



Spina Bifida

- Aetiology/Classification
- Incidence/ Prevalence/Prevention
- Initial assessment
- Clinical Description
- Management
 - Orthopaedic/Neuro/urology
 - Education/social
- Outcome

Spina Bifida

- Malformation of spinal cord and brain
- 28 days of development of embryo
- Failure of fusion of neural folds during neuralation
- Cause unknown?
- ?Genetic ?Metabolic ? Environmental
- 75% isolated
- 25% associated with other defects

Spina Bifida

- Partial paralysis of lower limbs
 - No weakness
 - Partial paralysis
 - Wheelchair dependence
- Joint deformity
- Spinal abnormalities
 - Scoliosis
 - Kyphosis/lordosis

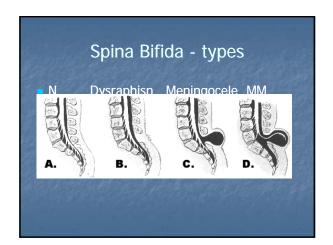
Spina Bifida

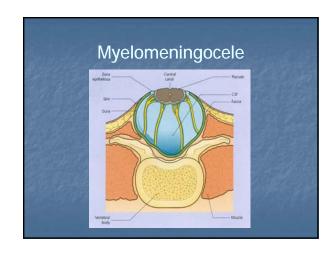
Neurogenic bladder
Urinary incontinence
renal and bladder abnormalities
Neurogenic bowel
Faecal incontinence
Skin

anaesthetic – pressure sores

Spina Bifida Hydrocephalus Arnold Chiari Type 2 CSF shunt Learning problems Upper spinal cord mild abnormalities Mild upper limb problems







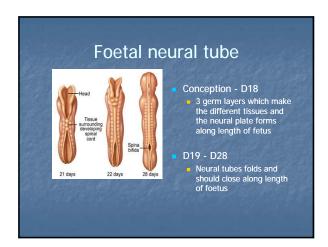














Spina Bifida- Classification

- Neural tube defect
 - Anencephaly

50% (lethal)

- Spina Bifida Cystica

45% (2/3 - 90% live) 5% (all live)

- Myelomeningcele Meningocele
- Encephalocele
- Lipomyelomenigocele
 - Clinically similar but no Hydrocephalus
- Spina Bifida Occulta
 - ?10 % back pain, constipation

Myelomeningocele

Cervical	0.5%	mild
Thoracic only	0.5%	Severe
Low thoracic	27%	Severe
High Lumbar L1-2	23%	WChair
Low Lumb, Upper sacral	45%	Braces
Lower sacral	4%	Independ

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)

Spina Bifida- Aetiology

- Unknown multifactorial
 - Abnormal gene interacts with other genetic loci and or environmental factors to modulate the incidence or severity of the defect.
- - Increased in chromosomal abnormalities i.e. trisomy
 - Increased in siblings and other relatives
 - Abnormal genes

 - SomVANGL1 (mouse, 3/166) (NEJM 356,(14)2007,

Genetics

- Multi-factorial
 - 1 affected child

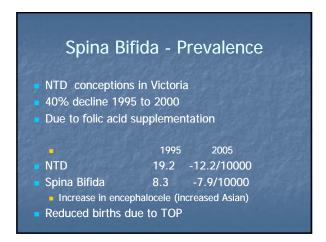
1:50 Recurrence Risk

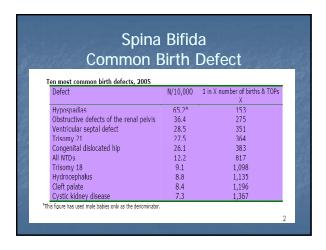
2 affected children 3 affected children 1:10 RR 1:4 RR

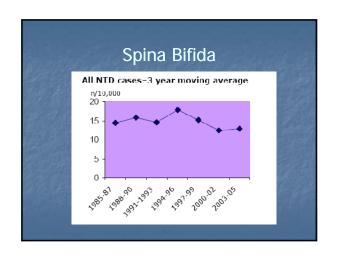
- Risk of any child born with a major defect 1:30 (3%)
- If an adult with Spina Bifida has a child risk is 1:25

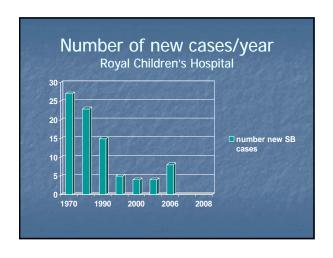
Spina Bifida- aetiology Drugs Valproate (1%) Methotrexate Clomiphene Folic acid antagonists Trimethaprim, Anticonvulsants Environment Maternal diabetes, Hyperthermia, Paternal- Agent orange

Incidence				
AustraliaWestern Europe	0.95 1.5-3	/1000 /1000		
Male	58%			
Incidence has decreased significantly in Victoria, largely due to termination, secondarily due to folate.				









