Disclosures

None
Objectives

1. Discuss considerations for all internists in care of YACC
2. Review types and organ system issues of Spina Bifida and Cerebral Palsy
3. Describe common pitfalls to avoid in care of patients w/ SB and CP
4. Discuss background and resources of STEP Program
#1 How comfortable are you with caring for patients with Spina Bifida?

A. Very comfortable
B. Somewhat comfortable
C. Not comfortable
#2 How comfortable are you with caring for patients with Cerebral Palsy?

A. Very comfortable
B. Somewhat comfortable
C. Not comfortable
Why is this important?
Nearly 1/3 of children in U.S. have been diagnosed with at least one chronic illness

90% of individuals born with a disability will live to be 20 years old

More than 500,000 children with special healthcare needs transition to adult care each year

Less than 50% children with complex medical needs receive recommended health care transition guidance
Adulting is Hard

Emerging Adulthood: 18 – 25yo

Significant barriers to healthcare:
- Insurance status
- Higher unemployment rates
- Lower incomes
- Lack of primary care
- Adult provider discomfort
They are scared

“...We have spent so many nights worried about what transition would look like. Will they be comfortable with my daughter? Will we lose our care and something horrible happen?”
They have had difficulty navigating adult healthcare.

“That was one of the things that was the most heartbreaking. They treated [my son] as if he wasn’t a human being. They didn’t explain things to him.”

“You know, it’s his body. He has [a medical device] implanted, so if they’re adjusting it, if something is going on, he needs to know because he’s the one feeling it. It’s his body, so I totally agree with the fact that a lot of, I don’t think they do it on purpose, but they look at us as parents to tell us what’s going on, and I want to know, but [my child] needs to know also. He may not understand it completely, but he’s in the room and you can’t talk about him like he’s not there.”
They have complex multidisciplinary needs

“ I had eight doctors at childrens. Will I still need a GI, Rehab, Renal, Neuro, Pulmonary doctor at UAB?”

“ Will I be able to get my equipment?”

” Will they understand that I am my child’s medical decision maker?”
They are at risk for preventable hospital stays and death

• Spina bifida- most common, permanently disabling birth defect in the U.S – 1500 new diagnoses per year
• 34% hospitalizations preventable -> 35% of these resulted in death
• 2.5X mean 30-day hospital stays compared to general population
• 1 mil lifetime healthcare cost for 1 patient with CP
They are underserved

• “There is a marked disparity of health between persons with intellectual and developmental disability and the general population” due to
  1. Genetic factors
  2. Communication difficulties
  3. Discrimination
  4. Social circumstances
  5. Access to Care
Check your bias

Healthcare workers' perception of QOL is worse than patients' actual self-reported QOL.
Case #1

Jackie is a 21yo w/ Cerebral Palsy presenting to the primary care office to establish care. She was last seen by her pediatrician 2 years ago. She has recently left school and moved back home after a 2 week hospital stay for pneumonia.

Jackie says she often feels frustrated and angry. She can’t be as independent as she wants to be due to her wheelchair and deconditioning after her hospital stay. Even today, her mother is with her (in the waiting room), because she needs help. She also wants help finding adult subspecialists who are comfortable with cerebral palsy.
What are the basic things you need to know about Jackie and/or any patient with CP?

- Type
- Surgical history
- Equipment/Devices
- Medications
- Living
- Work/School/Employment
Cerebral Palsy

- Most common motor disability in childhood → most live into adulthood
- Risk factors: prematurity, low birth weight
- 1-4 per 1,000 children → Estimated half a million adults with CP
- Nonprogressive motor impairment
- Benefit from Multidisciplinary approach: PM&R, Seating Clinic, Neurology, PT/OT, GI
Spectrum of CP

1. Stiff muscles (spasticity)- 80%
2. Uncontrollable movements (dyskinesia)
3. Poor balance and coordination (ataxia)
4. Mixed
Gross Motor Function Classification scale (GMFCS)

- ~49% use wheelchair
Treatment

Dystonia
- Carbidopa-levodopa
- Benzodiazepines
- Baclofen

Spasticity
- Baclofen- starting dose 5 mg TID, increased 5-10 mg weekly
- 2nd line: tizanidine, dantrolene, gabapentin
- Botox Injections- peaks 4-6 weeks, wanes at 12 weeks
What are the basic things you need to know about Jackie and/or any patient with CP?

