Common Congenital Neurosurgical Diseases

Surgery Team
MED 433

MED 433

Objectives :

1. Introduction to Neurosurgery
2. Approaching neurological symptoms
3. Congenital diseases
    a. Hydrocephalus
    b. Neural Tube Defect
    c. Chiari Malformation
    d. Dandy-Walker Malformation
    e. Craniosynostosis
    f. Arachnoid Cyst

In this lecture we answered the doctor questions from "**Neurology and Neurosurgery illustrated" By Kenneth W. Lindsay** (Doctor A. AlAjlan recommended this book).

Doctor mentioned 2 topics which is not covered in the slides (ARACHNOID CYST & CRANIOSYNOSTOSIS)

Color Index: Slides & Raslan's () | Doctor's Notes | Extra Explanation | Additional

This work is based on doctor's Slides +Notes and Raslan's only (Does not include the book)

Neuroscience specialties

Basic science

- Neurophysiology
- Nerve conduction study

Clinical

- Neurology
- Neurosurgery
- Psychiatry

- Vascular
- Oncology
- Skull-Base
- Spine
- Pediatrics
- Functional
- Epilepsy
- Peripheral Nerves
- Critical Care
- Trauma

Not important, just for you

Mind map

- **The lecture parts**
  - Headache
    - **Primary**
    - Is the headache serious?
    - **Secondary**
  - **1.Hydrocephalus**
  - **2. Neural Tube Defect**
  - **3.Anencephaly**
  - **4. Other Congenital Abnormalities**
    - **1- Encephalocele**
    - **2- Arachnoid cyst**
    - **3- Chiari Malformation**
    - **4- Craniosynostosis**
    - **5. Diastemamyelia**

Headache

Primary

Are benign, recurrent headaches not caused by underlying disease or structural problems
Examples: Migraines, tension-type headaches, cluster headaches

Secondary

Are caused by an **underlying disease**

Vascular
Inflammatory/Infectious
Neoplastic
Degenerative/Deficiency/Drugs
Idiopathic/Intoxication/Iatrogenic
Congenital
Autoimmune/Allergic/Anatomic
Traumatic
Endocrine/Environmental
Metabolic

→ Certain "red flags" indicate a secondary headache.

Red flag:

-In general:

- People complaining of their "first" or "worst" headache
- Progressively worsening

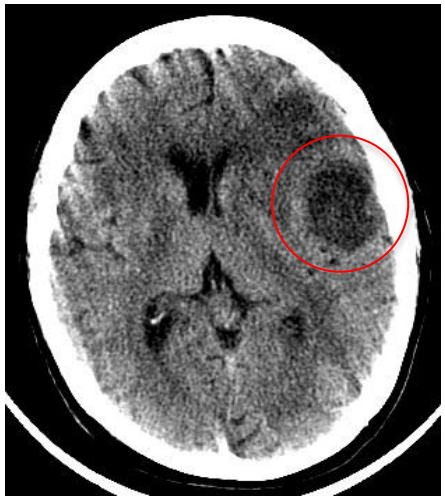
The American Headache Society recommends using "**SSNOOP**", a mnemonic to remember the red flags for identifying a secondary headache:

1. **S**ystemic symptoms (fever or weight loss)
2. **S**ystemic disease (HIV infection, malignancy)
3. **N**eurologic symptoms or signs
4. **O**nset sudden (thunderclap headache)
5. **O**nset after age 40 years
6. **P**revious headache history (first, worst, or different headache)

Is the headache serious?

The American College for Emergency Physicians published criteria for **low-risk headaches**:

-Age **younger than 30** years
-Features typical of **primary headache** (e.g. a Female 20-year-old has a headache affects half of the head, triggered by light, she has a family history of migraine etc.)
-**History** of similar headache
-**No abnormal findings** on neurologic exam
-**No concerning change** in normal headache pattern
-**No high-risk comorbid** conditions (for example, HIV, lymphoma)
-No **new concerning** history or physical examination findings (e.g. a patient complaining of global headache for a 10 years and changed recently to local area > serious problem)



CT-scan of a patient presented with a headache. What is your differentials?

