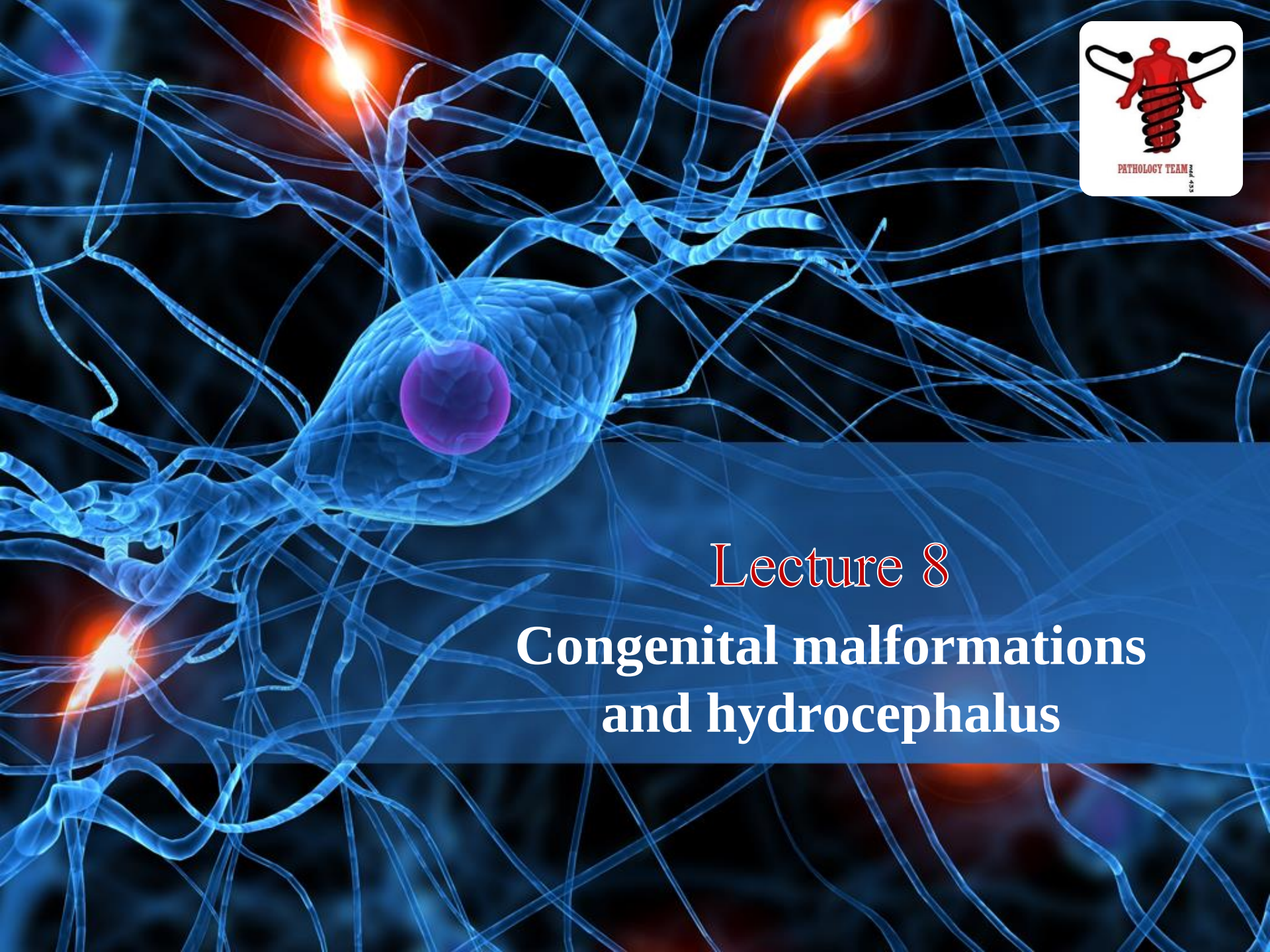


Lecture 8

Congenital malformations and hydrocephalus

OBJECTIVE

- Know the common types of congenital malformations of the CNS and have a basic knowledge of their pathological features.
- Correlate CNS normal development with the classification of congenital CNS malformations.
- Appreciate the role of folate deficiency as an etiological factor in neural tube defects and understand the role of Alpha feto-protein measurement and ultrasound in antenatal diagnosis of neural tube defects.
- Understand the various mechanisms that lead to the development of hydrocephalus.
- List and classify the main causes of hydrocephalus.

