Antenatal counseling in Spina Bifida and Neural Tube Defects

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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 image shows a bar chart that tracks the number of referrals over the years from 1995 to 2012. The chart is color-coded to differentiate between antenatal, termination, and new baby referrals. The y-axis represents the number of referrals, ranging from 0 to 30, while the x-axis lists the years from 1995 to 2012.
Neural Tube Defects, facts

• Neural tube defects (NTDs):
  o more severe can be seen as early as 11-12 weeks
  o most are diagnosed at the routine fetal anatomy scan (FAS) at 18-20 weeks

• Classification:
  o Anencephaly
  o Encephalocele
  o Spina Bifida
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
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
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
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
  o Mobility
  o Continence
  o 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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