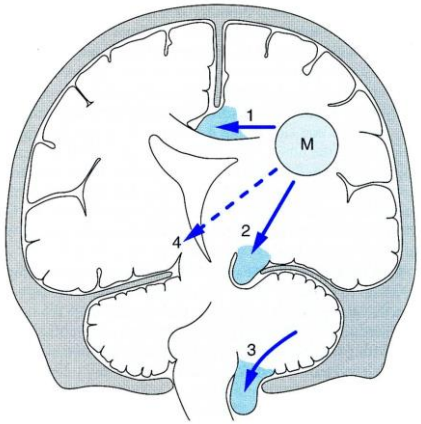
Vascular
Inflammatory/Infectious
Neoplastic
Degenerative/Deficiency/Drugs
Idiopathic/Intoxication/Iatrogenic
Congenital
Autoimmune/Allergic/Anatomic
Traumatic
Endocrine/Environmental
Metabolic

Most common in brain:

- Neoplastic (primary & secondary)
- Vascular
- Congenital
- Inflammatory
- Infectious

Any lesion in the brain can be presented in 3 ways:

1. **Local compression** (e.g. local compression on the motor cortex will lead to weakness of the opposite site)
2. **Mass effect & Herniation** (Mass pushes brain parenchyma results in midline-shift).
   - There are 4 types of herniation (see the picture).
     1. SUBFALCINE 'MIDLINE' SHIFT
     2. Tentorial Herniation (lateral): a unilateral expanding mass causes **tentorial (uncal) herniation** as the medial edge of the temporal lobe herniates through the tentorial hiatus. As the intracranial pressure continues to rise, 'central' herniation follows.
     3. TONSILLAR HERNIATION
     4. TENTORIAL HERNIATION (central)



Will be discussed in more details in ICP lecture

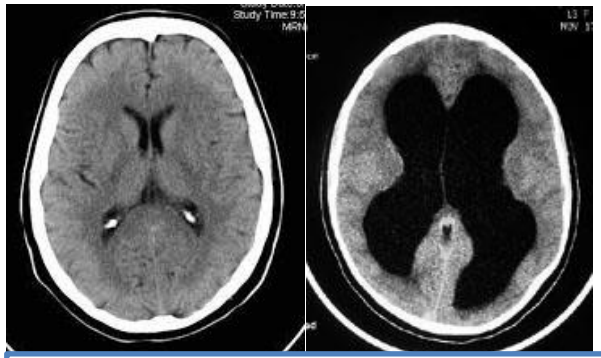
3. **High Intracranial pressure.**
   - Headache
   - Vision disturbance
   - vomiting
   - Altered level of consciousness

1. Hydrocephalus

Hydrocephalus is an **increase in the CSF volume, associated with increased ventricular size**
Not the same as Ventriculomegaly (increase in ventricular size without increasing the pressure)

Causes or types of Hydrocephalus:

1. **Communicating Hydrocephalus**
   a. Overproduction of CSF
   b. Under absorption of CSF **(common)**
2. **Non-communicating hydrocephalus=obstructive hydrocephalus**
   a. Obstruction of CSF flow
      i. Congenital
      ii. Acquired

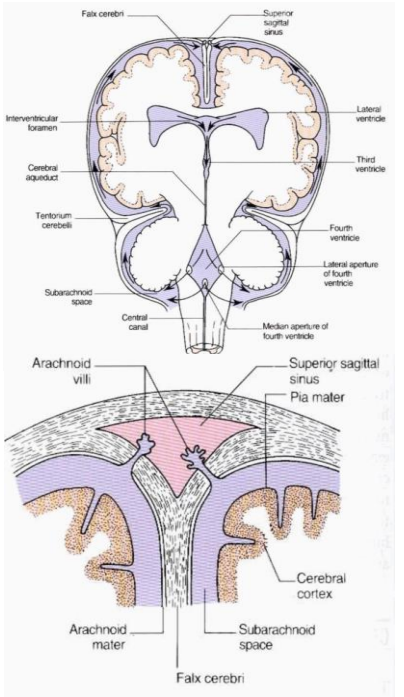


Left: normal. Right: Hydrocephalus

Physiology:

