

CNS - Pathology

Malformations during embryonic development of CNS are rare BUT serious

Sometimes isolated but more in multiple

Earlier malformation (early in embryonic life) → more severe insults

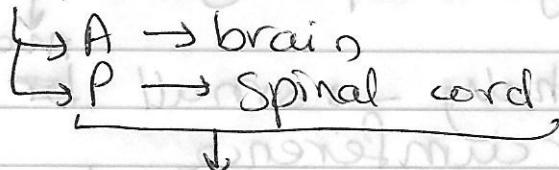
Etiology: insults that happen during embryonal life

- Gene mutations
- Chemicals (ex: certain drugs)
- Some infections (ex: rubella)

Not all insults can cause a gross/microscopic malformation, some result in functional problems, ex: disability

Types of malformations:

① Neural tube defects - most common



defects are more common (in born babies)

Causes:

- Genetic (recurrence 4-5%)

- Folate deficiency (folate taken early in pregnancy ↓ risk by 70%)

no risk for patients ← Spina bifida occulta → some herniation of spinal cord ~~not~~ covered by meninges + bone defects

neural tissue covered by meninges
cyst (going outside body)

More severe → myelomeningocele
→ Spinal cord goes out of the vertebral canal (bone defect)

→ can cause problems depending on its site

↓
most common
→ lumbosacral region

Defects in ant. part of NT

→ problem in brain:

- Less common ← {
- ① Anencephaly absence of brain ⊕ skull covering it
 - ② Encephalocele → herniation of brain through defect in cranium

② Forebrain malformations

[A] → related to volume of brain / number of neurons

large

↓
megalencephaly

small

↓
microencephaly

↓
more common

Microcephaly → small ~~of~~ head circumference

Microencephaly → due to ↓ generation of neurons of cortex (↓ number of neurons) → NOT all are retarded but will have certain problems

[B] → related to problems with neuronal differentiation

problems with gyri → problem in structure of cortex

- Agyria
- poly microgyri (small but large in number)
- Patchy gyri

- ③ Posterior fossa abnormalities (cerebellum)
 severe ← a) Arnold-Chiari malformation (Chiari type 2)
 b) Chiari type 1 → milder form of "a"
 c) Dandy-Walker → large posterior fossa ⊕ no vermis

④ Spinal cord abnormalities:

Most common 2 problems:

- Hydromyelia → expansion of central canal of spinal cord → mild + subtle
- Syringomyelia → same but with fluid-filled clefts

Brain edema:

↑ fluid in extracellular space of brain

from cells from blood vessels

Usually they coexist

↓ cytotoxic

↓ vasogenic

initial problem within neurons or glial cells

initial problem in BBB

↓ due to toxins, hypoxia

↓ + vessels of brain
 ↓ generalized vs. localized
 ↓ more dangerous

Shiny smooth surface + ↑ brain weight

Hydrocephalus:

↑ CSF in ventricles

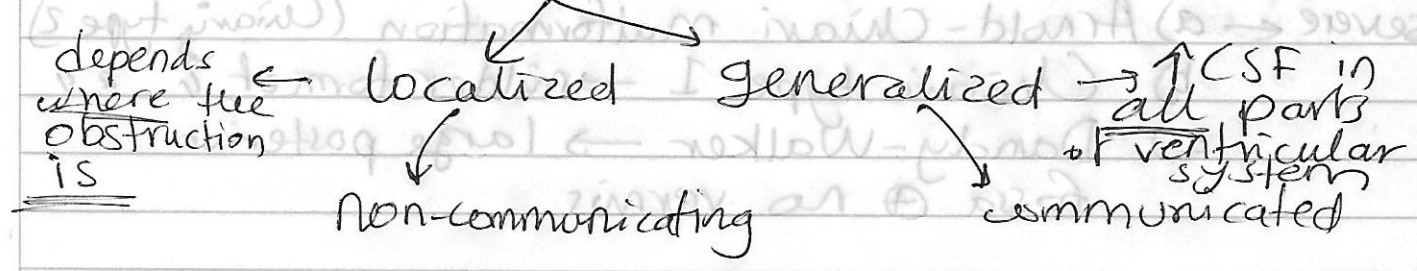
↓ due to ↑ production or ↓ resorption

↑ production: rare (tumors in choroid plexus)

↓ resorption: more common due to obstruction

many cases

resorption of CSF



→ Enlarged ventricles

Herniation:

Brain → in skull (protection) → but any ↑ in size will ↑ intracranial pressure

⇒ expansion of brain tissue

⇒ 3 sites where this can happen:

