SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA

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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 AUTOGRRAFT
  • 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 $\rightarrow$ 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
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
  – ARTHRGRAM INTRAOP
  – VALGUS CORRECTION KEY
    • 15° LESS THAN ADDUCTION
SKELETAL DYSPLASIAS

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