

# DIASTROPHIC DYSPLASIA

Vernon Tolo, MD  
ICEOS 2018



# DISCLOSURE

- EDITOR EMERITUS, JBJS

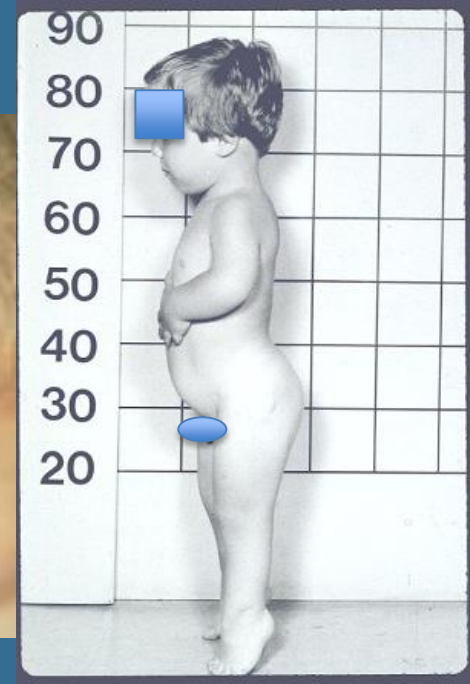


# DIASTROPHIC DYSPLASIA

- CHROMOSOMAL SITE 5q31-q34
- DEFECT IN DD  
SULFATE TRANSPORTASE
- DIFFERENT GENOTYPES
- RARE, EXCEPT IN FINLAND

- CLINICAL FEATURES

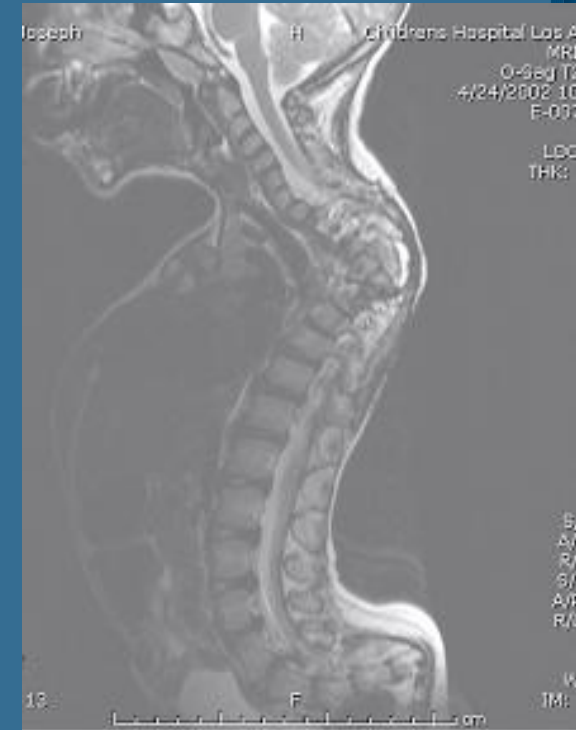
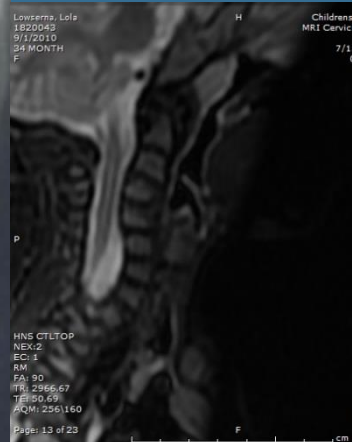
- SHORT, STIFF LIMBS
- CLUBFEET
- CAULIIFLOWER EAR
- HITCHHIKER THUMB
- VERY SHORT



# DIASTROPHIC DYSPLASIA

## — SPINAL ABNORMALITIES

- CERVICAL KYPHOSIS
- SEVERE KYPHOSCOLIOSIS
- LUMBAR LORDOSIS AND STENOSIS



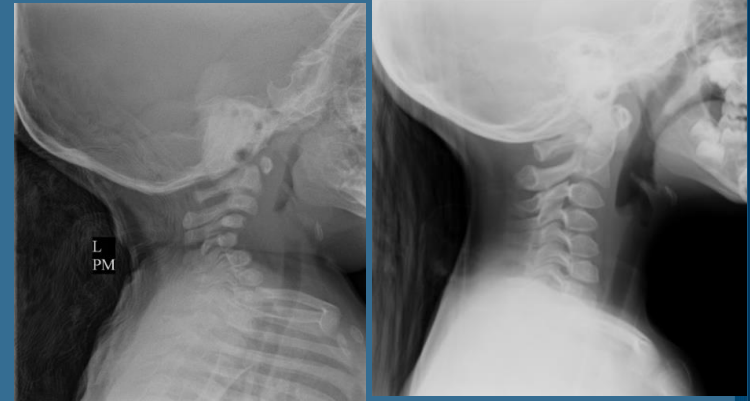
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Southern California

Children's  
Hospital  
LOS ANGELES  
CHILDREN'S  
ORTHOPAEDIC CENTER



# DIASTROPHIC DYSPLASIA

- CERVICAL SPINE KYPHOSIS
  - PRESENT AT BIRTH
  - AFFECTS C-3 TO C-5
  - MOST RESOLVE WITHOUT TREATMENT
    - USUALLY BY AGE 6 YEARS
  - SMALL NUMBER WITH PROGRESSIVE KYPHOSIS
    - MAY DEVELOP MYELOPATHY



# DIASTROPHIC DYSPLASIA

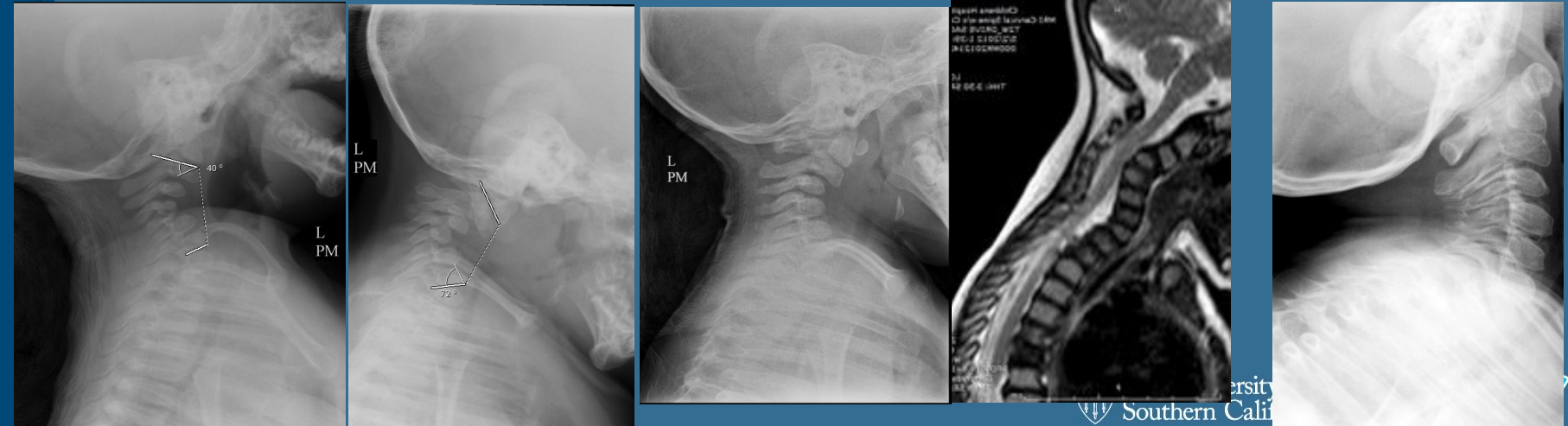
- CERVICAL KYPHOSIS UNTREATED
- NEUROLOGIC NORMAL
- DEVELOPMENTAL MILESTONES NORMAL FOR DD