Congenital Malformations

- The incidence of CNS malformations, giving rise to mental retardation, cerebral palsy, or neural tube defects, is estimated at 1% to 2%

- Prenatal or perinatal insults may either cause:
  - Failure of normal CNS development
  - Tissue destruction

  - CNS malformation can be caused by mutations affecting molecules in pathways of neuronal and glial:
    - Development
    - Migration
    - Connection
  - Additionally, some toxic compounds and infectious agents are known to have teratogenic effects

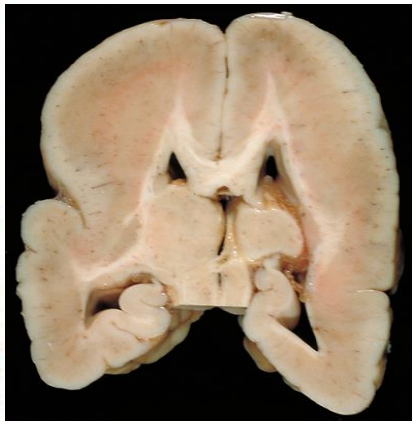
Forebrain Malformations

1-Megalencephaly: The volume of brain may be abnormally large

2-Microencephaly: brain is small and usually associated with a small head (more common)

- It can occur in a wide range of clinical settings, including:
  - Chromosome abnormalities
  - Fetal alcohol syndrome
  - Human immunodeficiency virus 1 (HIV-1) infection acquired in utero

•All causes are associated with a decreased number of neurons destined for the cerebral cortex.



3-Lissencephaly (agyria): Is characterized by an absence of normal gyration leading to a smooth-surfaced brain. The cortex is abnormally thickened and is usually only four-layered. Single-gene defects have been identified in some cases of lissencephaly.

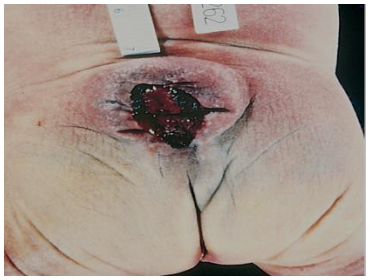
***Note : Encephaly = brain Cephaly =bone**

Neural Tube Defects

• Failure of a portion of the neural tube to close, or reopening after successful closure, may lead to one of several malformations. All are characterized by abnormalities involving some combination of neural tissue, meninges, and overlying bone or soft tissues

•Most frequent CNS malformations

1-Myelomeningocele: Is an extension of CNS tissue through a defect in the vertebral column.



• They occur most commonly in the lumbosacral region

• Patients have motor and sensory deficits in the lower extremities and problems with bowel and bladder control

• The symptoms derive from the abnormal spinal cord in this region, and are often compounded by infections extending from thin or ulcerated overlying skin.

2-Anencephaly is a malformation of the anterior end of the neural tube, with absence of the brain and top of skull

3-Encephalocele: is a diverticulum of malformed CNS tissue extending through a defect in the cranium it most often involves the occipital region or the posterior fossa

Posterior Fossa Anomalies

- The most common malformations in this region of the brain result in either misplaced or absent cerebellum
- Typically, these are associated with hydrocephalus.
- The Arnold-Chiari malformation (Chiari type II malformation) consists of:
  - A small posterior fossa
  - A misshapen midline cerebellum
  - Downward extension of *vermis* through the foramen magnum
  - Hydrocephalus
  - A lumbar Myelomeningocele

