MUSCULOSKELETAL ISSUES OF PRADER-WILLI SYNDROME: A FEW THINGS TO KNOW

United in HOPE
2025 PWS CONFERENCE

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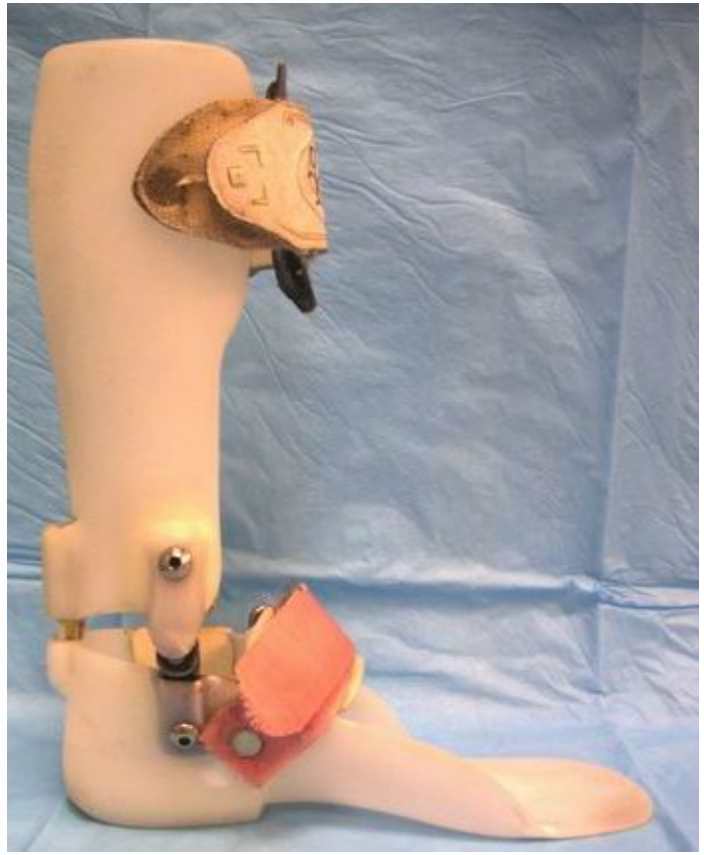
Orthopaedic Issues in PWS

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

DEVELOPMENT DELAY

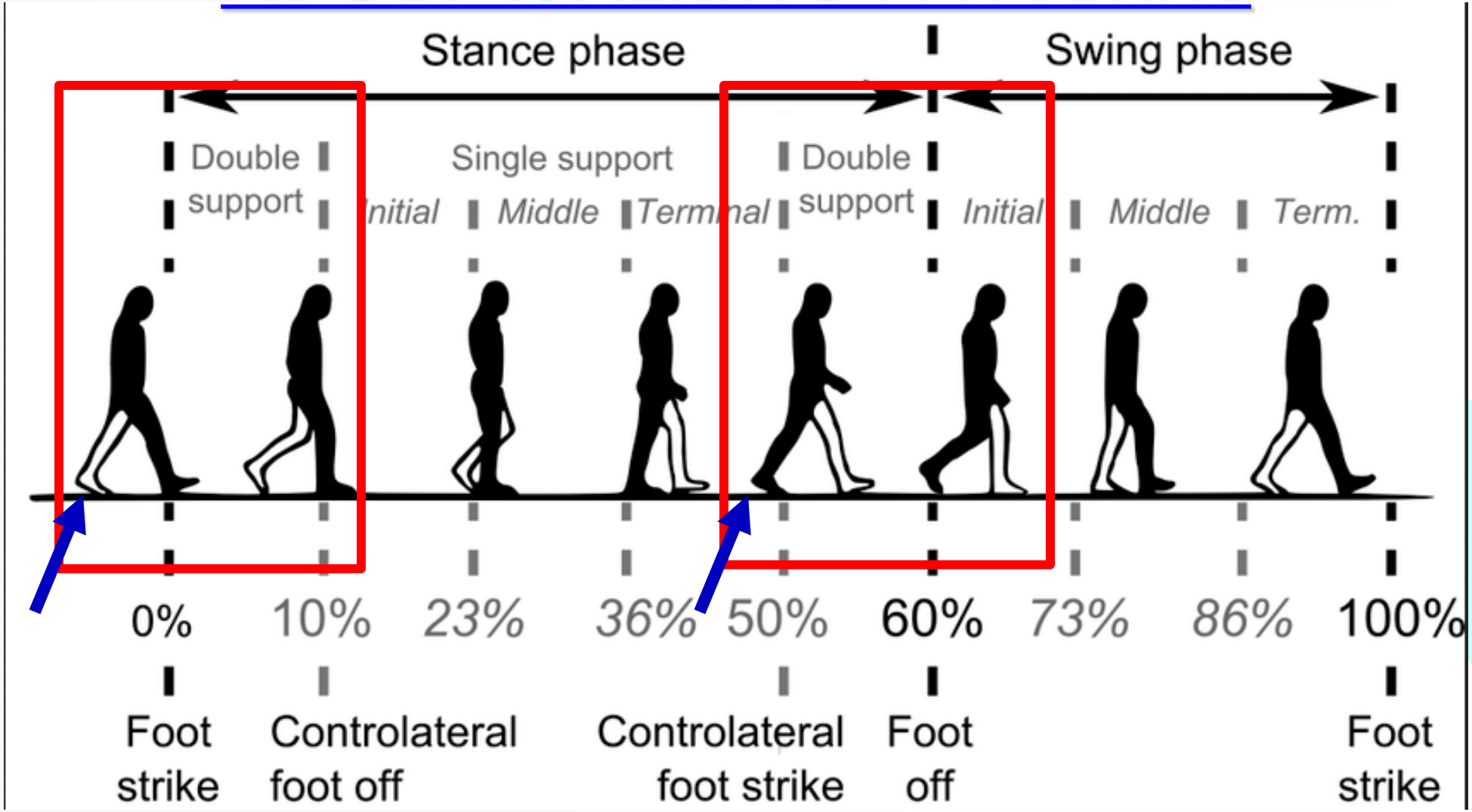
- **Milestones can take twice as long**
  - Sitting at 12 months, walking at 27 months
- **Therapy, Therapy, Therapy**
  - Physical, occupational and speech therapies
- **Bracing**
  - If not making significant gains toward walking by 16 months
  - Ankle-Foot Orthosis (AFO)
    - Solid ankle braces – stable foundation
  - "First we get them up walking any way possible… then we work on points for style"

Ankle Foot Orthosis (AFO)



- **Solid versus hinged**
  - **More ankle stability with solid braces**

ADULT WALKING PATTERNS



- Double support stance phase
- Toe off or Push off

ADULT WALKING PATTERNS

- Characteristic walking patterns of adults with PWS
  - Similar walking speeds and patterns to obese adults or elderly
    - Short steps, reduced speed, more time in double support gait
    - Decreased knee and ankle motion during gait
  - Poor push off
    - Decrease plantarflexion strength (weaker toe off/push off)
    - Smaller gastrocnemii compared to controls by ultrasound
    - Flat-footed gait pattern in late stance
  - Decreased gait efficiency and walking speed
- Exercises: work on muscular force and power
- Multisensory: reliance on vestibular system for balance

PES PLANUS (FLAT FOOT) DEFORMITIES

Pes Planus Deformities

- Flat feet occur frequently in PWS ~41%
  - Laxity in ligaments and low-tone musculature
  - Poor foot positioning for walking/running
  - Prolonged cruising, delayed reaching with hands and running
- Treatment
  - Bracing
    - Supramalleolar orthotics (SMO)
    - University of California Berkley Laboratory (UCBL)

SMOs and UCBLs

Adults

- **Pes planus (flat footedness)**
  - Unstable standing platform
  - Decreased push off strength
  - Consider UCBLs or plantarflexion assist AFOs
  - Possible surgical correction

OSTEOPOROSIS AND PWS

- **Bone mineral density (BMD)**
  - Measurement of calcium and other minerals in bone
- **Osteopenia**
  - BMD within 2 ½ standard deviations of normal
- **Osteoporosis**
  - BMD decreased by more than 2 ½ standard deviations
- **Studies show varying rates in adults**
  - 9% osteoporosis by survey
  - 29% and 45% history of fractures
    - Decreased pain sensitivity

Bone Mineral Density (BMD)

- **Conflicting data in childhood/adolescence**
  - **Childhood: BMD equal to *or* lower than peers without PWS**
    - **Growth hormone (GH) improves BMD, need to start in early childhood**
    - **Normal weight pre-pubescents with PWS have lower BMD even on GH**
    - **Exercise program (> 24 weeks) increased spine BMD in children**
  - **Adolescence: gradual BMD decrease despite growth hormone**
    - **Possibly related to incomplete pubertal development**
- **Treatment**
  - **Growth hormone, Vitamin D and calcium**
  - **Activity**
  - **Awareness (injury appreciation, surgical planning)**

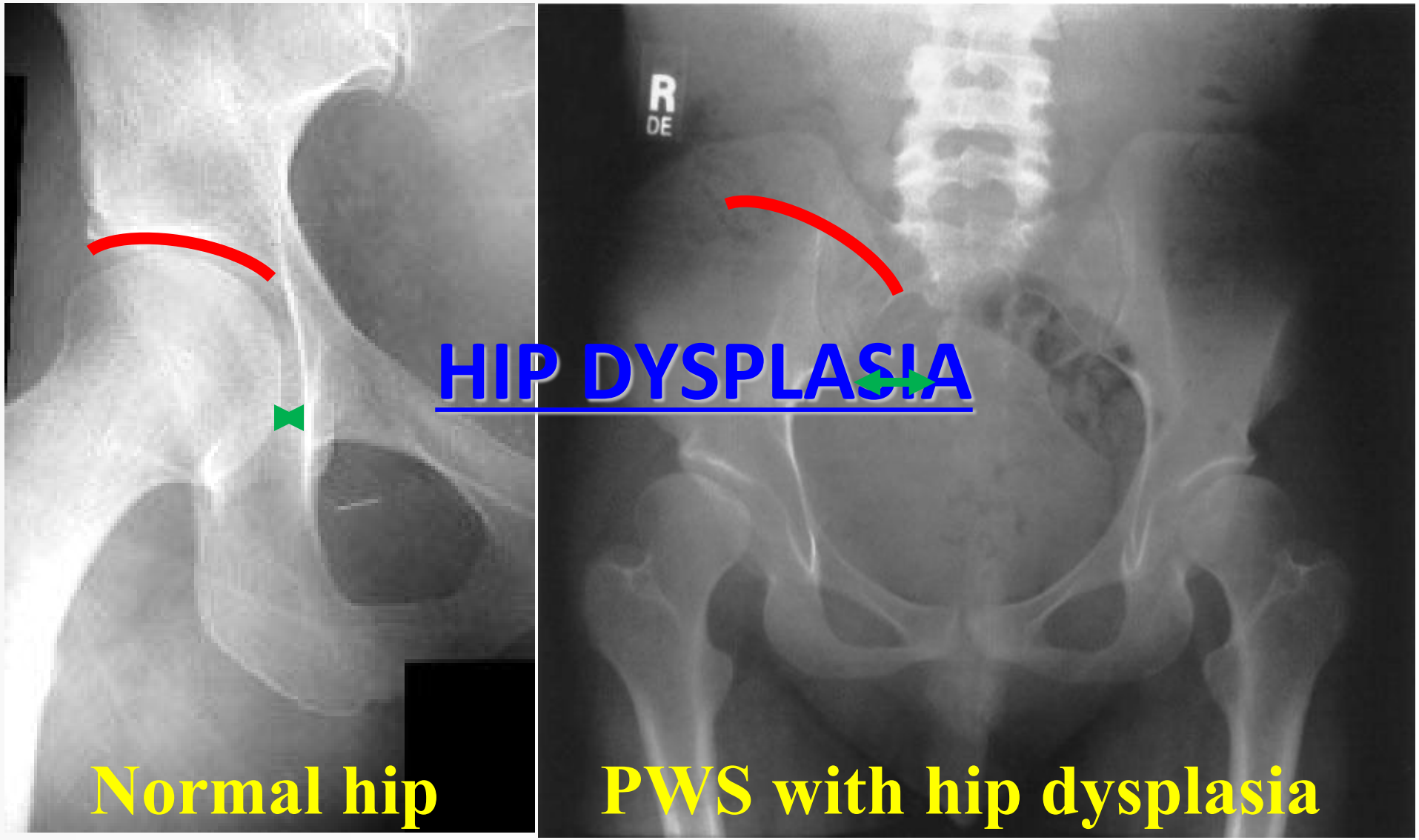
Bone Mineral Density (BMD)

- **Adults**
  - Recent study: 54% osteopenia, 14% osteoporosis
  - Osteoporosis more prevalent in males than females
    - Fractures more common in males
  - Adults on GH: no increased BMD, but better bone architecture
    - Starting GH as adult did not help BMD
- **Sex hormone replacement + GH**
  - Increases BMD and muscle mass in females with hypogonadism
  - BMD declined in males possibly due to insufficient replacement

Bone Mineral Density (BMD)

- **Vitamin D averaged 9ng/ml (Low normal 20ng/ml)**
  - Low levels most associated with low oral intake
    - Also associated with increased BMI and fat mass
  - Cognitive consequences of low vitamin D in *all adults*
- **Recommendations**
  - Continue GH in adulthood
  - Screen for hypogonadism in adolescence
    - Sex steroid replacement therapy for hypogonadism
  - Screening for osteoporosis in adults very 2-5 years with DEXA
  - Vitamin D (calcium) supplementation

