

Neural tube defects and spina bifida

A spectrum of defects that result from abnormal neurulation

Epidemiology

- Affects ~1/1000 pregnancies (USA)
- 1.2-1.7 times more common in girls
- Combination of genetic and environmental factors implicated
 - Highest rates in Hispanic women
- Incidence decreasing due to screening

Clinical features

- Motor & sensory deficits in legs
 - Bony deformities
 - Bowel and bladder dysfunction
 - Hydrocephalus
 - Chiari malformation
- Often associated with learning disabilities and executive dysfunction

Management

- Antenatal diagnosis and parental counselling
- Specialised obstetric care and planned birth
- Foetal surgery (between week 19 and 25)
- Neonatal management
 - Neurosurgery for open spina bifida
 - Monitor for hydrocephalus +/- shunt
 - Supportive care
 - Bladder management
- Follow-up by a MDT is required

Classification

- Can affect the brain (anencephaly, encephalocele) or any level of the spinal cord
- Myelomeningocele:** when the spine + meninges are exposed (open)
- Meningocele:** only the meninges are exposed
- Spina bifida occulta:** involving the vertebral arches only- affects 10% of the population and usually causes no issues

Diagnosis

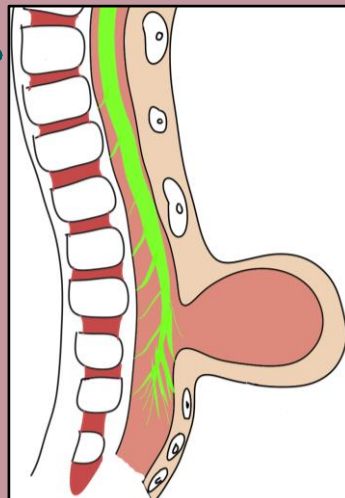
- Most are diagnosed antenatally or at birth
- Antenatal triple testing (15-20 weeks gestation)**
 - Alpha-fetoprotein, unconjugated oestradiol and hCG
 - Primarily for Down's Syndrome but high AFP suggests NTD
- Antenatal ultrasound**
 - Lemon sign (hydrocephalus)/ banana sign (Chiari malformation)
- Foetal MRI**
 - Defines structural brain anomalies associated with NTD
- Others:** Cranial/ spinal ultrasound, CT head

Prevention

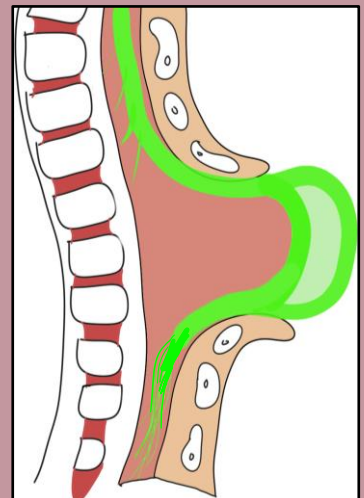
- Preventable by maternal folate supplementation
 - 400 micrograms/ day before pregnancy and the first 12 weeks**
- Higher dose (5 milligrams)** if there is a higher risk of NTD

Risk factors

- Inadequate maternal folate/ B12 intake
- Previous pregnancy affected by NTD
- Personal/ family history
- Foetal chromosomal abnormalities
- Ante-natal exposure to anti-epileptics
- Maternal obesity
- Maternal diabetes



Meningocele



Myelomeningocele