NEUTRAL

FLEXION

2 YEARS

6 YEARS

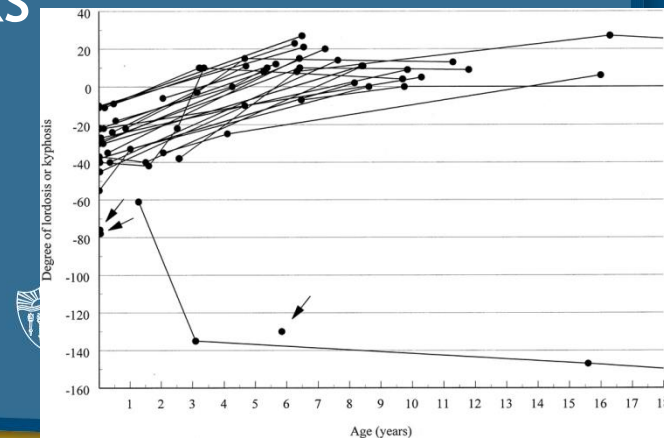


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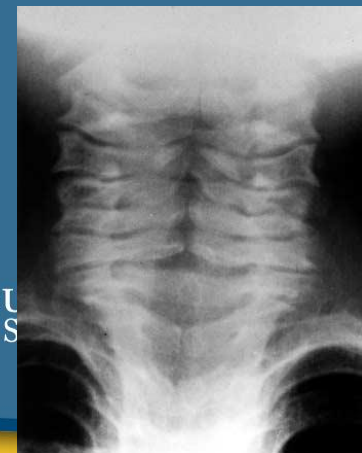
# DIASTROPHIC DYSPLASIA

- NATURAL H/O CERVICAL KYPHOSIS (*Remes, et al., 1999*)
  - 120 PATIENTS IN FINLAND
    - NEWBORN TO 63 YEARS
  - 29 WITH KYPHOSIS OVERALL
    - 4/120 WITH SEVERE KYPHOSIS
  - 24/25 WITH XRAYS BY 18 MONTHS WITH KYPHOSIS
    - RESOLVED IN 24 BY MEAN 7.1 YEARS
    - KYPHOSIS < 60° SHOULD RESOLVE



# DIASTROPHIC DYSPLASIA

- CERVICAL SPINE XRAY FINDINGS *(Remes, et al. 2002)*
  - 122 PATIENTS
  - AVERAGE LORDOSIS 17 DEGREES
  - FLAT VERTEBRAL BODIES
  - SAGITTAL CANAL NARROWED WITH AGE
    - DECLINE BEGINS AT AGE 8 YEARS
  - 79% WITH SPINA BIFIDA OCCULTA



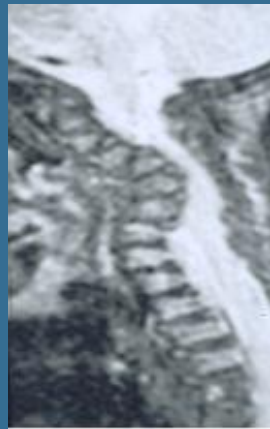
# DIASTROPHIC DYSPLASIA

- C-SPINE MRI FINDINGS *(Remes, et al. 2000)*
  - 90 PATIENTS AGED 3 MONTHS TO 50 YEARS
    - VERY WIDE FORAMEN MAGNUM
    - NARROWED SPINAL CANAL BELOW C-3
    - ABNORMAL DISCS IN ALL, BEGINNING AT EARLY AGE
      - CERVICAL SPINE OFTEN STIFF EVEN IN YOUNG
    - EARLY DEGENERATIVE DISC CHANGES
    - RAPID PROGRESSION OF DISC CHANGES
    - CORD COMPRESSION IN 2 WITH SEVERE KYPHOSIS

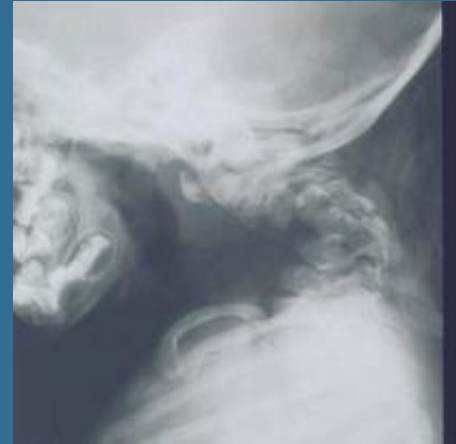
# DIASTROPHIC DYSPLASIA

## – CERVICAL KYPHOSIS EXAMPLE

- DID NOT RESOLVE AS EXPECTED



→posterior fusion



2-level  
vertebrectomy +  
ant/post fusion  
with rib strut  
→  
neuro function ok



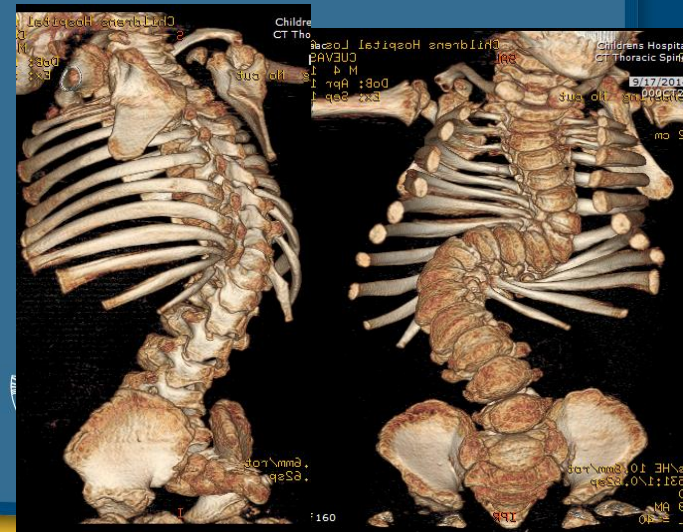
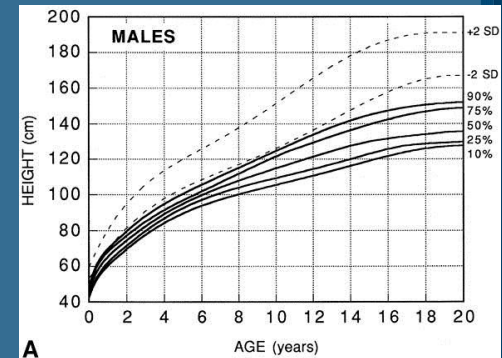