Spina Bifida Incidence

- So why have the numbers dropped
- Antenatal diagnosis with ultrasound
- Termination of pregnancy
- Folate small impact
- Small drop in prevalence

Spina Bifida- Prevention

- 1960's Laurence in Wales ?Vitamin Deficiency.
- 1970's Smithells

Periconceptual multivitamins Reduced conception rate if previous NTD Methodological flaws

- MRC Study 1992
 - Multi centre trial
 - Stopped at 1400 (n = 2000)
 - 76% reduction recurrence risk for NTD
 - Folate rather than other Vitamins the cause

Folate

- Folate supplementation reduces recurrence by up to 72% Lancet 1991
- Recommend -
 - 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

SB -Folate Prevention

How does it work – unknown

Generalizable.?

Berry NEJM 2004

130000 Chinese women 0.4 mg folate,117000 controls

- 41% reduction NTD

Risks - nil reported? Twins

How much? 0.4mg – 36% 5mg - 85%

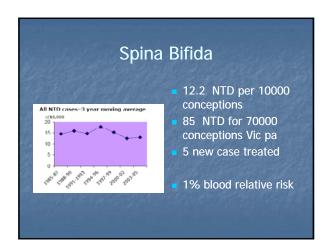
Peri-conceptual folic acid Awareness post 1996,1999

	Before	1997	2000
15-24 yrs	5%	6%	4%
25-34 yrs	19%	24%	29%
34yrs+	12%	16%	26%

NTD Recurrence Risk

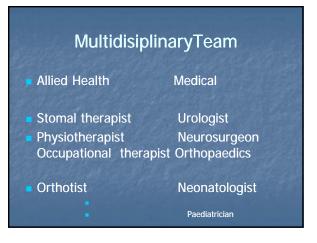
One child 2% Two Children 10% Parent NTD 4% **Blood** related 1%

Remarry 0.6%



SB – Folate to flour Folate added to flour Average daily consumption Supplement to 0.4 mg USA and 38 other countries 36 % reduction in NTD Australia about to start





Multidisciplinary team Neuropsychologist Social Worker Clinical Psychologist Education General practitioner Spina Bifida Association -social

Spina Bifida Antenatal diagnosis Ultrasound Lesion vertebral column Hydrocephalus – lemon sign Amniocentesis AFP, open lesion only Maternal Serum Screening AFP non specific (twins, down etc)

Spina Bifida Antenatal Ultrasound Can visualise at 15-16 weeks Vertebra have 3 ossification centres







Termination 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

Spina Bifida Mode of Delivery Improved outcome with elective LUCS Use if leg movement seen on US Rationale - reduces damage to neural plaque 2 neurosegmental levels better Bladder and bowel status unchanged (LUTHY et al NEJM 1989)

Spina Bifida Initial Assessment

- Refer specialist centre
- Neurosurgical assessment
- Paediatric assessment
- Discussion with family
- Facts not biases
- If severe non treatment option
- Family's wishes respected
- Palliative care 10% survive

Spina Bifida Initial Assessment

- Clinical exam chromosomal/other
- Extent of lesion can it be closed
- Motor level
- Hydrocephalus
- cranial US
- Musculoskeletal
- Hip US
- Joint deformity
- Spine deformity
- Renal

Renal US

Spina Bifida Initial Assessment

- "to treat or not to treat"
- Prior 1955 most diedHydrocephalus, Meningitis, Renal
- 1955 CSF shunt (VA), Ileal conduits
- 1955-70 many with severe abnormalities
- Treatment Criteria
 - Lorber 1971
 - Smith and Smith 1973