- Total volume of CSF in the ventricles varies from 5-15 ml in neonates to 150 ml in adults.
- **Produced mainly by choroid plexus.and the extracellular fluid of the brain.**
- Rate of production is 0.3-0.4 ml/minute.
- CSF production: (Overproduction)
  - Lateral ventricles has Choroid plexus (Site of CSF production)
- CSF Pathway: (Any obstruction will lead to non-communicating hydrocephalus)
  - Then CSF moves down to third ventricle through interventricular foramen
  - Then travels down to fourth ventricle through cerebral aqueduct
  - It leaves the ventricular system through the 3 apertures of the 4th ventricle ;
    - median foramen of Magindi &
    - 2 lateral foramina of Leushka
- CSF absorption: (Under absorption)
  - Arachnoid villi, and
  - Arachnoid granulation

Everyday the plexus produces 500ml of CSF



Notes from Raslan's

Hydrocephalus

- It is an accumulation of CSF within the cerebral ventricle and is usually associated with altered ICP
- The pressure is usually high, and sometimes normal, but rarely low (negative pressure hydrocephalus)
- When the ventricles are large but the patient is asymptomatic, that is not hydrocephalus; it's just hydrocephalus exvacuo "old name" or ventriculomegaly. So when you see large ventricles, it does not indicate hydrocephalus UNLESS there are symptoms of pressure changes of the brain

CSF production

- Rate of CSF production is 0.3-0.4 ml/minute → 500 ml CSF/day – 150 ml in the CNS → 350 CSF absorbed every day.
- It is a process of active formation and it does not stop it
- It may be affected by ↑ ICP BUT it does not stop it
- If there is any problem affecting absorption or the pathway of CSF → accumulation of CSF and ↑ ICP (which decreases CSF production)

Absorption of the CSF into the blood stream takes place in the superior sagittal sinus through structures called arachnoid villi .

Types Of Hydrocephalus

Communicating

- All ventricles are dilated
- Overproduction or under absorption of CSF
- No obstruction in the pathway of CSF within the ventricles (the ventricles can communicate with each other)

Non-communicating

More details in the next slide

- Partial dilatation
- Blockage of the flow of CSF
- Obstruction within ventricles or the pathway of CSF
- Congenital, since birth
- Acquired, develops after birth as a result of injury, tumors or meningitis.

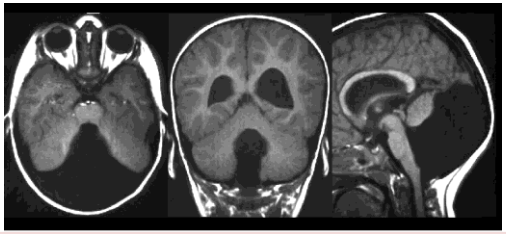
Non-Communicating Hydrocephalus Causes

Congenital (Primary)

More details in the next slide

1. Aqueductal anomalies = **Aqueductal stenosis** **(most common Causes of Congenital obstructive hydrocephalus)**
2. Dandy Walker malformation (cyst in posterior fossa causes obstruction in the ventricular pathway with small cerebellum)
3. Chiari II malformation (Will be discussed in details in slide 10).
4. Myleomeningocele
5. Intrauterine viral infection
6. Toxoplasmosis
7. Congenital tumors
8. Vein of Galen aneurysms (massive aneurysmal dilatation).
9. Chromosomal anomalies (trisomy 13 and 18)
10. Congenital or primary hydrocephalus.

important



Huge cyst in the **posterior fossa causing** Obstruction with small cerebellum (Dandy Walker malformation)

Acquired (Secondary)

1.Germinal plate hemorrhage: "immature blood vessel walls''
Leads to intracranial hemorrhage inpremature babies <1500 gm (30%-40%)

- In premature baby the blood vessels are weak. So, any spike in blood pressure can cause rapture and Communicating + non-communicating hydrocephalus.

2.Post-meningitis **Communicating Hydrocephalus**
(because The fibrosis after the inflammation →↓absorption of CSF
Especially G-ve organisms (i.e. E. coli)

3. Tumors **(Most common cause)**
4.Sub-arachnoid hemorrhage (Due to trauma)
5.Severe TBI (Traumatic Brain Injury)

Doctor said: 'Just memorize the highlighted in red.

