SPINAL DYSRAPHISM (SPINA BIFIDA)

Failure of the neural tube to close fully, produce one of many pattern of neurospinal dysraphism (spina bifida).

SPINA BIFIDA OCCULTA

**Epidemiology:** it is found in 10%-20% of population, most common at L5 or S1

**Definition:**
Congenital absence of spinous process and variable amount of lamina (posterior vertebral arch), no visible exposure or herniation of meninges or neural tissue.

**Etiology:** failure of fusion of the posterior neural arch.

**Clinical features:**
no obvious clinical signs, presence of lumbosacral cutaneous abnormalities (dimple, sinus, port-wine stain, or hair tuft) should increase suspicion of an underlying anomaly (lipoma, dermoid, diastomatomyelia). the defect may be palpable.

**Investigation:**
Plain film (absence of the spinous process along with minor amount of the neural arch, U/S, MRI to exclude spinal anomalies

**Treatment:** requires no treatment

hairy patch, an area of pigmentation, fatty lump or the dermal sinus), often an incidental finding

usually of no clinical importance when it occurs alone however it may be occasionally be associated with diastematomyelia, tethered cord, lipoma, or dermoid, epidermoid tumors with thicking of the filum terminale.

MENINGOCELE (SPINA BIFIDA APERTA)

**Definition:**
Herniation of meningeal tissue and CSF through a defect in the spine, without associated herniation of neural tissue

**Etiology:** Primary failure of neural tube closure.

**Clinical features:**
Most common in lumbosacral area, usually no disability, low incidence of associated anomalies and hydrocephalus.

**Investigation:** Plain films, CT, MRI, U/S, echo, genitourinary investigation.

**Treatment:** Surgical excision and tissue repair (excellent result)
**MYLOMENINGOCELE**

**Definition**: herniation of meningeal and CNS tissue through a defect in the spine

It is the most common significant birth defect involving the spine, **1-2/1000 live births**

**Etiology**: same as meningocele ......?

- History of affected pregnancy should take 4 mg of folic
- All women of childbearing age who have not had a fetus or infant with a neural tube defect should consume at least 400 μg/day of folate
- Embryologically the abnormality manifested between 3-4 weeks of gestation

Hydrocephalus develops in 80% of patients with mylomeningocele

**Diagnosis**

Prenatal diagnosis

- Alpha-fetoprotein measurements
- Ultrasound

**Management**

**ADMISSION**

1. Assessment and management of the lesion
   - Measure size of defect
   - Assess whether the lesion is ruptured or not; in ruptured one start antibiotics which is not necessary in unruptured MM
   - Cover the lesion with sterile gauze soaked in lactated ringers or normal saline
   - Surgical closure within 36 hr if there is no contraindication. To preserve neurological status and prevent CNS infections

2. Neurological assessment and management:

   Lower limbs examination for spontaneous movements, deformity, response for painful stimulus, reflex and autonomous activity.

3. Additional assessment and management: management of MM need team work neurosurgeon, neonatologist to assess for other abnormality, urologist and orthopedic consultation for severe kyphotic or scoliotic spine deformities and for hip or knee deformities

**Outcome**

Without any treatment, 14-30% of MM infants survive infancy but with modern treatment, 85% survive.

70%-80% have normal IQ.
3-10 have normal urinary continence.

Some may use wheelchairs

The most common cause of mortality and morbidity are shunt complications and complications from Chiari malformation.
ANENCEPHALY:

Due to failure of fusion of the anterior neuropore. Neither cranial vault nor the scalp covers the partially destroyed brain. Parts of the brain stem and spinal cord may be missing or malformed. Infants are stillborn or die within days or weeks.

ENCEPHALOCELE:

Is a protrusion of nervous tissue and meninges through a skull defect. The defect is caused by incomplete closure of the cranial vault (cranium bifidum). Encephaloceles usually occur in the midline and protrude anywhere along a line from the occiput to the nasal passages but can be present asymmetrically in the frontal or parietal regions. Small encephaloceles may resemble cephalhematomas, but x-rays show a bony skull defect at their base.

HYDRANENCEPHALY

is an extreme form of porencephaly in which the cerebral hemispheres are almost totally absent. Usually, the cerebellum and brain stem are formed normally, and the basal ganglia are intact. The meninges, bones, and skin over the cranial vault are normal. Often hydranencephaly is diagnosed by prenatal ultrasonography. Neurologic examination is usually abnormal, and the infant does not develop normally. Externally, the head may appear normal, but when transilluminated, light shines completely through.

CT or ultrasound confirms the diagnosis. Treatment is supportive, with shunting if head growth is excessive.

CHIARI MALFORMATION

caudal displacement of cerebellum with tonsillar herniation below the foramen magnum

DANDY-WALKER MALFORMATION:

atresia of the foramina of Magendie and Luschka. This result in agenesis of the cerebellar vermis with a large posterior fossa cyst communicating with an enlarge 4th ventricle. Hydrocephalus occurs in 90% of cases.

Figure. Typical features of Dandy-Walker variant on brain MRI: (A) large fourth ventricle in axial T1-weighted image; (B) small cerebellar vermis in axial T2-weighted image; (C) hydrocephalus in axial T1-weighted image; and (D) dysgenesis of the corpus callosum in sagittal T1-weighted image.

