

# SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA

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ICEOS 2018



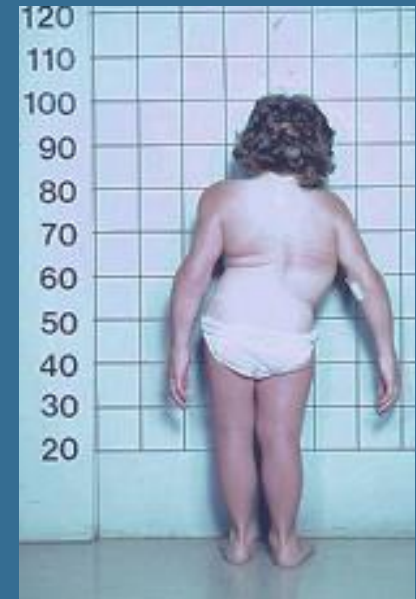
# DISCLOSURE

- EDITOR EMERITUS, JBJS



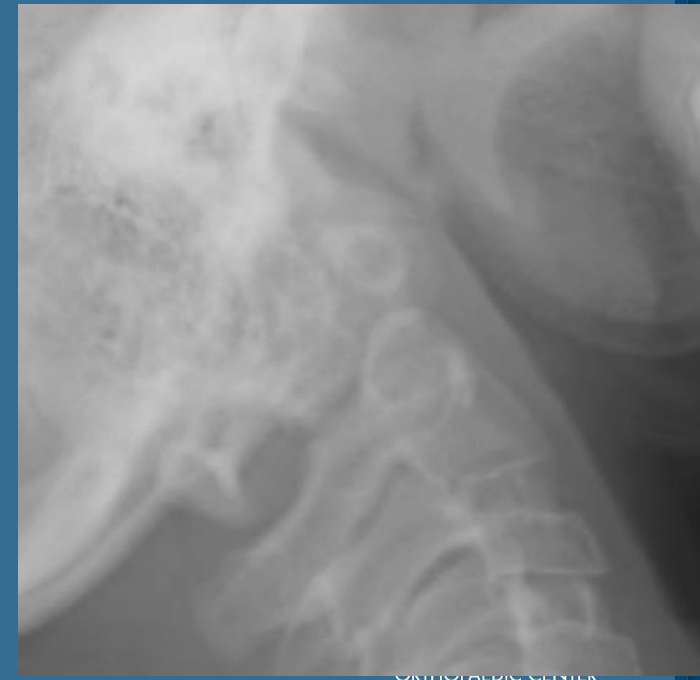
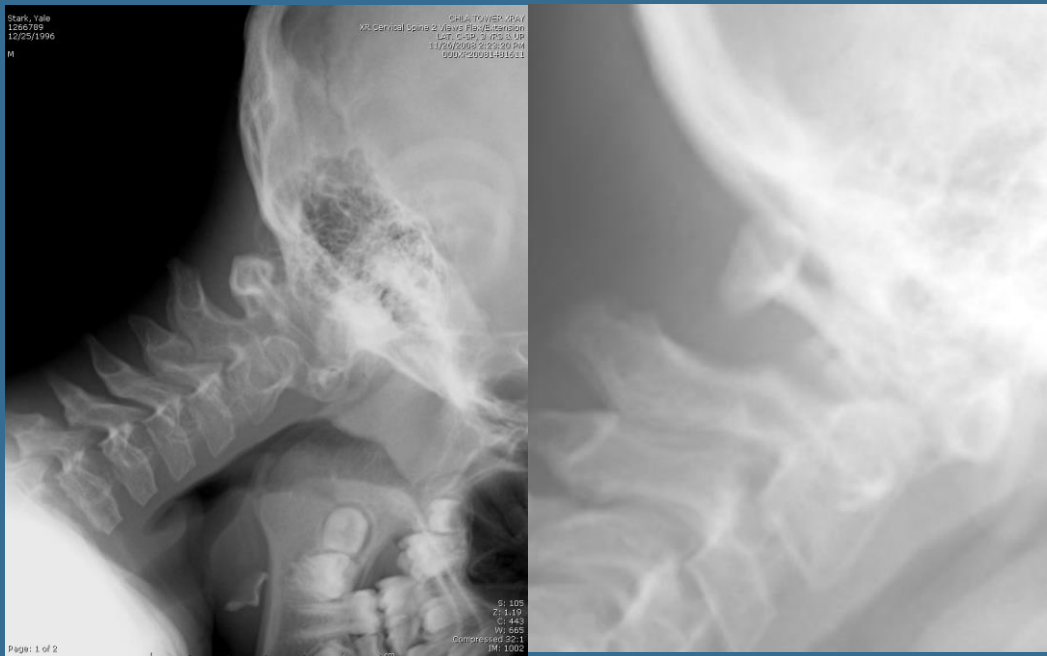
# SED CONGENITA

- CHROMOSOMAL SITE 12q13.11-q13.2
- TYPE II COLLAGEN DEFECT
  - COL2A1 MUTATION
- CLINICAL PICTURE
  - SHORT TRUNK AND LIMBS
  - HANDS/ FEET FAIRLY NORMAL
  - LUMBAR LORDOSIS
    - FROM SPINE AND HIPS
  - WADDLING GAIT, EXT. ROTATION
  - ABOUT 50% WITH C1-C2 INSTABILITY



