June 21 – 24, 2023
Orlando, FL
ORTHOPAEDIC ISSUES AND

PWS

Harold J. P. van Bosse, MD, FAAOS
Pediatric Orthopaedic Surgery
SSM Health Cardinal Glennon Children’s Hospital
Saint Louis, Missouri
Introduction

- Most common genetic obesity syndrome
- 1887 Langdon-Down
  - Adolescent girl
- 1956 Prader, Labhart and Willi
  - Series of patients
- 1981 Ledbetter, Riccardi and Airhart
  - Microdeletions of chromosome 15
Prader-Willi Syndrome

- Genetic syndrome
- Chromosome 15
  - Prader-Willi region
  - Lacking from father’s chromosome
  - If lacking from mother’s chromosome: Angelmann syndrome
- Genetic syndrome
  - Unlikely to “run in the family”
  - Most causes of PWS run <1% risk
  - “Imprinting defect” runs about 50% risk
  - Accounts for <5% of PWS cases
Musculo-skeletal Characteristics

- **Hypotonia**
  - Especially in the infant
  - Developmental delay
  - Contributes
    - Scoliosis
    - Hip dysplasia
    - Flat-footedness (pes planus)
“Why Doesn’t My Practitioner Know PWS?”

Rare: Occurs about 1:10,000 to 1:30,000 births

Compare with more “common” orthopaedic conditions

- Cerebral palsy 3:1000
- Spina bifida 7:10,000
- Hip dysplasia 1:1000
- Clubfoot 1:2000

Generalized treatment thinking

- But children with PWS follow different rules
- Need to treat as a “child with PWS who also has scoliosis, hip dysplasia, pes planus, etc”
- Not “Child with scoliosis who also has this PWS thing”
Musculo-skeletal Issues in PWS

- Developmental delay (milestones)
- Flatfoot deformity (pes planus)
- Osteopenia (low bone calcium)
- Frequent fractures
- Hip dysplasia
- Spine deformities
Hypotonia early on

Pre-natal and delivery history
- Decreased fetal movement
- Often Cesarian section for delivery

Newborn
- Poor sucking ability
- Weak cry
History

Infants and Toddlers

- Poor head control
- Poor postural control (sitting)

Milestones can take twice as long

- Sitting 12 months average (typical 6 months)
- Walking 27 months average (typical 12 months)
Treatment

Therapy, Therapy, Therapy

- Balance and motor abilities

Bracing

- Ankle-knee control
- Ankle-Foot Orthosis (AFO)
  - Solid ankle braces – stable foundation
- Purpose – “Get them up walking any way possible... then work on points for style”
Ankle Foot Orthosis (AFO)

- Solid versus hinged
- More ankle stability with solid braces
PES PLANUS (FLAT FOOT) DEFORMITIES
PES PLANUS DEFORMITIES

Flat feet occur frequently in PWS ~41%
- Laxity in ligaments and low-tone musculature
- Children generally “fall” into pronation
- Poor foot positioning for walking/running

Mechanical Effects
- Poor foot/ankle position, heel rolls to the outside
- Hard to generate “push off” or get up on toes
- Shortening of the Achilles tendon
- Develop a crouched position, knees slightly bent
Pes Planus Deformities

Residual Effects

- Poor balance, wide stance and walking base
- Prolonged cruising, delayed reaching with hands and running
- Leg fatigue
- Forward leaning posture with high guard (hold hands up)

Treatment

- Bracing
  - SMOs or UCBLs
  - If using AFOs, transition once walking well
Supra-malleolar orthotics (SMO)
- Start just above ankle malleoli
- Usually not full foot length
- Soft malleable plastic
My bias against SMOs

- Forefoot abduction
  - Lack of full lateral border
  - Front of foot still drifts out

- Ankle motion
  - Design inhibits full ankle motion

- Heel stability
  - Sub-optimal control of heel valgus
Arch support won’t work
Osteoporosis – weak bone

- Low bone mineral density

Different studies show different rates

- 9% osteoporosis (PWSA-USA survey)
- 29% history of fractures (British adult study)
- 45% fractures (Philadelphia adult study)