Important 3 causes of communicating hydrocephalus:
- Post-hemorrhagic
- Post-meningitic
- Post-traumatic

3 posts for acquired causes!
Remember that the cause of acquired hydrocephalus is usually outside the ventricles

Notes from Raslan's Not important

Congenital (Primary)

Aqueductal Anomalies

- Aqueduct is the passage of CSF between the 3rd & 4th ventricles-passes in the midbrain .It is still know as aqueductal stenosis, however by definition, the correct term is "aqueductal atresia" because no aqueduct is found in most of cases - but in some cases (who develop hydrocephalus late in their childhood) it allows a small amount of CSF to pass through so it is usually called stenosis

Dandy Walker Malformation (Large Dandy Walker Cyst)

- By definition it is congenital hypoplasia or even aplasia of cerebellum associated with formation of a large CSF cavity within the posterior fossa due to the obstruction of CSF flow by a large cyst (which doesn't allow CSF to pass from the 4th ventricle and circulate around the brain)
- There are different types of Dandy Walker cyst according to the volume of cerebellum that's involved Most of cases of Dandy Walker malformation are associated with hydrocephalus
- In short: large CSF cyst on posterior fossa due to agenesis of the cerebellum that communicates with the 4th ventricle, and causes hydrocephalus

Dandy-Walker malformation is characterized by agenesis or hypoplasia of the cerebellar vermis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa
Presentation: Incoordination, ataxia, nystagmus

Vein Of Galen Aneurysms

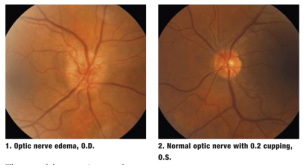
A large vascular malformation where there is a direct communication between the arterial system and venous system (shunting), leading to dilatation of the **Vein of Galen** (one of the deep venous structures in the brain) and to **obstructive hydrocephalus**.

What is the clinical manifestation for such cases in neonates?

o 1st and most important is heart failure (the size of the arteriovenous shunt that can steal 80% or more of the cardiac output), then symptoms of hydrocephalus (developmental delay, seizures, headaches)

Rx: treat the cause, which is the aneurysm (stop passage of blood from artery to vein by embolization) - and no need for shunt to treat the hydrocephalus o Also shunts can burst the aneurysms and cause fatal hemorrhage

important Clinical features of hydrocephalus important

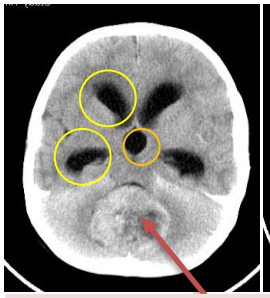
Infants	Adults
1. **Increasing head circumference.** 2. Irritability, lethargy, poor feeding, and vomiting. 3. Bulging anterior fontanelle. 4. Widened cranial sutures. 5. McEwen's (cracked-pot) sign with cranial percussion. 6. Scalp vein dilation (collateral venous drainage). 7. Sunset sign (downward deviation of the eyes). 8. Episodic bradycardia and apnea.	1. Headaches (**Most common symptom)** (Usually in the early morning. Why? Because during sleep, the patient will hypo ventilate, which will lead to ↑ CO2 → ↑ vasodilation → blood stasis & ↑ intracranial pressure) 2. Nausea 3. Projectile Vomiting (The symptoms are usually relived after vomiting) 4. Decreased level of consciousness 5. Focal neurological deficit (rare) 6. Papilledema 7. Seizures  1. Optic nerve edema, O.D. 2. Normal optic nerve with 0.2 cupping, O.S.

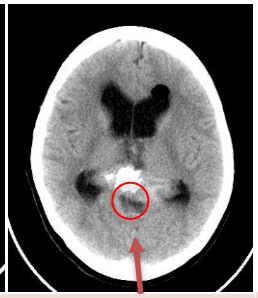
Investigations

CT or MRI

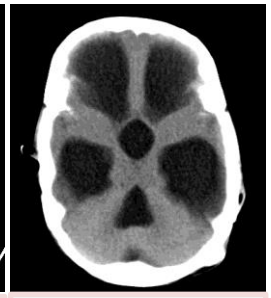
The pattern of ventricular enlargement can help delineate the cause:

- Lateral & 3rd ventricle dilatation
  - With normal 4th ventricle: suggests aqueduct stenosis
  - With deviated or absent 4th ventricle: suggests posterior fossa tumor
- Generalized dilatation: suggests a communicating hydrocephalus.





Dilated lateral ventricle (yellow), dilated third ventricle (orange) with normal 4th ventricle (red).