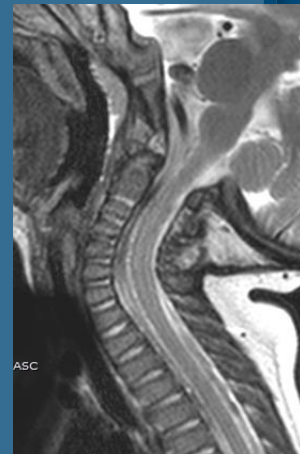
# SED CONGENITA

- **ODONTOID HYPOPLASIA IN MOST**
  - **FLEX-EXT NECK XRAYS**
    - **FLEXION OR EXTENSION INSTABILITY MAY BE SEEN**
  - **FLEX-EXT MRI IN YOUNG**



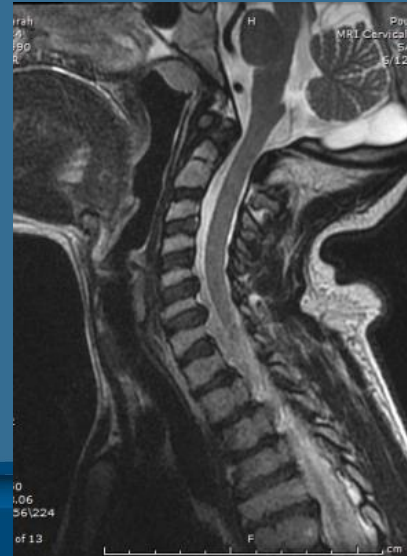
# SED CONGENITA

- **FLEXION-EXTENSION C-SPINE MRIs** *(Mackenzie, et al. 2013)*
  - **31 SKELETAL DYSPLASIA PATIENTS**
    - AVERAGE AGE 3 YEARS
    - VARIETY OF DIAGNOSES
  - **6 WITH XRAY INSTABILITY WITH NORMAL MRI**
  - **9 WITH NORMAL XRAYS HAD ABNORMAL MRI**
  - **14 HAD SURGERY**
  - **17 CONTINUED OBSERVATION**
  - **NO COMPLICATIONS FROM MRI**



# SED CONGENITA

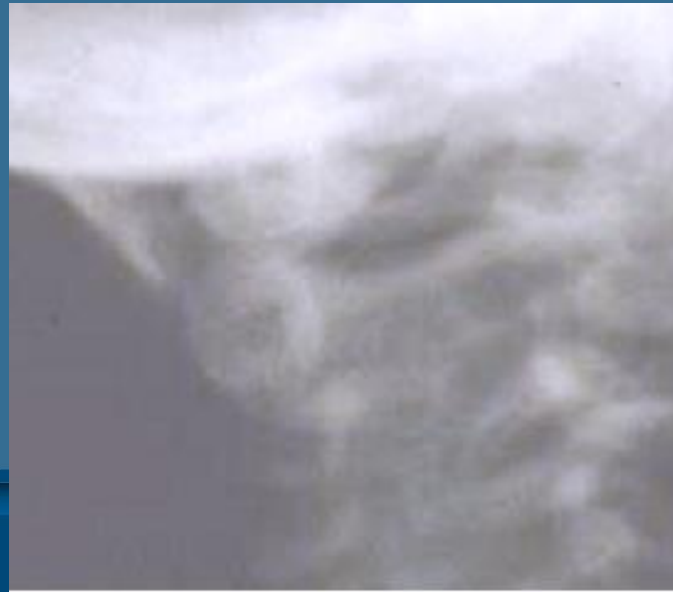
- FLEX/EXT MRI USEFUL
- HELPS IN SURGERY DECISION
- 11 year old with 7 mm C1-C2 motion
- No surgery recommended after MRI



# SED CONGENITA

## – ATLANTO-AXIAL INSTABILITY TREATMENT

- PRESENT AT YOUNG AGE OFTEN
- FUSION UNINSTRUMENTED OR INSTRUMENTED
  - ALWAYS ILIAC CREST OR RIB AUTOGRAFT
- FUSE OCCIPUT TO C2 IF NO IMPLANTS
- MAY DO C1-C2 OR O-C2 WITH IMPLANTS