- Decreased pain sensitivity
- No recent studies

- Probably better results due to growth hormone
Osteoporosis and PWS

20% lower Bone Mass Density (BMD) vs. typical (2004)

- Dual Energy X-Ray Absorptiometry (DEXA)

Significantly lower BMD

- Lumbar spine
- Pelvis
- Lower extremities

Not significantly lower BMD

- Head, upper extremities, thoracic spine
Osteoporosis and PWS

Treatment

- Growth hormone
- Vitamin D and calcium
- Activity
- Awareness
  - Injury appreciation
  - Especially with characteristic PWS higher pain threshold
- Surgical planning
Normal hip

PWS with hip dysplasia

HIP DYSPLASIA
**Hip Dysplasia**

**Dysplasia** - Abnormal development or growth
- Presumably normal during early development
- Deforms with growth in late pregnancy or after birth
  - Hip needs movement and muscle forces to develop correctly
  - Hip dysplasia can lead to hip arthritis

**Congenital Dislocation of Hip**
- Hip born out of socket
- Very rare in PWS (to me)
  - I only know of 3 cases
- Other centers have reported higher incidence
Hip Dysplasia

- **Incidence 8 – 30%**
  - ~1% for typically developing children

- **Prevention**
  - Early screening of hips (when start sitting independently)
  - Activity
  - Continue to screen every 1-2 years for at risk hips

- **Hip arthritis**
  - No reported cases
  - Too few cases per surgeon
National Hospital Discharge Database

- 2004-2014: 3.1 million (weighted) THAs
- 39 total with PWS
- 68% under 50 years old
- THA incidence rate in PWS was 1:80,000
- PWS incidence 1:30,000
- People with PWS ~2½ times less risk of hip replacements
- If dysplasia rate so high, why is arthritis rate so low?

REMODELING!
5.5 year old girl, PWS/UPD

5.5 years old

10 years old

7 years old
13 month old girl

5 years old
SPINE DEFORMITIES
Spine Deformities

- **Scoliosis**
  - Any curve seen from behind

- **Kyphosis and Lordosis**

- **Hyperkyphosis**
  - Exaggeration of the normal postural upper back curve, seen from the side

- **Hyperlordosis**
  - Exaggeration of normal lower back curve (small of back)

![Diagram showing normal and deformed spines]
Let’s do the numbers!

Spine deformity prevalence in PWS
- Approximately 60%-70%

Bimodal age distribution
- 23% of children before 4th birthday
- Second (bigger) peak is in the adolescent period

15% of PWS children will need spine surgery

Complication rates from surgery ~56%
More numbers

Treatment rationale for scoliosis

At maturity

Curves $\leq 40^\circ$: 95% will not progress in adulthood
Curves $\geq 50^\circ$: 95% will progress
Between $40^\circ$ and $50^\circ$ - a grey zone

Curves under $25^\circ$ - observe
Surgery is indicated for curves $> 40^\circ$ to $50^\circ$
SCOLIOSIS
Treatment Rationale

Cardiopulmonary Compromise

Pulmonary insufficiency

- Lungs too squooshed to get enough oxygen into the bloodstream for the body

Cor pulmonale

- Heart has to work too hard to push blood through the squooshed lungs: overwork

Curves over 80° to 90°

Smaller curves can cause breathing problems (curves over 60°)
Hidden Spine Deformities

8 ½ year old boy

99°
Hidden Spine Deformities

4 year old with 30° curve
Scoliosis

**Obesity**

- No differences in Body Mass Index
  - Between children with or without scoliosis
- More than 50% of all curves start before obesity onset
  - 2/3 of all severe curves start before obesity

More likely due to **hypotonia**

**Obesity control**

- Detection
- Bracing
Gender
- Females have ~10% higher chance of developing scoliosis
- Curves behave the same (risk of progression) for both genders

Genetics
- UPD had a slightly higher risk of developing scoliosis
- No PWS type has a higher risk of progression

Looking forward to FPWR online registry results
Treatment

- Difficult to draw conclusions
- Few reported operative cases in literature
- These are my opinions based on accumulated experience
Prevention

- Delay upright sitting until baby can pull to sitting position themselves
  - Prevents hypotonic slouch
  - Seating devices tilted back about 30°

- Emphasize tummy time activities as much as possible

1 ½ year old girl
Screening

Yearly screening/radiographs, once starts sitting

Physical therapy

Casting

Usually start before patients reach 3 years old

Bracing

For curves larger than 25°
Prevent curve progression when upright

Surgery

For curves larger than 45°
Reality Check

Important to note – some curves are destined to progress even with the best treatment.