# DIASTROPHIC DYSPLASIA

## – KYPHOSCOLIOSIS

- MOST SPINE GROWTH BY AGE 8
- RIGID, MID-THORACIC CURVES
  - SEVERE KYPHOSIS ALSO
- IF SEVERE, ONSET < 4 YRS

2 YEAR OLD





# DIASTROPHIC DYSPLASIA

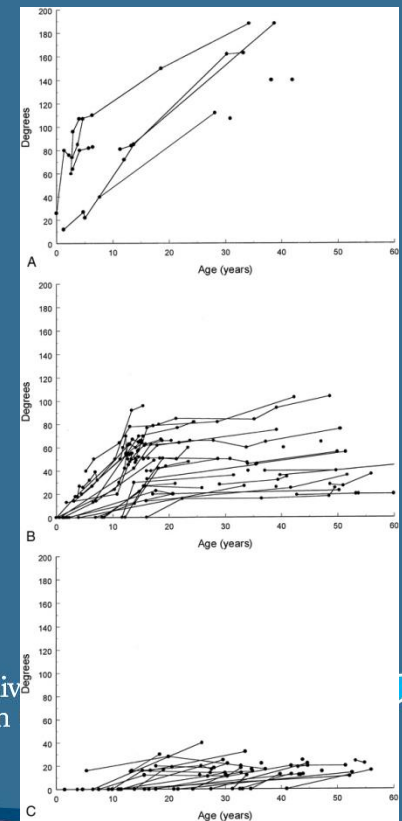
- SPINAL DEFORMITY *(Tolo and Kopits, 1979)*
  - 46 PATIENTS
    - AGES 1 TO 52 YEARS
  - 71% SCOLIOSIS, 50% KYPHOSIS, 41% BOTH
    - 17% WITH NO SPINAL DEFORMITY
  - IF SEVERE, PRESENT BY 4 YEARS
    - 40% WITH SEVERE THORACIC CURVE
    - APICAL VERTEBRA WEDGING
  - OF 7 WITH SURGERY, 4 WITH NEURO DEFICIT
    - ONE PARAPLEGIA, REST RECOVERED
    - MINIMAL CORRECTION
    - ANT/POST SURGERY RECOMMENDED



# DIASTROPHIC DYSPLASIA

- **SCOLIOSIS CLASSIFICATION** *(Remes, et al., 2001)*

- **98 PATIENTS**
- **88% WITH SCOLIOSIS**
- **3 SUBTYPES**
- **EARLY ONSET PROGRESSIVE (11)**
  - MEAN SCOLIOSIS  $134^{\circ}$ , KYPHOSIS  $97^{\circ}$
- **IDIOPATHIC-LIKE (41)**
  - MEAN SCOLIOSIS  $49^{\circ}$ , KYPHOSIS  $29^{\circ}$
- **MILD NON-PROGRESSIVE (33)**
  - MEAN SCOLIOSIS  $19^{\circ}$ , KYPHOSIS  $25^{\circ}$



# DIASTROPHIC DYSPLASIA

- **SCOLIOSIS SURGERY** *(Matsuyama, Winter, Lonstein, 1999)*
  - **21 PATIENTS OVER 42 YEARS**
    - UP TO 1996
  - **17 WITH DOUBLE CURVE**
    - MEAN 79° UPPER, 97° LOWER
    - MEAN KYPHOSIS 101°
  - **ALL PSF, 17 WITH ASF TOO**
    - 2 WITH PEDICLE SCREWS, REST HOOKS AND RODS
    - MOST WITH BRACE POST-OP
  - **CORRECTION AT 2 YEARS F/U**
    - FOR SCOLIOSIS 1°
    - FOR KYPHOSIS 7°



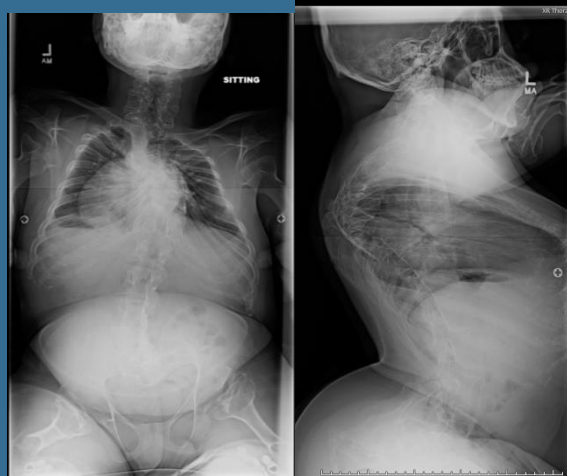
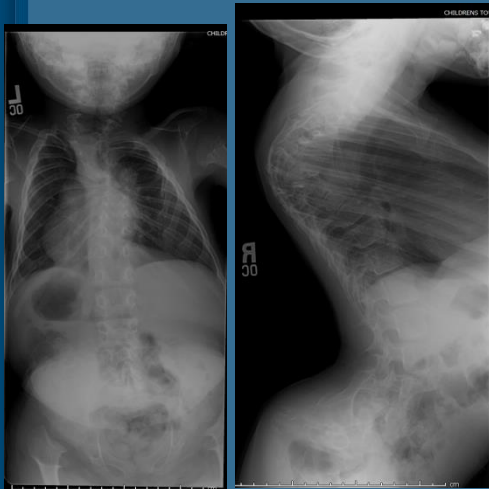
# DIASTROPHIC DYSPLASIA

- **SCOLIOSIS TREATMENT** *(Jalenko, et al., 2009)*
  - **OVER 45 YEARS, 18 OF 180 HAD RX**
  - **8 WITH BRACE TREATMENT**
    - NOT EFFECTIVE
  - **12 WITH SURGERY**
    - 4 WITH SOME PEDICLE SCREWS, REST HOOKS
    - PSF CORRECTION 13%
    - ANT/POST FUSION CORRECTION 40%
    - MEAN OVERALL CORRECTION 24%
    - 42% COMPLICATION RATE



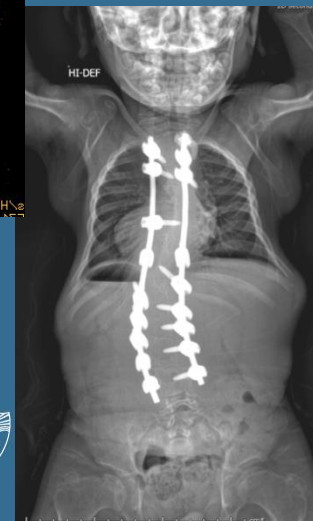
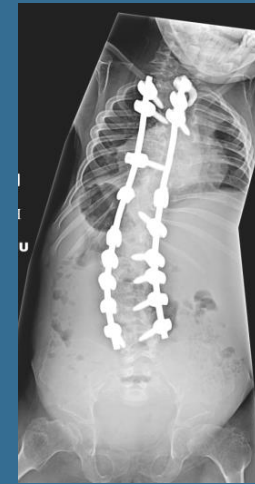
# DIASTROPHIC DYSPLASIA

- SCOLIOSIS UNTREATED MAY BE OK  
NEUROLOGIC EXAM INTACT
- HOUSEHOLD AMBULATOR (limited by feet and knees)
- HYPERLORDOSIS IF SEVERE KYPHOSIS
- AGE 5
- AGE 21



# DIASTROPHIC DYSPLASIA

- NO CURVE AT 7 MONTHS
- LARGE CURVE AGE 3
- PSF AGE 4 ½
- PJK 2 YEARS POST-OP





# DIASTROPHIC DYSPLASIA

- SCOLIOSIS SURGERY EXAMPLE
  - BRACE AGE 3 TO 8
    - CONTINUED PROGRESSION
  - PSF AT AGE 8
    - MAINTAIN LUMBAR LORDOSIS





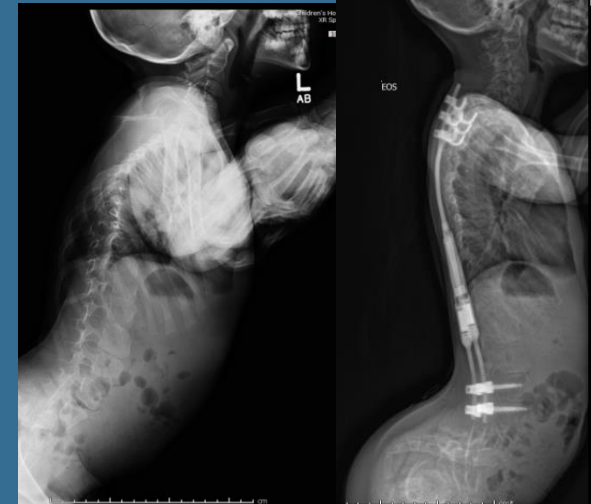
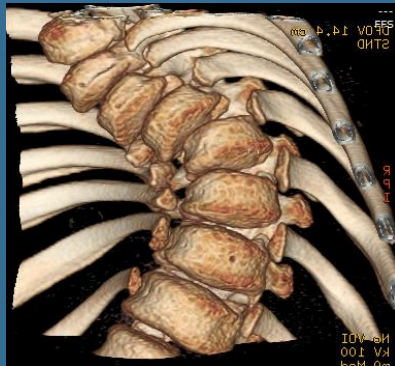
# DIASTROPHIC DYSPLASIA

