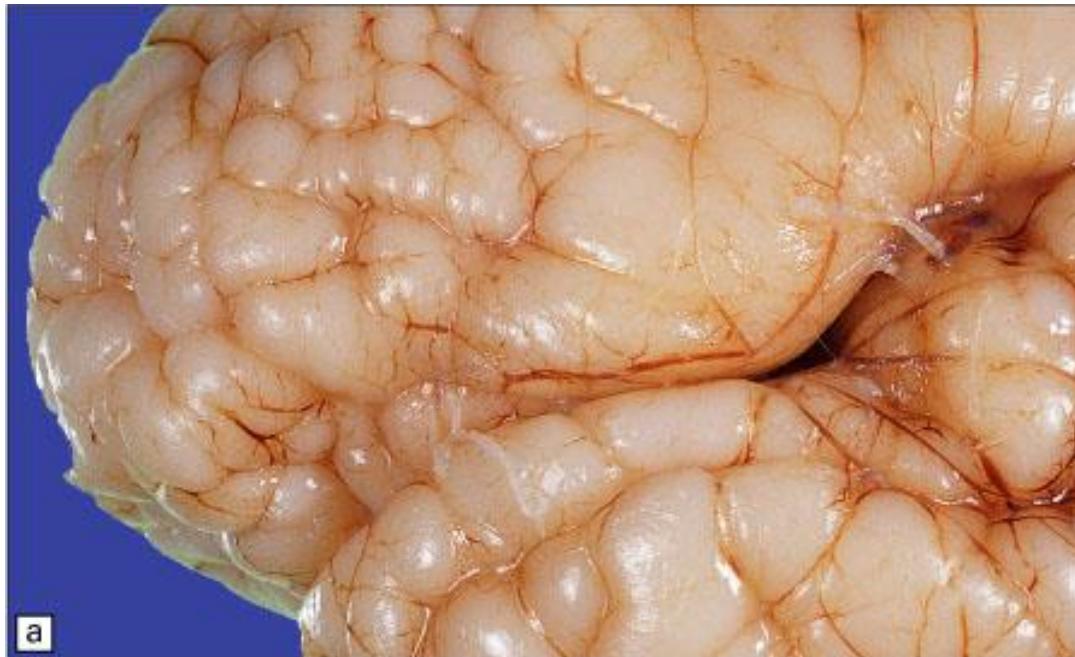


# Forebrain malformations

## Disrupted neuronal migration

### Polymicrogyria – focal/widespread

- Increased number of irregularly formed gyri → cobblestone-like surface, fused molecular layers



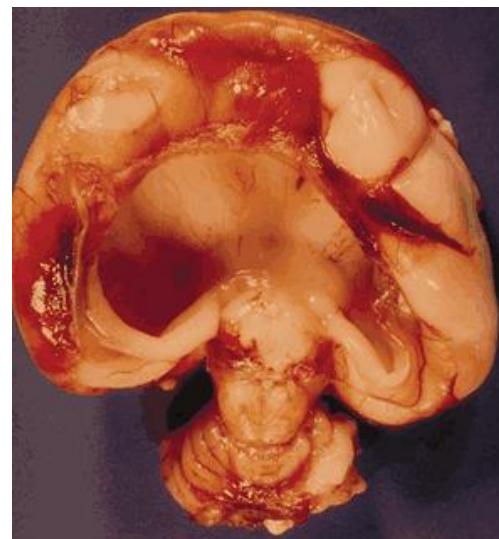
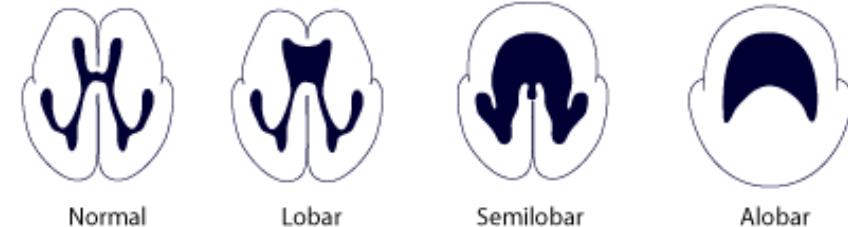
# Forebrain malformations

## Disrupted neuronal migration

### Holoprosencephaly (e.g. mutation in SHH)

- various degrees of incomplete lobation of the brain hemispheres
- Severe → no hemispheres or lobes +/- cyclopia, proboscis
- Arrhinencephaly – absence of olfactory bulbs

Holoprosencephaly: degree of severity



### 3. Posterior fossa anomalies

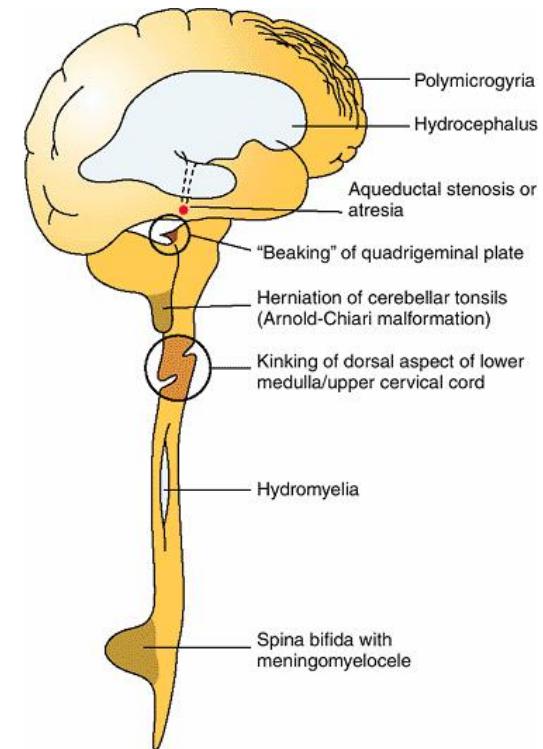
#### Arnold-Chiari malformation (Chiari type II)

- Small posterior fossa
- Downward extension of the vermis
- +/- hydrocephalus, myelomeningocele



#### Chiari type I

- Low lying cerebellar tonsils



# Posterior fossa anomalies

## Dandy-Walker malformation

- Enlarged posterior fossa
- Absence or hypoplasia of the vermis
- Cystic dilatation of the 4th ventricle
- Hydrocephalus

