Spina bifida
Early management and long-term care

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Perspective

• Spina bifida is the most common birth defect involving the Central Nervous System
• Often *preventable*
• Initial management has a *profound* effect on survival and the handicaps that they may suffer
• Hope and support make this a manageable condition which culminates in a productive and meaningful life for many...

*Meningomyelocele is the most complex congenital abnormality compatible with a *normal* life (David McLone)*
This process is complete within the first month post-conception;

This has clear implications for the concept of prevention (see later)
This is a real process-not just a hypothetical construct!

Scanning Electron Micrograph of Chick Neural Tube
Important points to remember about neural tube closure

• Occurs by **day 28** postconception........ this is critical for understanding strategies for prevention!

• This process is called **primary neurulation** and gives rise to virtually the entire CNS; the terminal part of the spinal cord arises through caudal regression or **secondary neurulation**

• Abnormalities cause conditions that are termed neural tube defects (NTDs) or **dysraphism** and may occur at either end .....
Classification of Neural Tube Defects

Cranial dysraphism

open  anencephaly

closed  encephalocele

Spinal dysraphism (Spina bifida)

open  spina bifida aperta
  meningomyelocele (myelomeningocele)

closed  spina bifida occulta
Encephalocele

- **Anencephaly** is probably the commonest NTD, but seldom seen clinically as it usually results in a miscarriage or termination if diagnosed antenatally.

- **Encephalocele** is usually obvious at birth and may be:
  - **Anterior** long term prognosis usually good
  - **Posterior** long term prognosis poor in most cases due to extent of disruption of brain development; some may do well though
  - **Basal** very rare
Spinal Dysraphism
Meningomyelocele (open spina bifida)

Patulous anus and buttock atrophy

Neural placode
Open neural tube

Enlarged ventricles
Cerebellar vermis
Syrinx
Myelomeningocele
Open spinal cord
Bone (vertebrae)
There is a **very** wide variation in size, level and appearance...
Perinatal management

- Antenatal diagnosis
  - NTD incidence varies by country
    - 2/1000 live births in Cape Town (Buccimazza)
    - Family history and many other risk factors...
  - Investigations
    - Screening: maternal serum αFP
    - Diagnostic: ultrasound

- Labour and delivery
  - NVD vs Caesarean Section

- The folate story... pre-conception reduces the risk by >70%!
Decline in the Prevalence of Neural Tube Defects Following Folic Acid Fortification and Its Cost-Benefit in South Africa

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Figure 1. Proportion of NTD deaths to all other congenital anomalies, <1 year, South Africa 1998–2005.

What about 2014?
Surgical repair of meningomyelocele

Goals:
1. freeing of the neural placode
2. neurulation
3. watertight closure of the dura
4. skin closure

Neural placode (Open Spinal Cord)

Dura mater
and that’s not all folks.....
Postoperative management

- Keep flat for a week to avoid CSF leak
- Normal feeds- ensure no bulbar palsy
- Daily head circumference (watch for hydrocephalus)
- Counsel parents about folate prophylaxis in future

- Multidisciplinary follow-up....

  Neurosurgery, Orthopaedics, Urology, Radiology, Developmental Paediatrics, Genetics, Stomatherapists, Physiotherapists

  Community Nurses, Social Workers....
Is non-operative management an option?

The placode may epithelialise....

If a decision is made not to treat a severely affected newborn, this must be thought through very carefully as the child may survive, culminating in a much worse situation with a massive sac, sepsis etc.
Changing philosophies of management

- Pre-1950 survival <10%
- Shunts
- 1970s Lorber “Selective treatment”
- 1990s Groningen protocol

Deliberate termination of life of newborns with spina bifida, a critical reappraisal

T. H. Rob de Jong
Long term issues

• **Neurosurgical**
  – Chiari malformation
  – Hydrocephalus
  – Tethered spinal cord

• **Orthopaedic**
  – Feet, ankles, knees, hips...
  – Spinal deformity

• **Urological**

• **Cognitive**
Hydrocephalus

- A hydrodynamic disorder of CSF circulation leading to an increase in intracranial pressure

- Treatment options
  - Ventriculoperitoneal shunt
  - Endoscopic third ventriculostomy

Kandasamy BMJ 2011
Orthopaedic issues

• Lower extremity
  – Equinovarus (clubfoot) in >50%
  – Other foot and ankle, knee and hip deformities

• Spine
  – Incomplete posterior arch
  – May have other bony anomalies
  – Scoliosis
  – Kyphosis
Urological issues

• Neurogenic bladder dysfunction
  – Incontinence
  – Vesico-ureteric reflux (3-5% .... 40-50% if not correctly managed)

*Proactive management*
  • urodynamics
  • anticholinergics
  • **clean intermittent catheterisation** (CIC: Lapides 1971)
  • Vesicostomy, bladder augmentation etc

• Don’t forget
  – Constipation
  – Sexual function
Cognitive outcome

- Mean IQ of patients with spina bifida is within the normal range, but usually lower than that of the general population.
- Weaker in visuo-spatial and motor domains.
- Discrepancy between Verbal IQ and Performance IQ.
- Numerous factors play a role, including hydrocephalus and shunt complications, lesion level, other complications, social circumstances and expectations etc.

Spennato and Cinalli 2008
Chicago series: the gold standard!

118 children managed actively from 1975-1979

19 lost to follow-up
28 died
71 reviewed at 20 – 25 years

63% normal schooling (37% special classes)
45% actively employed (others volunteers)
Fig. 1. The number of shunt revisions completed on the study cohort.

Fig. 4. This figure demonstrates the continual increase in death rate as the cohort ages.
Long term outcome

- Most are able to attend **school** although may have particular learning disabilities
- **Ambulation** possible if neurological level **below L3**
- Social **continence** achievable in most with CIC (Clean intermittent catheterisation)

*Multi-disciplinary follow-up is the key to optimising their long term potential*

- The **transition** through **adolescence** is particularly challenging as there are complex psycho-sexual issues to deal with
- They in turn will may one day have children- NB need to counsel about **folate** prophylaxis!
- Remember this condition can be prevented in most...
Spina bifida: A multidisciplinary perspective on a many-faceted condition

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