Adults with Spina Bifida and transition of care

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Objectives

- Review spina bifida embryology, epidemiology and prevention
- Review medical conditions in spina bifida
- Discuss management of secondary health issues
- Discuss transition and transfer of care, with a goal of optimizing participation
- MOMS study outcomes: fetal surgery
Spina bifida

- “split spine”
- Myelomeningocele
- Meningocele
- Lipomeningocele
- Spina bifida

- Occulta: may be neurogenic or failure of mesodermal layer to close (bony union of posterior elements)
Embryology – neural tube

- Closure of caudal neuropore 21-28d
- Sometimes before pregnancy recognized
Neural Tube Defects

Cross section of neural plate and tube
Embryology – brain

- At the same time, brain cell differentiation and migration occurs
- Chiari II, corpus callosum and posterior fossa size, volume and morphometry differences
- Thinner white matter, larger frontal and smaller occipital surface area and volume.
- Suggestion of reduced dendritic branching and connectivity

Epidemiology and Prevention

- Worldwide average is 1/1000 live births
- Racial and geographic variation
- Irish/Welsh, UAE and South America have had higher rates
- **About 75% reduction with folic acid 1st 6wks**
  
  Multivitamin/folic acid supplementation in early pregnancy reduces the prevalence of neural tube defects. *JAMA. 1989; 262(20):2847-52. Milunsky A; Jick H; Jick SS; Bruell CL; MacLaughlin DS; Rothman KJ; Willett W*

- Recommended dose is **0.4mg** [or **4mg** for 1st degree relatives]
Risk factors

- Related to folic acid or increased metabolism
  - Partly genetic

- Gest DM, DM and obesity

- Fever, hot tubs/saunas in 1st 6 weeks

- Anti-epileptic drugs (VPA, CBZ), ovulation inducing medication

- FDA 1998 enriched grains with daily 0.1mg intake with decline in incidence
Changing expectations

- Survival changed from ~0-10% to >80%:
  - Antibiotics WWII
  - Ventricular shunt (1950s)
  - Intermittent Catheterization (1970s)

- Function changed by mobility and accessibility which continue to improve
  - From Mechanization to computer aided controls
Rehab goals

- Maximize ADLs including mobility
- Maximize IADLs (household duties)
- Optimize health care education and management, healthy lifestyle, preventive care
- Transition care from parent/caregiver to patient
- Optimize Community Participation = QOL
Primary conditions in MM

- Combination of medical and cognitive impairments
- Hydrocephalus
- Brainstem Chiari II malformation
- Paraplegia, cauda equina type
- Neurogenic bowel and bladder
- Insensate skin areas
- Latex allergy in 20-40%
Arnold Chiari Malformation

Arnold Chiari type II:  
- cerebellar hypoplasia  
  (hypoplasia = reduced growth)  
- with caudal displacement of the hindbrain through the foramen magnum
- usually associated with hydrocephalus

Brainstem- Chiari II:  
- Swallowing issues: dysphagia, texture sensitivity  
- Gag reflex +/-  
- Central apnea  
- Obstructive Sleep apnea
Hydrocephalus

- Average of 3 revisions by adulthood
- Initially thought >90% need a shunt
- Morbidity and mortality are shunt related, independence
- Now <70% need shunt
- **Shunt symptoms can be subtle**, worsening of Chiari brainstem symptoms, back pain, walking and bladder issues → **coning risk** with detethering
Hydrocephalus

- Overlap with tension, sleep apnea and depression symptoms
- May have slow onset:
  - Lethargy, headache, worsening attention
- Consider OSA screening for adults with daytime sleepiness
- Non-compliant ventricles in some with no apparent change in imaging
- KNOW YOUR PATIENT, KNOW THEIR SYMPTOMS
Neurogenic bladder

- Early bowel and bladder training predicts success and independence

- Hydronephrosis due to reflux (abx)
- Detrusor-sphincter dyssynergia
- Detrusor instability
- Sphincter incompetence
Neurogenic bladder

- Renal u/s q6-24 months for hydro, stones, scar
- Consider bladder augmentation or diversion for storage
- Goal pressure <40mmHg
- Mitrofanoff procedure (umbilical stoma)
- Leaking: sling, collagen injection, Botulinum toxin
Mitrofanoff
Long term uro outcome

- Cost effectiveness of renal function monitoring unclear
- Not cost effective to do cytology or cystoscopy screening
- Long term ICP with ++ inflammation or ileal pouch augmentation may be at risk
- Case series of bladder ca in young MM

Neurogenic bowel

- Early independent management predicts continence
- cauda equina type lesion
- Food “allergy” frequent – 3 Cs
- 20% have voluntary control (patchy) assoc’d with low sacral reflex
- Methods vary: dig stim, suppository, irritant, enema, biofeedback, extraction
- MACE: Malone anterograde cecostomy/continence enema
Typically takes 45-60 mins to complete bag via gravity to clear. q24-48h

www.Aboutspinabifida.blogspot.com
Paraplegia

- Clubfeet at birth
- Progressive lower limb deformity during growth from imbalanced muscles
- Hip dislocation: not like in CP (not painful)
- Scoliosis: kypho, rotational, may need correction
- LOL determines walking
Mobility

