FRACP: Spina Bifida

Dr Kate Thomson
Paediatrician
Spina Bifida Clinic, RCH

Dr Kevin Dunne, Dr Catherine Marraffa and Ms Judy Wells.
Neural Tube Defects

- Congenital
- Abnormal closure of the neural tube (which forms the spinal cord, vertebrae and skin coverings)
Embryology: Conception - D18

- Germ layers
  - Endoderm (incl. notochordal plate) and
  - Intraembryonic mesoderm
  - Ectoderm to form neural plate
- Neural plate forms along length of fetus

27_12bFrogNeuralTube_3-L.jpg
doctoraliblog.wordpress.com
Foetal neural tube: D19 - D28

- Neural tube folds
- Begins in cervical region and closure extends both rostrally (D24) and caudally (D28)
Neural Tube Defects

The term *spinal dysraphism* includes the overall group of defects derived from the maldevelopment of the ectodermal, mesodermal, and neuroectodermal tissues, and its sequelae may affect brain, bones, extremities, and bowel and bladder functions.

Spectrum of the NTDs

- Anencephaly
- Spina Bifida Cystica
  - Myelomeningocele
  - Meningocele
  - Encephalocele
- Lipomyelomenigocele
  - Clinically similar but no Hydrocephalus
- Dermal Sinus, Syringomyelia, Diastematomyelia
- Tethered cord
- Spina Bifida Occulta
  - Back pain, constipation
Spina Bifida - types

Normal                       Dysraphism                   Meningocele             Myelomeningocele
Lipomyelomeningocele

- Lipoma
- Grows in the spinal canal or outside
- Press on the spinal cord
- Nerves traverse and become nonfunctional
- Partial weakness of lower limbs
- Neurogenic bladder and bowel
- Spine abnormality
- No hydrocephalus - normal cognition
Lipomeningocele

- Nerves
- Dimple in Skin
- Lipoma - abnormal collection of fat

MRI of Underlying Abnormality (Lipoma)
## Incidence

- **Australia**: 0.95 /10,000
- **Western Europe**: 1.5-3 /10,000
- **Canada**: 2.6 /10,000
- **USA**: 6-7 /10,000

- **Male**: 58%
Spina Bifida - incidence

- Varies from country to country
- Range 1-7 /10,000 live births
- High in Celts (Ireland, Wales)
- Low in Northern Europe.
- Encephaloceles higher in SE Asia
Spina Bifida
Common Birth Defect

### Ten most common birth defects, 2005

<table>
<thead>
<tr>
<th>Defect</th>
<th>N/10,000</th>
<th>1 in X number of births &amp; TOPs X</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypospadias</td>
<td>65.2*</td>
<td>153</td>
</tr>
<tr>
<td>Obstructive defects of the renal pelvis</td>
<td>36.4</td>
<td>275</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>28.5</td>
<td>351</td>
</tr>
<tr>
<td>Trisomy 21</td>
<td>27.5</td>
<td>364</td>
</tr>
<tr>
<td>Congenital dislocated hip</td>
<td>26.1</td>
<td>383</td>
</tr>
<tr>
<td>All NTDs</td>
<td>12.2</td>
<td>817</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>9.1</td>
<td>1,098</td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td>8.8</td>
<td>1,135</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>8.4</td>
<td>1,196</td>
</tr>
<tr>
<td>Cystic kidney disease</td>
<td>7.3</td>
<td>1,367</td>
</tr>
</tbody>
</table>

*This figure has used male babies only as the denominator.
Spina Bifida

All NTD cases—3 year moving average

n/10,000

Incidence of new cases

- 12.2 per 10,000 births
- 1 per 817 births and TOP

So expect:
- 85 NTD for 70000 conceptions

But
- Now seeing ~5 new cases/y at RCH, same at MMC

- Incidence has decreased significantly in Victoria, largely due to termination, secondarily due to folate.
RCH annual first admission infants <12 months
RCH patients - from the new Registry 1993-2015

Patients with NTD by year of birth
Aetiology

- Presumed multifactorial
  - Abnormal gene interacts with other genetic loci and or environmental factors to modulate the incidence or severity of the defect.