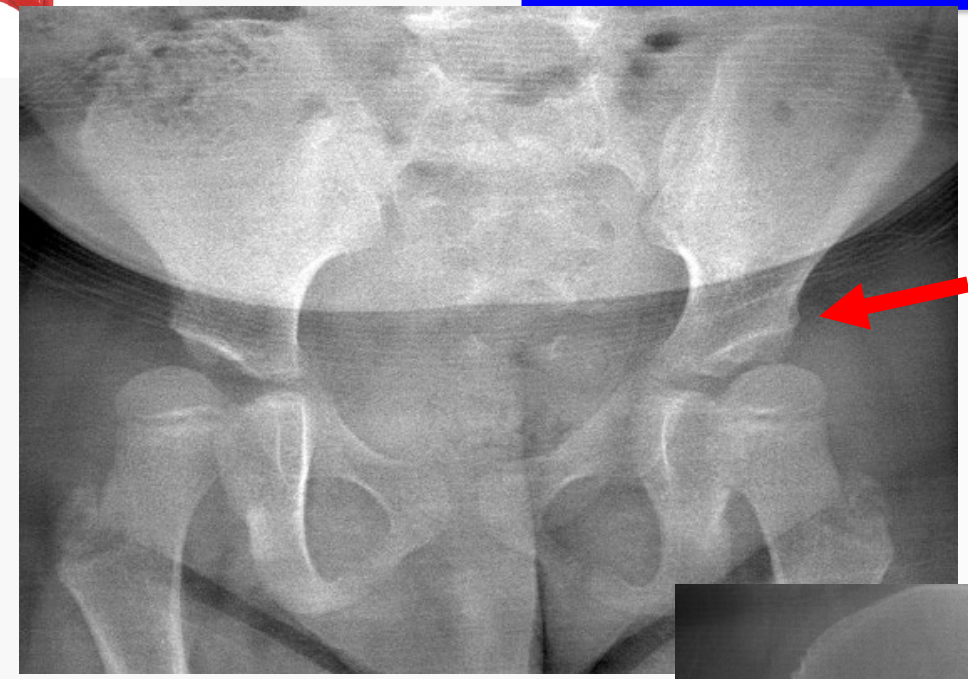
HIP DYSPLASIA



Hip Dysplasia

- **Incidence of dysplasia 8 – 30%**
  - Higher rates in babies with UPD type versus deletion type
  - Hypotonia is a risk factor
  - Incidence of hip dysplasia appears lower in GH treated group
- **Incidence of congenital hip dislocation very low**
- **Consequence of hip dysplasia - early arthritis**
  - Main reason for total hip replacements in general
    - Adults with PWS: 2½ times lower risk for total hip replacements
    - Probably due to high potential for remodeling with growth

5.5 year old girl, PWS/UPD

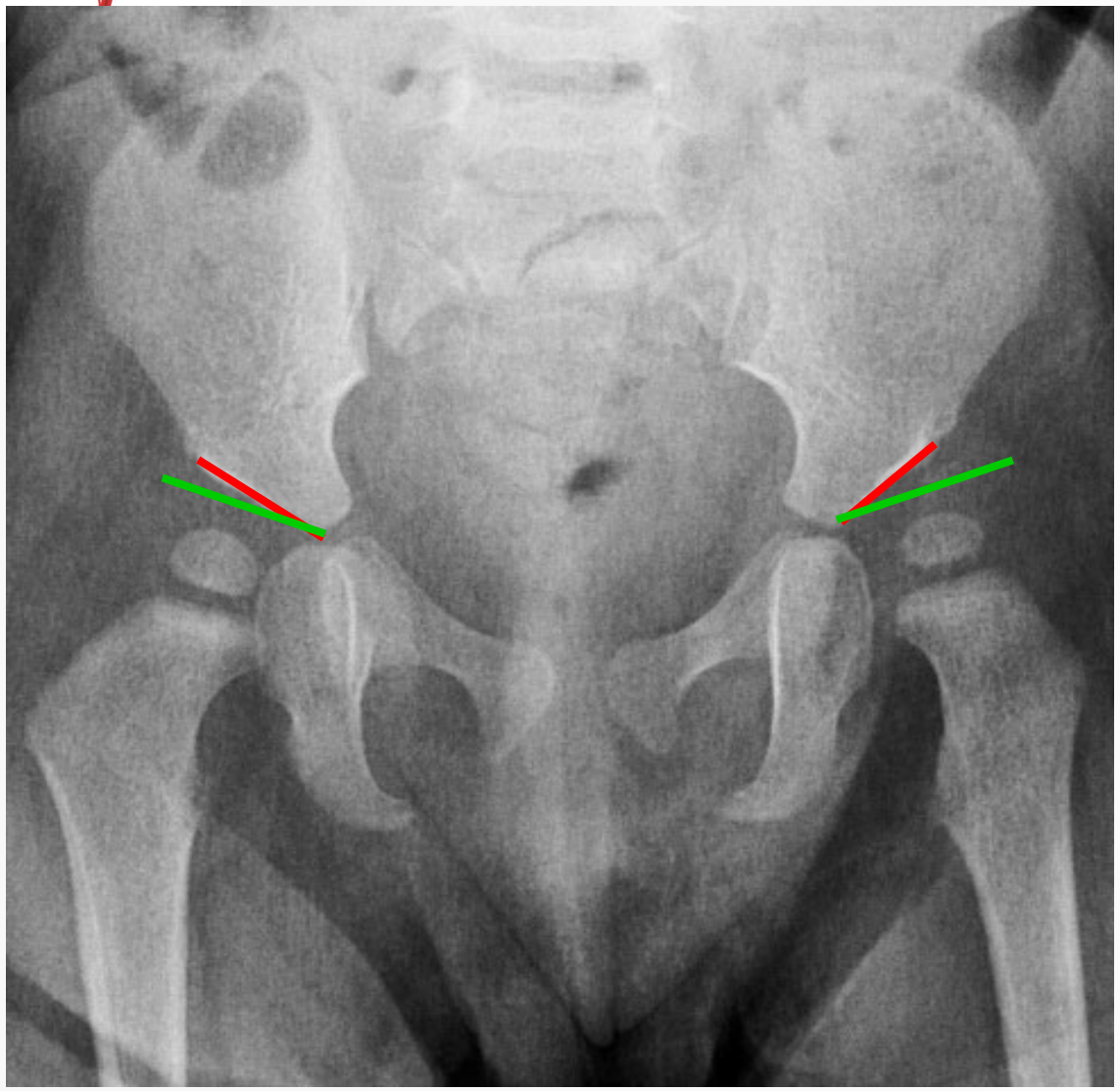


5.5 years old

7 years old

10 years old

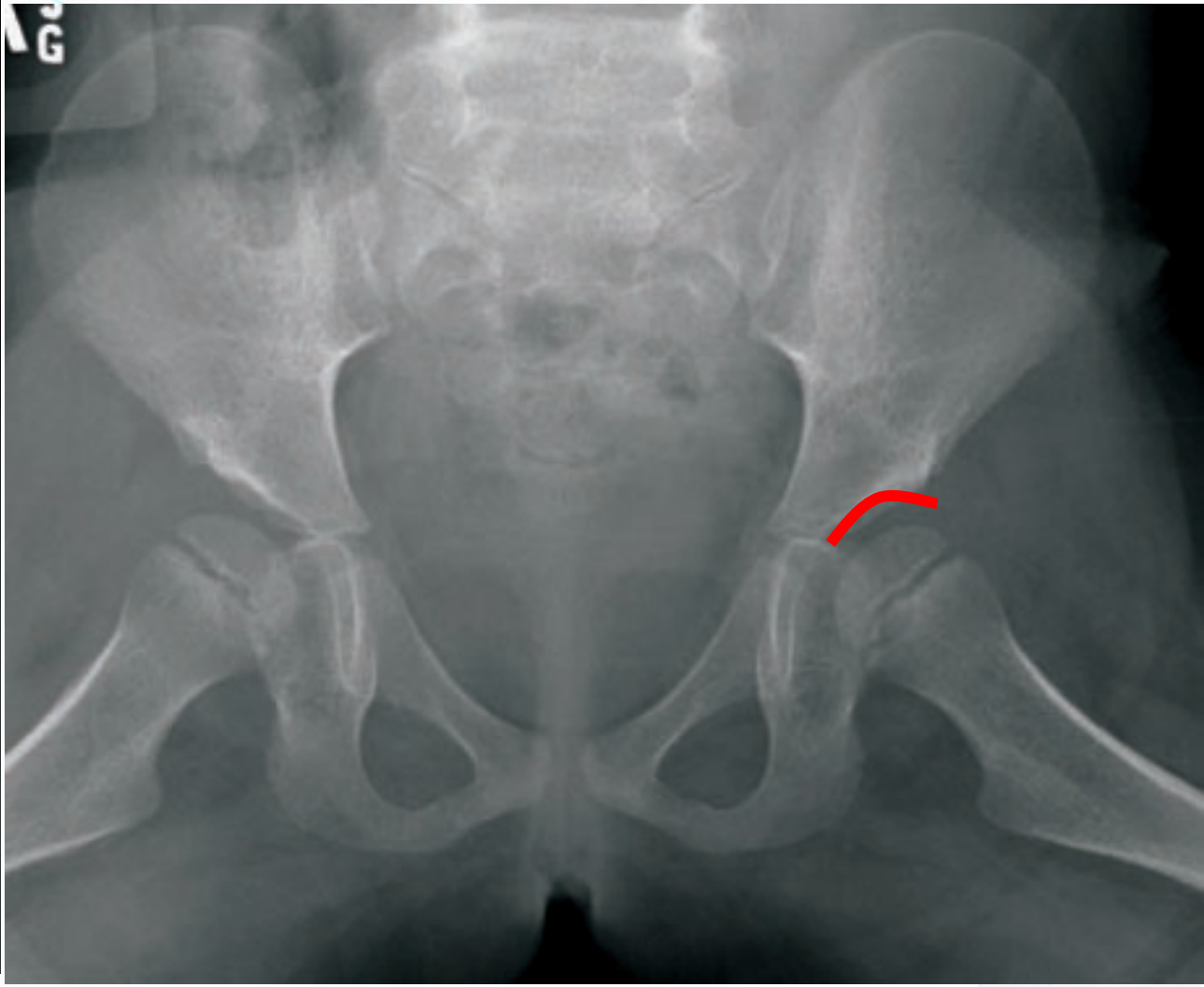
13 month old girl



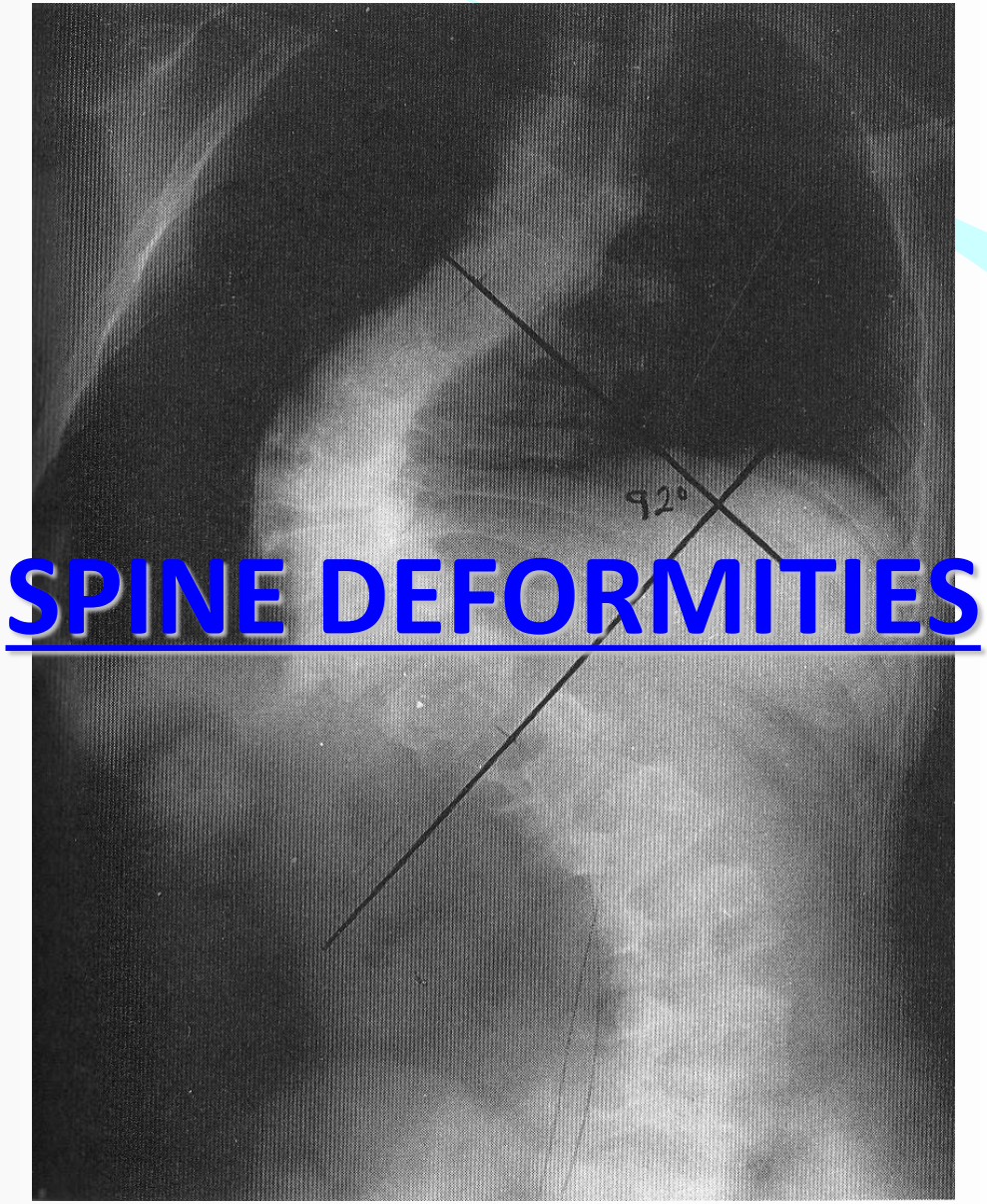
5 years old

Asymptomatic 2 year old child

Hip subluxation (outward drifting)



SPINE DEFORMITIES



Let's do the numbers!

- **Spine deformity prevalence in PWS: 60%-70%**
  - 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%
- **Thumbnail sketch of treatment algorithm**
  - Routine sitting xrays when sitting independently
  - Curves under 25° - observe
  - Curves ≤ 40° - most will *not* progress in adulthood
  - Curves ≥ 50°: most *will* progress, plan surgery

Hidden Spine Deformities

4 year old with
30º curve

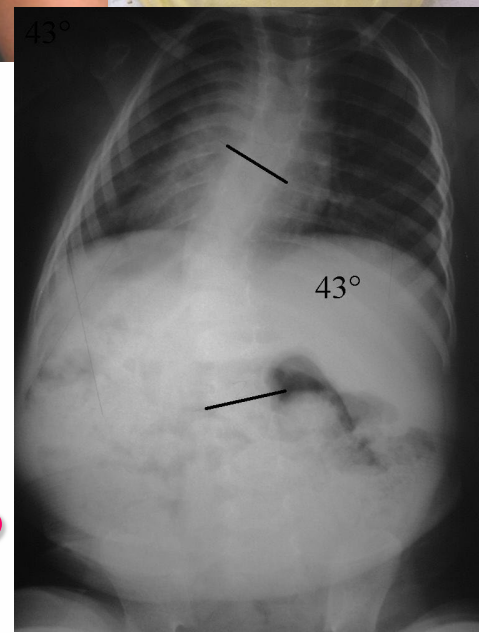
SCOLIOSIS

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 80º to 90˚**
  - **Smaller curves can cause breathing problems**
    - **Curves over 60º**

Prevention

- Delay upright sitting
  - Until baby can pull to sitting position
  - Prevents hypotonic slouch
  - Seating devices tilted back about 30°
- Emphasize tummy time *activities*
- Physical therapy and physical activity
- Growth hormone
  - No adverse effects on prevalence or severity of scoliosis in children with PWS
- High ghrelin levels under 1 y.o. predict scoliosis?



