

# Antenatal counseling in Spina Bifida and Neural Tube Defects

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# The Sydney Children's Hospitals Network Westmead Campus



## Historical Changes

- Late 1980's - Introduction of MRI scanning
- 1990's - Development of antenatal Ultrasound.
- 2000's - Significant decrease in affected newborns and less severe disability.
- About 5000 people in Australia have Spina Bifida, currently maximum number of persons living with Spina Bifida in the community.

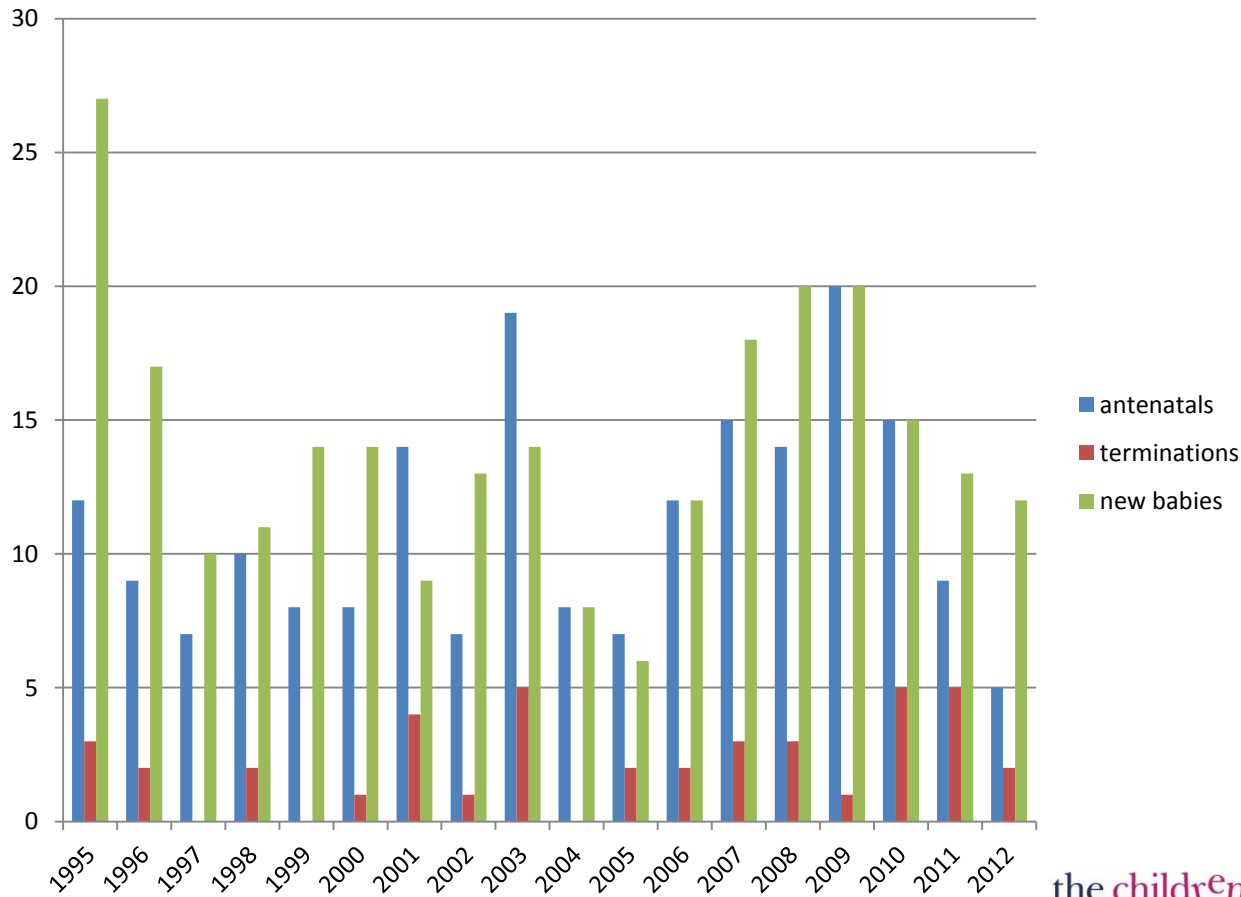


# Improvements in Quality of Life Medical

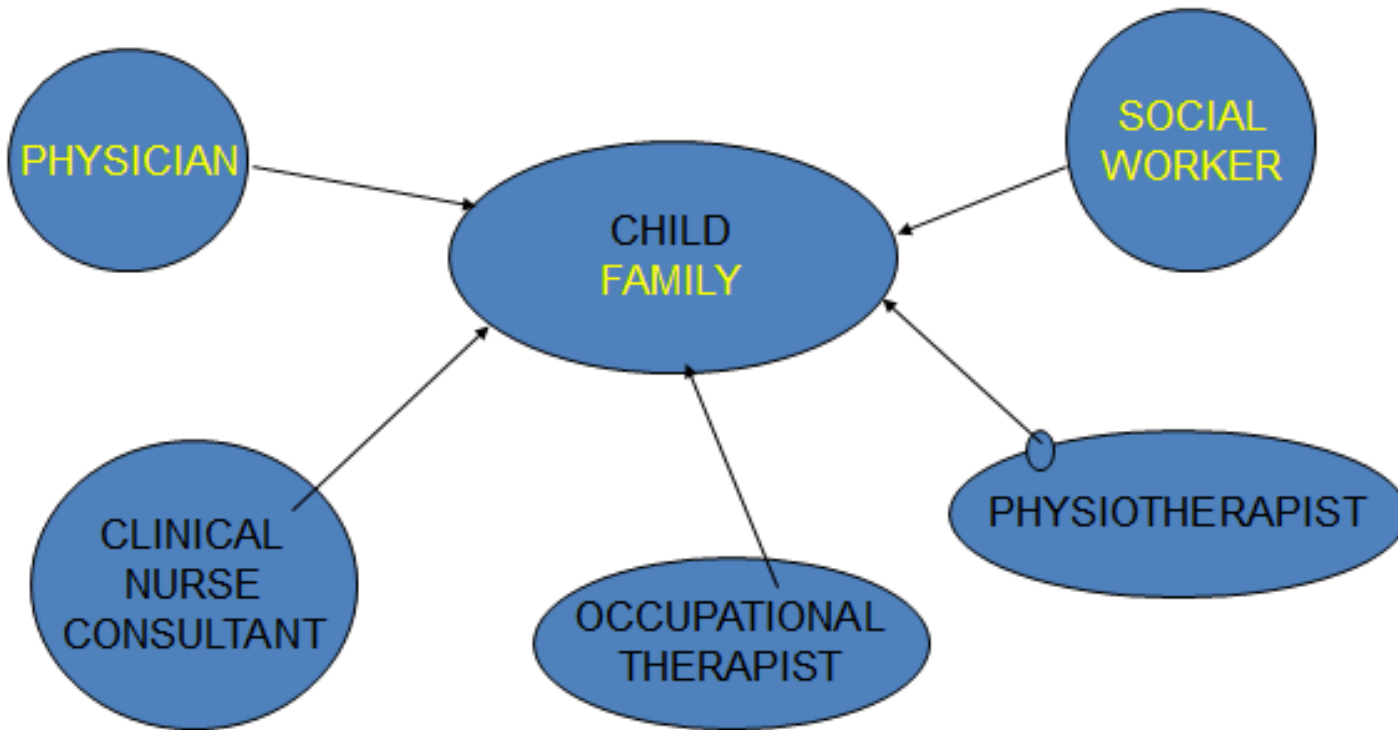
- Treatment of hydrocephalus