Spina Bifida



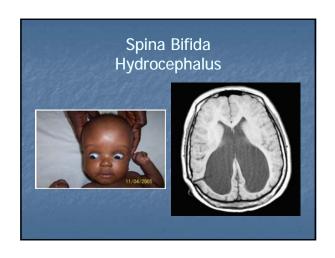
- Treatment considerations
- Severe paralysis
- Severe hydrocephalus
- Kyphosis
- Other congenital abnormalities

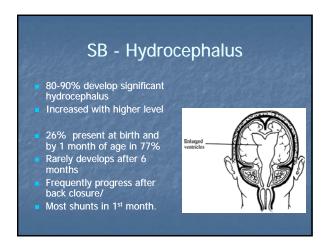
Spina Bifida Initial Treatment

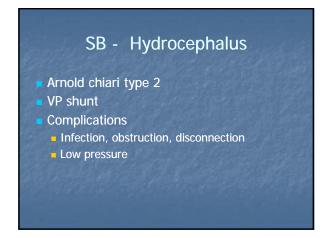
- Back closure within 24 hours (infection)
- Prophylactic antibiotics
 - Delay closure for 72 hour
- Monitor for hydrocephalus
 - Back closure head circumference increases
- Shunt insertion about 1 week later
- Hospital 1-3 weeks

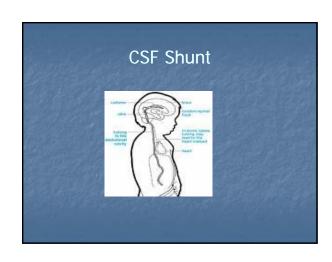
SB - Neurosurgical

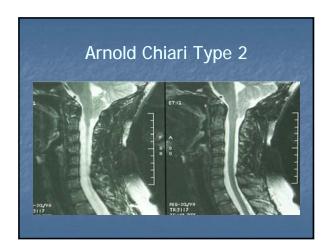
- Back Closure
 - Within 24 hours
 - Preserve nerves
 - Motor deterioration post surgery
- Investigations US, CT MRI
- Back closure hydrocephalus

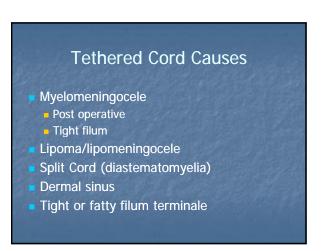


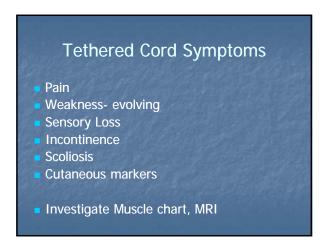




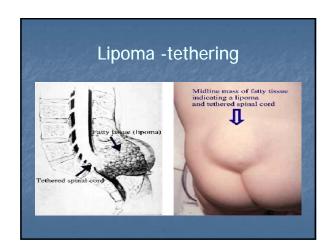


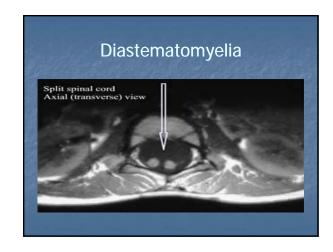


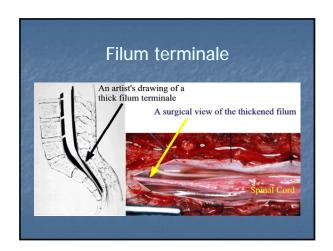














Spina Bifida

Orthopaedic

SB Orthopedic Aims

- Pattern of motor development near
- Maximize mobility
- Stable posture if standing
 - Centre of gravity over feet
- Prevent pelvic obliquity
- Correct spinal deformity
- stability of skin"

SB Orthopaedics Principals

- Developmental knowledge
- Natural history of condition
- Monitor progress
- Time interventions
- Investigations
 - Imaging
 - Muscle Charts
 - Gait analysis

SB - Orthopaedic Hip

- Hip Flexion contracture
 - Anterior hip release
 - Femoral or pelvic osteotomies
- **Abduction External Rotation Deformity**
- Limitation of Abduction
- Hip surveillance prevent dislocation

SB Orthopaedic Hip dislocation

33%

20%

- Thoracic L1, L2, L3

- Bilateral
- Operative benefit ?marginalPain
- Prevent hip flexion contracture or fit an orthosis (RGO) Unilateral
- - Leg length discrepancy, pelvic obliquity