# SED CONGENITA

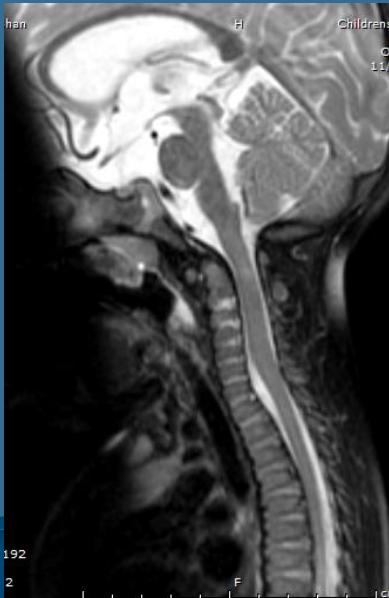
## UNINSTRUMENTED FUSION

### – SIGNAL CHANGE IN CORD AT AGE 3

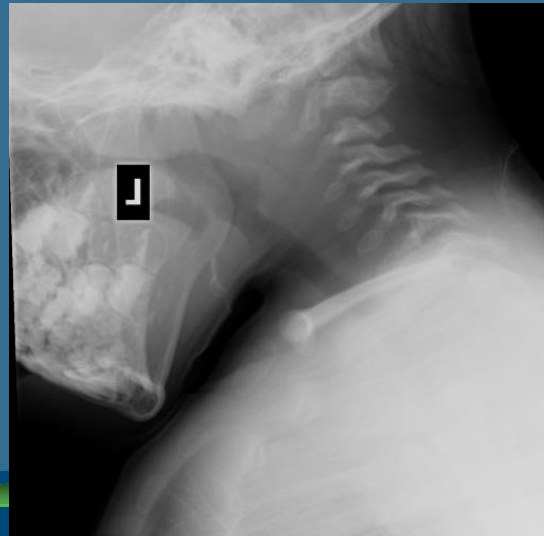
- ILIAC CREST GRAFT AND HALO FOR 3 MONTHS

### – IMPROVED MRI AT 7 YEARS POST FUSION

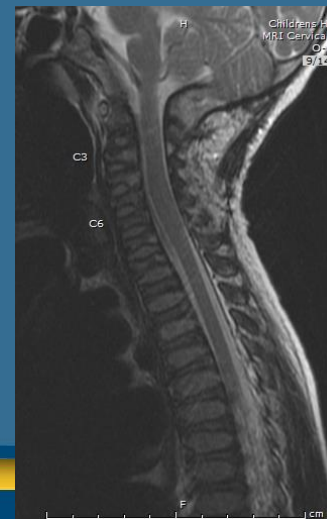
• 2000



2002 nonunion?



2007 fusion





# SED CONGENITA

- UNINSTRUMENTED FUSION
  - DECOMPRESSION AND FUSION AT 4
  - HALO 3 MONTHS

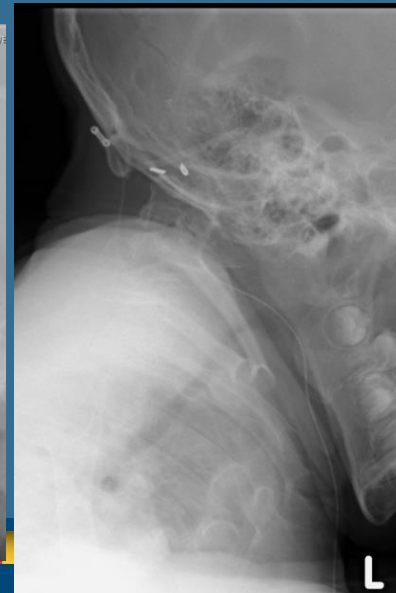
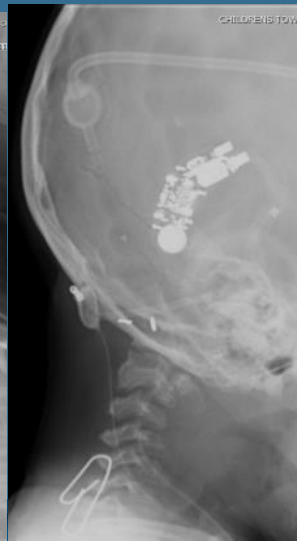
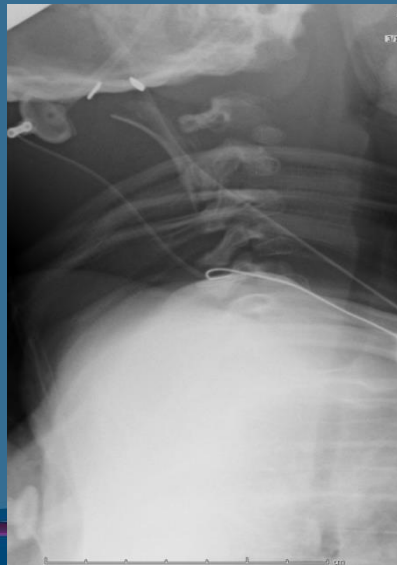
—

post-op

2 yrs

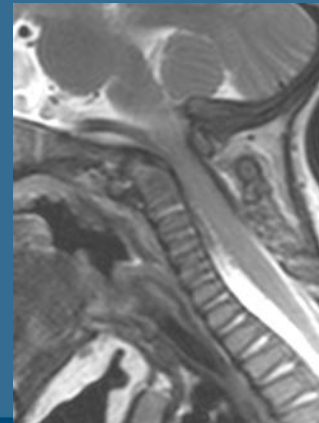
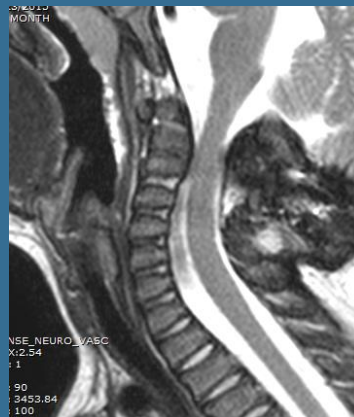
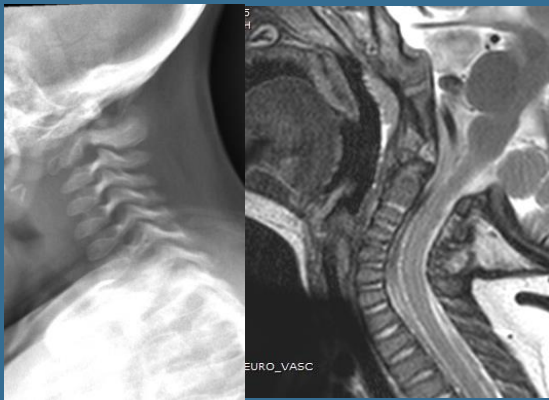
5 years