Treatment

- **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°

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

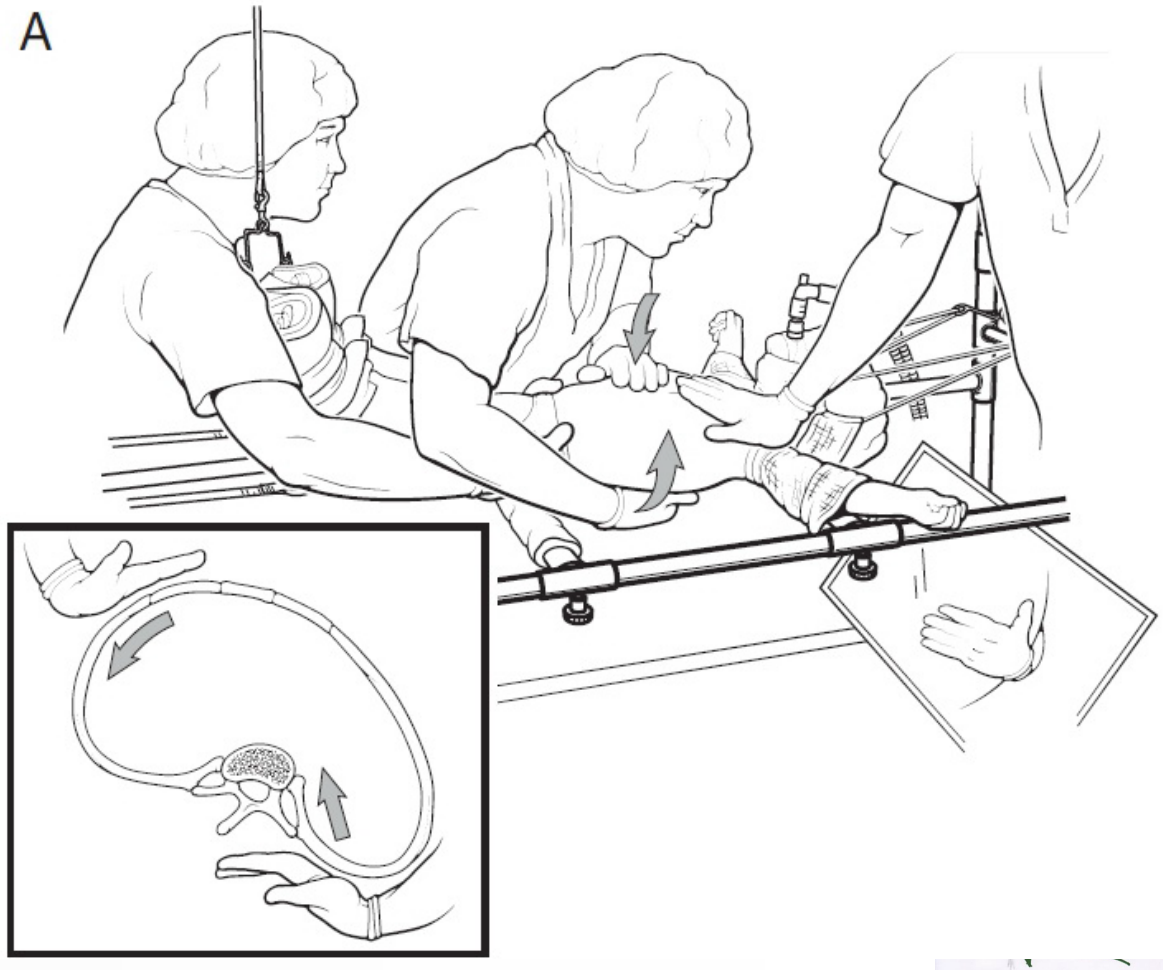
Spine Casting

Casting

- From sitting age to as old as 7 years of age
- 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
  - Delay tactic before other treatments options: have problems
- Post-treatment bracing

Head halter traction

Criss-cross pelvic band traction

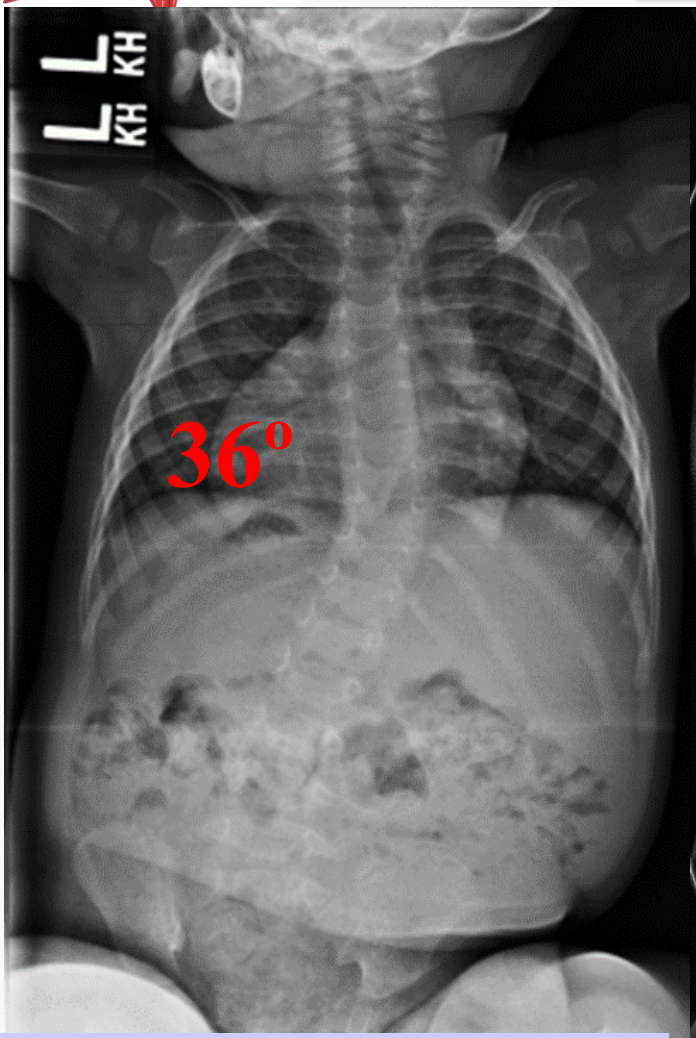


Sanders *et al*, JPO 2009

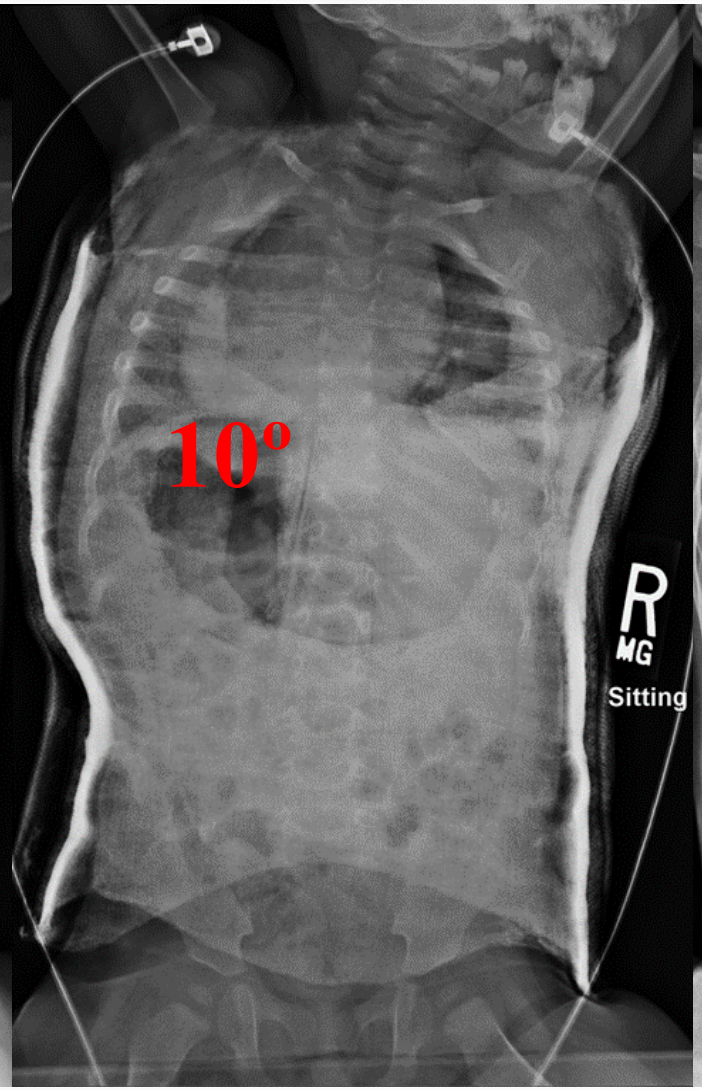
Outcomes of Spine Casting in PWS

- **Cured: curves decreased to less than 20º**
  - One third of patients
  - Curves went from 44º → 17º over 6 casts (17 months)
- **Braced: curves decreased to 20º-50º and over 5 years old**
  - About half of the patients
  - Curves went from 55º → 35º over 7 casts (27 months)
- **Controlled: delayed surgery 22 months to 72 months**
  - Curves went from 85º → 54º after casting