 - Operate on low lesion

SB Orthopaedics Knee

- Valgus deformity
- Flail undeformed knee
- Undeformed knee with reduced quadricep
- Fixed flexion
- Treatment

 - Braces Muscle transfers

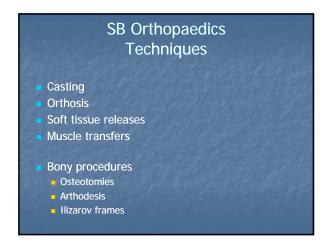


Deformities in Ankle Feet The deformities include; Equinus deformity Clubfoot or talipes equino varus deformity. Calcaneal deformity. Cavus or cavo-varus deformity. Plano-valgus deformity.

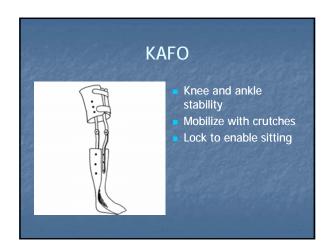












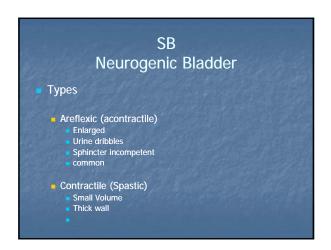


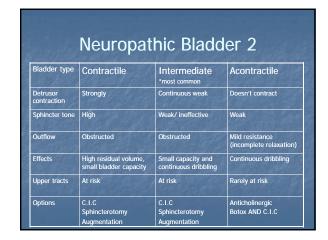
Muscles and movement 3 Physiotherapist In community provide regular therapy Equipment assessment and funding Independent skills - transfers/muscle strength RCH service 6-12mly muscle mapping Orthopedic surgeries Major reason for admission

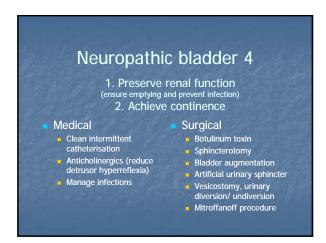


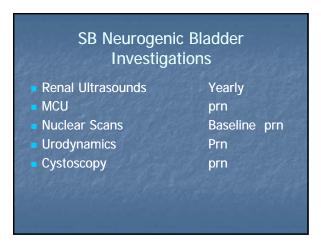
SB Neurogenic Bladder 90% plus affected Inervation S2-4 Associations VU reflux Renal Anomalies (horseshoe kidney) Bladder wall trabeculation UTI/pyelonephritis

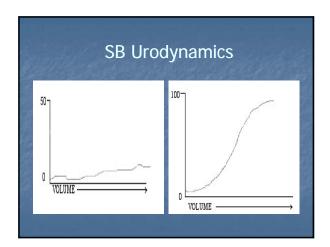
SB Urology Treatment Principles Protect the kidney's from damage Social continence Management Evaluate renal tract (imaging, urodynamics) Regular imaging (deterioration silent) Prevent infection (Bactrim, keflex, macrodantin, cranberry) Treat infection Investigate deterioration Surgery

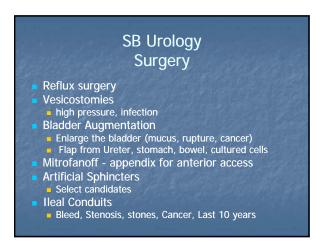


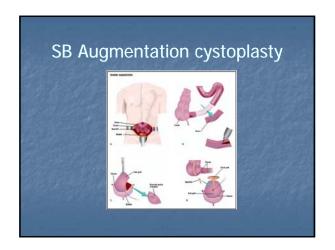


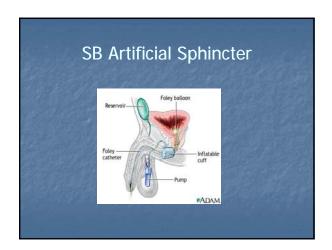












SB Urology Recent Developments

- Botox
 - High pressure bladder
 - Detrusor instability
- Nerve Transplants
 - Chinese research
 - Improved function
 - Trials underway in USA

SB Urology Continence

- Initially nappies
- Prophalactic antibiotics
- Clean intermittent catheterization 3-5/day
- Pads
- Bokka Pants
- Condom drainage
- Medication eg oxybutinin
- Surgery

