CONGENITAL ABNORMALITIES – NEURAL TUBAL DEFECTS

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LEARNING OUTCOMES

- Definition/overview
- Types
- Pathophysiology
- Causes/ aetiology
- Diagnosis
- Management
- Complications

DEFINITION/OVERVIEW

- Neural tube defects (NTD) are significant birth deformities of the central nervous system
- They occur due to a defect in the neurulation process of embryogenesis.
- They cause birth defects of the brain, spine, or spinal cord
- They happen in the first month of pregnancy, often before a woman even knows that she is pregnant.
- The neural tube forms the early brain and spine.

DEFINITION/OVERVIEW…

- The two most common neural tube defects are spina bifida (spinal cord defect) and anencephaly (brain defect).
- In spina bifida, the fetal spinal column doesn't close completely.
- Because the neural tube is ultimately formed from the migration and fusion of the neural plate, the type and severity of malformation varies based on the location of the defect.
- This includes both cranial and spinal cord malformations

TYPES OF NEURAL TUBAL DEFECTS

- Spina bifida

Anencephaly

TYPES OF NEURAL TUBAL DEFECTS..

- NTDs can be classified as **“open”** or **“closed”** types
- This is based on embryological considerations and the presence or absence of exposed neural tissue
  - i.e., failure of incomplete fusion of the neural plate.
- Open NTDs frequently involve multiple aspects of the CNS (e.g., associated hydrocephalus, Chiari II malformation)
- They are due to failure of primary neurulation

TYPES OF NEURAL TUBAL DEFECTS..

- Thus the neural tube fails to appropriately close along the dorsal midline.
- Neural tissue is completely exposed, or covered by a membrane
- It is associated cerebrospinal fluid (CSF) leakage.
- Open NTD's represent roughly 80% of all NTD's
- The most common being meningocele (spina bifida), myelomeningocele, encephalocele, and anencephaly.

TYPES OF NEURAL TUBAL DEFECTS..

- Closed NTDs are localized and confined to the spine (the brain is rarely affected)
- They result from a defect in secondary neurulation.
- Neural tissue is not exposed and the defect is fully covered by epithelium
- However the skin covering the defect may present as a tuft of hair, dimple, birthmark, or other superficial abnormality.

TYPES OF SPINA BIFIDA

- **Spina bifida cystica:**
- Is a bony defect in the vertebral column that causes a cleft in that column.
- The meningeal membranes that cover the spinal cord and part of the spinal cord protrude through this cleft, and are clearly visible.
- The opening can be surgically repaired, usually shortly after birth.

TYPES OF SPINA BIFIDA..

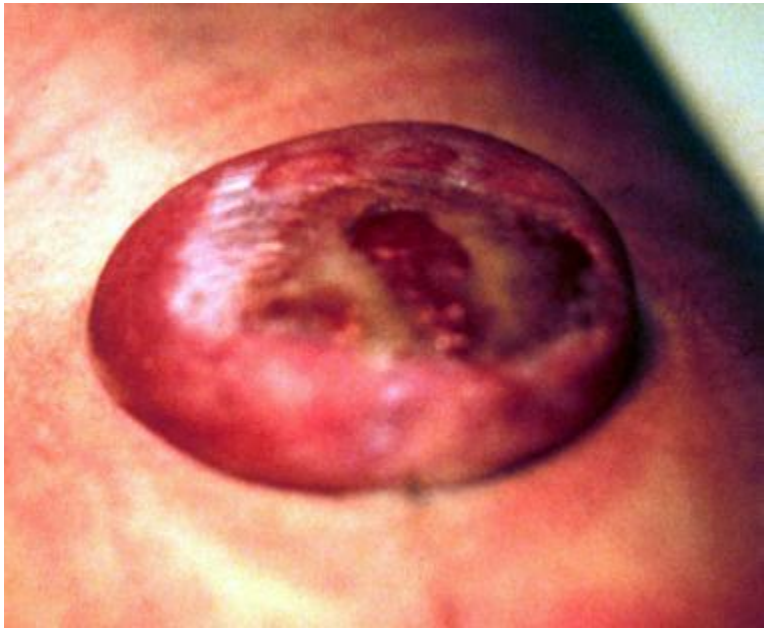
- **Spina bifida occulta** is when a baby's backbone (spine) does not fully form during pregnancy.
- The baby is born with a small gap in the bones of the spine, but is not visible to the eye.
- Spina bifida occulta is common and happens in about 1 out of 10 people.
- Usually, spina bifida occulta causes no health problems.