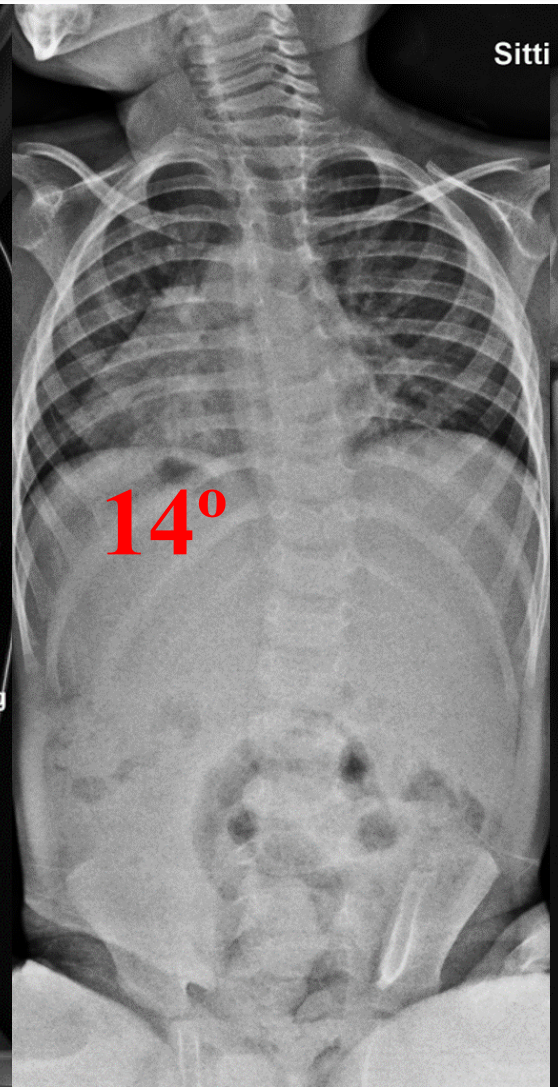
Timeline of A Cured Curve



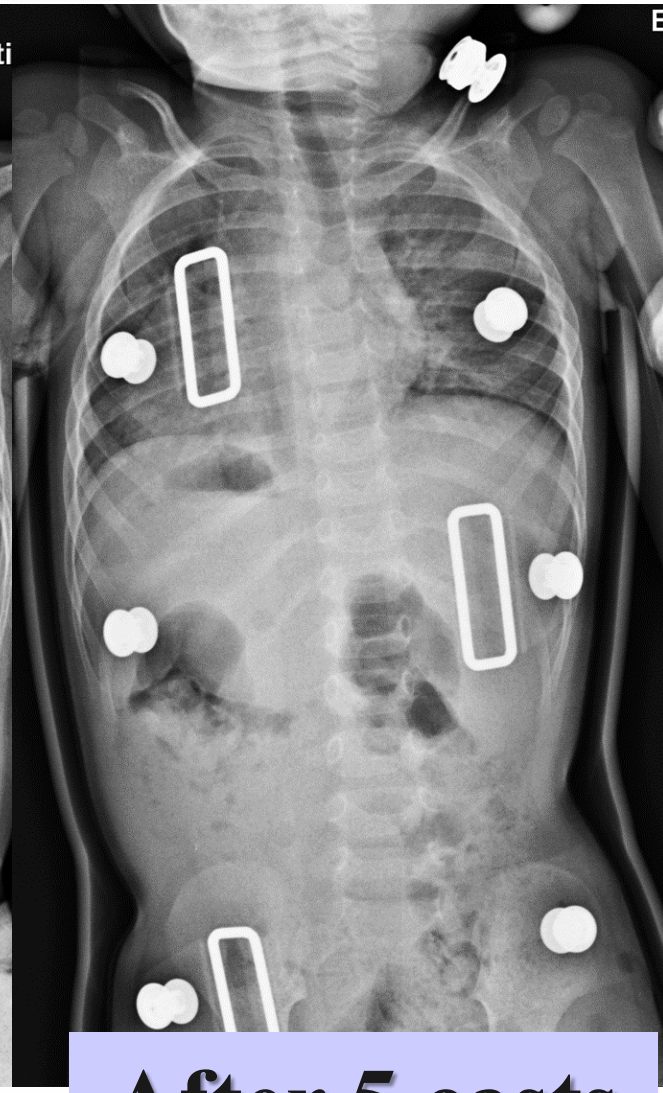
17 mos old boy
UPD



1st cast

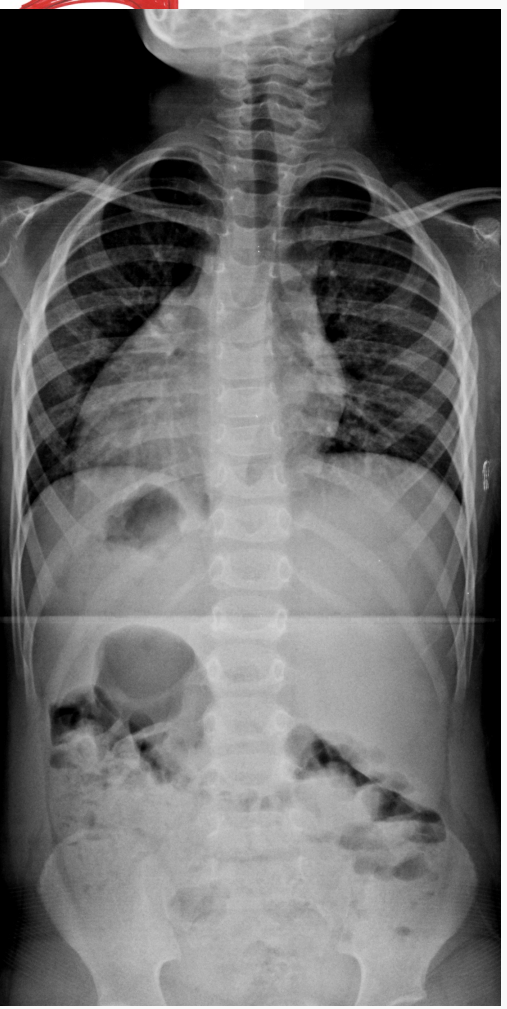


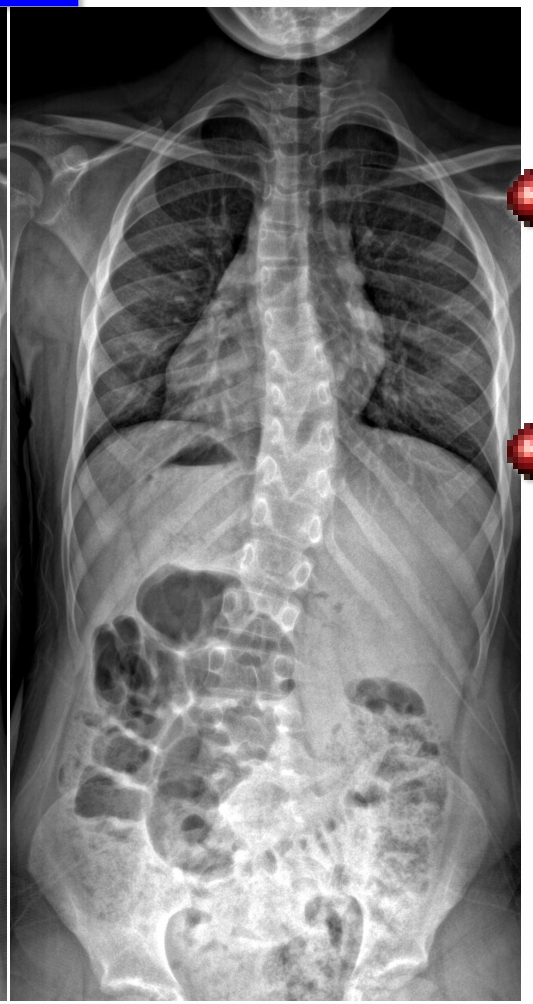
After 4 casts



After 5 casts
20° curve

Followup

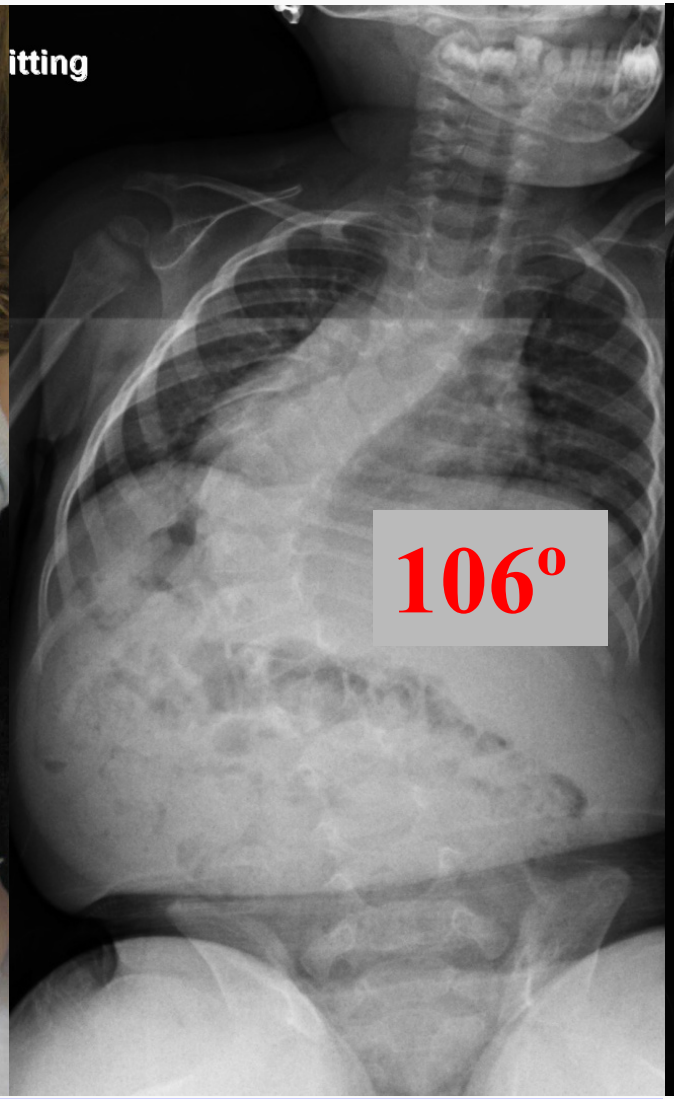


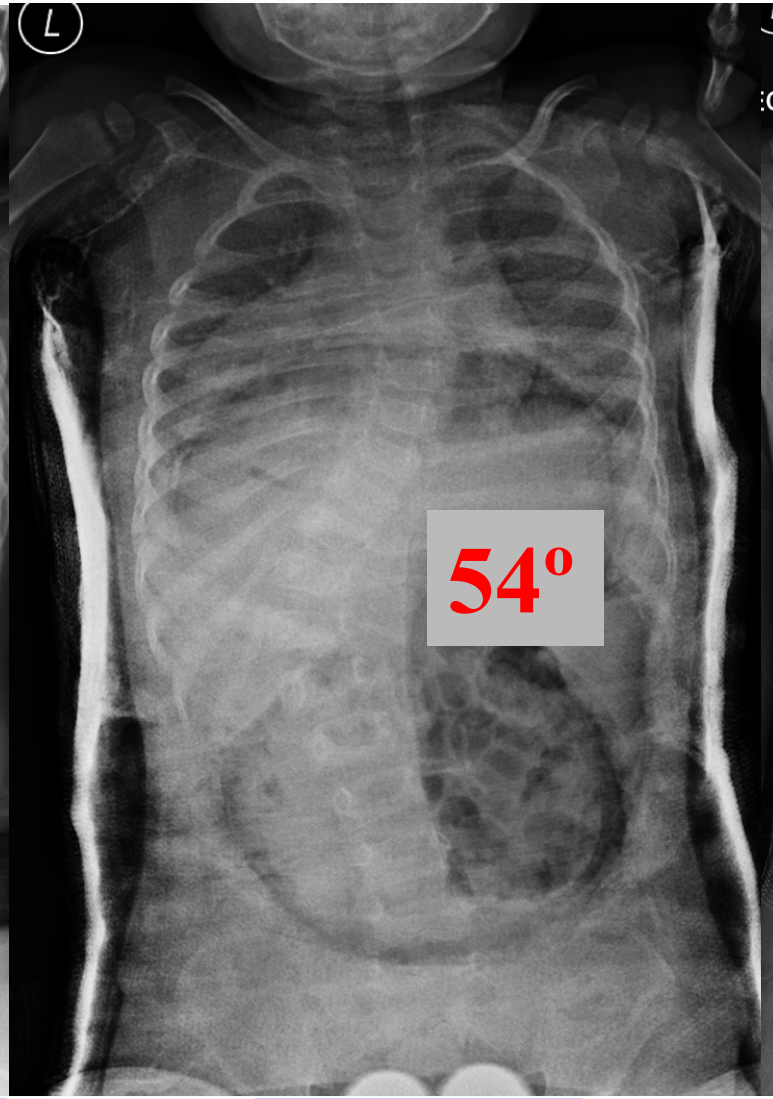


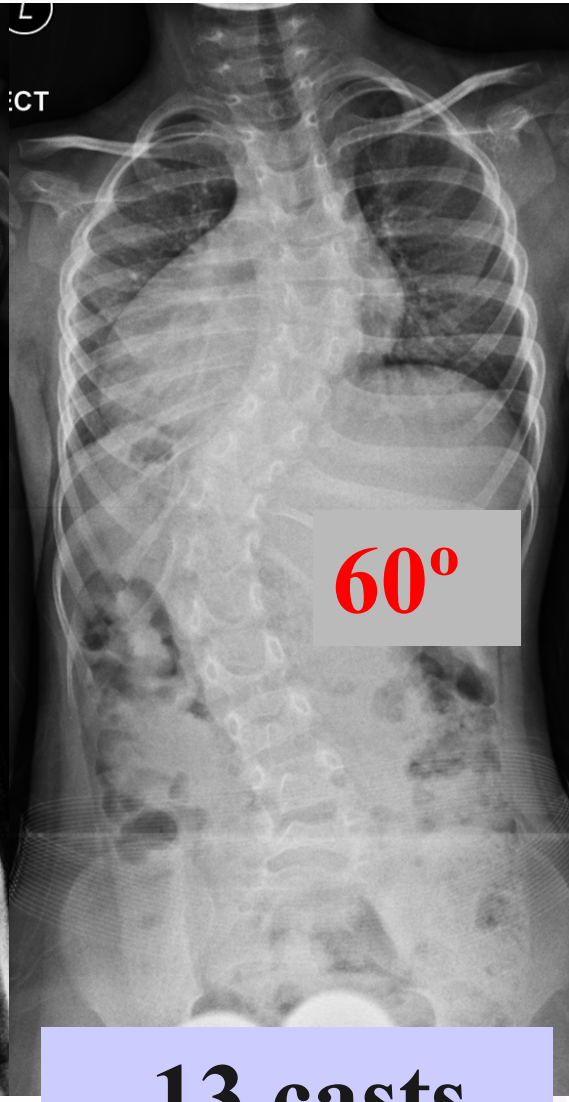
- New technique
- Adolescent growth phase

4 years old 2 ½ years post-casting	8 years old 6 years post-casting	9 years old 25º, restart bracing	11 years old 30º curve