7 years



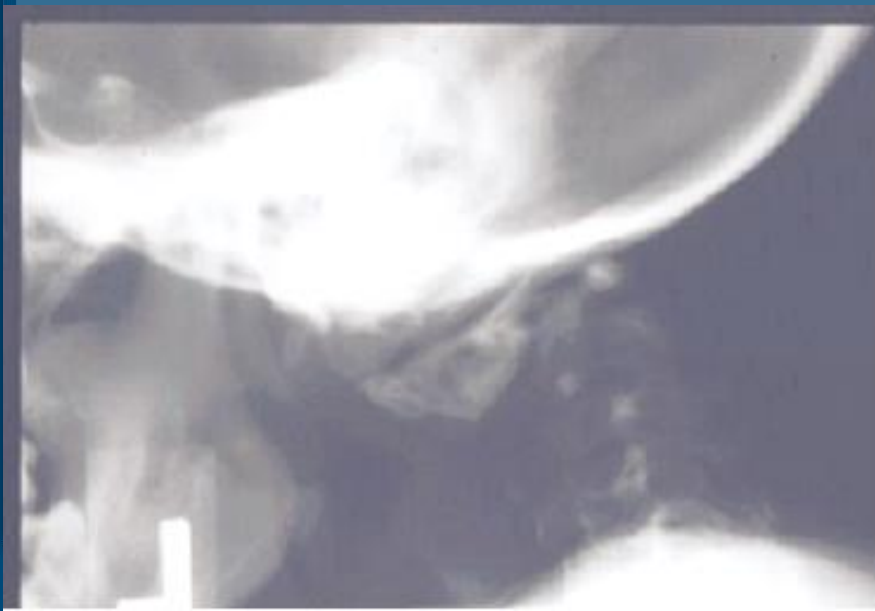
# SED CONGENITA

- UNINSTRUMENTED FUSION
  - INSTABILITY AT AGE 2
  - MRI WITH COMPRESSION ON FLEXION
  - OCCIPUT TO C2 FUSION IN HALO
    - NOW 1 YEAR POST-OP



# SED CONGENITA

- SIMPLE INSTRUMENTED FUSION
  - 15 Y.O. WITH C1-C2 INSTABILITY
    - REDUCED IN HALO
    - HALO ON FOR 3 MONTHS
    - POSTERIOR WIRING AND ILIAC AUTOGRAFT

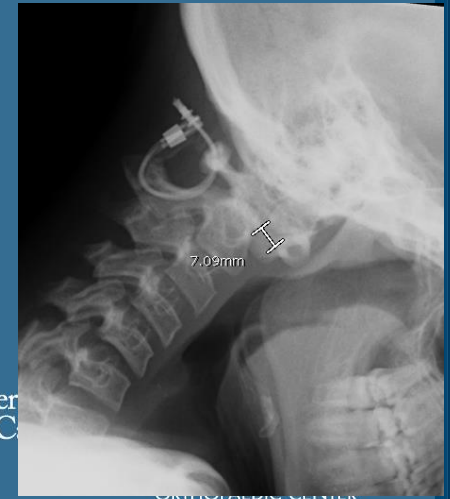


# SED CONGENITA

- **SKELETAL DYSPLASIA CERVICAL FUSION** *(Ain, et al,2006)*
  - **25 PATIENTS**
    - 7 WITH SED
  - **INDICATIONS: >8 mm ADI AND <14 mm SAC**
    - INTERNAL FIXATION IN 13/25
    - HALO VEST FOR MEAN 3.8 MONTHS IN 23/25
  - **92% FUSION RATE**
    - 20% COMPLICATION RATE
      - 4 NONUNION
      - 2 IMPLANT LOOSENING

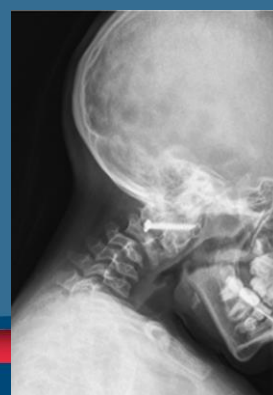
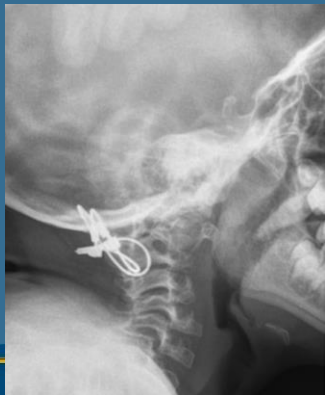
# SED CONGENITA

- FAILURE OF INSTRUMENTED FUSION
  - 16 y.o. WITH 8 mm ADI → FUSED IN HALO
  - ILIAC CREST GRAFT
  - CABLES FOR INSTRUMENTATION
  - NON-UNION WITH 7 mm ADI