- Genetic
  - Increased in chromosomal abnormalities i.e. trisomy 13 and 18.
  - Increased in siblings and other relatives
  - Abnormal genes
    - Som
    - VANGL1 (mouse, 3/166) NEJM 356,(14)2007,
Type of NTD by year of birth (can have >1)

Number of NTD by Year of Birth

- Myelomeningocele
- Lipomeningocele / Lipomyelomeningocele
- Fatty/thickened filum
- Split cord malformation
- Sacral agenesis
- Bifurcated cord
- Other dysraphism
Genetics

- Multi-factorial BUT:

  1 affected child       1:50 Recurrence Risk
  2 affected children    1:10 RR
  3 affected children    1:4 RR

  Adult with Spina Bifida, offspring risk is 1:25

Note

- Risk of any child born with a major defect 1:30 (3%)
Folate

- Folate supplementation reduces recurrence by up to 72% \textit{Lancet 1991}

- Recommendation:
  - 1 month pre- and 3 months post-conception
  - Routine 0.5mg daily
  - High risk pregnancy 5mg oral daily
    - Parent with SB
    - Parent with a previously affected child
    - Mother on Anti-Epileptic Drugs
    - Mother with Diabetes Mellitus
Antenatal diagnosis

- Failure of closure - α-fetoprotein (AFP) excreted into amniotic fluid
- Prenatal Ultrasound
  - 18-20 weeks
  - Detailed foetal anatomy
  - Feto-maternal obstetric outpatients for discussions and counselling
  - Termination at 20-22 weeks should the parents choose
Location and extent

- This is a disease with multi-system consequences - nerves, muscle, bone, bladder, bowel, skin, intellectual function, executive function (planning, motivation), emotional development and independence

- Which tissues are involved?
- Where the lesion is, in the spinal cord?
- Extent of lesion?
i. Which tissues are involved?

- **Spina bifida occulta** (bone only)
  10% adult spines

- **Meningocele** (bone and meninges)
  6% SB, 11% survivors

- **Myelomeningocele** (bone, meninges and nerves)
  94% SB, 89% survivors
ii. Where is the lesion?

- 1% Cervical only (Level 1)
- 1% Thoracic only (Level 1)
- 6% Lower thoracic (Level 2) and Upper lumbar (Level 3)
- 92% occur at L3 and below
  - [42% Lumbosacral jct (Level 4)]
  - Lower sacral (Level 5)
iii. Size of lesion

- 89% of SB survivors have myelomeningocele
- 92% of SB lesions are at L3 or below
- The extent of the lesion (size) varies greatly and contributes to the severity of effect
Systems
Hydrocephalus

- 72% develop significant hydrocephalus
  - 83% levels 2 and 3
  - 60% levels 3 and 4
- 26% present at birth and 77% by 1 month of age
- Rarely develops after 6 m
- Shunts are inserted in the first 2 months of life mostly.
Tethered cord syndrome

- Traction, damage to neural tissue
- Common at birth
- Highest recurrence at peak longitudinal growth (puberty) but can occur any time.
- Monitor with serial MRI
- Symptoms and signs - Changes in:
  - Bladder/bowel function
  - Foot position/
  - Back or lower limb pain
Neuropathic Bladder

- Urinary tract innervation - sacral segment

- All children would be expected to have neuropathic bladder BUT long term studies show 25% are continent
  - These patients had *little or no* sensory loss