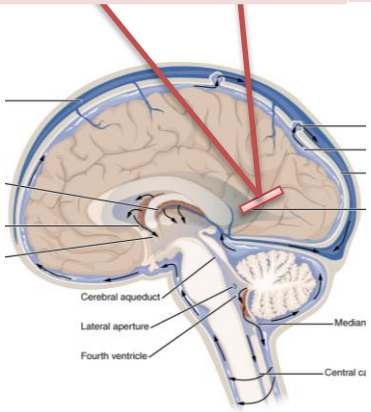


Generalized dilation (communicating)

X-ray is not a diagnostic procedure for hydrocephalus now You will see **widening of the suture of skull** secondary to increased intracranial pressure

Classical Radiological Sign:

-Separation of sutures -Pressure of the gyri on the bone



Management

- **Communicating:** Medical or surgery
- **Non-communicating:** surgery

More details in the next slide

Notes from Raslan's Not important

❖ Surgical Treatment

Endoscopic choroid plexectomy was tried but found unsuccessful. Not done now because all children die during the operation

Replaced now by **Choroid plexus coagulation** We use endoscope, introduce telescope in the ventricle, and it helps in reduction of CSF formation

Endoscopic third ventriculostomy: 1st line treatment now

❖ Intracranial Shunts

In obstructive hydrocephalus where the subarachnoid spaces are still patent

1.Endoscopic Third Ventriculostomy

The endoscope is passed through a burr hole to the third ventricle where the floor is fenestrated just anterior to mamillary bodies. (So we bypass the obstruction in the aqueduct or posterior fossa).The hole is enlarged by introducing the endoscope or an inflatable balloon

2. Ventriculocisternostomy: Shunt between lateral ventricle and the cisterna magna.

It has high morbidity and mortality. (Not done any more)

❖ Extracranial Shunt:

- From the ventricular system, usually the lateral into another body cavity; the peritoneal cavity (VP Shunt), right atrium (VA Shunt) and occasionally pleural cavity.
- **Aim** is to normalize the intracranial pressure
- Specially designed shunt valve with the appropriate rate of flow and pressure. Regulate the CSF flow in a unidirectional way.
- Shunts are made of silicon which is well tolerated by the body. It causes minimal or no tissue reaction or intravascular thrombosis

Extra-cranial Shunt: if you have communicating hydrocephalus or if you don't have the facility or capability to do endoscopic third ventriculostomy

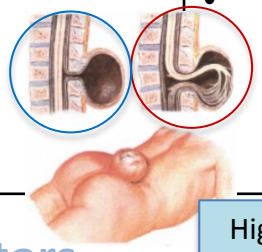
Complicationsof VP SHUNTS: *For more details check Raslan*

- Obstruction
- Infection
- Or less commonly :
  - subdural collection from over drainage
  - slit ventricle syndrome "causes collapse of ventricles"
  - disconnection or fracture of shunt tubing
  - seizures .

2. Neural Tube Defect

Spinal Dysraphism: Failure of closure of posterior neural arch

Types of Myelodysplasia		
1. Spina bifida occulta	**2. Meningocele**	**3. Myelomeningocele**
• 5-10% of population • not clinically significant • tuft of hair, dimple sinus or port wine stain • high incidence of underlying defect • **no treatment required**	• Cystic CSF-filled cavity lined by meninges • no neural tissue (see the blue circle) • communicates with spinal canal • look for other cong. Anomalies • Seldom any neurological deficit • U/S or MRI	• Spinal cord and roots protrude through the bony defect, • lie within cystic cavity (see the red circle) • observe limb movements (degree & level of neurological damage) • Note dilated bladder & patulous annual sphincter • U/S or MRI • Look for any associated congenital anomalies, e.g. hydrocephalus, scoliosis, foot deformities.



Higher level = more neural deficit i.e. if the lesion is in L2 the patient won't be able to walk. But if the lesion is in L5 the walking will be intact.