CP related comorbidities

- Epilepsy
- Intellectual Disability/ ASD 50%
- Vision issues: Strabismus, Visual Impairment
- Speech/ Hearing: Dysarthria, Aphasia
- Dental caries
- Pulmonary: Aspiration Pneumonia
- GI: GERD, Constipation, Reflux, Dysphagia, GT dependance
- Bladder Dysfunction: UTIs
- Bone Health: Scoliosis, Hip dislocation, Osteoporosis, Chronic Pain (up to 75%), Contractures
- Skin ulcers
- Nutrition: Malnutrition, Obesity
- Drooling
- Mental Health: Anxiety, Depression, ADHD, Sleep issues
What are the basic things you need to know about Jackie and/or any patient with CP?

CP related comorbidities

- Epilepsy
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- Pulmonary: Aspiration Pneumonia
- GI: GERD, Constipation, Reflux, Dysphagia, GT dependance
- GU: UTIs
- MSK: Scoliosis, Hip dislocation, Osteoporosis, **Chronic Pain** (75%), Contractures
- Skin ulcers
- **Nutrition**: Malnutrition, Obesity
- Drooling
- **Mental Health**: Anxiety, Depression, ADHD, Sleep issues
ASSOCIATED IMPAIRMENTS

- Sleep Disorders
- Intellectual Impairment
- Breathing Difficulties
- Bladder Control Problems
- Unable to Walk
- Behavior Disorder
- Vision Impairment
- Saliva Control Problems
- Unable to Talk
- Epilepsy
Case continued

Jackie reports being frustrated because she is not able to do her day to day activities of daily living (ADL) by herself; she needs assistance for walking, bathing, getting dressed, toileting, and transferring from one place to another.

Jackie reported feelings of anger, isolation, sadness, and anxiety. Jackie wants to stay home all the time and does not enjoy interacting with her family and friends like she used to; in addition, this has affected her social and emotional well-being of maintaining relationships and interacting with friends and family.
Quiz

What is your next recommendation?

A. CBT
B. SSRI
C. Refer to PT
D. All of the Above
Case continued

Using shared decision-making, you start an SSRI and refer Jackie to a local counselor who specializes in adults with physical disabilities.

You will look into referrals for adult neurology, orthopedics and physical medicine and rehab.

By the end of her visit, she is hopeful that she will be able to receive adequate care as an adult.
Common Pitfalls Caring for a Patient with CP

• Not screening for STIs - sexual health should be routinely addressed at visits
• Neglect, Abuse, domestic/sexual violence on the ddx → 60% increased risk
• Discuss QOL: school, work, exercise, mental health, nutrition
• Low threshold for DEXA- nutrition, nonambulatory, immobility
• All adults w/ disability can access Vocational Rehabilitation
• Never stop baclofen without discussion- withdrawal is life threatening!
Case #2

Danielle is a 21yof born with spina bifida. She recently established in your outpatient STEP clinic.

She called clinic today with reports of confusion and was told to come to the ER.
Case Introduction

In the ER, Danielle’s mother was with her and reported she had 2-3 days of progressive confusion.

At triage, she was found to have Temp: 100, HR 110, BP 90/60.

What are the basic things you need to know about Danielle and her disease process?
What are the basic things you need to know about Danielle and/or any patient with SB?