# SED CONGENITA

- **IMPLANT vs. UNINSTRUMENTED** *(Serhan Er, et al., JPO, 2017)*
  - **20 CHILDREN WITH SED**
    - AVERAGE AGE 6 YEARS
  - **15 INSTRUMENTED, 5 NOT**
    - ALL INSTRUMENTED FUSED
    - ALL BUT 4 IN HALO 3 MONTHS
    - 3 OF 5 WITHOUT INSTRUMENTS → NONUNION
  - **DJK IN 13% WITH IMPLANTS**

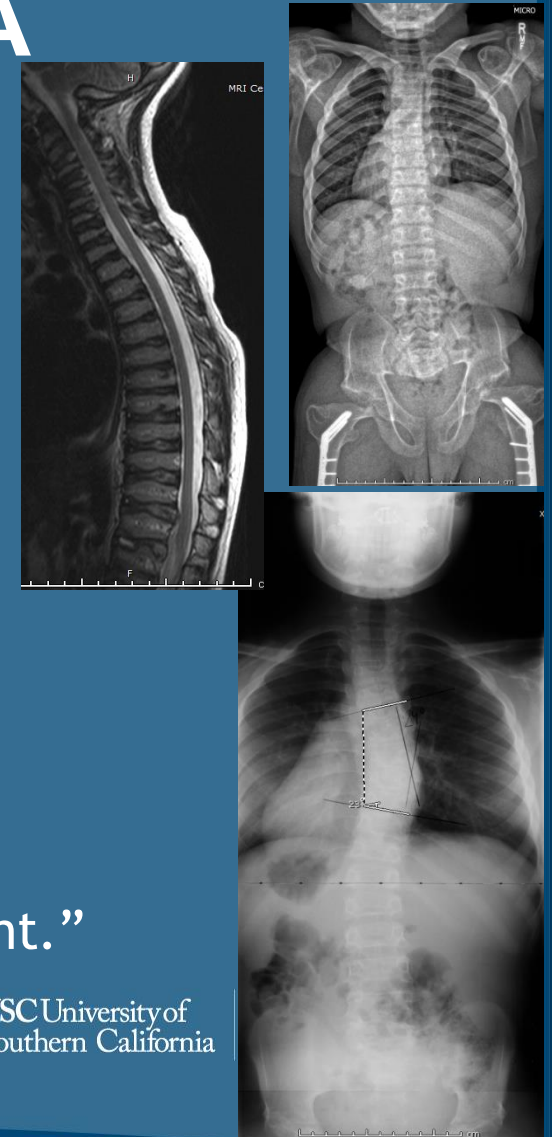


# SED CONGENITA

- THORACIC AND LUMBAR SPINE
  - PAUCITY OF LITERATURE
  - PLATYSPONDYLY
  - LUMBAR HYPERLORDOSIS IN SOME
  - 93 PATIENTS WITH COL2A1 MUTATION
    - VARIETY OF DIAGNOSES
    - 48% WITH SCOLIOSIS
    - NO NOTE ON TREATMENT

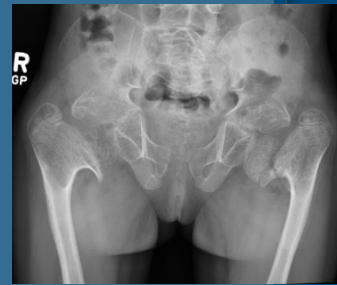
“Six children had thoracolumbar scoliosis or kyphoscoliosis which required surgical management.”

(Serhan Er, et al., 2017)



# SED CONGENITA

- PERSONAL EXPERIENCE
  - NOT COMMON
  - TREAT AS IDIOPATHIC
  - INSTRUMENTS IN CANAL OK
  - EXTENSION WITH HIP OSTEOTOMY
    - TO HELP LUMBAR LORDOSIS