Incidence & Risk factors

- 2 affected in 1000 birth
- Low in western countries but very high in KSA (antenatal care).
- **Folic acid deficiency**
- Teratogens
- Genetic (5% if siblings are affected)

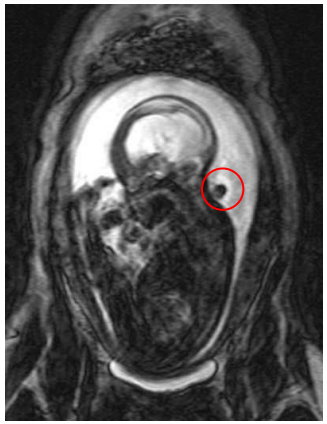
Prevention

Give folic acid supply during pregnancy

Antenatal diagnosis

- Maternal U/S
- MRI
- Serum/amniotic fluid for **alpha-fetoprotein & acetylcholinesterase**
- Contrast enhancing amniography
- possibility of therapeutic abortion

important



3.Anencephaly (No brain)

Defective closure of the **rostral** neural tube results in anencephaly or encephalocele Neonates with anencephaly have a rudimentary brainstem , or midrain , no cortex or cranium
Rapidly fatal condition if born alive

Clinical Manifestations

According to the level of spinal bifida (Meningomyelocele)

- **Lower Lumbar** (L5,S1) → Distal weakness in the feet and sphincters incontinent
- **Upper lumbar** (L1) → Complete paraplegia of the lower limbs

Incidence

Teratogens; Sodium Valporate
Associated with Hydrocephalus, Chiari II and aqueduct forking

Causes

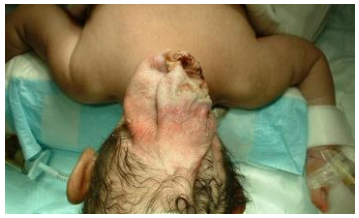
1- The main cause is Folic acid deficiency; since the development of the nervous system end in day of 28 of pregnancy. The woman should take folic acid when they plan to get pregnant.
2- Teratogens; Sodium Valporate (antiepileptic drug , if you start giving this medication, once she becomes pregnant she may develop baby with spina bifida > so you need to change it before pregnancy)

Myeloschisis or rachischisis: where there is defect in the bone and the spinal cord exposed outside no skin covering > type of Meningomyelocele Sever neurological deficit in the lower limbs.

4. Other Congenital Abnormalities

Encephalocele **Large cyst in the skull contains CSF or brain**

- Same pathophysiology as neural tube defect but usually happens in the occipital
- may contain occipital lobe, or cerebellum
- often associated with hydrocephalus
- Immediate treatment if ruptured
- outcome depends upon contents



Arachnoid cyst important

- Cystic collections of CSF-like fluid of developmental origin
- Arachnoid cysts may gradually increase in size, either due to CSF being driven in through a valve-like opening or by active secretion of fluid from the cyst wall.
- Occasionally patients present with mass effects, or in children with asymmetric cranial enlargement, macrocephaly and/or psychomotor retardation. **More often they are discovered incidentally** on CT or MRI.

Chiari Malformation **Cerebellum herniation through foramen magnum** important

When part of the cerebellum is located below the foramen magnum, it is called a Chiari malformation.

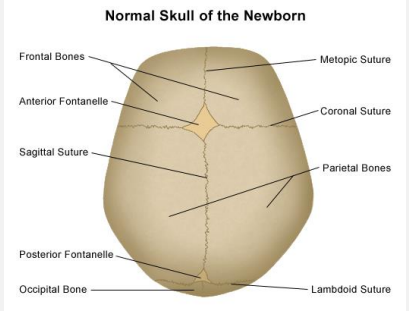
Type I	Type II
• Extension of the cerebellar tonsils only into the foramen magnum, without involving the brain stem. • Primary or secondary (big cyst, hydrocephalus & post lumbar puncture) • Presentation: headache, ataxia, nystagmus and hydrocephalus. • Benign form	• Type II is a triad composed of: 1. Extension of both cerebellar and brain stem tissue into the foramen magnum **2. Myelomeningocele** 3. Hydrocephalus • Malignant

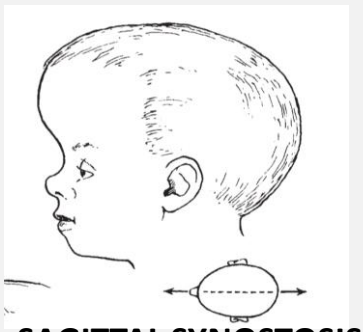
3. Other Congenital Abnormalities

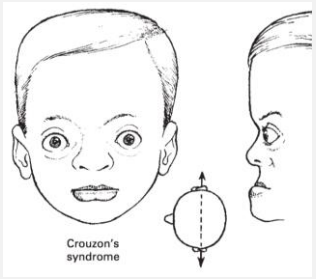
Craniosynostosis *For more details check Raslan*

Premature fusion of one or more sutures results in restricted growth of bone perpendicular to the suture and exaggerated growth parallel to the suture. The effect depends on the site and number of sutures involved.