PRESENTATION OF NTDS

- Spinal presentations include the following:
  - Spina bifida occulta
  - Spina bifida in relation to a dermoid cyst
  - Spina bifida aperta (meningocele, myelomeningocele, meningomyelocele, myeloschisis)
  - Split-cord malformations
  - Diastematomyelia
  - Diplomyelia
  - Caudal agenesis
  - Lipomatous malformations (lipomyelomeningocele)

PRESENTATION OF NTDS…

- Myelomeningocele in a newborn



PRESENTATION OF NTDS…

- Cranial manifestations include the following:
  - Anencephaly
  - Encephalocele (meningocele or meningomyelocele)
  - Craniorachischisis totalis
  - Congenital dermal sinus

PATHOPHYSIOLOGY

- Two distinct and critical processes are involved in the formation of the neural tube:
  - primary neurulation and
  - secondary neurulation (i.e., canalization).
- The neural plate and the notochord are formed during early embryonic development.
- The neural groove develops by the third gestational week
- Primary neurulation (weeks three and four during embryogenesis, forming the early brain and spinal cord)

PATHOPHYSIOLOGY…

- Secondary neurulation (canalization: weeks five and six, forming the early sacral and coccygeal cord)
- By 8 weeks after conception, spinal cord tissue runs the entire length of the spinal cord
- Open NTDs have been suggested to result from defective primary neurulation
- While defective secondary neurulation gives rise to closed NTDs
- However there is still debates on these findings

CAUSES

- Can be genetic or environmental
- The most common historical cause of NTDs globally is folate deficiency in the maternal diet
- Consanguineous marriages, have suggested a genetic basis for NTDs.
- Chromosomal abnormalities (trisomy 13, 18, 21) are also associated with NTDs.
- Some of the environmental factors include
  - geographic location,
  - season of conception,

CAUSES..

- socioeconomic class,
- maternal diabetes,
- maternal age,
- zinc and folate deficiencies,
- maternal alcohol abuse,
- maternal use of valproate, and
- intrauterine hyperthermia.

- A cohort study by Jentink *et al* suggests that carbamazepine monotherapy in the first trimester produces fetal malformations specific to spina bifida; however, the risk is lower than for valproic acid.

CLINICAL PRESENTATION

- Open NTDs are readily visible at birth, with the majority being discovered during pregnancy.
- Closed NTDs may remain undetected for years, even decades, especially in the absence of cutaneous markers
- NTDs are commonly discovered during prenatal screening and are often associated with poor prenatal care.

CLINICAL PRESENTATION…

- The most common presentation of a closed NTD is
  - an obvious abnormality along the spine such as a fluid-filled cystic mass,
  - area of hypopigmentation or hyperpigmentation,
  - cutis aplasia,
  - congenital dermal sinus,
  - capillary telangiectasia/hemangioma,
  - hairy patch (hypertrichosis),
- A closed NTD can also present without a cutaneous marker.

CLINICAL PRESENTATION…

- The second most common reason for seeking medical attention is asymmetry of the legs and/or feet.
- Other children exhibit progressive spinal deformities such as scoliosis.
- Some children present with a picture of progressive neurological deficits that may include
  - weakness in one distal lower extremity,
  - sensory loss in the same distribution, and
  - bladder or bowel dysfunction.