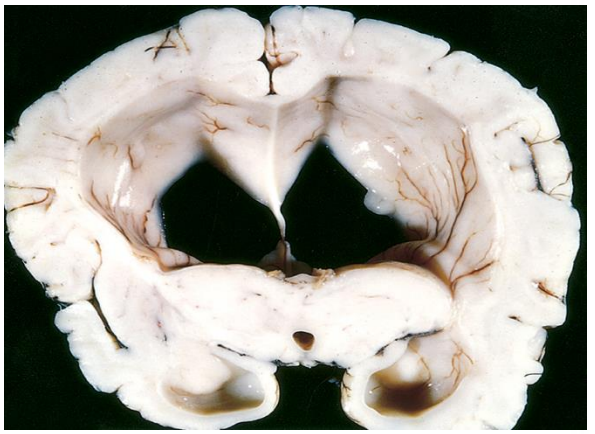
Hydrocephalus

• Hydrocephalus refers to the accumulation of excessive CSF within the ventricular system

• Most cases occur as a consequence of impaired flow or impaired resorption of CSF
• In rare instances (e.g., tumors of the choroid plexus), overproduction of CSF may be responsible

• When hydrocephalus develops in infancy before closure of the cranial sutures, there is enlargement of the head

• Hydrocephalus developing after fusion of the sutures, in contrast, is associated with expansion of the ventricles and increased intracranial pressure, without a change in head circumference



• Noncommunicating hydrocephalus: obstacle to the flow of CSF within the ventricular system, then a portion of the ventricles enlarges while the remainder does not. and is most commonly seen with masses at the formamen of Monro or aqueduct of Sylvius

• Communicating hydrocephalus: all of the ventricular system is enlarged; here the cause is most often reduced reabsorption of CSF

What can cause hydrocephalus?

- Hypersecretion of CSF: e.g. choroid plexus tumor

- Obstructive hydrocephalus (Not important)
  - Obstruction of the foramina of Monro (colloid cyst)
  - Obstruction of the 3rd ventricle (Pilocytic astrocytoma)
  - Obstruction of the aqueduct (aqueductal stenosis or atresia and posterior fossa tumors)
  - Obstruction of the foramina of Luschka or impairment of flow from the 4th ventricle (Chiari malformation, meningitis, subarachnoid hemorrhage, posterior fossa tumors).
  - Fibrosis of the subarachnoid space (meningitis, subarachnoid hemorrhage, meningeal dissemination of tumors)

- Defective filtration of CSF: postulated for low-pressure hydrocephalus.

Summary from Robbins

SUMMARY

Congenital Malformations and Perinatal Brain Injury

- Malformations of the brain can occur because of genetic factors or external insults.
- The developmental timing and position of the injury determine its pattern and characteristics.
- Various malformations stem from failure of neural tube closure, improper formation of neural structures, and altered neuronal migration.

Homework

Meningocele: protrusion of the membranes of the brain or SC through a defect in the cranium or spinal column.

Polymicrogyria: characterized by increased number of irregularly formed gyri that result in a bumpy surface.

What is the difference between microcephaly and microencephaly?
Microcephaly: Small head (skull).
Microencephaly: Small brain.

Hydrocephalus Ex Vacuo: an enlargement of cerebral ventricles and subarachnoid spaces, and is usually due to brain atrophy (as it occurs in dementias)

MCQs

Q1: A pregnant female comes to you for her ultrasound. On the screen you can see that the brain is shrunken compared to the size it should be at this point of her pregnancy. You run an HIV test and it is (-). you revise the history and it turns out that she "enjoys" the occasional evening drink after work. Which of the following based upon this case would you expect to see within the fetus' brain:

A) Spina bifida as well
B) A decrease in the number of neurons
C) Anencephalus

Q2: During your OB/GYN rounds, a female gave birth to a baby with arnold chiari. what you would expect to see in that baby:

A) Anencephalus
B) Myelomeningocele
C) Microcephaly
D) Myeloschisis

Q3: If a female gives birth to a baby with microenccephaly, the first infectious cause that would come to you mind would be:

A) HIV
B) TB
C) Brucella
D) Toxoplasmosis

Q4: The pattern of CNS malformation depends on the region of the brain which primarily depends on:

A) Location of injury
B) Timing of development
C) Alcohol consumption

1- B
2- B
3- A
4- B

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