- Young children use equipment
  - Stander
  - Swivel walker/parapodium
  - RGOs with crutches
  - KAFOs or AFOs for low lumbar lesions

- May lose walking with age
  - Contractures, center of gravity
  - Obesity
  - Lack efficiency and fitness
  - Lower priority
  - Loss with hospitalization/immobility
  - Pain at knees or shoulders/wrists
Insensate skin

- Lack of sensation, orthopedic deformity → high risk of pressure sores
- Obesity, skin folds, lymphedema
- Teach to monitor daily
- Risk of burns, injury (hot food on lap, sleeping near radiator)
- Ambulators get foot ulcers
- Wheelchair users get ischial and sacral ulcers (infections, incontinence risk factor)
Pressure sores

- Often ++ exudative, can benefit from antimicrobials (Silver)
- Lymphedema may need specialized reductive treatment to prevent cellulitis ~10%, increase with obesity, age
- Risk of osteomyelitis and potential amputation
- ALWAYS check feet and braces
- Pressure mapping of seating can help
Other health conditions

- **Syringomyelia**
  - May be due to tethering, or not

- **Tethered cord**
  - Always looks tethered on MRI
  - Look for loss of strength, mobility, back pain, bladder changes on Urodynamics (but r/o shunt)

- **Shunt infection**

- **Fracture risk** – Vit D low (<17% of RDI), Calcium intake (<70%RDI)

- **Charcot joints**
Secondary health issues

- Hypertension
- Loss of mobility
- Obesity
- Renal function
- Sleep apnea
- Pressure sores
- Lymphedema
- Amputation
- Metabolic syndrome (DMII)?
Obesity

- How best to measure?
  - BMI required, but scoliosis, contractures, wheelchair weigh scale needed

- Study of body composition showed higher body fat compared to BMI

- Abdominal circumference (morning) can be a good way to track metabolic risk

- Ongoing study to determine if metabolic risk like SCI
What is Transition?

- A hot topic across pediatrics
- As chronic condition management improved, survival improved
- Expectations of independence and community living
- Parents learn to take care of the child → expert care providers
- When does the child/teen take over?
  - Peer pressure
  - Expectations /role models
  - When has the skill to do so, parents will allow it
  - About 50% age 14y (Holmbeck et al)
Transition

- Start early, educate child and parents of shared goal that needs **preparation and practice**
- Bladder catheter with beading, transfer skills
- Timing with time management skills
- Assessment of own bladder and urine health

Recipient of care ➜ shared management ➜ CEO
Integration issues

- **Learning disability:**
  - Visual-spatial processing, reading, math, organization

- **Health care management:**
  - Executive task skills, auditory comprehension, memory

- **Vocational adaptation:**
  - Access, adaptations, acceptance

- **Community:**
  - Accessibility, activity options, acceptance
QOL and participation

- WHO participation = involvement in life situations
- Several studies across diseases have shown that QOL ~ participation
- A study across clinics at LCH showed community participation was assoc’d with independence
- Tonack 2007 Life satisfaction after SCI in Canada.
  - Participation~QOL consistently
  - Self described health was the second important factor
  - SWLS ~ health, participation, demographics
Initial assessment of cohort

59 respondents, age 13-30

- Walking: 34/59 (all below L3, most L4+) 57.6%
- Obese 15/37 (by BMI- underestimates) 40.5%
- Catheterizes Indep 43/59 (73%) cueing in 2
- Bowel incontinence 16/58 (27.6%) >than q 2mos
Cohort results:

- Chores 46/59 (78%) (22% do none)
- Meets Friends 40/57 (70.1%)
- Dating 26/57 (45.6%)
- School 47/57 (82.4%)
- Working 5/54 (9.3%)
- Unemployed 7/54 (13%)
Transitioning patients

- Obtain records or “sign-over”
- Assess knowledge
- Educate about condition, care and future issues
- Assess ADL and IADL, optimize (devices, therapy)
- Discuss bowel, bladder, skin prevention
- Discuss emergent care
- PCP for metabolic disease care
Future questions

- Frequency of sleep apnea
- Frequency of shunt malfunction in adults
- Management of obesity
- Risk of metabolic syndrome
- Renal health trajectory
- Aging and self-care function
- Life expectancy?
MOMS fetal surgery trial

- Feb 2011 published NEJM
- Stopped early due to prelim results of reduced need [RR 0.48] for shunt and improved motor outcome at 30 mos
- Maternal and fetal complications: including PROM, oligohydramnios, preterm birth, and prematurity (respiratory distress) in 1/5 prenatal surgery
- Consider with caution (risk to mom and baby)

Summary

- SB/MM is the most complex congenital disability compatible with life
- Cognitive and executive function issues
- Multiple organ system involvement, mobility issues
- Subtle shunt malfunction symptoms
- Few centers for specialty care for adults
- Physiatrists are well equipped to be a medical home for the MM care, with urology and neurosurgery and ortho support
Thanks!

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