- “IDEAL” TREATMENT OF EARLY ONSET OF SCOLIOSIS  
– FROM 5 MONTHS TO 32 MONTHS to 46 MONTHS



# DIASTROPHIC DYSPLASIA

- “IDEAL” EARLY ONSET TREATMENT
  - EDF CAST AGE 3-4
  - GROWING RODS AGE 5-8
  - *(patient of Lindsay Andras, MD)*



# DIASTROPHIC DYSPLASIA

## — LUMBAR SPINE LORDOSIS

- PARTLY FROM HIPS, PARTLY FROM SPINE
- DO NOT OVERCORRECT WITH SURGERY

## — LUMBAR STENOSIS

- ONLY IN LOW LUMBAR AREA
- SPINAL CANAL LEAST AT L5
  - 40% LESS THAN L2/L3
- LAMINECTOMY RARE



# DIASTROPHIC DYSPLASIA POINTS

- 1) CERVICAL KYPHOSIS COMMON AT BIRTH
  - MOST RESOLVE WITHOUT TREATMENT BY AGE 6
  - SURGERY ONLY IF PERSISTENT AND MYELOPATHY
- 2) SEVERE KYPHOSCOLIOSIS STARTS BY AGE 2
  - EARLY EDF CAST AND GROWING RODS
  - FUSION AFTER 8 SINCE LITTLE GROWTH AFTER
- 3) LATER ONSET SCOLIOSIS LESS OF A PROBLEM
- 4) HYPERLORDOSIS FROM SPINE AND HIPS
  - HIP FLEXION CONTRACTURES COMMON



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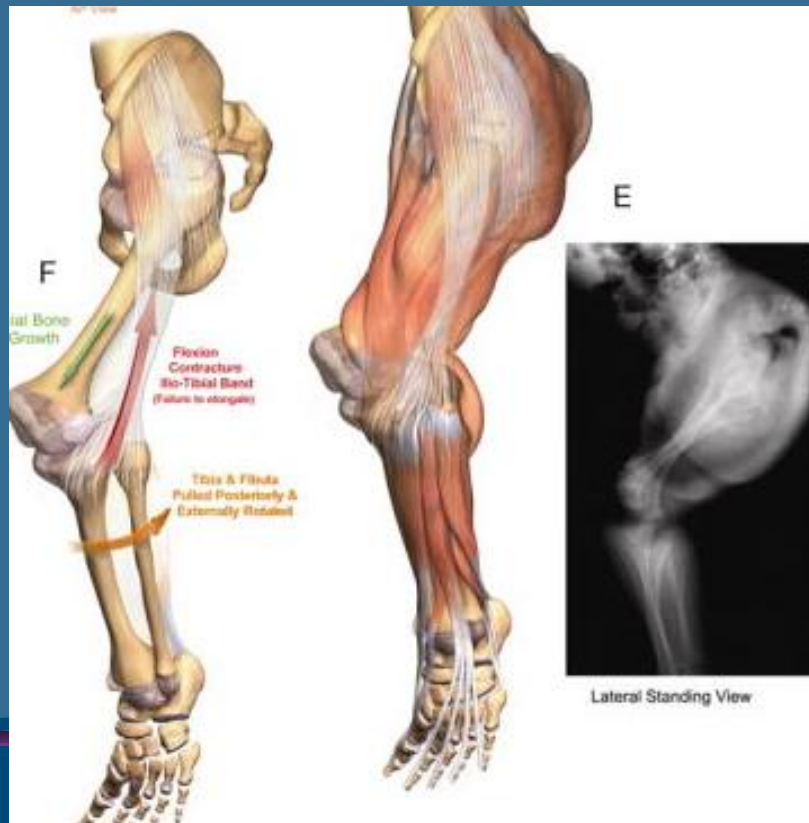
# DIASTROPHIC DYSPLASIA

- PREMATURE HIP DJD
  - **SUBLUXATION EARLY**
  - **VDRO +/- PELVIC OSTEOTOMY**
    - **CHANGE NATURAL HISTORY?**
      - » UNKNOWN
  - **THR AS ADULT**