- Treatment of the neurogenic bladder
- Independence in personal care
- Improvement in skin care
- Understanding learning issues



# Perinatal referrals – 1996-2012



THE CORE SPINA BIFIDA TEAM



# Neural Tube Defects, facts

- Neural tube defects (NTDs):
  - more severe can be seen as early as 11-12 weeks
  - most are diagnosed at the routine fetal anatomy scan (FAS) at 18-20 weeks
- Classification:
  - Anencephaly
  - Encephalocele
  - **Spina Bifida**

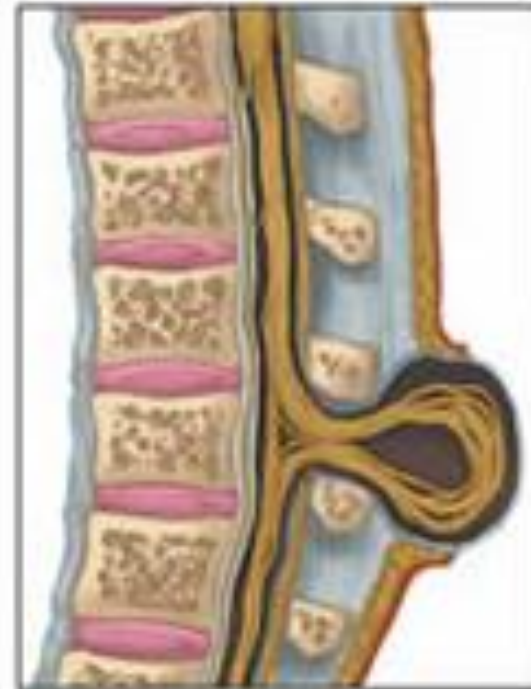


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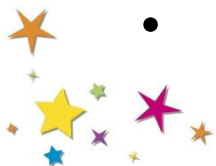


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# Neural Tube Defects, facts cont.