1. Type of spina bifida
2. Surgical history
3. Equipment/ Devices
4. Spina bifida-related comorbidities
   - Continence
   - Mobility
   - OSA
   - Hydrocephalus/Chiari malformation
   - Intellectual Disability
The spectrum of spina bifida
Myelomeningocele

Clinical complications depend on lesion location and surgical outcome

1. Chiari II malformation with associated hydrocephalus (80%)
   - The Chiari malformation can cause brainstem dysfunction

2. Neurogenic bowel and bladder
   - Almost all patients have this to some degree

3. OSA, Restrictive lung disease, chronic hypercapnia, aspiration PNA

4. Weakness, Paralysis, Club foot, scoliosis

5. Intellectual Disability, Malnutrition, Obesity, Anxiety, Depression
Treatment of Hydrocephalus
Treatment of Neurogenic Bowel

Medications: Stool softeners + Enemas

Malone Antegrade Continence Enema (MACE)

CHAIT Cecostomy
Treatment of Neurogenic Bladder

CIC q 4 hours

Mitrofanoff bladder stoma

Ileocystoplasty
Why is bowel and bladder regimen important to discuss inpatient and outpatient?

Increase risk of preventable hospital stays:

- Urinary tract infections
- Kidney stones, CKD
- Shunt malfunction
- Potential for skin breakdown d/t incontinence
- Decreased quality of life
- Worsening anxiety/depression
What basic questions should you ask Danielle?

1. What type of spina bifida do you have?
2. What surgeries have you had? Do you have a shunt? When was it last revised? Do you know if it is programmable or not?
3. How do you manage your bowel and bladder? If you self-catheterize, how often? Does anyone help you?
4. How do you get around your house and your community?
5. When you were under pediatric care, what subspecialists did you see?
6. What supplies and equipment do you need?
Danielle’s responses

1. Danielle had a lumbar myelomeningocele, repaired at birth.
2. She has a ventriculoperitoneal shunt (last revised six months ago).
3. Her mother helps her catheterize her bladder via the urethra intermittently every 4 hours. She uses a Malone antegrade continence enema channel (MACE)
4. For most of the day, Danielle is in a manual wheelchair.
5. As a child and adolescent, she saw a neurosurgeon, a urologist, a physical medicine and rehab specialist, and an orthopedic surgeon.
6. Her supplies and equipment include: catheters, pull-ups, cone enema, manual wheelchair, walker, bath/toilet adaptive chair.
In the ER, Danielle’s mother was with her and reported she had 2-3 days of progressive confusion.

At triage, she was found to have HR 110, BP 90/60.

What are you most worried about with this patient?
Pop Quiz

A. Urosepsis
B. Shunt malfunction
C. Pneumonia
D. Skin/ Soft tissue infection
E. All the above
What are the most common causes of hospital stays?

1. Urinary tract infection
2. Complication of device/implant/graft
3. Skin and soft tissue infection
4. Pneumonia
5. Chronic skin ulcer
6. Sepsis
7. Fluid/electrolyte disorders
Case continued...

Exam w/o neurologic deficits

Shunt imaging pending

UA revealing 2+ leukocyte esterase, + WBCs, RBCs, bacteria.

Pt is admitted to hospital medicine on abx for presumed urosepsis.
In the EMR...

Her urine culture is growing *E. faecalis*

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Case Continued...

Next day, pt has temp 101, HR 80, BP 120/80. Shunt series with normal ventricle size, no concern for hydrocephalus.

Mom is at bedside today and says "she is still confused. Could there be something wrong with the shunt?"
What do you do next?

A. Tell her the imaging is normal and this is nothing to worry about
B. Consult NSGY to tap the shunt
C. Lumbar puncture
D. MR brain
E. No clue!
True or False: Normal size ventricles rules out shunt malfunction
False

- Imaging is NOT always conclusive
- Intermittent draining, low compliance can still result in small/normal size ventricles
- Do NOT ignore clinical symptoms- new swallowing issues, snoring, apneas, behavior changes
- Discuss with NSGY directly
What to do next?
What to do next?

- CT head with shunt series
- Blood culture, CBC
- CSF cultures are done by neurosurgery
Patient follow-up

- NSGY consulted → Patient found to have abdominal abscesses in shunt catheter and CSF w/ + pyuria

- Transferred to NICU for shunt externalization, IV antibiotics

- After 1 month prolonged hospital stay, patient improved, shunt internalized and pt discharged home to follow-up in multiD Spina Bifida Clinic
Could this have been prevented?