# DIASTROPHIC DYSPLASIA

- HIP AND KNEE DISORDERS LINKED
- PATELLAR DISLOCATION COMMON
  - EXTERNAL TIBIAL TORSION COMMON





# DIASTROPHIC DYSPLASIA

## CLUBFEET

- RIGID
- NEED TO FULLY CORRECT AT SURGERY
- REMOVE CARTILAGE?
- SPLIT SYNDESMOSIS?
- BRACE





# THANKS!



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# LOLA LOWSERNA..1820043

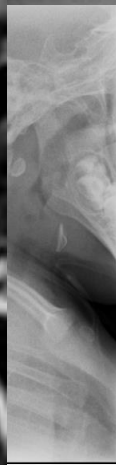
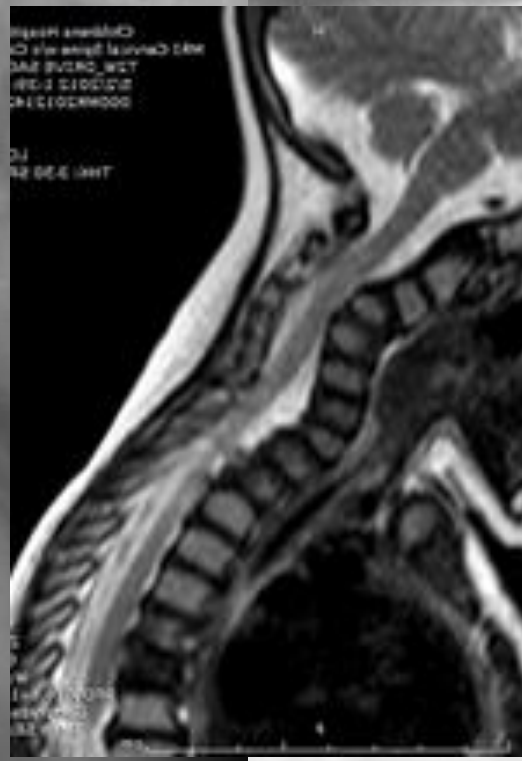
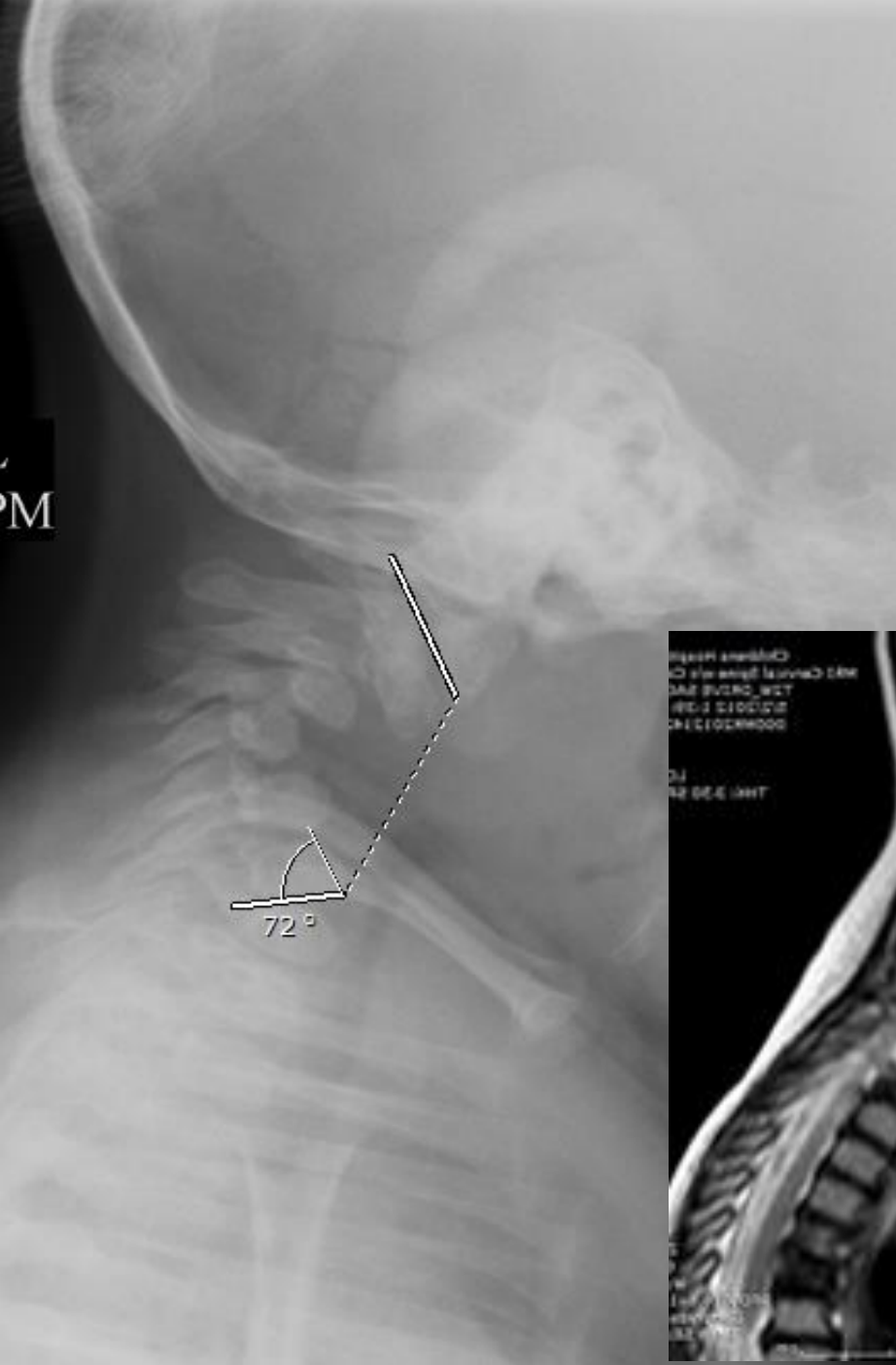
## DIASTROPHIC DYSP WITH RESOLVED KYPHOSIS

- 6 months



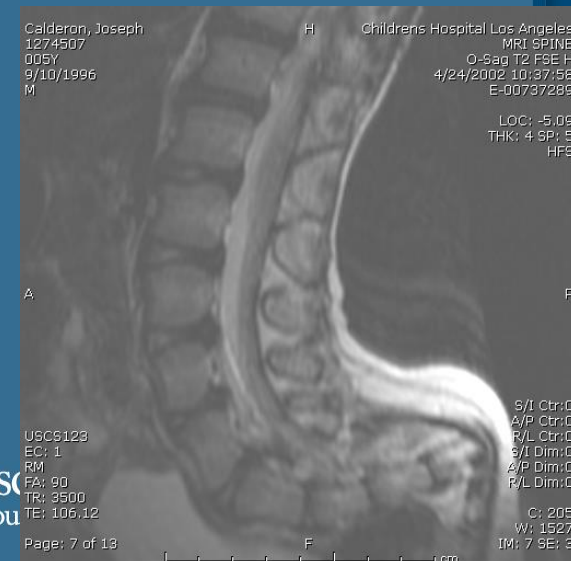
46 months





# SKELETAL DYSPLASIA SPINE

- DIASTROPHIC DYSPLASIA
  - LUMBAR SPINE LORDOSIS
    - PARTLY FROM HIPS, PARTLY FROM SPINE
  - LUMBAR STENOSIS
    - ONLY IN LOW LUMBAR AREA
    - LAMINECTOMY RARE

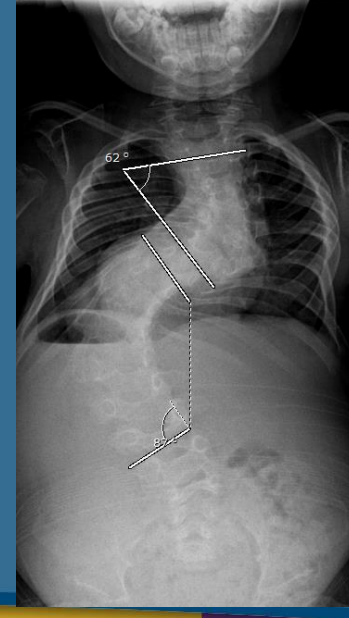


ORTHOPAEDIC CENTER



# SKELETAL DYSPLASIAS

- DIASTROPHIC DYSPLASIA
- EXAMPLE OF EARLY ONSET OF SCOLIOSIS
  - FROM 5 MONTHS TO 48 MONTHS
  - RAPID PROGRESSION, CONSIDER EARLY APICAL FUSION



# SKELETAL DYSPLASIAS

- DIASTROPHIC DYSPLASIA
  - KYPHOSCOLIOSIS
    - SPINE GROWTH DONE BY AGE 8
    - RIGID, MID-THORACIC KYPHOSCOLIOSIS IN 30%
    - IF SEVERE, ONSET < 4 YRS

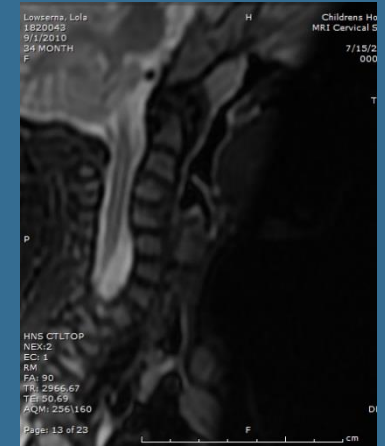


# SKELETAL DYSPLASIAS

- **DIASTROPHIC DYSPLASIA**

- **SPINAL ABNORMALITIES**

- BIFID C-SPINE
- CERVICAL KYPHOSIS, USE MRI
- SEVERE KYPHOSCOLIOSIS
  - **RARE NEUROLOGIC DEFICIT**





# SKELETAL DYSPLASIAS

- DIASTROPHIC DYSPLASIA
  - **KYPHOSCOLIOSIS**
    - SPINE GROWTH DONE BY 8
    - RIGID, MID-THORACIC
    - IF SEVERE, ONSET < 4 YRS