- NTDs: overall incidence of 1:1000 pregnancies (250/y)
  - number is declining with earlier ultrasound diagnosis and the increasing use of folic acid intake by women of child bearing age
- About half of these are Spina Bifida
- > 80% are now diagnosed and many terminated antenatal period
- Blood screening for raised alpha fetoprotein is helpful if positive.
- Incidence at birth now less than 1 in 2000

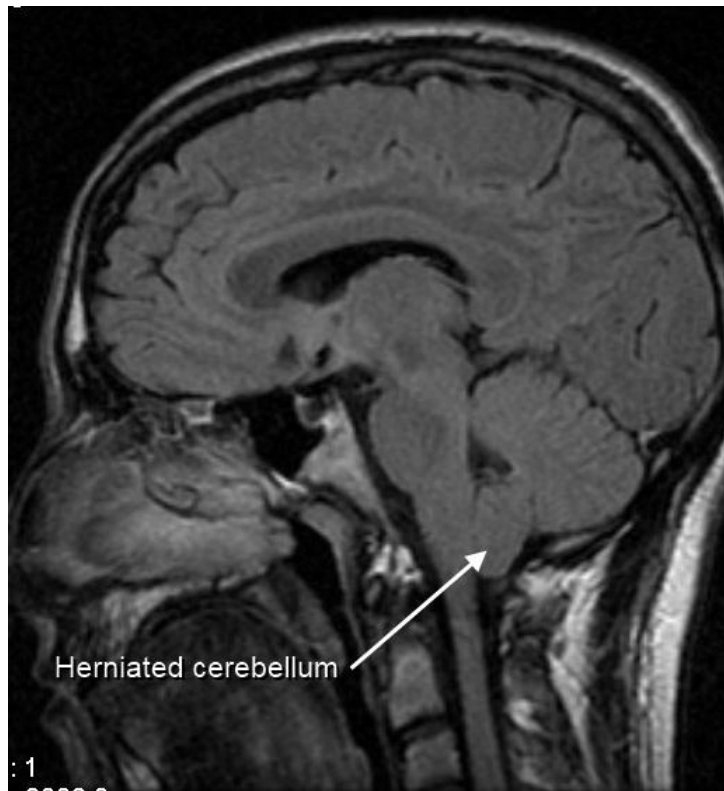


# Neural Tube Defects, facts cont.

- Most spinal defects in the lower lumbar to sacral region.
  - can occur at any level
  - most are located posteriorly, some do rarely occur anteriorly
- Greater than 97% of cases of Spina Bifida are associated with Chiari II malformation in the brain
  - displacement of the cerebellum, fourth ventricle and medulla through the cisterna magna resulting in a banana shaped cerebellum
  - concavity of the frontal bones resulting in lemon shaped skull and ventriculomegaly



# Neural Tube Defects, facts cont.

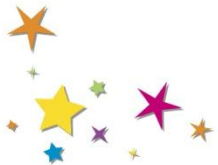


# Neural Tube Defects, facts cont.

- Ascertainment of the level of the spinal lesion is the most important predictor of functional outcome
  - MRI may be used to aid in this, however a thorough high resolution US examination is more effective at determining the level of the lesion
- Associated deformities of the lower limbs such as **club feet** and hip dislocation
  - Most NTDs are isolated malformations
  - Up to 15% association with other abnormalities (VSD, renal, IUGR)
  - 3% association with chromosomal abnormalities



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## Other NTDs

- Anencephaly:
  - Natural history - 67% stillborn - 33% neonatal death
- Encephalocoele:
  - Natural History - 23% - stillborn - 43% neonatal death
  - Many of the survivors have a significant disability - spastic quadriplegia, epilepsy, hydrocephalus and intellectual disability



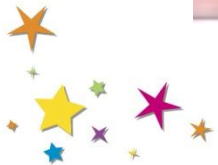
## Other NTDs, cont.

- Spinal Dysraphism
  - potential for neurological deficit
- Lipomyelomeningocele
  - usually no Chiari malformation
  - mobility issues harder to predict, may be progressive
  - often have incontinence





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# Antenatal referrals

- Obstetricians can refer to SB service when NTD is diagnosed antenatally
- Usually urgent referral
- Purpose – to discuss nature and effect of NTD in their baby
- Usually seen within 2 days of referral