Strategy then is to control the curve as long as possible.
Physical Therapy

Physical therapy
- Trunk strengthening
- Sensory integration
- Keep the young child down to develop normal gross motor skills
- Children with PWS develop their extremities before their trunk
Janice Agarwal’s Favorite Therapy
Hippotherapy

- Child who needs head, trunk, leg control
- Movement of the horse
  - Encourages the child’s body to exercise
  - Correct alignment
- Therapist can provide
  - Extra support
  - Resistance
Casting

Appropriate age has been increasing

- Even effective in older children (up to even 7 years old)
- Delay tactic before other treatments options: have problems

Cast under anaesthesia

Casting schedule

- Under 2 years, change every 2 months
- Over 2 years, change every 3 months
- Over 3 years, change every 4 months

End casting when reach goal, or curve reaches a plateau

Post-treatment bracing
Head halter traction

Criss-cross pelvic band traction

Sanders et al, JPO 2009
Derotate Chest to Correct Curve

Lamina thinner, vertebral canal narrower on concave side

Rib pushed posteriorly; thoracic cage narrowed

Vertebral body distorted toward convex side

Rib pushed laterally and anteriorly

Convex side

Concave side
PWS Spine Casting Study

PWS spine casting

- 34 PWS spine casting patients
  - 2008-2018
  - >24 months followup

Criteria for starting casting

- Curve greater than 25°
- Age: Sitting age to 5 y.o.
Endpoints

“Cured”
- Curve under 15° out of cast
- Curve progression is likely halted
- Transition to brace for 1 year, hopefully brace free afterwards

Braced
- 20°-50° and over 5 years old – brace expectantly

“Controlled”
- Reached surgical criteria before started casting
- >50° and over 5 years old – discuss expandable implants
Results

- **Average age at first cast**: 32 months (range: 14-64 months)
- **Average number of casts**: 8 casts (range: 3-18)
- **Average followup**: 57 months (range: 25–103 months)
### Results

**Cured ~1/3 of the group**
- PWS type: 7 UPD and 5 deletion
- Curves went from 44° to 17° over 6 casts (17 months)

**Braced ~50% of the group**
- PWS type: 10 deletion, 7 UPD, 1 methylation defect
- Initial curve 55° improved to 35° over 7 casts (27 months)

**Controlled 4 patients**
- PWS type: 3 deletion and 1 UPD
- Pre-cast curve 85° improved to 54°
- Surgery delayed ~ 4 years (from 22 months to 72 months)
Timeline of A Cured Curve

17 mos old boy
UPD

1st cast

After 4 casts

After 5 casts
20° curve
Followup

New technique

Adolescence

- 4 years old
- 2 ½ years post-casting

- 8 years old
- 6 years post-casting

- 9 years old
- 25°, restart bracing

- 11 years old
- 30° curve
Timeline of a Controlled Curve

18 month old with deletion

106°

1st cast

54°

13 casts

4 years old

60°
Followup

- 18 casts
- 4 years later
- Curves below 60º in cast
  - About 65º out of cast
- At a good age for expandable implants

13 cast
4 years old

18 cast
6 years old
No differences seen between those with “Cured” curves and any of the other groups

- Gender
- Sidedness (right versus left)
- Curve region (thoracic versus lumbar)
- Age at cast initiation

Initial curve <50°

- Odds Ratio 9, p=0.008
Casting Is Survivable

17 month old boy with 55º curve
5 casts over 15 months, braced for 12 months
Now, 4 years old with 13º curve, no brace
Casting is survivable!
Bracing