Timeline of a Controlled Curve





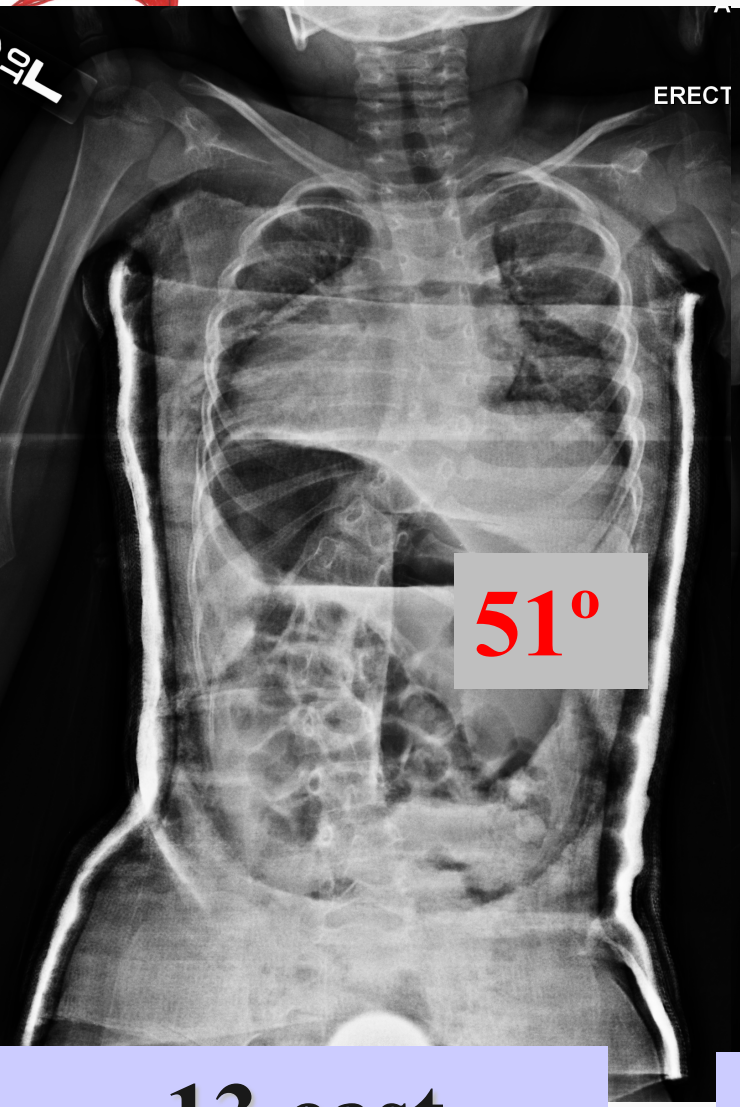


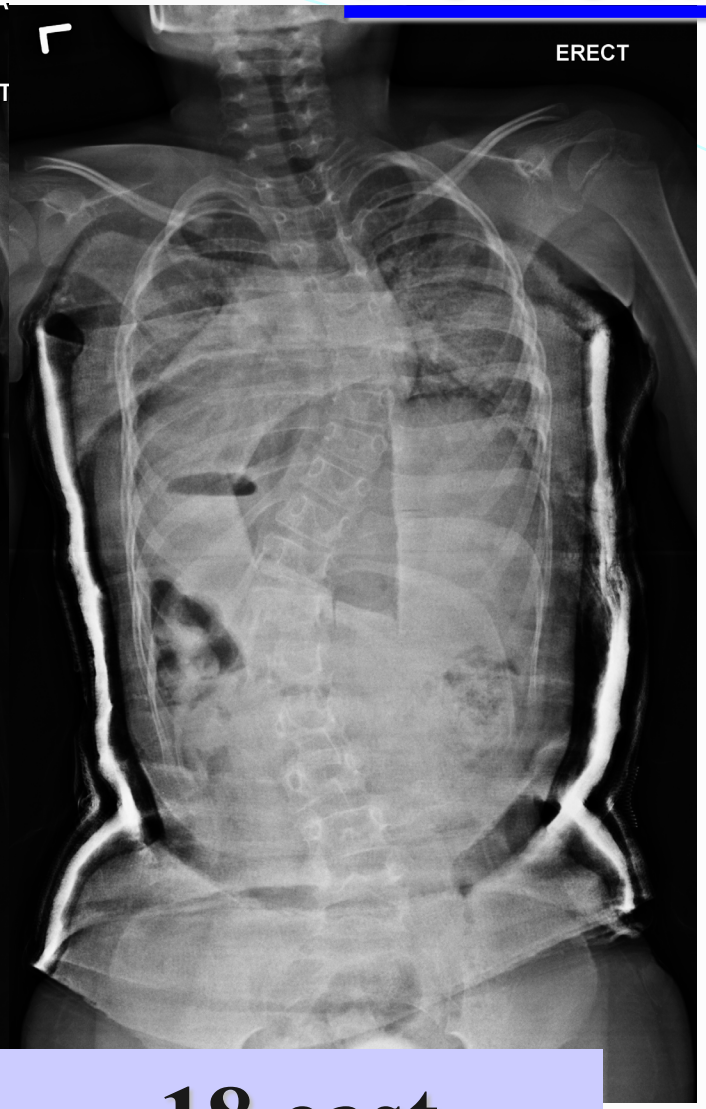
18 month old with deletion

1st cast

13 casts
4 years old

Followup



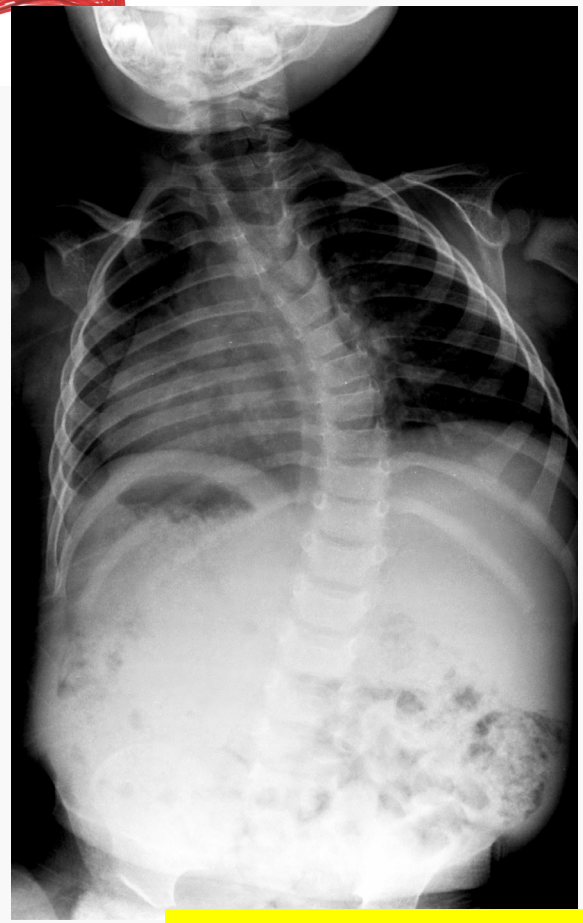


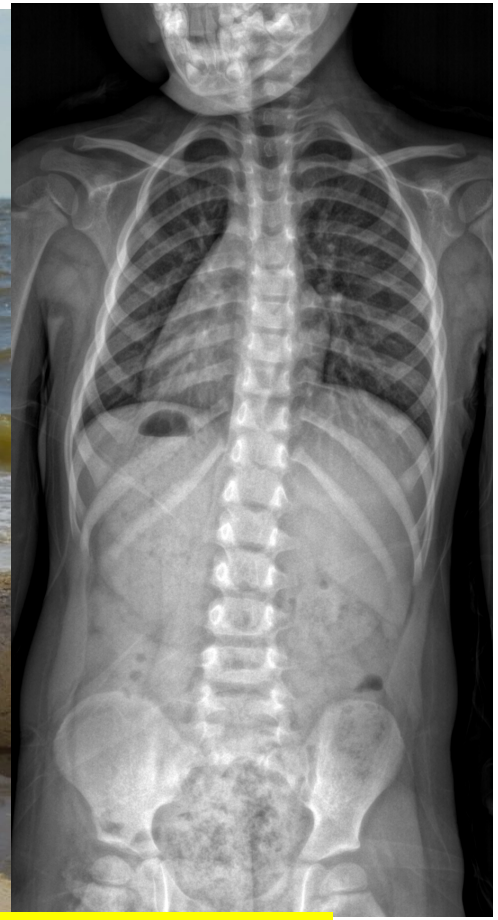
13 cast
4 years old

18 cast
6 years old

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

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

Bracing

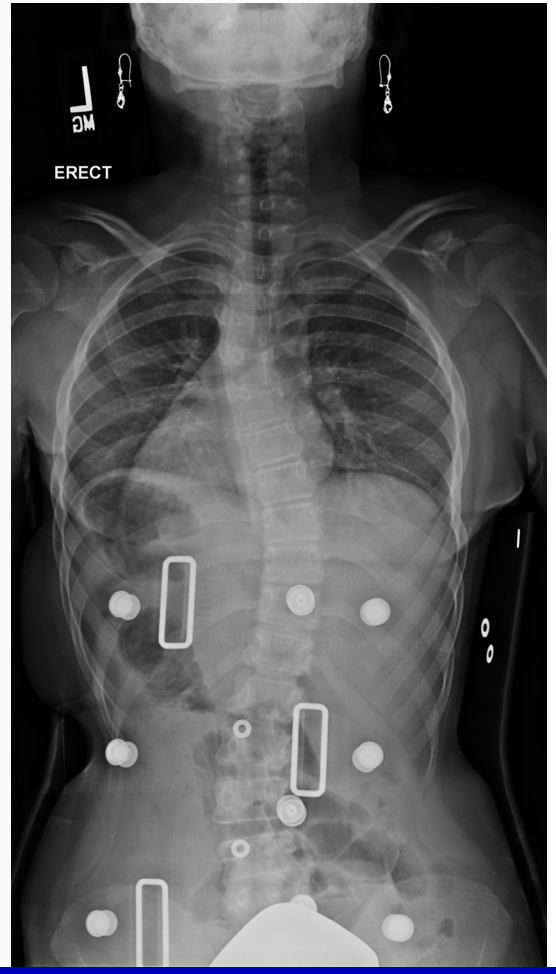
Bracing

- For curves larger than 20º - 25º
- Prevent curve progression
- Conventional wisdom: does not make curves smaller
  - Personal experience and some small studies
- Weight control important for well fitting brace
- My protocol
  - Curves 30º or less – nighttime “bending” brace
    - Maintain curve flexibility
  - Curves over 30º - add daytime brace
    - Correct curve alignment when standing