- **Sagittal Synostosis**
  Lateral growth is restricted, resulting in a long narrow head with ridging sagittal suture (scaphocephaly).
- **Coronal Synostosis**
  - Involvement of several sutures (**oxycephaly**) results in skull expansion towards the vertex.
  - Expansion occurs in a superior and lateral direction (**brachiocephaly**). (This produces a short anterior fossa, shallow orbits and hypertelorism (widening of the interocular distance)).
  - Bilateral coronal synostosis commonly occurs as one of several congenital defects incorporated in Crouzon's and Apert's syndromes.
- **Pansynostosis**
  (all sutures affected) results in failure of skull growth with a symmetrical abnormally small head and raised intracranial pressure.
- **Posterior plagiocephaly**
  (flattening of the back of the head) An increasing number of infants present with this condition perhaps resulting from the 'back to sleep' campaign.




SAGITTAL SYNOSTOSIS



CORONAL SYNOSTOSIS
Exophthalmos, elevated ICP and visual impairment from papilloedema may result.

3. Other Congenital Abnormalities

Not important

Diastemamyelia (from Raslan's)

A bone or fibrous band divides spinal cord in two longitudinal sections

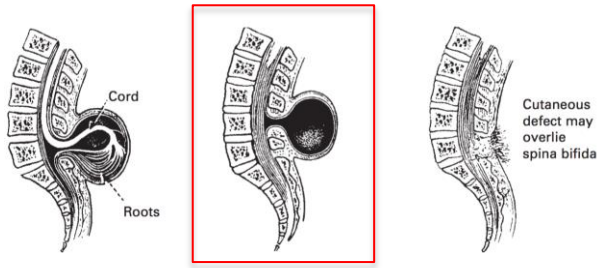
Associated lipoma may be present, which tethers cord to vertebra

Signs &Symptoms include weakness, numbness in feet, urinary incontinence, decreased or absent reflexes in feet

Dx: CT

Rx - surgery to free cord

MCQs

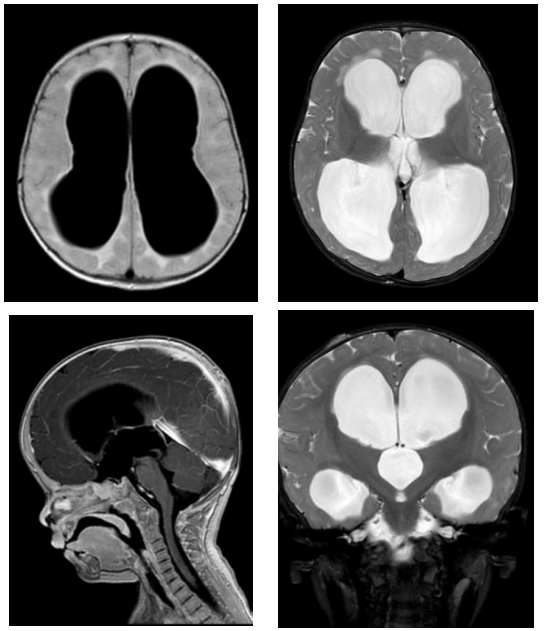


Q1: What is the abnormality labeled in red?

a. Spina bifida occulta
b. Meningocele
c. Myelomeningocele

Q2: 1-year-old patient presented with increase head circumference. From the history which was given by her mother she had a vomiting. Clinical examination revealed sunset eye. An MRI was done. What is your judgment?

a. Aqueductal stenosis
b. Dandy Walker malformation
c. Chiari Malformation type II
d. Communicating Hydrocephalus



Q3: Which of the following is NOT ASSOCIATED with Chiari type II?

a. Hydrocephalus
b. Brain stem herniation into the foramen magnum
c. Meningocele
d. Myelomeningocele

Ans: 1:b 2:a 3:c

Thank You..

Done By :

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