CLINICAL PRESENTATION…

- Low back pain sometimes without neurological deficit in older children or adolescents
- A patient with a closed NTD such as a congenital dermal sinus with an intraspinal dermoid cyst or a neuro-enteric cyst can present with symptoms of spinal cord compression due to enlargement of the mass.
- A patient with a dermal sinus also can present with bacterial meningitis or spinal abscess.

CLINICAL PRESENTATION…

- The prevalence of medical comorbidities depends on the level and severity of the lesion e.g. UTI

DIAGNOSIS

- Ultrasonography is used antenatally for neural tube defect (NTD) screening
- MRI is the study of choice for imaging neural tissue and for identifying contents of the defect in the newborn.
- CT scan allows direct visualization of the bony defect and anatomy.
- CT scan is also used to determine the presence or absence of hydrocephalus or other intracranial anomalies

DIAGNOSIS..

- Maternal serum alpha-fetoprotein can be measured in maternal serum (MSAFP), amniotic fluid, and fetal plasma.
- It is typically measured around 16–18 weeks' gestation.

MANAGEMENT

- The new-born with an open neural tube defect (NTD) should be kept warm and the defect covered with a sterile wet saline dressing.
- Give prophylactic antibiotics
- The baby should be positioned in the prone position to prevent pressure on the defect
- Prompt closure of the defect is indicated, ideally within the first 72 hours after birth for myelomeningocele.

MANAGEMENT…

- Closed NTDs typically do not warrant urgent surgery
- For closed defects associated with cord tethering, surgery involves removal of structures that are anchoring the cord.
- In children born with severe hydrocephalus, a ventriculoperitoneal shunt placement should be considered at the time of myelomeningocele closure.

MORBIDITY AND MORTALITY

- Anencephaly is incompatible with life.
- No neural tissue is present, and the brain stem consists of nests of poorly differentiated neural elements.
- The brain stem is not sufficiently developed.
- The survival of these new-borns is limited to a few hours (rarely >2 days).
- Other NTDs lead to neurological deterioration, which may present early after birth or later in life.

MORBIDITY AND MORTALITY…

- The neurological deficits may be due to
  - accompanying hydrocephalus,
  - a Chiari II malformation,
  - tethering of the cord,
  - cystic mass, or fibrous band compressing the neural elements.
- Another possible complication is meningitis (infectious or chemical), especially in open NTDs
- The most common NTD compatible with life and a positive prognosis is myelomeningocele

MORBIDITY AND MORTALITY…

- Paralysis, bladder and bowel incontinence, and hydrocephalus are the most common clinical complications.
- Severe intellectual disability is present in 10–15% of these patients.
- Neurologic deficits are overall difficult to predict based on the level of the lesion
- Some segments of the spinal cord may retain central connections and maintain partial function, allowing voluntary control or sensation in affected limbs.

MORBIDITY AND MORTALITY…

- Bowel and bladder function are affected in roughly 90–95% of patients with myelomeningocele
- They manifest as neurogenic bladder and/or fecal incontinence
- Despite aggressive treatment
  - 10–15% of these children die prior to reaching the first grade.

MORBIDITY AND MORTALITY…

- Most children with isolated myelomeningocele (without major anomalies of other organs) survive to adulthood, and life expectancy is nearly normal.
- Sixty percent have normal intelligence, although of these, 60% have some learning disability
- Hydrocephalus is present in 85% of children

- Anencephaly has a female preponderance, especially among premature births, with a female-to-male ratio of 3:1

PREVENTION

- Nutrients seen to decrease incidence of NTDs are rich in
  - folic acid,
  - vitamin C, and
  - riboflavin
- Increased intake of folate in first month of pregnancy is seen to help with neurulation
- The metabolic pathways and role of folate in neurulation remains unclear, however, studies have demonstrated that folate has a direct role in neural tube closure

PREVENTION..

- It is recommended that all pregnant mothers are given 400mcg of folate during pregnancy