- Renal failure was, historically, a major cause of death in people with Spina Bifida.
<table>
<thead>
<tr>
<th>Bladder type</th>
<th>Contractile</th>
<th>Intermediate</th>
<th>Acontractile</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Strongly</td>
<td>Continuous weak</td>
<td>Doesn’t contract</td>
</tr>
<tr>
<td>Detrusor contraction</td>
<td>High</td>
<td>Weak/ ineffective</td>
<td>Weak</td>
</tr>
<tr>
<td>Sphincter tone</td>
<td>Obstructed</td>
<td>Obstructed</td>
<td>Mild resistance (incomplete relaxation)</td>
</tr>
<tr>
<td>Outflow</td>
<td>Obstructed</td>
<td>Obstructed</td>
<td></td>
</tr>
<tr>
<td>Effects</td>
<td>High residual volume, small bladder capacity</td>
<td>Small capacity and continuous dribbling</td>
<td>Continuous dribbling</td>
</tr>
<tr>
<td>Upper tracts</td>
<td>At risk</td>
<td>At risk</td>
<td>Rarely at risk</td>
</tr>
<tr>
<td>Options</td>
<td>C.I.C Sphincterotomy</td>
<td>C.I.C Sphincterotomy</td>
<td>Anticholinergic Botox AND C.I.C Augmentation</td>
</tr>
</tbody>
</table>
Monitoring and Investigation

- Ultrasound
  - Growth
  - Architecture esp. dilatation upper tracts, post-void residual, trabeculation of the bladder wall
- Urodynamics
- Pressure studies
  - Cystometrogram
  - Cystourethrogram
- Catheter specimen urine MCS
- Creatinine, Cistatin-C
1. Preserve renal function
(ensure emptying and prevent infection)
2. Achieve continence

- Medical
  - Clean intermittent catheterisation
  - Anticholinergics (reduce detrusor hyperreflexia)
  - Manage infections

- Surgical
  - Botulinum toxin
  - Sphincterotomy
  - Bladder augmentation
  - Artificial urinary sphincter
  - Vescicostomy, urinary diversion/ undiversion
  - Mitroffanoff procedure

Note: up to 35% acquire and IgE-mediated LATEX allergy
Neuropathic bowel

- 25% of adults with Spina Bifida are bowel continent
- Most who are incontinent have poor sensation AND either
  - Increased bowel outlet resistance: Constipation and overflow diarrhoea
  - Decreased bowel outlet resistance: Frequent stools throughout the day
Bowel Management

- Diet, fluids
- Regular sit, cough, push on toilet
- Laxatives
- Enemas, washouts
- Anal plugs
- Biofeedback
- Malone Procedure
  - Antegrade washouts

(exclude other causes diarrhoea)
Continence Nurses

- Advise: management of the bladder and bowels
- Training (CIC, washouts): parents, carers, aides and patients
- Applications for funding
- Sourcing equipment (catheters, pads, nappies, specialised underwear)

- Also Stomal Therapists and Wound Care experts!
Muscle and movement

- Neonatal priorities
  - DDH
  - Clubfoot

- Childhood
  - Independent mobility
  - Optimise joint position
  - Maintain muscle function (includes body habitus)

- Teenage
  - Monitor scoliosis

Ankle foot orthoses (AFOs) are the most common aid.
Mobility

- The higher the lesion, the less the chance of independent walking
- BUT the anatomical level does not always predict function accurately
- Assess functional level
- Give upright experience to lightweight, young with low C.O.G
- Assisted walking may not be maintained into teenage years

- Kaye walker
  - Especially - pre-schoolers with crouch gait (fail to fully extend knees) Allows upright experience.
Physiotherapist

- In community provide regular therapy
- Equipment assessment and funding
- Independent skills - transfers/muscle strength
- RCH service 6-12mly muscle mapping
Orthopedic Surgery

- Common reason for admission
- Abnormal pressures on joints
- Scoliosis
- VPRS
  - inpatient or OP
<table>
<thead>
<tr>
<th>Level</th>
<th>Mobility</th>
<th>Progress</th>
<th>Complication</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thoracic/ High Lumbar</td>
<td>No quads Early walking with RGO Speed 30%</td>
<td>Independence conferred by Hx of walking Wheelchair by 11-13y</td>
<td>Obesity</td>
</tr>
</tbody>
</table>
| Low Lumbar   | Weak gluteals w strong flexors/ adductors. AFOs, forearm crutches Speed 80% | 80% maintain adult walking  