Bracing

January 2009

32º



January 2009

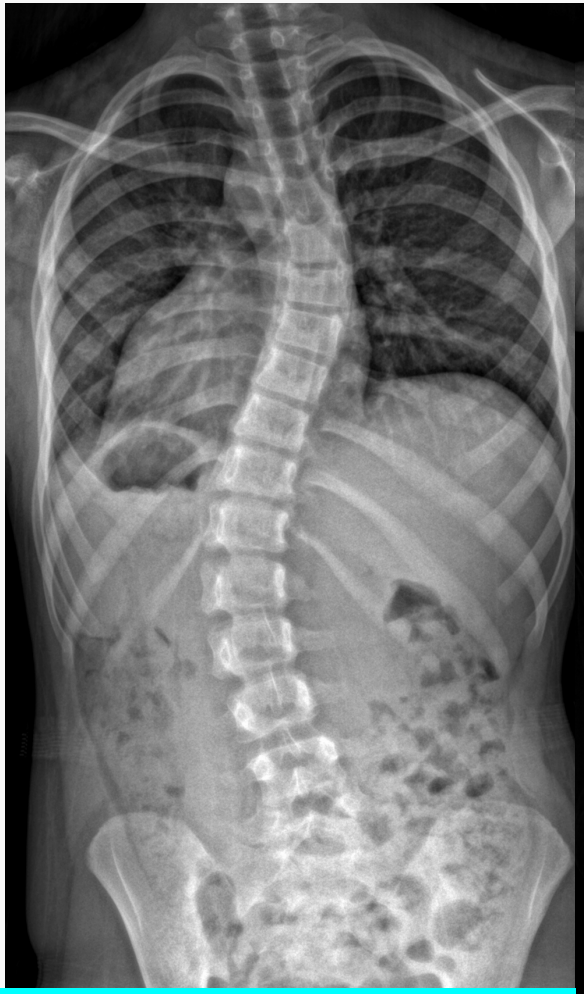
First brace

March 2013

44º

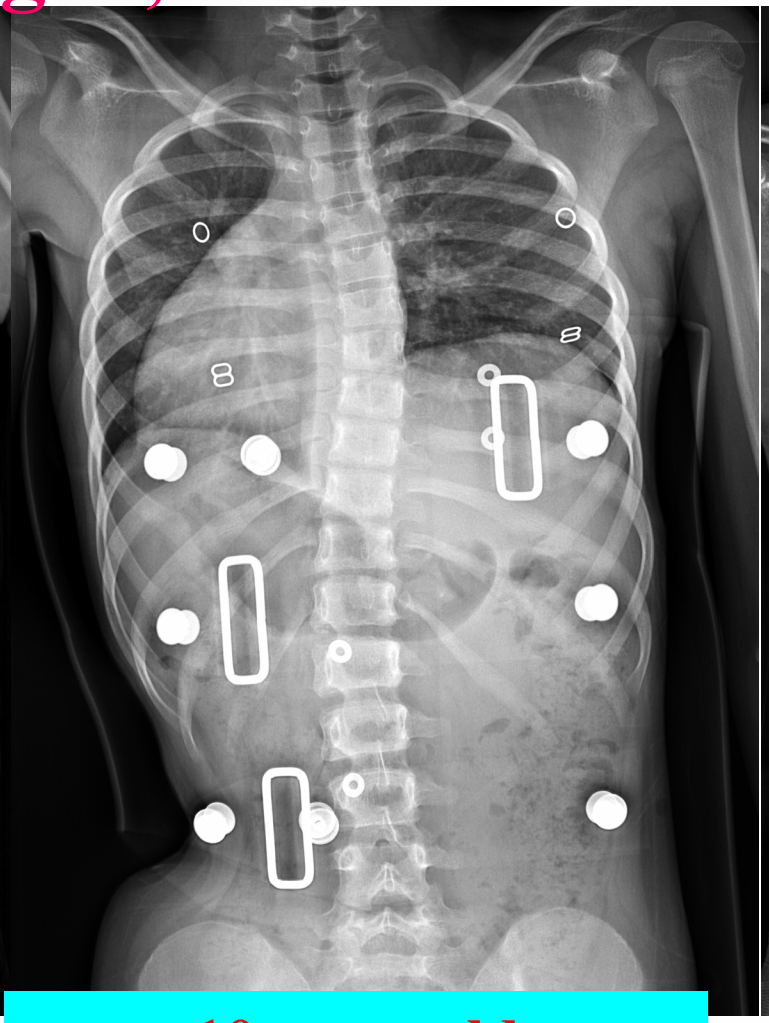
Bracing

10 year old girl, PWS/del

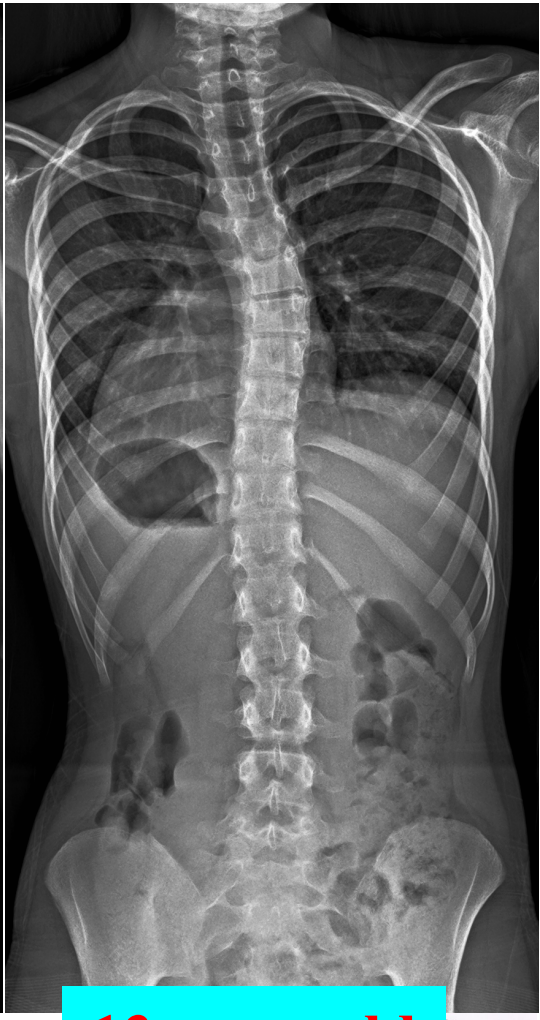


10 years old
37° and 41° curves

10 years old
in TLSO



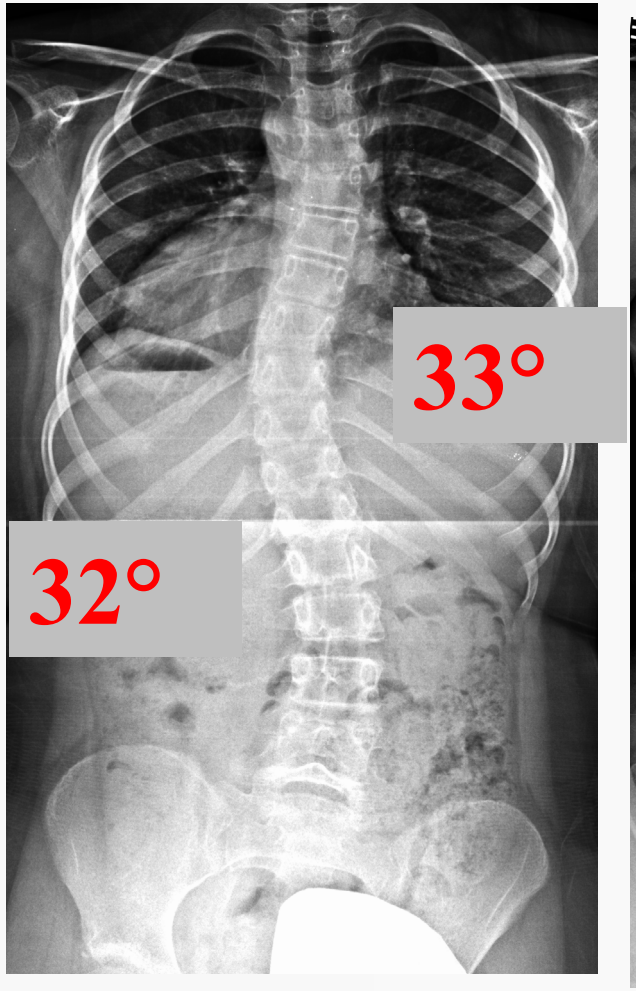
10 years old
in Providence brace



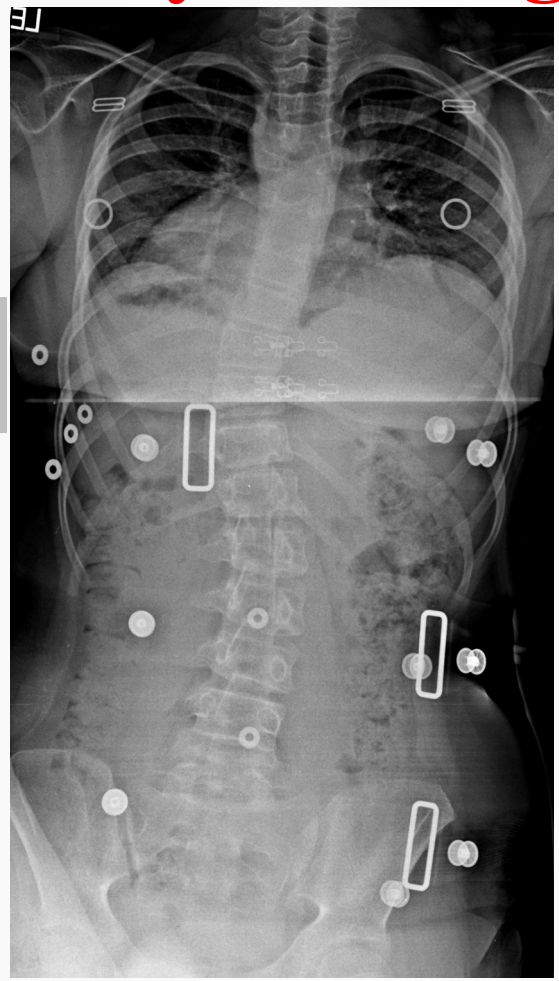
13 years old
25° curve

Bracing

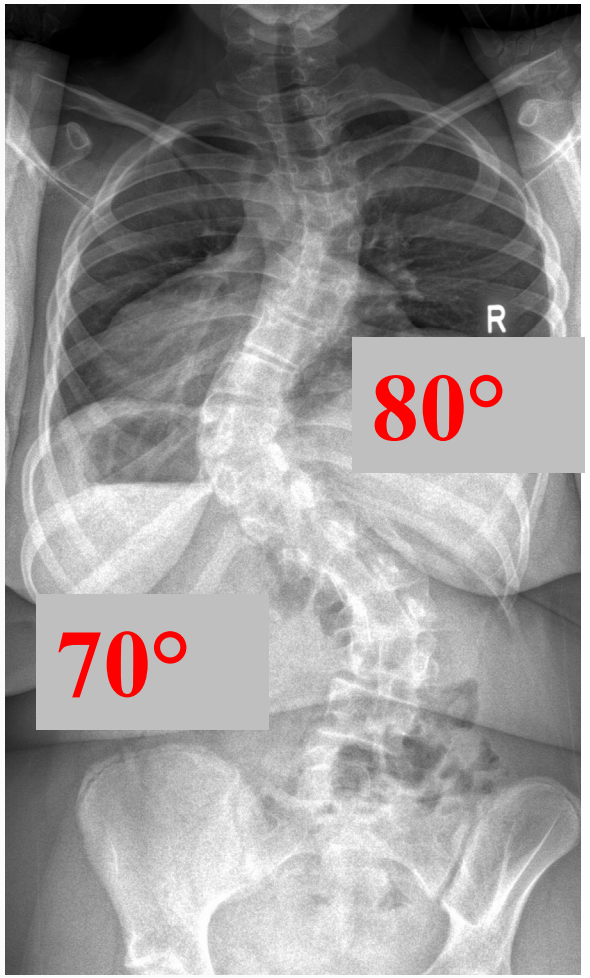
10 year old girl, PWS/UPD



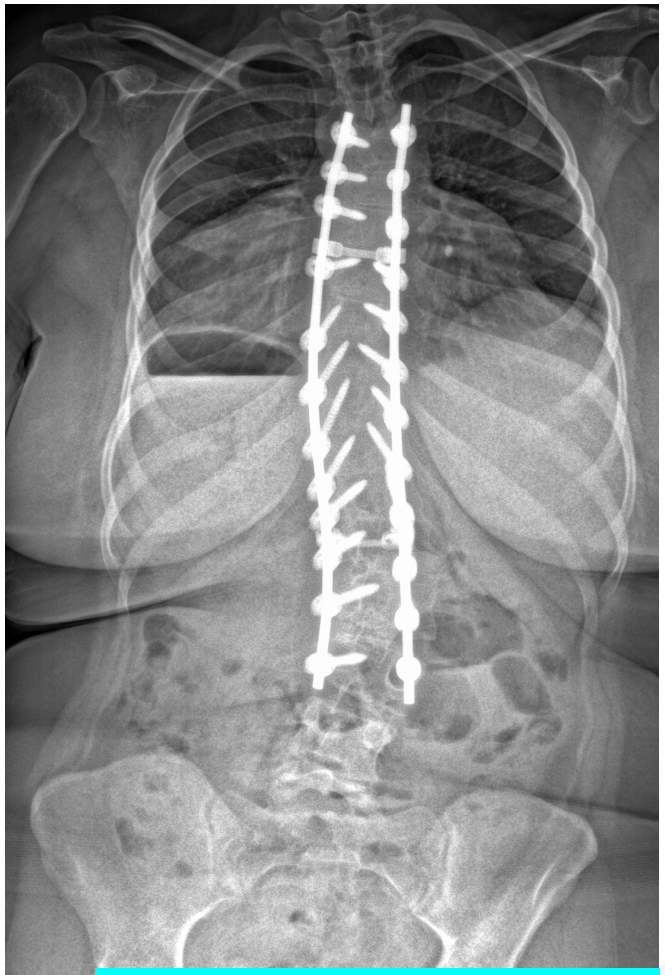
10 years old
33° and 32° curves



12 years old
In brace



15 years old
80° and 70° curves

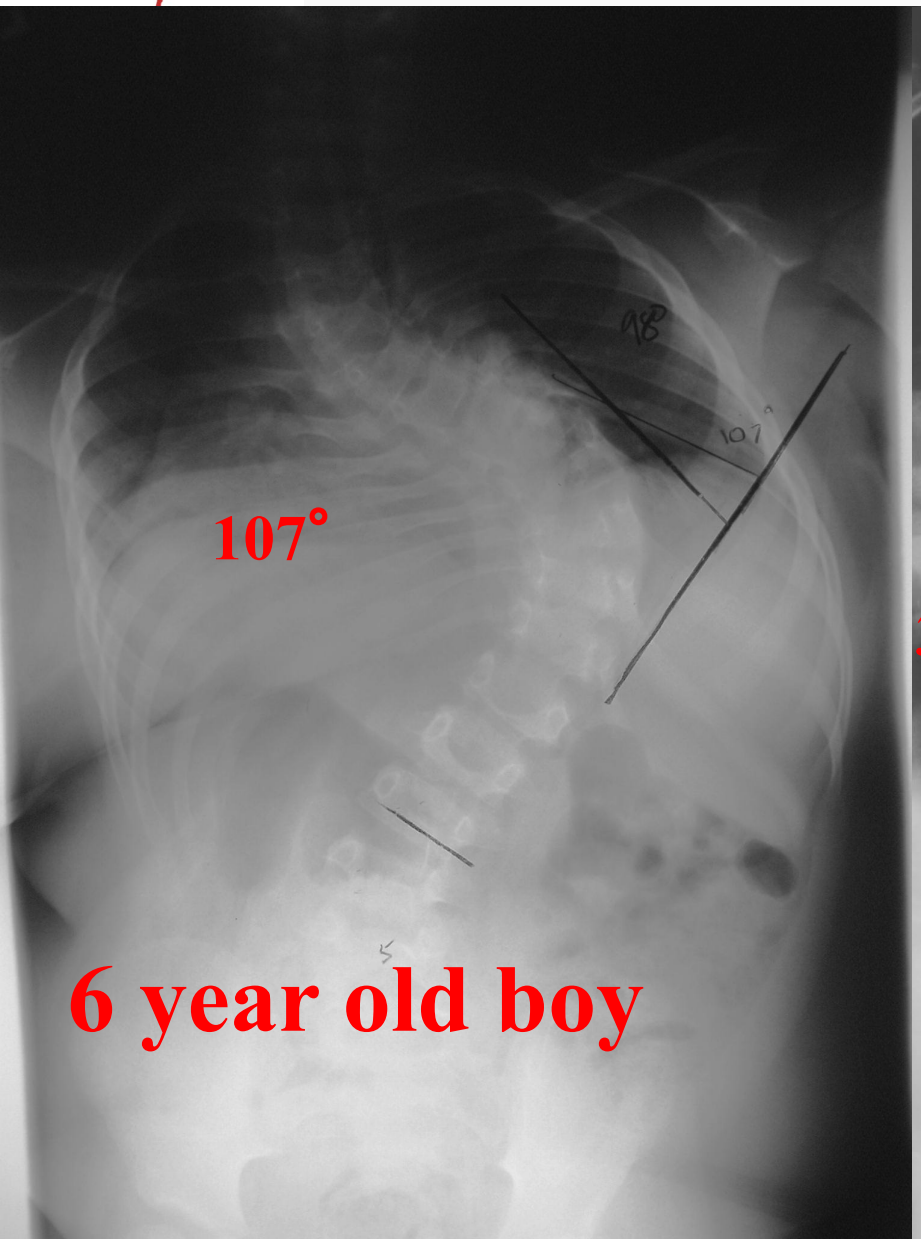


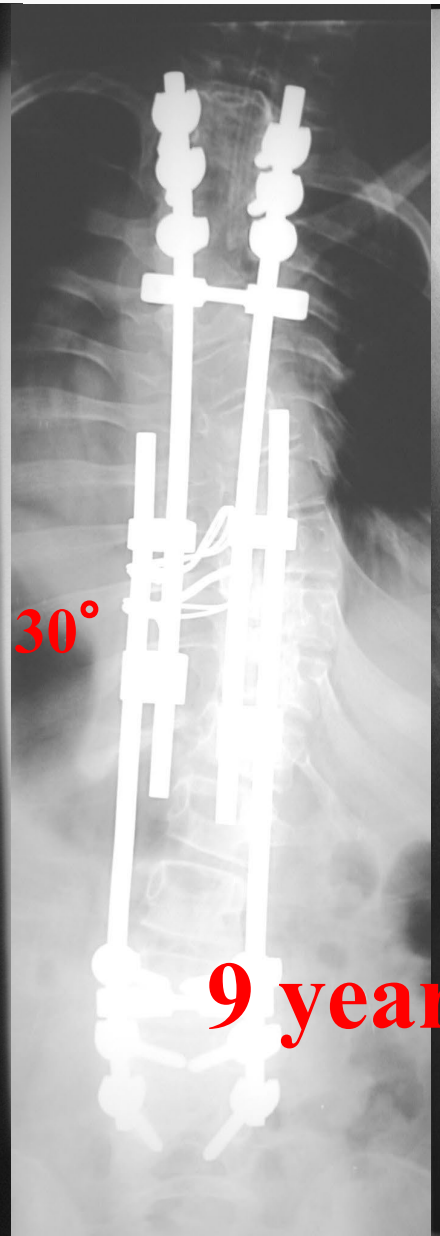
19 years old
4 years after fusion

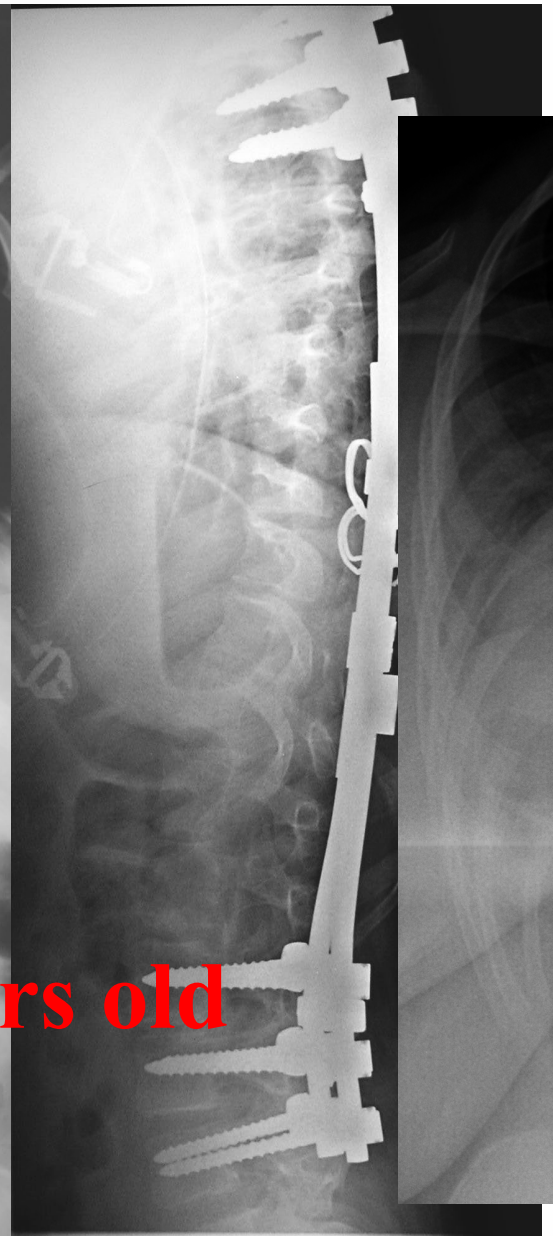
Surgery

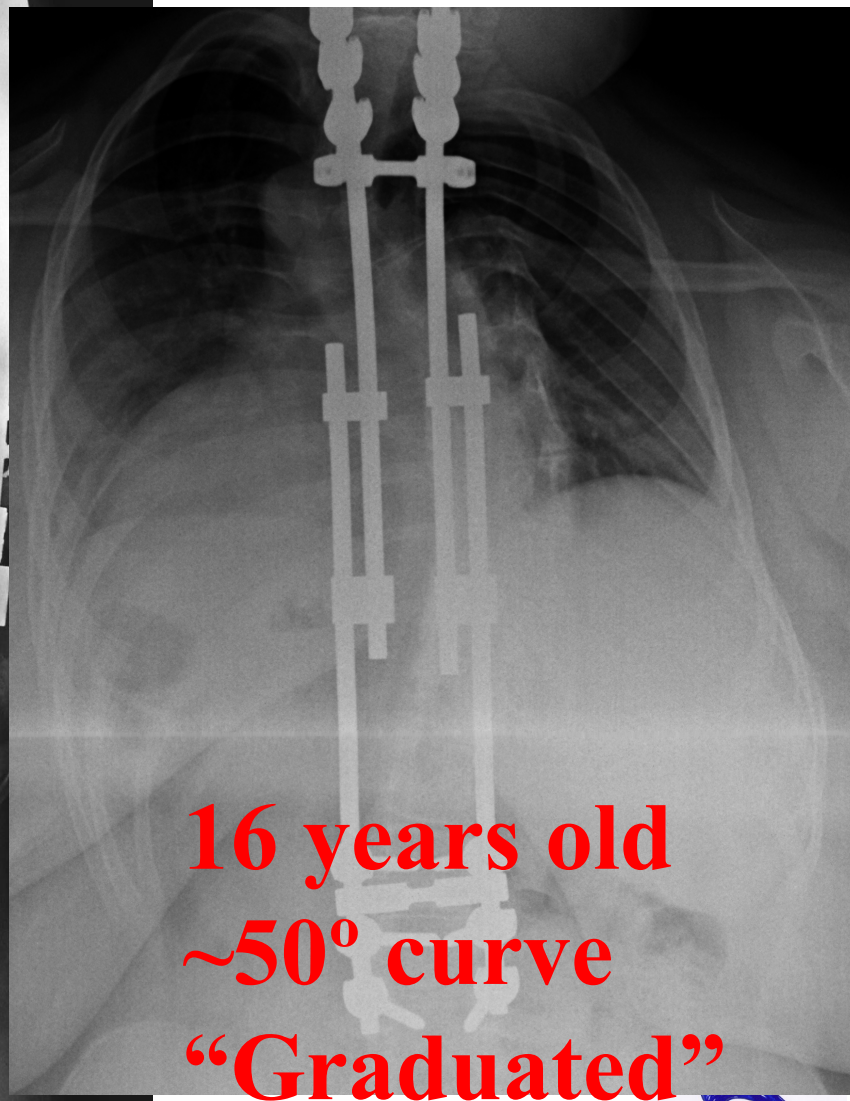
- Indicated for curves over 50º
- Goals
  - Align spine in best position
    - Side to side curve (scoliosis)
    - Front to back alignment (kyphosis/lordosis)
  - Prevent progression
  - Decrease curve size
- Hold in position
  - Rods
  - Hooks, wires, and screws

