DIASTROPHIC DYSPLASIA

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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
  - CAULIFLOWER EAR
  - HITCHHIKER THUMB
  - VERY SHORT
DIASTROPHIC DYSPLASIA

– SPINAL ABNORMALITIES
  • CERVICAL KYPHOSIS
  • SEVERE KYPHOSCOLIOSIS
  • LUMBAR LORDOSIS AND STENOSIS
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
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°, KYPHOSIS 97°
  – IDIOPATHIC-LIKE (41)
    • MEAN SCOLIOSIS 49°, KYPHOSIS 29°
  – MILD NON-PROGRESSIVE (33)
    • MEAN SCOLIOSIS 19°, KYPHOSIS 25°
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 PEDICILE 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
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!
LOLA LOWSERN.A..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
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
DIASTROPHIC DYSPLASIA

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

- 2 YEAR OLD
DIASTROPHIC DYSPLASIA

– KYPHOSCOLIOSIS

• SPINE GROWTH DONE BY 8
• RIGID, MID-THORACIC
• IF SEVERE, ONSET < 4 YRS
• 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
Joseph calderone 1274507
Adrian gasca

- BRACE AGE 3 TO 8
  - CONTINUED PROGRESSION
- PSF AT AGE 8
• 6 mo
• 32 mo
• DD