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California



# DIASTROPHIC DYSPLASIA

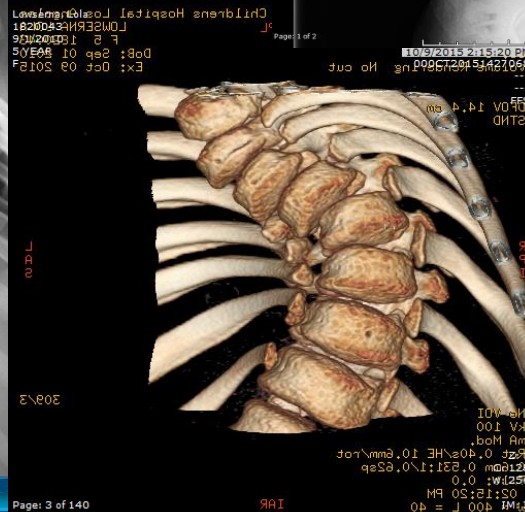
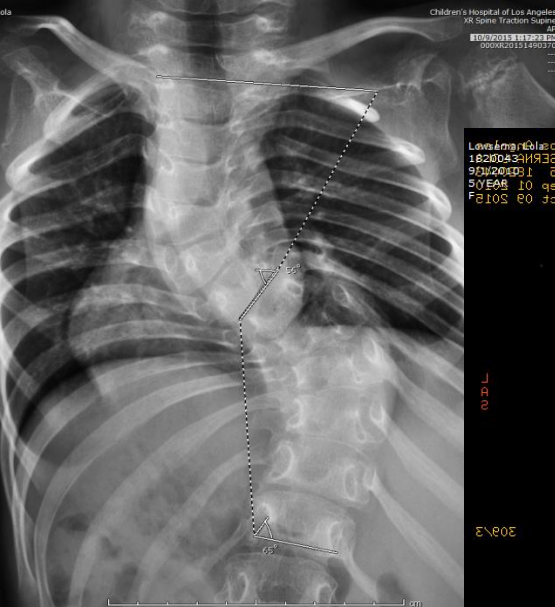
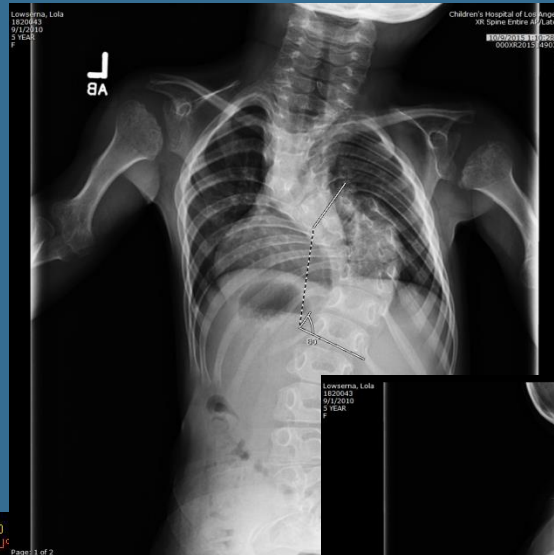
## – KYPHOSCOLIOSIS

- SPINE GROWTH DONE BY AGE 8
- RIGID, MID-THORACIC IN 40%
- IF SEVERE, ONSET < 4 YRS

- 2 YEAR OLD

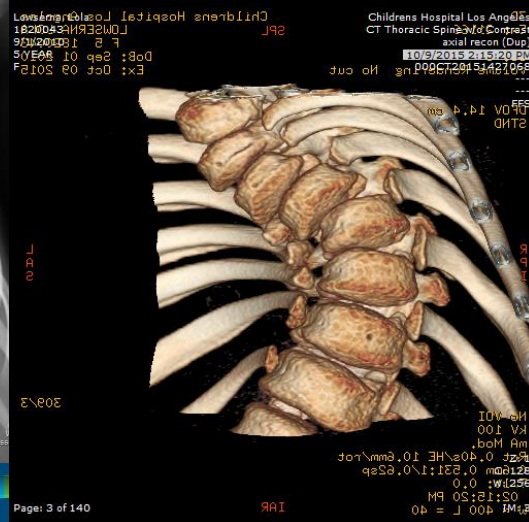
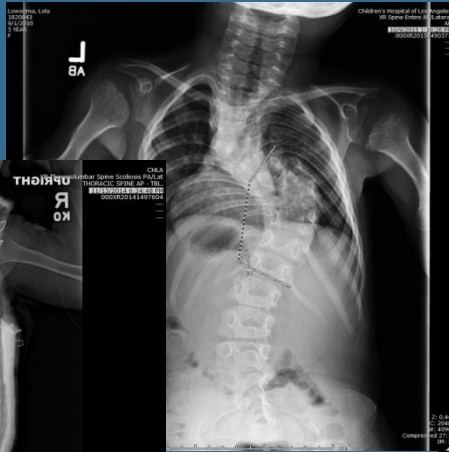


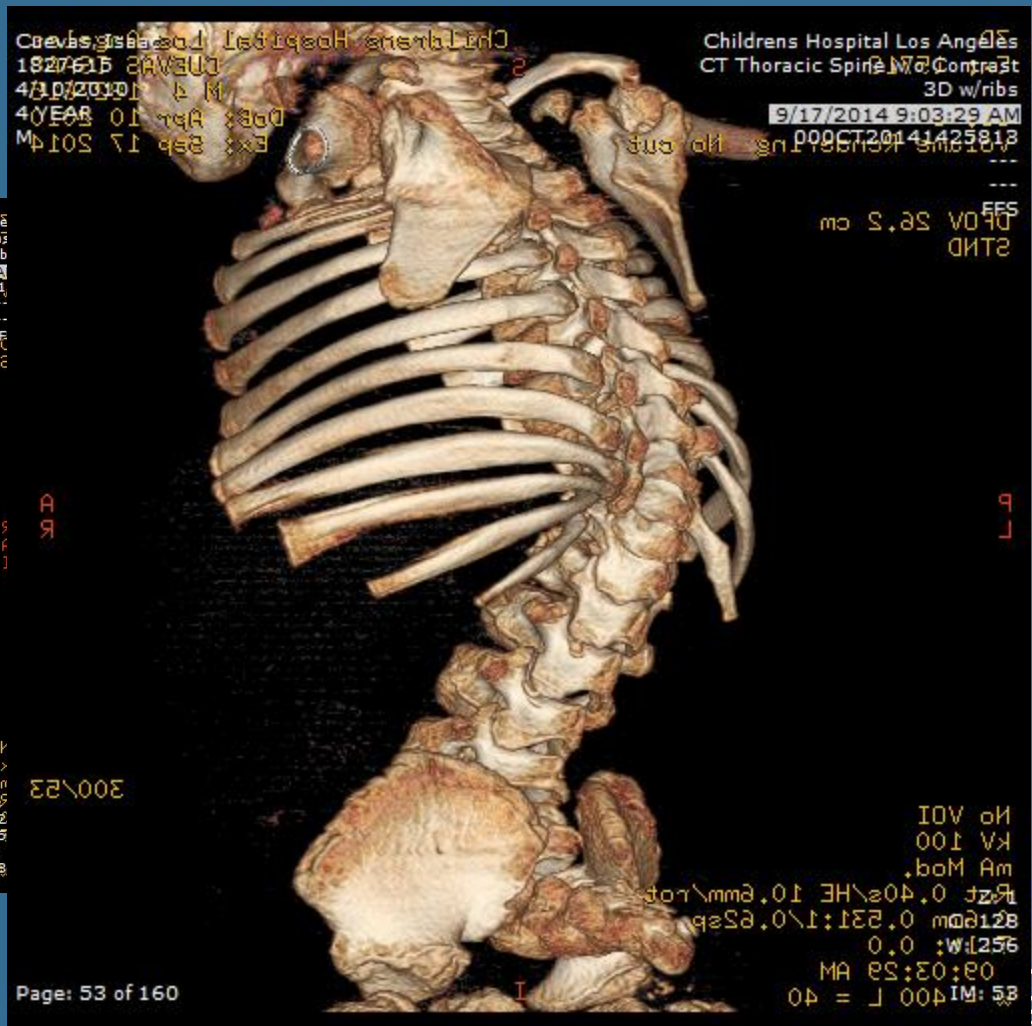
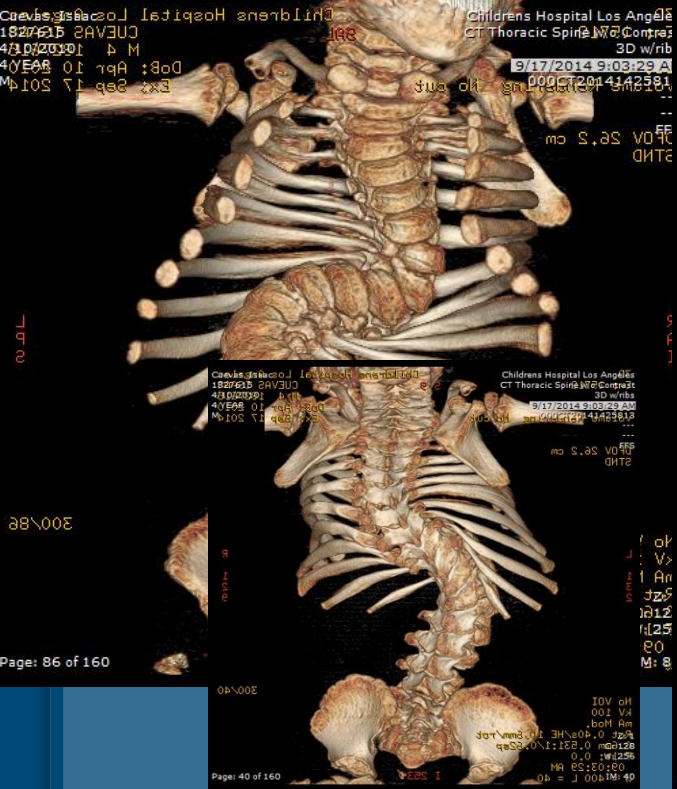
# Lola lowserna