- For curves larger than 20º - 25º
- Prevent curve progression
- Cannot (usually) make a curve smaller
- Difficult to fit on obese children
Bracing
Bracing

January 2009
32°

January 2009
First brace

March 2013
44°
Bracing
5 year old girl, PWS/imprinting defect

Dec 2014, 5 y.o. 46º

Jan 2015, 22º Daytime brace

Jan 2015, 2º Nighttime brace

Nov 2018, 9 y.o. 21º
Bracing

10 year old girl, PWS/del

10 years old
37° and 41° curves
in TLSO

10 years old
in Providence brace

13 years old
25° curve
Bracing

20 month old boy, PWS/del

- 20 months old
- 51° lumbar kyphosis
- 2 ½ years old, in brace
- 4 years old
- 15° residual kyphosis
Bracing

10 year old girl, PWS/UPD

10 years old
33° and 32° curves

12 years old
In brace

15 years old
80° and 70° curves

19 years old
4 years after fusion
Surgery

Surgery is indicated for curves between 40° and 50°

- Nearing maturity

Align spine in best position

- Side to side curve (scoliosis)
- Front to back alignment (kyphosis/lordosis)

Hold in position

- Rods
- Hooks, wires, and screws
Surgery

High rate of complications in PWS

- Infections
  - Skin picking
- Anaesthetic (intra or peri-operative)
- Pulmonary/Respiratory
  - Apnea
- Hardware failure/pseudoarthrosis
  - Osteoporosis

Need to continuously educate treating surgeons to these special risks
Curves in Young Children

6 year old boy, 90º curve
Non Fusion Spinal Instrumentation

6 year old boy

107°

9 years old

30°
6 year old with 107° curve

Followup

- 16 years old
- ~50° curve
- “Graduated”
MAGnetic Expansion Control
MAGEC System

10 year old boy with PWS After 2 lengthenings
10 y.o. girl, 103°

End-game

Graduation

15 y.o.
Spinal Fusion

- For curves over 50° at maturity
- Timing of surgery
  - Balance expected maturity with curve size
  - My preference in younger patients: delay until the curve is over 50° in brace
- Avoid anterior approach
- Newer pedicle screw instrumentation
  - Better in osteopenic bone
15 y.o. girl with 67º scoliosis
18 y.o. with kyphosis
25° scoliosis and 110° kyphosis
Surgery and PWS
Pre-operative

Pulmonology work-up
- Sleep study for apnea (need post-op CPAP or BiPAP?)
- Possible ENT evaluation

Anaesthesia evaluation
- Assess airway management
  - Ability to intubate
  - Thickened saliva
  - IV access – may need central line
Surgery and PWS

Other Factors

- Be aware of food seeking behaviors
- High pain threshold
  - May be difficult to awake
  - Often more compliant with post-op therapy
- Airway management, apnea
  - Consideration to delay extubation
  - PICU for observation, CPAP or BiPAP
  - Hypotonia – poor cough
Surgery and PWS

Other Factors

- Skin picking
- Infections
- GI complications
  - Gastro-motility slows down
  - Very gradual increase in post-op diet
Typical Sagittal Alignment

Posture of the spine

- A “plumb” line from the upper neck vertebra
- Passes just front of sacrum
- To the center of the hips

PWS
- More head forward posture
- Plumb line more forwards
- Need to preserve surgically
Cervico-Thoracic Kyphosis

- Exaggerated forward positioning of the head
- Forward head thrust
- Characteristic of PWS posture
- Can be significant problem
- Can worsen after spine surgery
Sagittal Alignment
Reconstructions

17 year old girl
Growth Hormone and Scoliosis

**Concerns of worsening scoliosis with GH**
- Turner syndrome
- Scoliosis worsens with GH

**PWSA 2007 Survey**
- For every month GH not started, risk of surgery increases 0.7%

**My “take” on it**
- Scoliosis will progress comparative to growth
- GH has many important benefits
People with PWS are living longer

We work together for a better quality of life