*Crutches decrease the forces causing knee valgus*  

*Untreated contracture > Pelvic obliquity > Scoliosis* |

<table>
<thead>
<tr>
<th>High Sacral</th>
<th>Weak gluteals AFOs Speed - fast!</th>
<th></th>
<th>Increased lumbar lordosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low Sacral</td>
<td>Strong gluteals Normal walking</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Sensation

- Sensory loss below level of lesion
  - Patchy or dense

- High risk of pressure areas/burns
  - Slow healing (poor blood supply)
  - Historically, common cause of death

- Occupational therapists
  - Seating advise (includes car)
  - Pressure care
  - Tools for function/ home modification (hoists, rails, handles, grips)
  - Upper limb function
  - Driver’s Licence
Cognitive function

- Variation in abilities
- Intellectual disability is highly associated with hydrocephalus
- Those without hydrocephalus have similar I.Q. and mainstream school attendance
- Even those without I.D. often have executive function problems
  - Following instructions
  - Planning
  - Motivation
- Psychological assessment
- Liaison with school, funding letters
Self-esteem
Dr Paige Church, MD Holland Bloorview Kids, CANADA, AACPDM, 2011

- Youth with Spina Bifida at risk for depression, anxiety and low self-esteem
  - Social factors
  - Continence
  - Financial independence
  - Partnership
  - Sexuality

- “Vunerable child”
  - Won’t grow up
  - Poorly informed

- Importance of self advocacy
  - The need to educate their adult health-care provider

- High rates of heart disease and suicide in adults with SB
Sexuality

- Sexuality is intimately related to sense of self, mastery, independence and self-esteem

- Transition: “Growing up ready”
  - Checklist through all ages
  - Solid anticipatory transition plan

- Independent consultations

- Access to information
  - Sex shops and suppliers of aides
  - UTIs and hygiene
  - Bowel washouts and anal plugs
  - Contraception (thrombotic risk, latex-free condoms)
  - Pregnancy (family planning, genetic counselling, 5mg folic acid)
  - Pregnancy effects on bladder, pelvic stability
Summary of effects of spina bifida

- Muscle weakness and joint deformities
- Sensory deficits *
- Hydrocephalus
- Cognitive impairment or executive function difficulties
- Neuropathic bladder *
- Neuropathic bowel
- Mental health
- Independence
- Inclusion
- Access
- Transition
A community to raise a child

- Parent
- Extended family
- Community
- Spina Bifida Foundation Victoria

- School
  - Friends
  - Teachers
  - Aides

- ECIS
- Physio
- Occupational Therapist
- Social worker
- Speech Pathologist

- GP
- Paediatrician
- Urologist
- Neurosurgeon
- Orthopedic surgeon
Case studies
Service provision
Patient A (Baby)

- Postnatal diagnosis
- Transfer to RCH
- Day 1 closure MMC
- VP shunt
- 3 debridements
- Meningitis
- Hips (no dysplasia)
- Renal US, urology
- Constipation
Early Childhood supports

RCH - NICU, inpatient
Neurosurgeon, SW and care mx
Spina Bifida paed and physiotherapist
Hip US (Ortho)
Renal US (Urology/Continence) → OPD
Orthotics

Flexible support packages (NGOs)
Private Allied Health
General Practitioner

MCHN growth, immunisations, support
ECIS (wait list 6-12m) physio (responsible for mobility devices until enrol at school) - OT, Speech Pathology (+/- psych, SW)
Community Continence support
Kinder Inclusion Support (council)

Health Care Card
Carer’s Allowance
Carer’s Payment
SWEP (catheters, mobility devices)
PBS (free enemas)

NOT eligible for Better Start

ECIS (wait list 6-12m) physio (responsible for mobility devices until enrol at school) - OT, Speech Pathology (+/- psych, SW)
Patient B starting school