# a lowserna

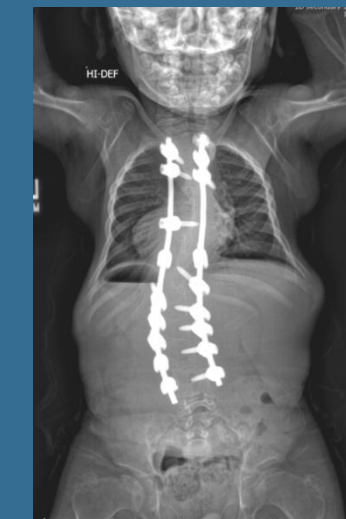
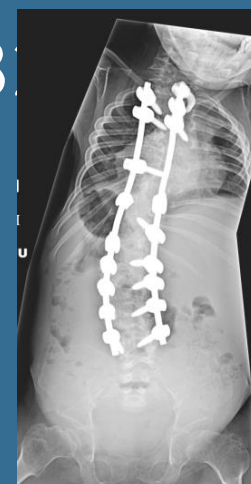








AC CUEVAS 18

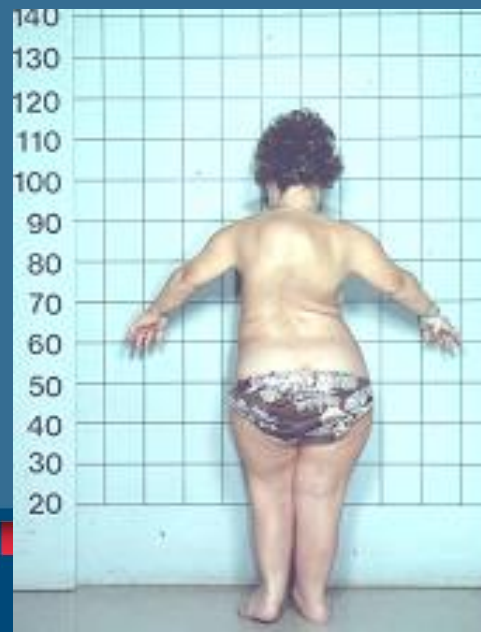




# DIASTROPHIC DYSPLASIA

## – KYPHOSCOLIOSIS

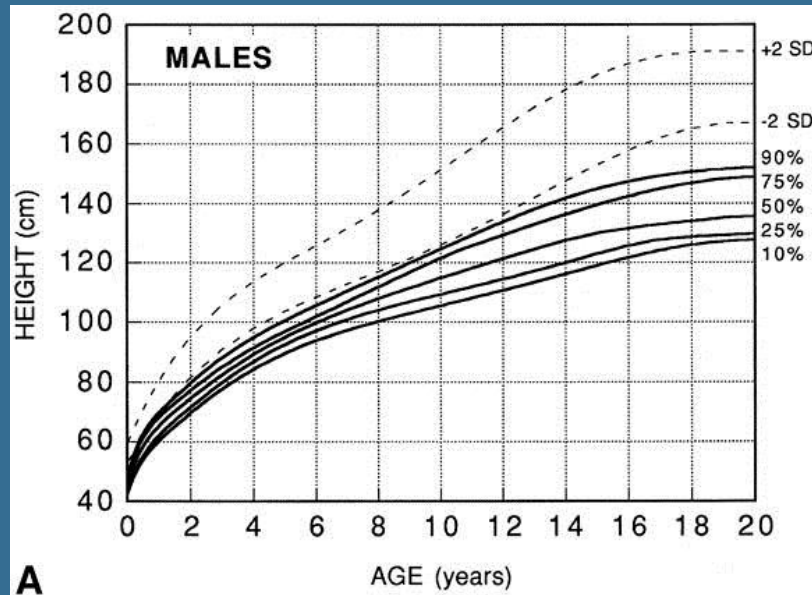
- SPINE GROWTH DONE BY 8
- RIGID, MID-THORACIC
- IF SEVERE, ONSET < 4 YRS



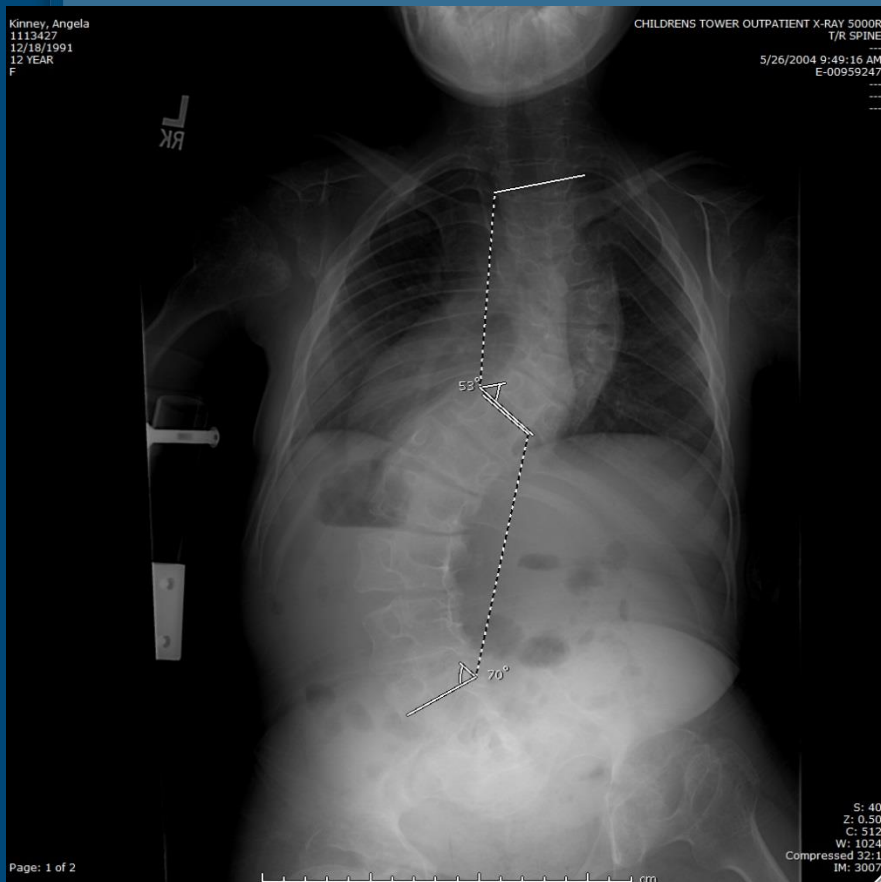
University of  
California



- Diagnosis with molecular genetic testing of
- SLC26A2..the only gene in which pathogenic forms of DD are known to occur