f  
nia



# SED CONGENITA

## – ANGULAR DEFORMITY

- MULTI-LEVEL OSTEOTOMY
- ARTHROGRAMS AT ALL JOINTS
  - ALLOWS VISUALIZATION OF JOINT LINE

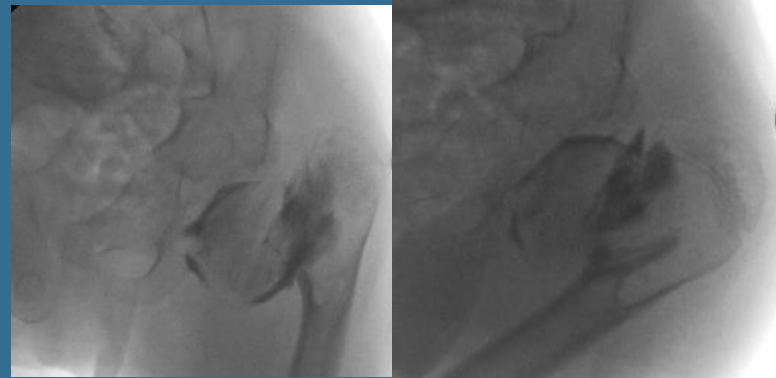
## – COXA VARA

- OSTEOTOMY PROX. FEMUR
  - VALGUS
  - INTERNAL ROTATION
  - EXTENSION



# SED CONGENITA

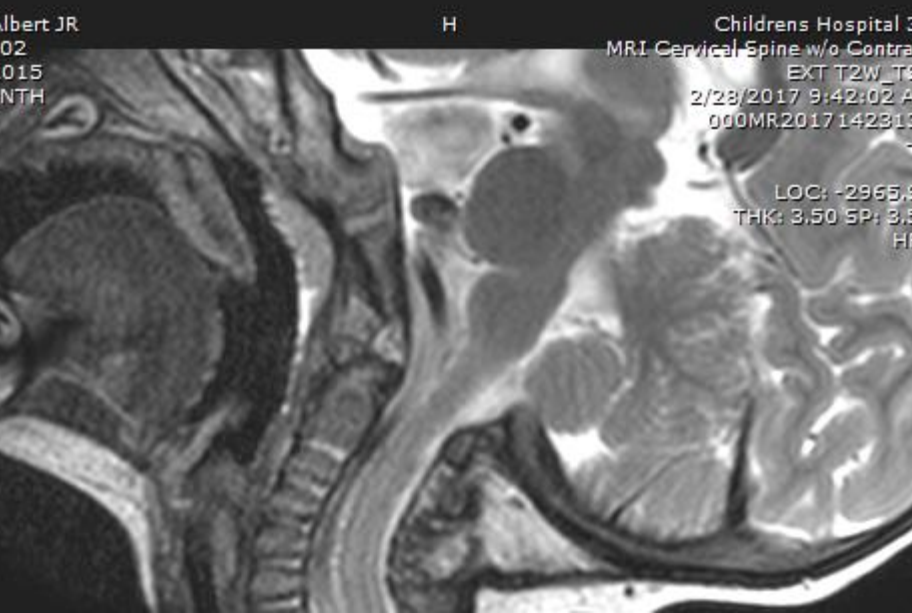
- TREATMENT OF COXA VARA
  - VALGUS, INTERNAL ROTATION, EXTENSION
  - ARTHROGRAM INTRAOP
  - VALGUS CORRECTION KEY
    - 15° LESS THAN ADDUCTION





 USC University of  
Southern California

  
CHILDREN'S  
ORTHOPAEDIC CENTER

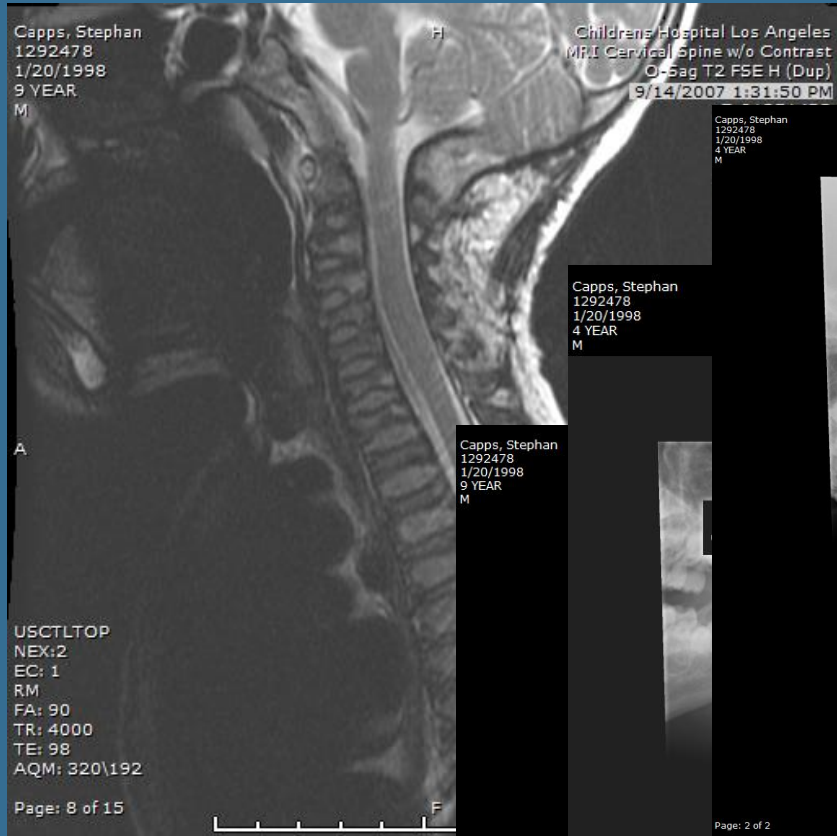


ES  
CENTER

# SKELETAL DYSPLASIAS

- SPONDYLOEPIPHYSEAL DYSPLASIA  
(SED CONGENITA)
  - CHROMOSOMAL SITE 12q13.11-q13.2
  - TYPE II COLLAGEN DEFECT
  - RELATED DISORDERS
    - KNIEST DYSPLASIA (PRIMARY ONE)
    - STICKLER SYNDROME
    - STRUDWICK TYPE SMED