SPLIT CORD MALFORMATION:

Defined as two hemicords either each within a separate dural tube separated by a bony septum (diastematomyelia) or consists of two hemicords within a single dural tube separated by a nonrigid fibrous median septum (diploomyelia).

CRANIOSYNOSTOSIS

Is the premature closure of a cranial suture, which causes abnormal calvarial growth. Incidence is 0.6/1000 live birth.
HYDROCEPHALUS

Physiology and circulation of CSF
The normal volume of circulating CSF is around 140ml
daily production is about 500ml/day
the CSF volume is replaced approximately ...?.......times daily

Function:
1. The CSF is both protects and support the brain and spinal cord.
2. transport medium for transmitters and as a method of removing the end-products of metabolism.

CSF is mainly produced by the choroid plexus

Definition of hydrocephalus
It is defined as a disproportionate increase in the amount of CSF within the cranium, usually in association with a rise in ICP.
(Ventricular enlargement with excessive CSF) And it should be differentiated from hydrocephalus ex vacuo (due to brain atrophy)
Incidence of congenital hydrocephalus is 1-2/1000 live births

Types of hydrocephalus
Communicating (non-obstructive): CSF circulation blocked at the level of the basal cisterns, the subarachnoid space or the level of the arachnoid granulation.
Non-communicating (obstructive): the normal pathways of CSF flow are for some reason occluded such as aqueduct stenosis or as a result of a local compression from a tumor.(enlargement of ventricles proximal to the block)
Further types of classifications

Acute and chronic

Congenital and acquired

Can be classified by etiology

*posttraumatic*,
*post hemorrhagic*
*postmeningitis*

Etiology

imbalance has occurred between the normal physiological production of CSF and its absorption

Congenital .....?

aqueduct stenosis
Chiari malformations
Dandy-Walker malformation

Acquired

infection (post-meningitis)
post – hemorrhagic(SAH,IVH)
secondary to masses( non- neoplastic like vascular malformation, tumors like posterior foss tumors, pituitary and colloid cyst )

Clinical features

*In neonatal period*

1. Cranium enlarges at a rate more than the facial growth (craniofacial disproportion).
2. irritability, poor head control, nausea and vomiting
3. Fontanel full, bulging and wide.
4. Thin and glistening scalp with enlargement and engorgement of scalp veins; due to reversal of flow from intracerebral sinuses due to increase intra cranial pressure.
5. Macewen’s sign (cracked pot sound on head percussion)
6. Sixth nerve (abducens) palsy, long intracranial course make this nerve very pressure sensitive.
7. Setting sun sign, (upward gaze palsy, from pressure on region of suprapineal recess.
8. Hyperactive reflexes.
9. Irregular respiration with apneic spells.
10. Separation of cranial sutures (sutures diastasis).

*In older children and adult (with rigid cranial vault)*

1. headache,
2. vomiting.
3. gait changes,
4. aducent palsy ,
5. upgaze palsy.( Parinaud’s syndrome )
6. Papilledema.
7. Loss of consciousness
Investigation

Ultrasonography to visualize the ventricular system (when the anterior fontanelle is patent). Also for antenatal diagnosis.

CT and/or MRI of the head; it is important to exclude any abnormal masses and to study the size and the shape of the ventricles, and some time needs contrast study.

LP in cases of communicating hydrocephalus for both diagnostic and therapeutic.....(NPH)

Management

Medical ..........ineffective

- Acetazolamide
- Frucemide

Surgical treatment

1. Think about the Surgical treatment
2. reducing the CSF production ..... 
3. bypassing the blockage to normal CSF flow ..... 
4. drainage of CSF externally ..... 
5. drainage of CSF to another absorptive viscus
6. Treat the cause (remove the obstruction)

Surgical treatment

removal of obstruction like posterior foss tumor

shunting: many types of shunts

1. ventriculo-peritoneal(VP), (the most common method)
2. ventriculo-atrial(VA)
3. Torkildsen shunt (overcame the aqueduct stenosis by passing a catheter from the lateral ventricles into the cisternal space)
4. Lumbo-peritoneal shunt.
5. External drainage , temporary for management for acute hydrocephalus but the risk of infection preclude this as a means of long term management.
6. miscellaneous : the lower end may inserted into the plural space, gall blader, ureter or bladder. (in past)

Cannulation of the aqueduct of Sylvius or third ventriculostomy (by endoscopy or open operation).....the most advanced one.

choroids plexectomy (in past)
Complications of shunts

In general shunts may have this complications

1. infection, usually apparent within the first few weeks or months following implantation, treatment is by removal of the device and antibiotics (intravenous or intrathecal) and usually it due to low grade organisms.

2. Obstruction.

3. Acute hemorrhages

4. shunt malfunction and over drainage resulting in low pressure symptoms

5. hematoma

6. disconnections

7. seizure

8. erosion of skin (in infants) and perforation of viscus

9. metastases of tumors

Normal-pressure hydrocephalus (NPH)

- Normotensive hydrocephalus
- Hakim's syndrome
- Hakim's-Adams syndrome
- Occult hydrocephalus

A broad based shuffling gait (gait apraxia)

Dementia (memory loss and a generalized slowing of thought)

Urinary incontinence

In addition to CT and MRI lumbar puncture (LP) is important in the diagnosis of NPH.

[See the slideshow for the great many images it contains.]