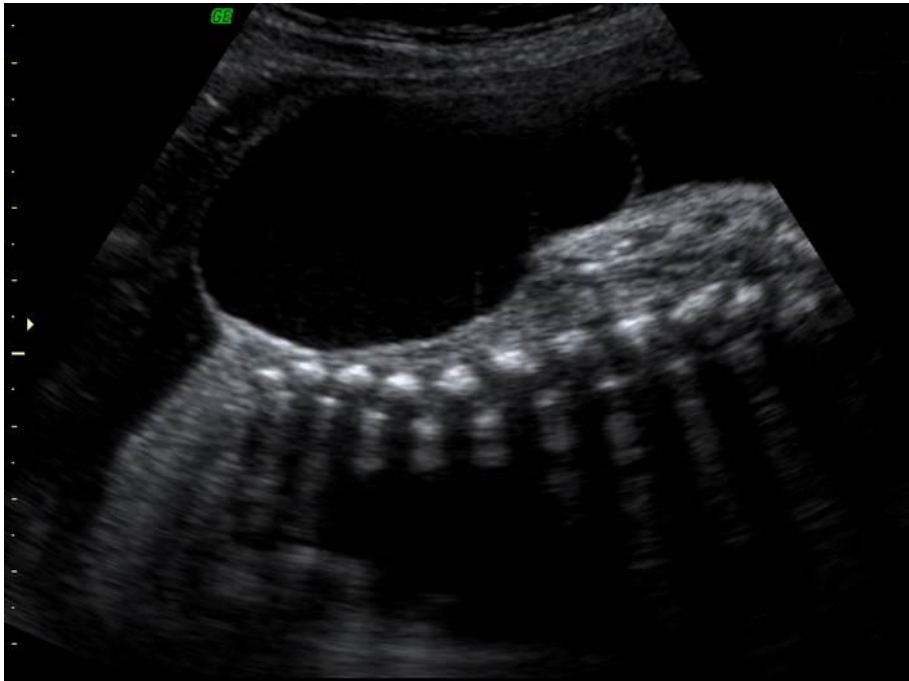


# Ultrasound - what signs?

- Chiari malformation and ventriculomegaly - often the alerting sign but not always present early in pregnancy
- Lemon and Banana signs
- Splayed vertebrae
- Soft tissue mass
- Bony deformity - scoliosis, kyphosis, talipes
- Leg movements
- Renal anomalies



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# Predicting functional outcome

- Is this Spina Bifida?
- Where is the lesion? How big? What type?
- Is there Chiari malformation?
- Is there hydrocephalus?
- Is there bony deformity?
- Are there leg movements?
- Any other anomalies?



# Antenatal counselling

- Aetiology
- Anatomy – what goes wrong
- Functional implications
  - Mobility
  - Continence
  - Cognition
- Estimation of extent of ability / disability
- Delivery issues and neonatal care
- Developmental / clinical management
- Future pregnancies



# Antenatal counselling

- Ultrasound findings confirm diagnosis
- Explore parents reactions to diagnosis to determine how to give further information
- Start with what they know
- Build on what they know about the anatomy
- What does this mean for function?



# Antenatal counselling

- How does this translate into daily life?
- Can it be fixed?
- What is the medical / surgical treatment in the neonatal period?
- What further treatment?
- How long in hospital?
- What about follow up?





# Antenatal counselling

- Can my baby be delivered normally?
- Where should my baby be delivered?
- Will my baby live?
- Will we be able to cope with a baby with a disability?
- Do you know any child with a disability?  
Understanding / stigma / misconceptions re disability



# Antenatal counselling

- Who in the family knows? How supportive are they?
- Other supports, friends etc
- Hospital supports, GP, community - baby health, therapists, Northcott Society
- Philosophical / cultural and religious beliefs.



# The future for the child

- What type of schooling
- What type of independence
- What type of work
- What about relationships
- What about parenting
- What other quality of life issues
- What about genetics



# The future for the parents

- How much additional caring
- How much medical / hospital care
- How expensive
- How limiting physically
- How about my other kids
- How about future kids
- How about ME, my marriage, my career



# Decision making

- Parents decision.
- What is best for their baby. No right or wrong.
- Offer support now and in the future whatever the decision.
- Give written handouts if parents want them.
- Offer to meet again if continuing with pregnancy
- Visit to Baby Ward.
- Provide telephone contact.



# What is the most appropriate decision?

- For this couple
- At this point in time
- In their circumstances
- With the limited information / forecasting we can give for the baby's future
- Do we have a “good enough” baby and “good enough” parents to make life worthwhile for this child?



# The future – ongoing pregnancy

- Preparing for delivery
- Organising the family to care for other siblings
- Family aware
- Hospital labour ward and baby ward at CHW aware.



# The future - after termination

- Period of adjustment
- Going back to work
- What to tell others
- Who is going to be their main support?
- Who can they ring for extra help?
- What information do they need for future pregnancies - who will give them this advice?
  - the incidence of spina bifida can be reduced by up to 70% by taking folate supplementation





## Contact details

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