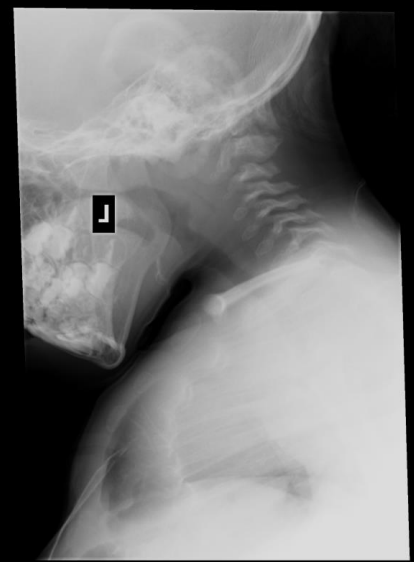


Capps, Stephan  
1292478  
1/20/1998  
4 YEAR  
M

CHILDRENS TOWER OUTPATIENT

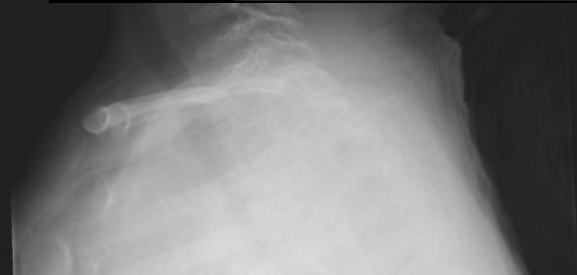
2/19/200

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1/20/1998  
4 YEAR  
M

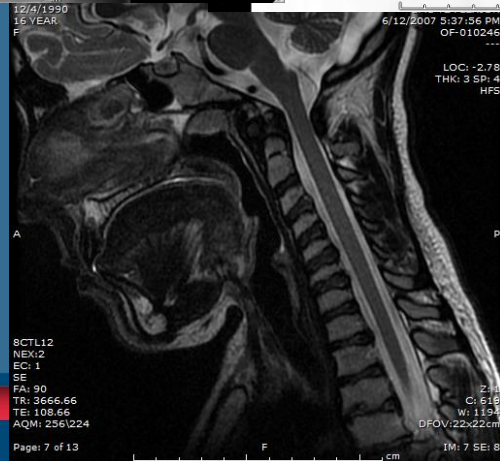
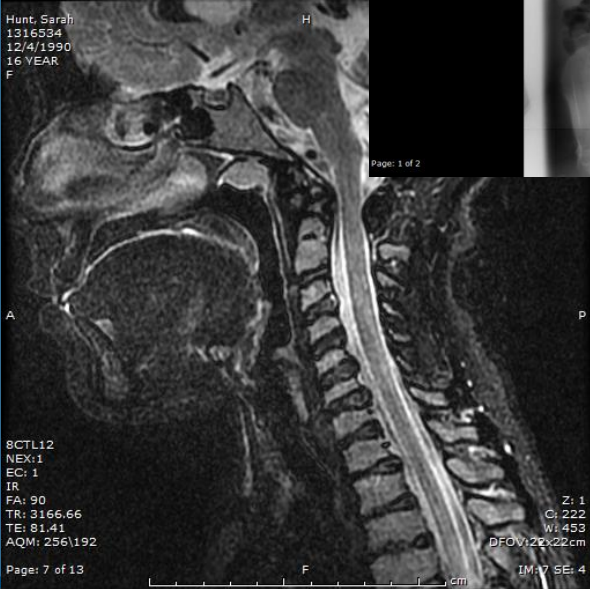
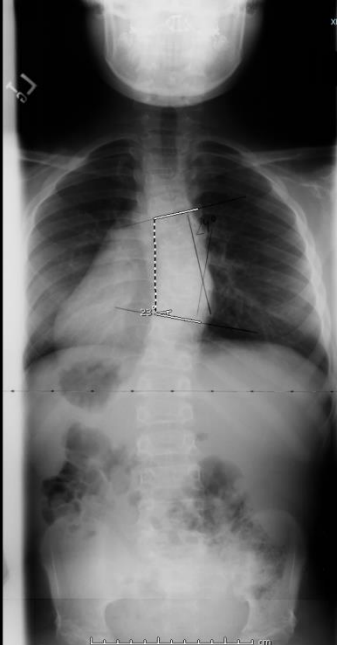


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Com



# SARAH HUNT SED





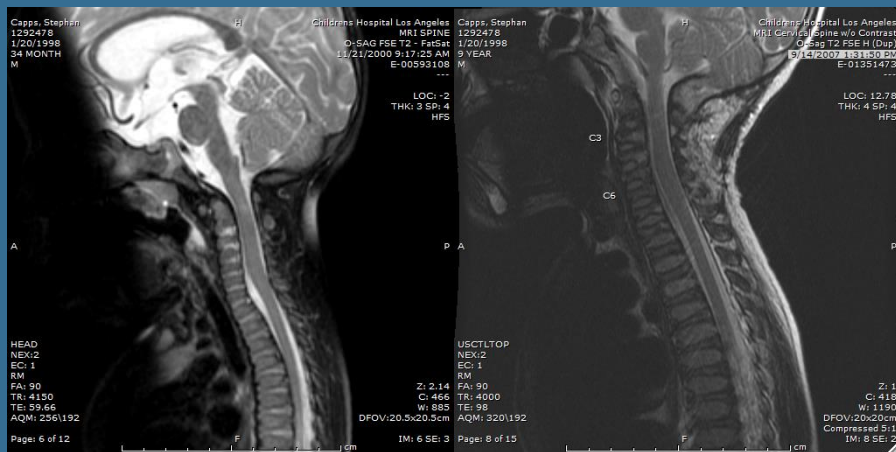
# Stephan capps...1292478

sed with signal change at C1-2 at age 34  
mo → fusion,,,,, 5 yr later mri without signal change