Non Fusion Spinal Instrumentation

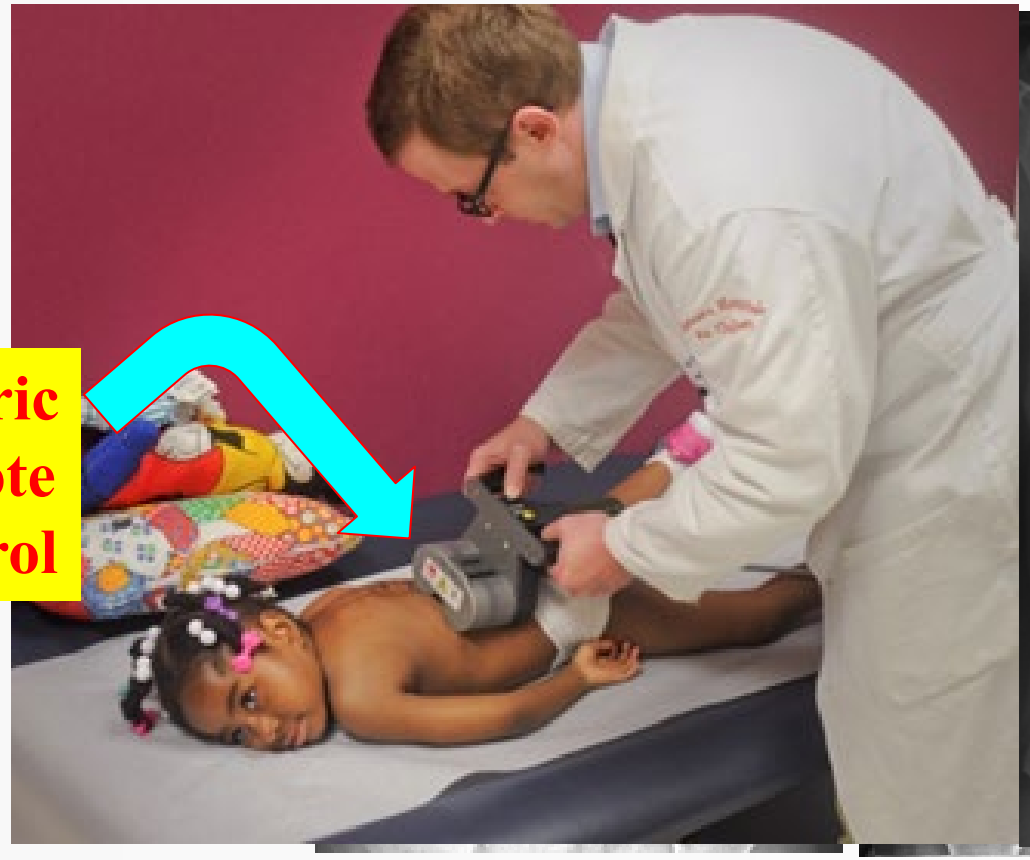


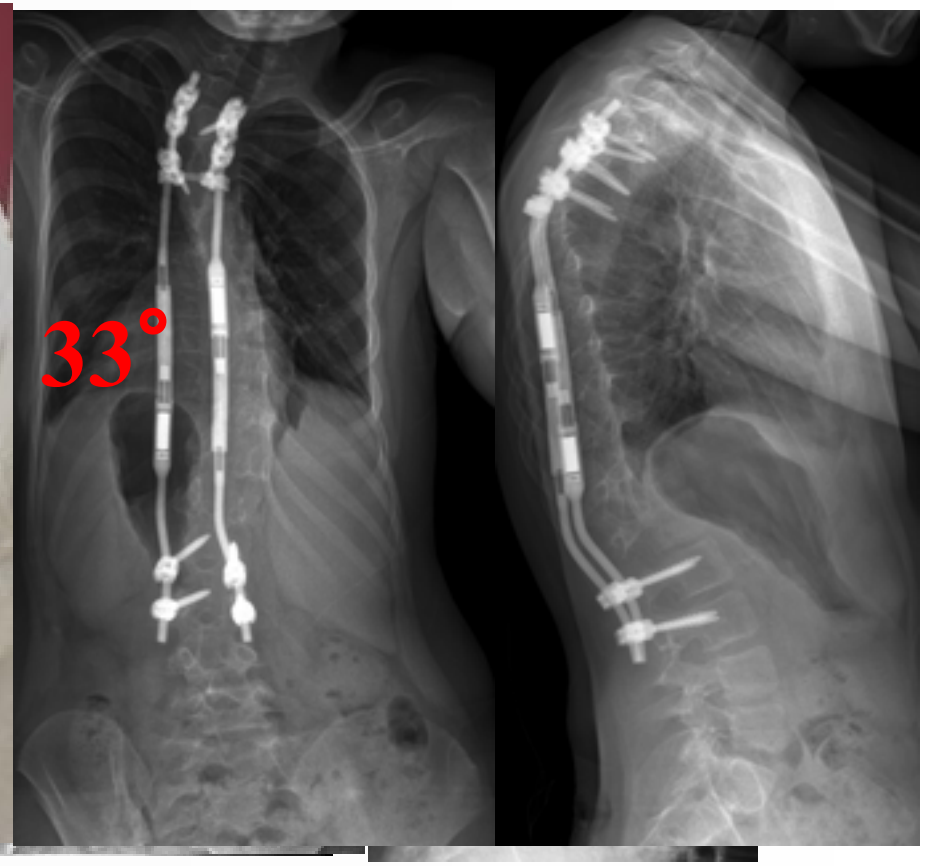






MAGnetic Expansion Control: MAGEC Rods



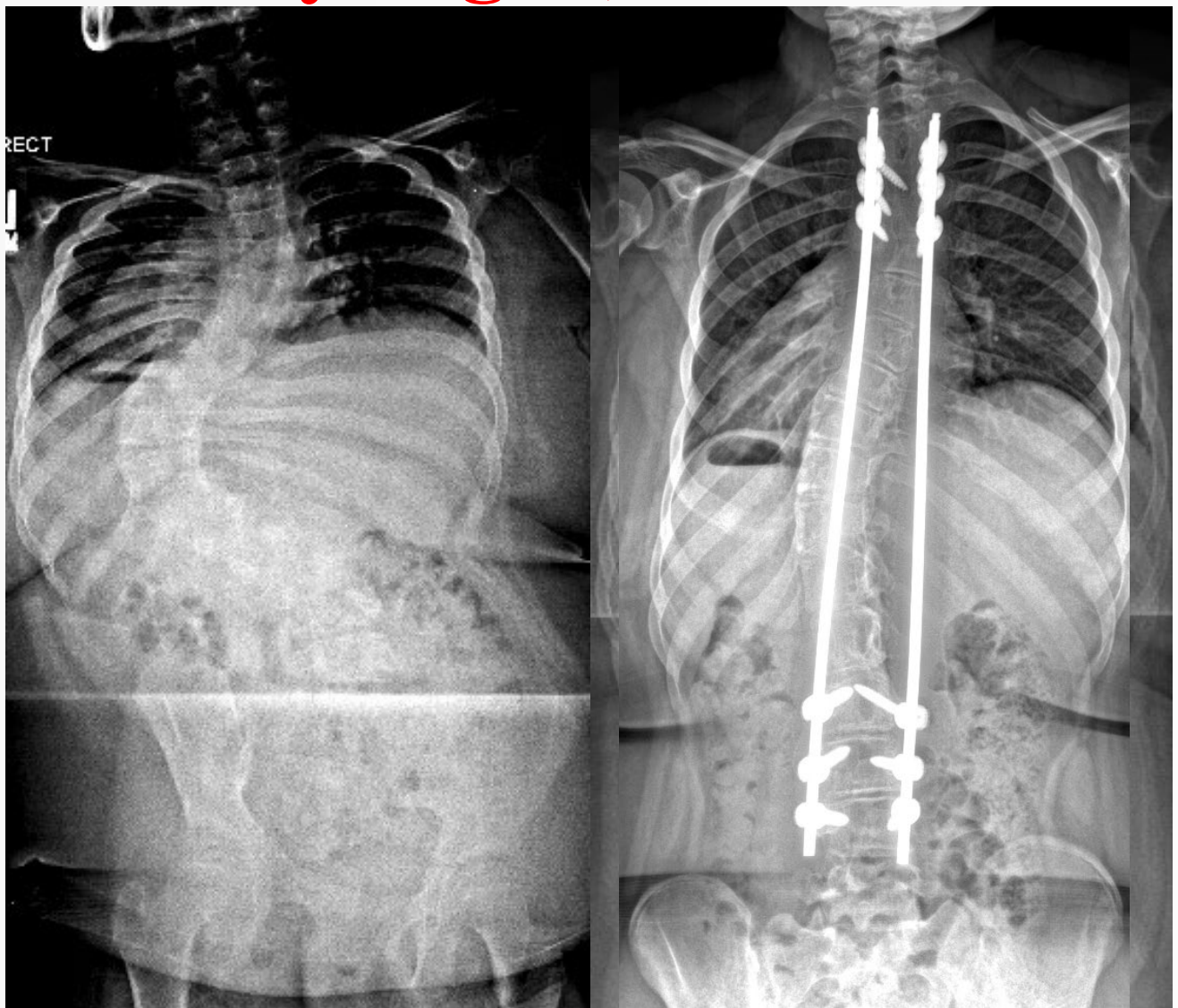


10 year old boy with PWS

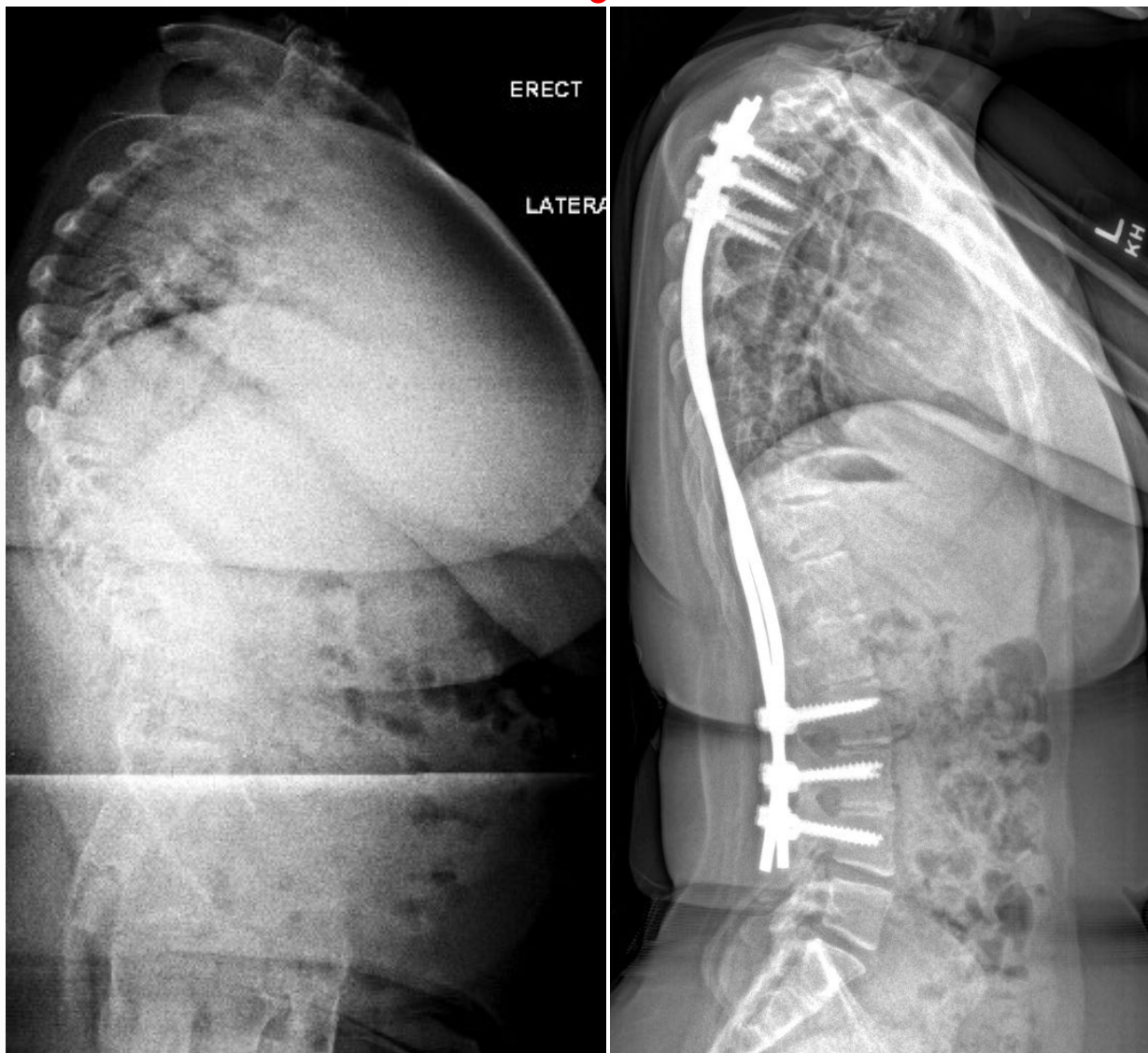
After 2 lengthenings

End-game: *Graduation*

10 y.o. girl, 103°



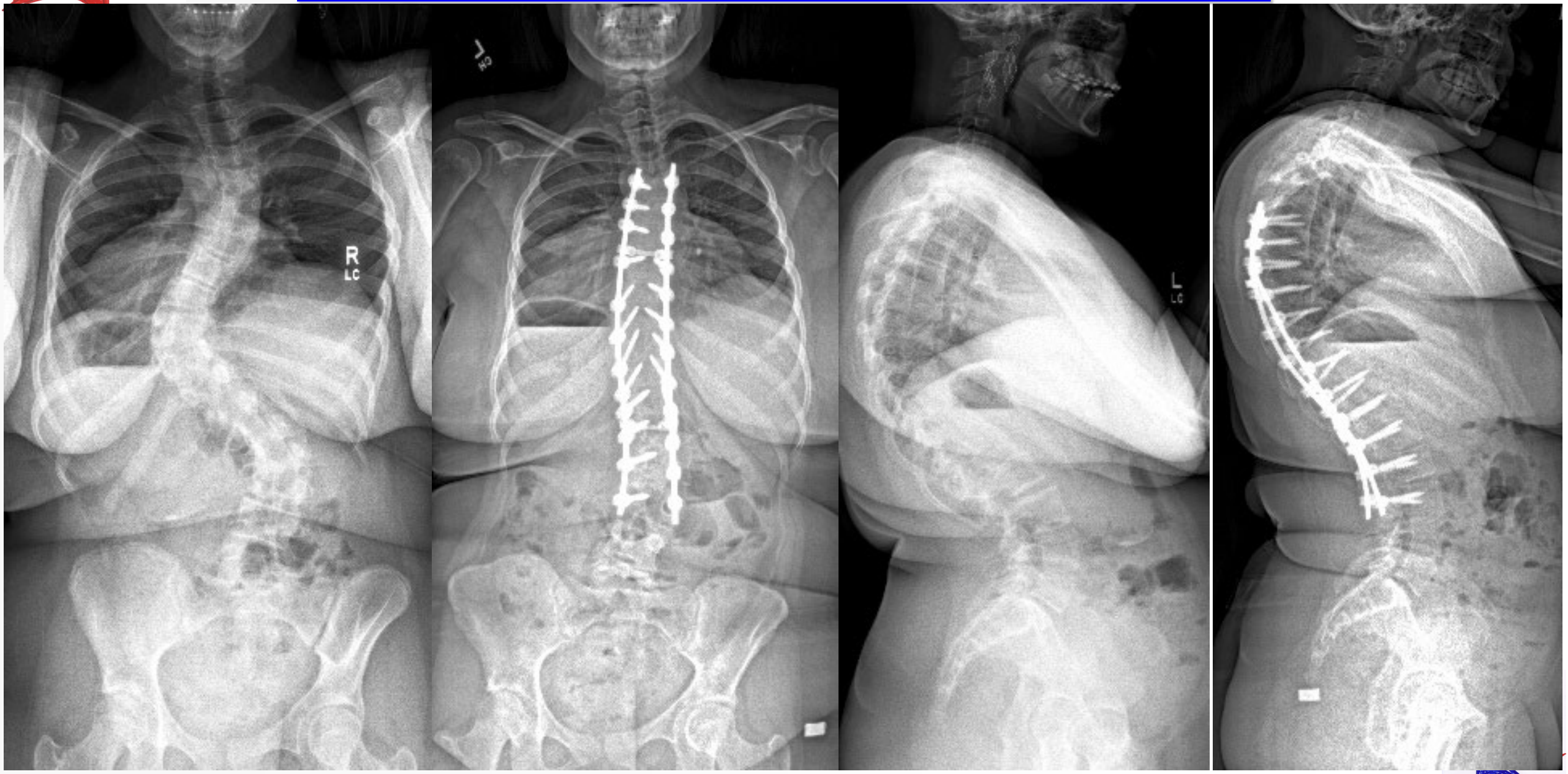
15 y.o.



Spinal Fusion

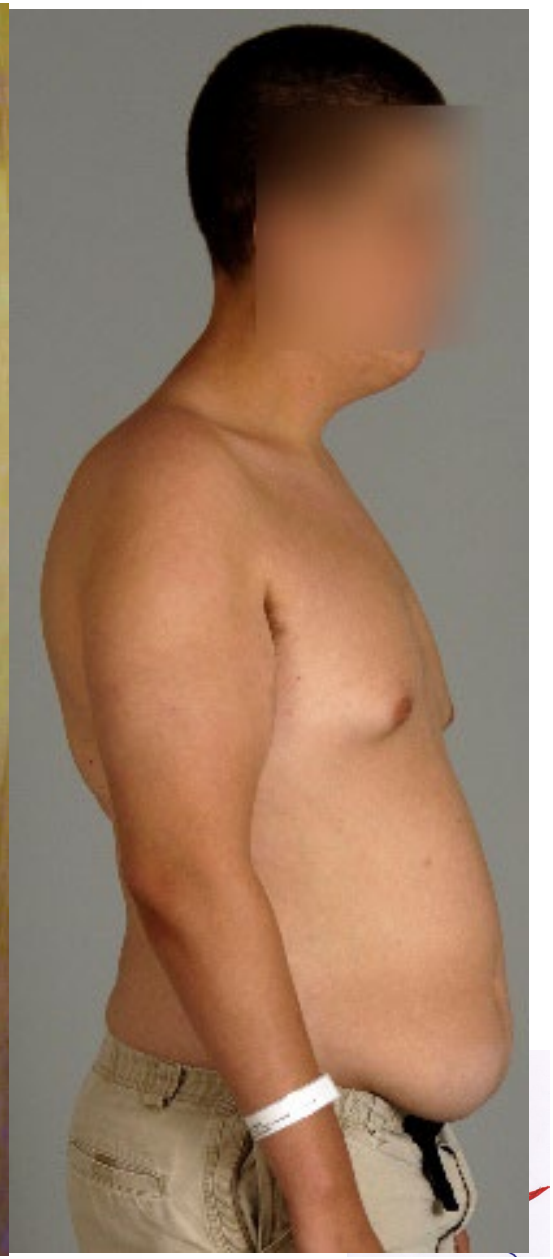
- For curves over 50º at maturity
- Timing of surgery
  - Balance expected maturity with curve size
  - My preference in younger patients:
    delay until the curve is over 50º in brace
- Avoid anterior approach
- Newer pedicle screw instrumentation
  - Better in osteopenic bone

15 y.o. girl with 67º scoliosis

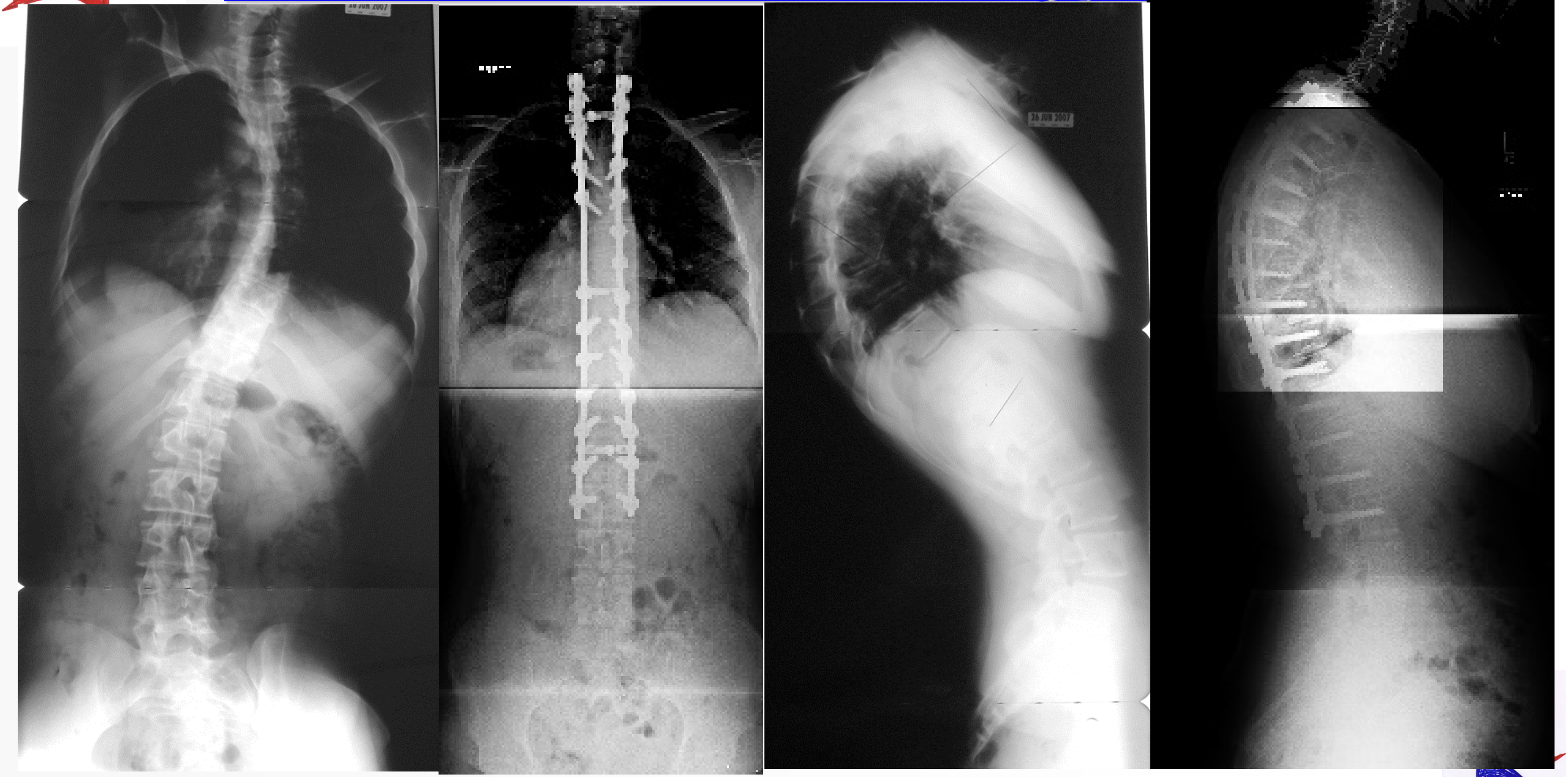


18 y.o. with kyphosis



25º scoliosis and 110º kyphosis



Special Considerations Surgery and PWS

- Skin picking
  - Infections
- GI complications
  - Gastro-motility slows down
  - Very gradual increase in post-op diet
- Profile (posture)
  - Spine alignment from the side
  - Head forward position in people with PWS
  - Hardware (screw) fixation failure

- Hardware too straight
- Spine pulling away from rods up top
  - Proximal junction kyphosis (PJK)
- Spine pulling away from rods below
  - Distal junction kyphosis (DJK)

High Complication Rate of Surgery

- Infections
- Anaesthetic (intra or peri-operative)
- Pulmonary/Respiratory
  - Apnea
- Hardware failure/junctional kyphosis
  - Osteoporosis
- Spinal cord compromise/paralysis
- Need to continuously educate treating surgeons to these special risks

Adults with PWS and Scoliosis