Spina Bifida Neurogenic Bowel

- 90% + have faecal incontinence
- Sensory nerve to colon, rectum, anus\$2-4
- Autonomic nerves
- Major social problem

Neuropathic bowel 1

- 25% of adults with Spina Bifida are bowel continent
- Most who are incontinent have poor sensation AND either

i. Increased bowel outlet resistance outlet resistance outlet resistance continuous frequent stools outlet resistance continuous frequent stools throughout the day

SB Neurogenic bowel Types

- Patulous anus and constipation
- Diarrhoa
 - Diet sensitive
 - **10-20%**
 - Loperamide
- Patulous anus and constant leakage
 - Seen in mobile (abdominal compression)

SB Neurogenic Bowel Principles

- Aim for social continence
- No single program that works
- Bowel management starts from birth
- Try to have a program by school
- Teenagers and adults work it out

SB Neurogenic Bowel Treatment

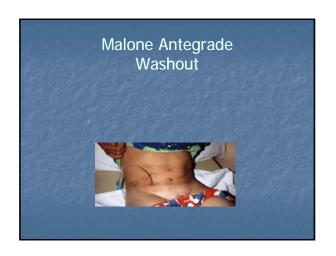
- Nappies initially
- Diet
- Laxatives often worsen but used.
 - Movicol
- Timed Toileting
- Pressure
- Digital stimulation
- Combinations

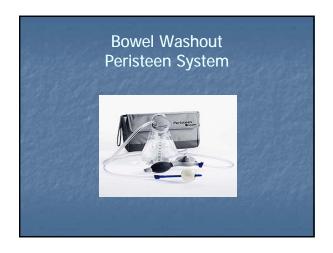
SB Neurogenic Bowel Treatments

- Suppositories
- Microlax enema's
 - Give 4-5 hour clean good for school
- **Bowel washouts**
 - 2 or 3 per week
- Malone antegrade enema
- **Anal Plugs**
- Work well in some 30%
- , expensive, last 6 hrs
- Buttock strapping eg for swimming

SB neurogenic Bowel Malone procedure

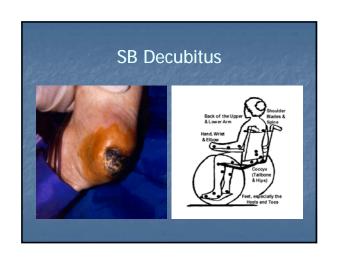


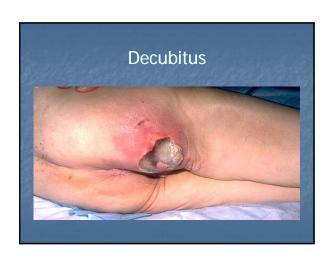




SB - Bowel Management Experimental Gracilis slings Electrical stimulation Cuff similar to Artifical Sphincter.





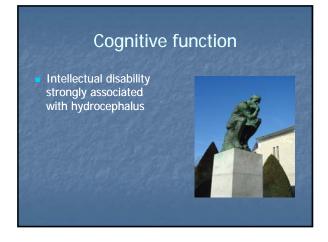


Spina Bifida Pressure Sores Prevalence 20 -25% 80 – 90% will have at some stage Cause Anaesthetic skin Excessive pressure Orthosis Urine, faecal soiling Friction 10%









Spina Bifida Cognitive General intelligence "normal range" Skewed to the lower end Specific Learning difficulties Verbal IQ > Performance IQ Relative Deficit Increases with age Primary school 5 points down - overestimates Secondary 10-15 points down Factors Hydrocephalus, shunt blockage, infection

Spina Bifida Cognitive profile

- **Executive Functions**
 - Difficulty completing task

 - Difficulty organizing task
 Overwhelmed by complex new information

 - Difficulty applying new knowledge
- Higher order Language
- Information Processing (memory)
- Attention Memory, New learning
 - Poor sequential and working memory
- Sequencing

Spina Bifida Cognitive profile

- Visuo motor Integration
 - Delayed laterality
 - Poor motor planning
 - Poor hand control
 - Slow mastery of writing
 - Untidy and disorganized presentation or written work