- ASD device closure
- Chronic suppurative lung disease on home oxygen
- Vesicostomy recently closed
  - training CIC
- Soiling - bowel washouts
- Mild I.D.
  - Mainstream school with aide
- AFOs
- FHx obesity
- Supports - family breakdown
Transition to school

RCH - Resp/ Cardiol
Neurosurg/Urology/Ortho
Spina Bifida paed, physio, orthotics
Continence - CIC, bowel
Weight Mx for brothers
Psychology Cognitive Ax

General Practitioner
Private Allied Health

Companion card
Disabled Parking Permit
Difficulty getting funding for gym membership
Housing commission house

Health Care Card
Carer’s Allowance
Carer’s Payment
CAPS
CSS
SWEP (catheters, peristeen, undies, bed protection)
Programme for students with disability - Aide for continence/learning/mobility
Funding for the Community Physio at School (~4 per year)
Some access to Dept of Ed OT, Speech Pathology, Psychology
Patient C transition

- Obesity
- OSA
- Latex Allergy
- Malone stoma - failed
- Bladder augmentation monitoring (cancer risk)
- Access to further training or employment
  - Drivers licence (OT)
  - Rural public transport
  - Vocation?
  - Isolation
- Adult Spina Bifida services
  - New team
  - Seating
  - Urologists
Transition to adult life

RCH/RMH - Transition
Summary and appointments
Continence support/equip
OT seating
Vocation support (SW)
2yr + for Neurosurg OPD
Private Ins for Urol?

Health Care Card
CAPS
(no more CSS)
SWEP
Own Medicare Card (16y)

Disability payment
Work placement support beginning during year 11

TAFE/uni

Private funds for gym programme

Driving - difficult to access Community OT
confusion b/t VicRoads and OT about testing

GP
Where to next?
Bladder Botox YES

- Botulinum A toxin injected into the detrusor muscle (bladder)
  - Under G.A.
  - Relaxes the bladder to improve capacity
  - Stops the Detrusor spasming, stops leaking
  - MUST use C.I.C to empty 4-6 times per day
  - Wears off after 6-9 months
  - Tiny risk of severe SE causing whole body weakness. (Has been seen in children with severe Cerebral Palsy)

- Used frequently now by Urologists at RCH
Bladder reinervation NO

- Reimplant nerves in a reflex arc: skin-CNS-bladder
- Spina Bifida and Spinal Cord Injury
- Initiate micturition by scratching skin.
- Reported 17/20 became over 12 months
- Ineffective in 3/20 children
- 5/20 had SE (muscle weakness - worst foot drop)

*NOT CONSIDERED EFFECTIVE*
*IDEA ABANDONED BY RCH NEUROSURG/UROLOGY*

Xiao CG et al. 2000
Social support and parental hope are more strongly associated with self-worth and health-related QOL than age, gender, diagnosis or physical impairment.

Sawin et al 2002
Future directions
Perineal sensation: an important predictor of long-term outcome in open spina bifida

<table>
<thead>
<tr>
<th>Sensory level in infancy</th>
<th>&lt;L3</th>
<th>L3–T11</th>
<th>&gt;T11</th>
<th>Asymmetrical</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole cohort</td>
<td>38</td>
<td>32</td>
<td>42</td>
<td>5</td>
</tr>
<tr>
<td>Perineal sensation*</td>
<td>24</td>
<td>6</td>
<td>0</td>
<td>3</td>
</tr>
<tr>
<td>Renal deaths*</td>
<td>0</td>
<td>4</td>
<td>15</td>
<td>0</td>
</tr>
</tbody>
</table>

Future directions

- Quality of Life of children using Peristeen bowel washouts
- Spina Bifida Register
  - Started pilot in 2015
  - Long-term cohort project
  - RCH patients
  - Considering Medical and Surgical Care
  - Consent for future research - Continence, Cognition, School and Community inclusion, Quality of Life