# Angela kinney 12 to 14

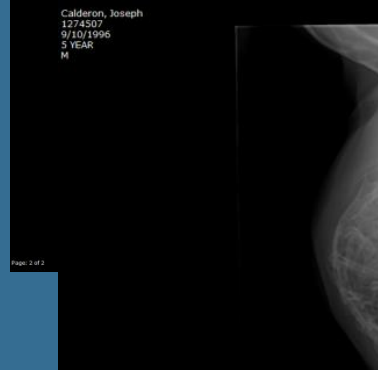


KINNEY, ANGELA, R  
12/18/1991  
12 YEAR  
F  
12/18/1991  
12 YEAR  
F  
12/18/1991  
12 YEAR  
F

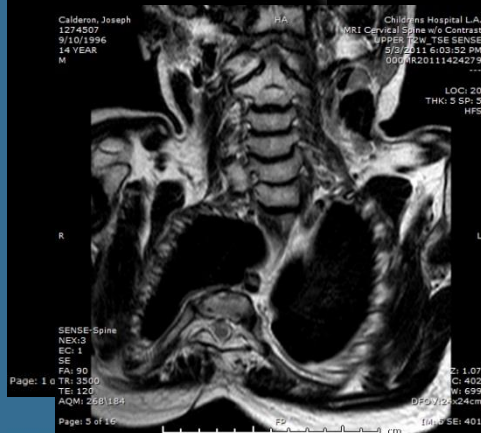
Kaiser Permanente Downey GMO  
Ref.: WEISER, WILLIAM MARTIN  
SIEMENS  
RADIS V



# Joseph calderone 1274507



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Calif

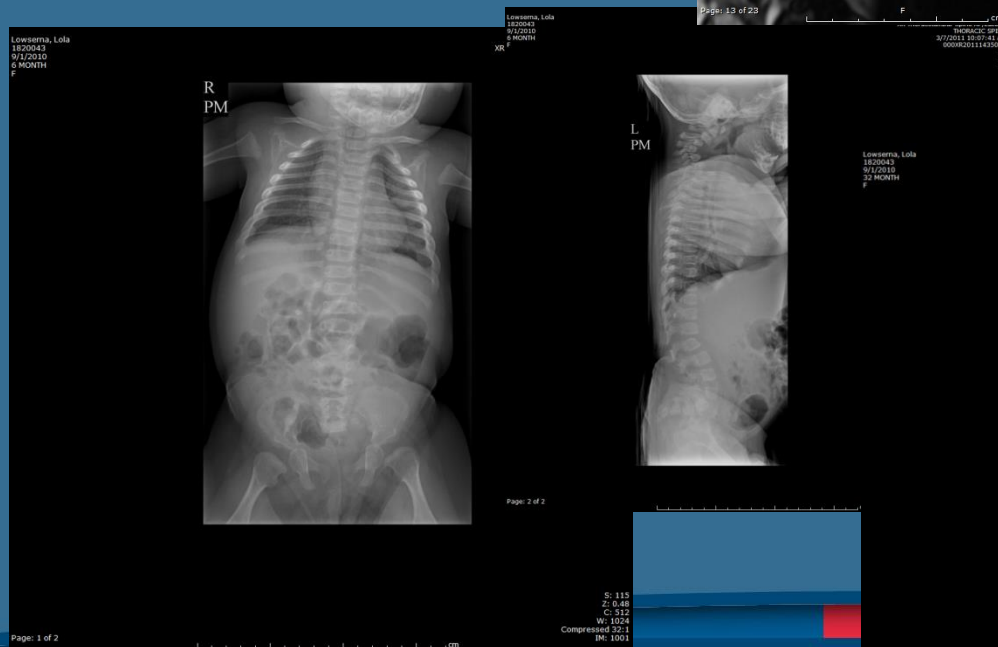
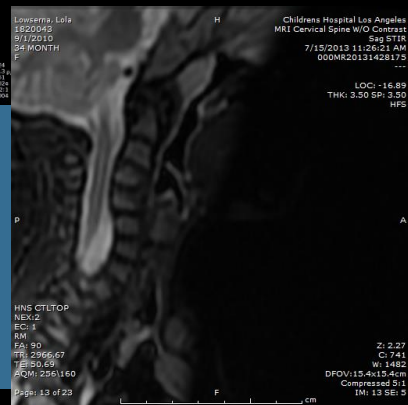
# Adrian gasca

- BRACE AGE 3 TO 8
  - CONTINUED PROGRESSION
- PSF AT AGE 8





- 6 mo
- 32 mo





• DD

Dino, Alexander  
1599181  
9/22/2005  
7 YEAR  
M

CH: Dino, Alexander  
XR Feet W/ Dino, Alexander  
1599181  
9/22/2005  
7 YEAR  
M

CHLA TOWER XRAY  
XR Feet WB Bilateral AP  
P  
5/26/2004 9:57:40 AM  
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Kinney, Angela  
1113427  
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12 YEAR  
F

CHILDRENS TOWER OUTPATIENT X-RAY 5000  
T/R SPINE

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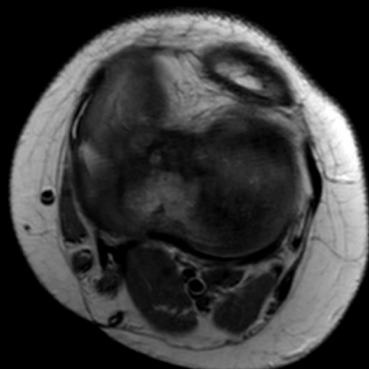


Dino, Alexander  
1599181  
9/22/2005  
5 YEAR  
M

A

Childrens Hospital 3T  
MRI KNEE W/O CONTRAST LT  
LT, PD, AX  
000MR20111427723

LOC: 42  
THK: 3 SP: 3  
PPS



SENSE-Knee-3  
NEX:2  
EC:1  
SE  
PA: 90  
TR: 4390.64  
TE: 30  
NQA: 763/276

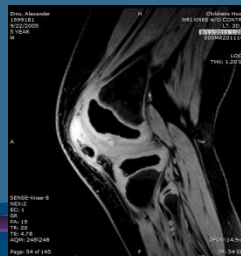
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IM: 15 SE: 1201

Page: 15 of 38  
No cut

DFOV 39.2 cm  
STANDARD  
135/10



No shutter  
Av: 120  
WB: 50  
2.5 mm/2.5sp  
11:11:00 PM  
01:28:14 PM  
Page 12 of 38



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C: 607  
W: 1024  
Compressed 32:1  
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