- Deletion type: 80% prevalence
- UPD type: 58% prevalence
- Increased thoracic kyphosis, worsened 3°/decade
- Bone health in adults, average age 31 years
  - 3% vertebral fractures
  - Scoliosis rates same regardless of GH supplementation as children or adults, sex hormone supplementation or hypogonadism, sex, BMI, genotype
- Recommend regular spine screening for adults

Strategy for Orthopaedic Issues in PWS

- Newborn and Infants
  - Full orthopaedic clinical evaluation of spine and hips
  - Plenty of stimulation and active tummy time
  - Physical and occupational therapies
  - No upright sitting until can pull to a sit
    - High chair tilted 60°

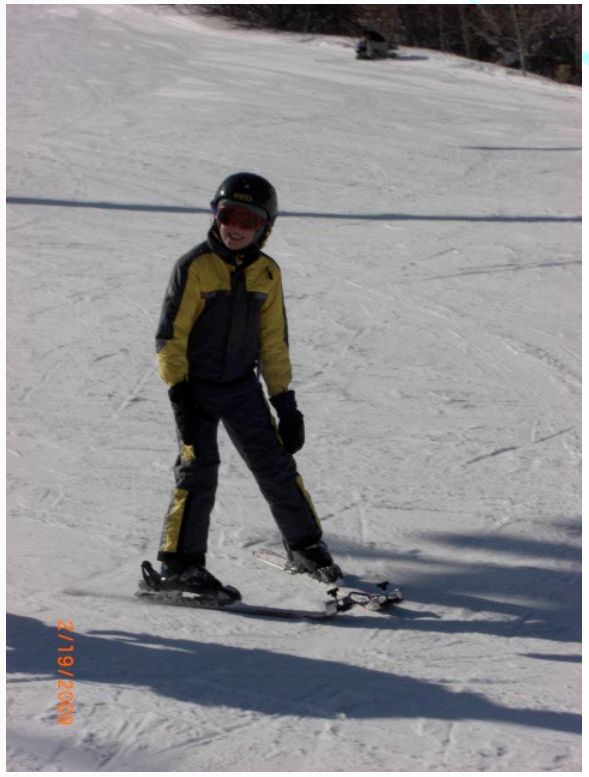
Strategy for Orthopaedic Issues in PWS

- **Sitting Milestone**
  - **Routine sitting spine radiographs**
    - **Curves under 25° - observe with periodic radiographs**
    - **Curves over 25° - serial spine casting**
  - **Routine supine hip radiographs**
    - **If hip dysplasia, yearly x-rays to make sure improving**
    - **If hip subluxation, watch carefully, may need surgery**
  - **AFOs if not walking by 16 to 18 months**
    - **Once walking, evaluate for flat-footedness, possible braces**

Strategy for Orthopaedic Issues in PWS

- **Scoliosis**
  - **Monitor**
    - Routine xrays until 4 years old
    - Resume monitoring at about 8-10 years old clinically
  - **Curves**
    - Casting: start between sitting age and 5 years old
    - Bracing for curves under ~50°
    - NFSI surgery: 5 year old until 10 years old
    - Fusion surgery: 12 years old and older

People with PWS are living longer



We work together for a better quality of life