Spina Bifida Educational

- Most attend normal school
 - Special on cognitive grounds (Yooralla)
- parent should visit schools
- Usually require an integration aide
 - Continence
 - Cognitive
 - Safety
- School modifications (access, toilets)
- **Program Support group**
- School therapy

SB School **Problems**

- Lack of understanding about cognitive problems
- Toileting expectations unrealistic
- Academically problems around Grade 4
- Teacher zealous disillusioned
- Child withdrawn, depression
- Socialization issues

Spina Bifida Secondary School

- Cognitive Difficulties more apparent
- Mobility/incontinence affect relationships
- Peer group issue isolation
- Poor body image
- Sexuality issues
 - Impotence in males
- Misery, low self esteem
- Depression

Spina Bifida Therapy

- Developmental program
 - Mainly physiotherapy
- OT many have upper limb/ cognitive issue
- Speech
- RCH initially or
- Specialist Children Services (home)
- Centre Based from 3y
- School based therapy

Spina Bifida Therapy

- Motor development depends on level
- L3 and quads for walking
- Motor developmental program
 - Standing frames
 - Walking frames
 - Braces
 - Crutches
 - Wheelchair training

Continence

- Continence nurses
 - Advise: management of the bladder and bowels
 - Training (CIC, washouts): parents, carers, aides and patients
 - Applications for CAAS funding
 - Sourcing equipment (catheters, pads, nappies, specialised underwear)
- Also Stomal Therapists and Wound Care!

SB Social problems

- Friendship difficulties
- Realization that they are different 7-8y
- Exclusion by peers
- Unable to keep up in playground
- Reduced self esteem
- Body image problems

Spina Bifida Post School

- Unrealistic expectations re careers
- Tertiary few
 - University a few.
 - TAFE
- **Employment**
- Independent Living

Spina Bifida

- CLINIC STAFF
- Monitoring
 - Medical 6 monthly
 - Orthopaedic varies Physio annually
 - Neurosurgery annually
 - Urology 6 -12 monthlyUltrasounds 1-2 years

Transition

- When complete high school
- Sometimes ongoing orthopaedics or urology
- **MECRS**
- Monash
- Bendigo, Ballarat, Geelong



Spina Bifida Adults Medical Urological Recurrent UTI 38% Hypertension 11% Stomal Problems 58% Review recommended 36% Orthopaedic

Spina Bifida Adults Medical Orthopaedic Backache 25% Knee Instability 15% Hip pain 14% Shoulder Pain 10% Arthritis 6%

Spina Bifida Adults Medical Neurosurgical Symptoms of cord tethering Shunt review Seizures 2%

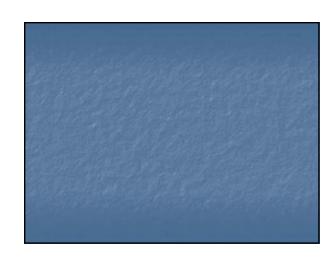
Depression	5%
Anxiety	3%
Psychosis	2%
Suicide attempts	7%
Substance Abuse	7%

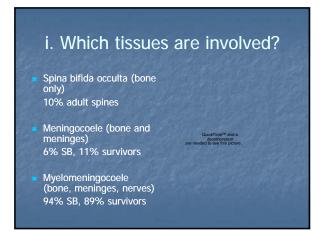
Spina Bifida Adults Education			
114 Seen in 1992TertiaryCompleted High SchoolSpecial school	8% 16% 28%		
England 42% completed high school compared to 75% Controls (Tew 1984)			

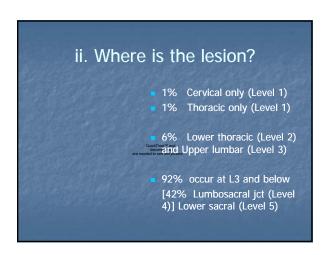
Spina Bifida Adults Employment - Home Competitive Employment 33% Not Working 54% Living with parent 62%

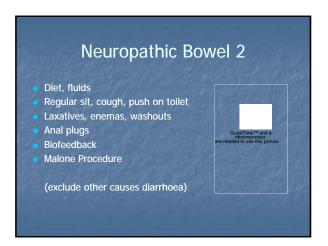












Myelomeningocele Functional Ambulation Non Ambulator Thoracic Household Thoracic – L3 Indoors only, braces Wheelchair Community L3 –Sacral Indoor/outdoor Braces Crutches Wheelchair long distances