Over 1/3 hospitalizations in adults with SB are due to **preventable** causes:

- Urinary tract infection
- Pressure ulcer
- Skin/soft tissue infection
- Septic arthritis
Common Pitfalls Caring for a Patient with Spina Bifida

- Do NOT forget bladder regimen (Cath vs Foley)
  - Many are caregiver dependent for caths
- MACE/CHAIT dysfunction or leaking → Call Interventional Radiology
- Do NOT do an LP → tap the shunt
- Assuming that a positive urinalysis means UTI
  - Almost all adults who have spina bifida will have a positive urinalysis - wait for the urine culture
- Do NOT forget the SHUNT
  - Headaches, apnea, choking, cognitive changes, nausea/vomiting, persistent fevers should ALWAYS prompt urgent NSGY evaluation;
Common Pitfalls Caring for a Patient with Spina Bifida

- Ignoring a loss of hand function
  - This could mean a syrinx has developed and the patient should have a complete spine MRI and an urgent neurosurgery referral
- Ignoring back or neck pain
  - This could be tethered cord syndrome
- Ignoring Elevated bicarbonate
  - Concern for chronic hypercapnia/ OSA- may need BIPAP
- Expecting cognitive function to be totally equal to an unaffected individual
  - People with SB can often have issues with attention, word-recognition, math, visuospatial reasoning
Multidisciplinary model of care at UAB
What is STEP PROGRAM?

Opened doors 2019

Med-Peds Physicians

Transition Consultation and/or Primary Care for ANY patient over 18 w/ complex disease of childhood

“Equal or better care for every patient”
What is STEP PROGRAM?

- Multidisciplinary Clinics for pts w/ CP, Epilepsy, Neuromuscular Disease, Technology Dependance, Renal Transplant
- Transition Champions
- SW and PT in every clinic
- Community Partnerships
- Statewide Outreach
Goals

Access

Individualized transition plan (ITP)

Advance Care Planning

Improve transition readiness

Telehealth

Emergency Planning
STEP Demographics

• >300 new patients from around the state of AL
• 67% require medical equipment
  • Including wheelchairs, CHAIT tube, gastrostomy tubes, ventriculoperitoneal shunts, etc.
• Most common diagnoses include cerebral palsy (25%), genetic disease (14%), neural tube defect (12%), and neurologic disease (11%)
• 28-33% verbal patients with severe depression or anxiety
• Only 3% patients with normal BMI
Takeaways
Lesson #1
They need Transition resources
Lesson #2

Communication is key

“Do not Assume I am Dumb”
Communication is KEY

- Do not ASSUME your patient cannot communicate or understand based on a diagnosis
- Address Patient directly
  If Patient cannot communicate for his/her/them self, clarify -
  - Who is the patients medical decision maker?
  - Do you have legal power of attorney?
  - Do you have legal guardianship?
  - If there is ID, does patient require assistance from caregiver to make medical decisions?
- Fight temptation to ask caregiver
- Acknowledge the patients right to consent
- Two-way communication with caregiver is CRUCIAL
- Establish Rapport
- Gradually discuss difficult issues: GOC, Guardianship
Lesson #3: Screen Mental Health

• Consider GAD7, PHQ9 in all patients
• Discuss sleep, mood, attention
• Psychiatric comorbidities are COMMON up to 33%
  • Depression, Schizophrenia, Bipolar, Anxiety, PTSD, ADHD
• 1/3 on psychotropic medications
Lesson #4
They need coordinated social work

- Insurance status, waiver support
- Guardianship
- Advanced Care Planning
- Vocational Rehabilitation
- School accommodations
Lesson #5: Nutrition is poor

- BMI can be difficult to interpret
- Discuss Ca, VitD, Iron intake, water intake, exercise w/ every patient
- Discuss swallowing, reflux, GERD
Lesson #6 These patients need each and every one of YOU
And you are not ALONE
STEP CLINIC Contact

UAB Medicine Refer a Patient: https://uabmedicine.org/web/medicalprofessionals/refer-a-patient

External Consultation: Cahaba Medicine, plans to expand

Email me directly: Chstein@uabmc.edu