• 2000

2007

2013, 6 yrs after hips, age 15



# SKELETAL DYSPLASIAS

- SED CONGENITA

- ANGULAR DEFORMITY

- MULTI-LEVEL OSTEOTOMY
    - ARTHROGRAMS AT ALL JOINTS
      - ALLOWS VISUALIZATION OF JOINT LINE



- COXA VARA

- OSTEOTOMY PROX. FEMUR
      - VALGUS
      - INTERNAL ROTATION
      - EXTENSION



# YALE STARK 1266789

Stark, Yale  
1266789  
12/25/1996  
15 YEAR  
M

CHLA TOWER XRAY  
XR Thoracolumbar Spine Scoliosis PA/Lat  
LATERAL SCOLIOSIS  
11/25/2003 2:25:06 PM  
000XR20081481610

Stark, Yale  
1266789  
12/25/1996  
15 YEAR  
M

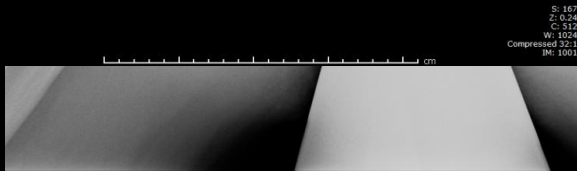
CHLA TOWER XRAY  
XR Hip w/ AP Pelvis Min 2 Views Bilat  
PELVIS, GENERAL  
000XR20121472274

CHLA TOWER XRAY  
XR Hip w/ AP Pelvis Min 2 Views Bilat  
PELVIS, GENERAL  
000XR20121472274



Stark, Yale  
1266789  
12/25/1996  
6 YEAR  
M

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S: 167  
Z: 0.34  
C: 512  
W: 1024  
Compressed 32:1  
IM: 1001

Page: 2 of 2



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W: 1024  
Compressed 32:1  
IM: 1002

Page: 2 of 2

Page: 1 of 1



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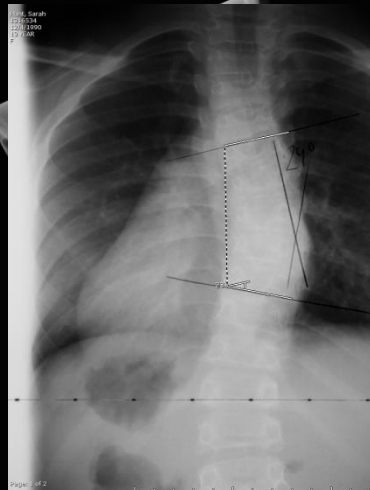
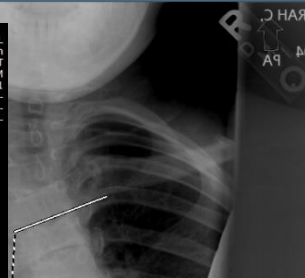


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Hunt, Sarah  
1316534  
12/4/1990  
11 YEAR  
F



Hunt, Sarah 06.01.2005



CHILDRENS TOWER OUTPATIENT X-RAY 5000R  
XR Cervical Spine 2 Views Flex/Extension



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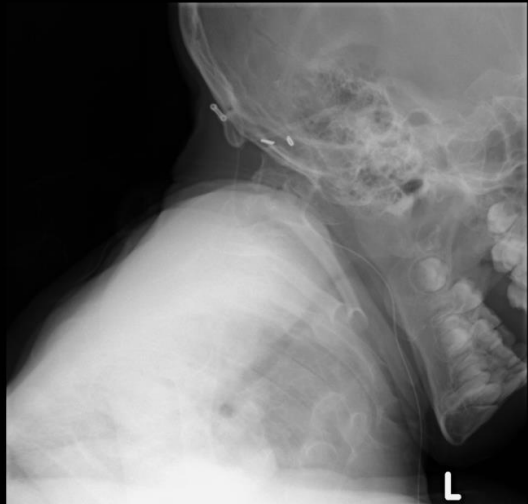
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CHLA TOWER XRAY  
XR Cervical Spine 2 Views Flex/Extension  
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E-01150720

STARK 12510

Stark, Julian  
1251090  
12/25/1996  
4 YEAR  
M

SPINE\_68MHZ.LUMBAR\_CHL



FLEXION

cut

Children

S

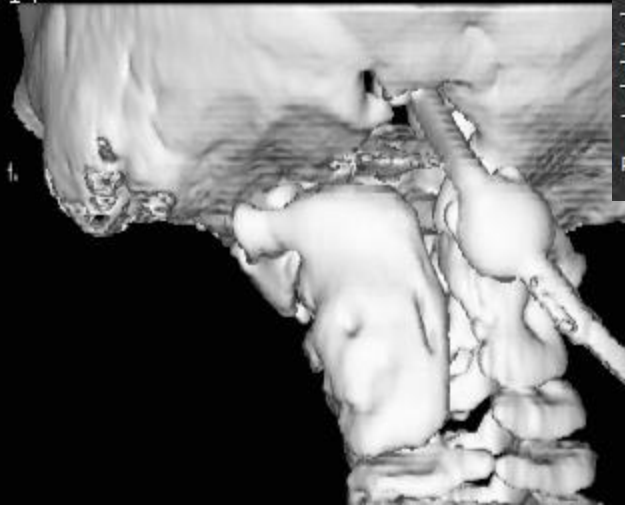
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612/14

RL

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IM: 2063



NEX:2  
TSE1  
TR: 4000  
TE: 103

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