Craniocervical Deformity in Osteogenesis Imperfecta
Whom to Screen, How to Diagnose and When to Treat

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The Spine in Patients With Osteogenesis Imperfecta

Abstract

Osteogenesis imperfecta is a genetic disorder of type I collagen. Although multiple genotypes and phenotypes are associated with osteogenesis imperfecta, approximately 90% of the mutations are in the COL1A1 and COL1A2 genes. Osteogenesis imperfecta is characterized by bone fragility. Patients typically have multiple fractures or limb deformity; however, the spine can also be affected. Spinal manifestations include scoliosis, kyphosis, craniovertebral junction abnormalities, and lumbosacral pathology. The incidence of lumbosacral spondyloysis and spondylolisthesis is higher in patients with osteogenesis imperfecta than in the general population. Use of diphosphonates has been found to decrease the rate of progression of scoliosis in patients with osteogenesis imperfecta. A lateral cervical radiograph is recommended in patients with this condition to examine for cervical spine abnormalities.
Etiology / Pathogenesis of OI Scoliosis

- Severity of disease
- Compression fractures
- Asymmetric growth
- Age

Weinstein, *The Pediatric Spine* 2001
Watanabe, *Spine* 2007
BI: What is it?

- Migration of the upper cervical spine and odontoid peg into the base of the skull
- Brainstem and cerebellum compression
- CSF flow disturbance
- Cranial nerve deficits
- Sudden death (?)
- Acquired disorder of bone-softening conditions
  - OI, rickets, hyperparathyroidism, Paget’s disease, SED, Morquio, Hajdu-Cheney syndrome
Basilar Invagination in OI

- Sillence DO, Pediatr Radiol 1994
- Overall – 25%
- Type IV OI and dentinogenesis imperfecta – 75%
  - Over half were symptomatic
- Delay independent sitting posture until 18 mos
Basilar Invagination in OI

- Platybasia – flattened skull base with a basal angle of more than 145 degrees
- Basilar Impression – Upward infolding of foramen magnum ring into the skull
- Basilar Invagination – odontoid process protrudes into intracranial space penetrating the foramen magnum
26-37% OI patients have craniocervical deformity
- Basilar invagination: 4-25%
- Basilar impression: 13-40%
- Platybasia: 16-29%

44% have BI, hydrocephalus and syringomyelia (Menezes)

Incidence increase with age
- younger cohorts → lower prevalence reported

Prevalance is related to severity of OI (height Z score)

Bisphosphonates: no signif effect

Sawin, Menezes J Neurosurg, 1997
Arponen J Bone Miner Res, 2012
Etiology

- Failure of weight bearing parts of the skull
- CVJ microfractures
- Aggravated by early childhood erect posture
- Why don’t all patients with OI get BI?
- Perhaps many do, but variably
- Only a few become symptomatic
- Sillence 30% of Type III/IV have BI
  - Only 1/3 of them are symptomatic
## BI Signs & Symptoms

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<tr>
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<tbody>
<tr>
<td>Headache</td>
<td>76%</td>
<td>50%</td>
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<tr>
<td>Cranial N Palsy</td>
<td>68%</td>
<td>65%</td>
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<tr>
<td>New Dysphagia</td>
<td>60%</td>
<td>45%</td>
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<tr>
<td>Hyperreflexia</td>
<td>56%</td>
<td>-</td>
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<tr>
<td>Quadriparesis</td>
<td>48%</td>
<td>10%</td>
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<tr>
<td>Ataxia</td>
<td>32%</td>
<td>50%</td>
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<tr>
<td>Nystagmus</td>
<td>28%</td>
<td>55%</td>
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BI – Who to Screen

- Baseline lateral skull/c-spine recommended in all OI patients before school age
  - MRI recommended if BI suspected on xray
  - Repeat imaging if symptoms occur (+/-)

- Definitions, radiographic “abnormal” vary

Basilar Invagination: *dens* above McRae’s line
Basilar Impression: *dens* above modified Chamberlain’s line

- >5mm
- >2.5 SD above controls
- >3 SD above controls = 10.5mm

Basilar Impression: 

dens above McGregor’s line

- >7mm
- >2.5 SD above controls
- >3 SD above controls  
  = 11.9mm

Landmarks are difficult to identify!!
Basilar Invagination

McRae

Chamberlain

McGregor

DM
Headaches / Neck Pain
...his Dad’s MRI scan
Two OI patients (severe scoliosis) without BI
Ibrahim & Crockard, *JNS* 2007

- Ventral decompression
- Elective tracheostomy / extended maxillotomy approach, remove clivus, anterior FM, dens and basal axis
- Posterior occipitocervical fixation Occ-C7 or Occ-T1
- Complications
- Surgical morbidity
- “benefits remain unclear” for asymp patients
Trans nasal approach:
BI: Treatment

- Indicated by symptomatology
- Surgical decompression + fusion mainstay
  - Minerva-type bracing proven successful in halting progression
- Pre-operative halo-type traction
  - Invagination **reducible** pre-op: posterior decompression and craniocervical fusion
  - Invagination **irreducible** pre-op: anterior + posterior decompression and craniocervical fusion
    - Mild radiographic improvement (ie 4mm) complete symptom resolution

Halo gravity traction
Preop CT / Postop X-ray
Preop OI Scoliosis
Clinical Photos
Basilar Invagination / Syrinx
Intraop Fluoro
Improved BI and syrinx
Postop PSF for Scoliosis
BI & Syringomyelia
Occipitocervical Fusion

Dual Cable fixation from Occiptal burr holes to C2 lamina with iliac crest or rib graft provides good alignment, stable fixation and reliable fusion

Sitoula et al. *Spine* 2014
Outcomes

- 100% fusion rate reported in all studies
  - ...but, progression is frequently seen in LT f/u

- 15% recurrence of symptoms seen 2-10yr post-op
  - Treat hydrocephalus + Minerva successful

- No improvement or decreased rates with bisphosphonate tx
Outcomes

- **BI**
  - Resolution or halt progression of symptoms (myelopathy or CN)
  - Improvement of syringomyelia
  - Improvement of radiographic parameters

- **Scoliosis**
  - Improvement in function (mobility) or sitting tolerance (pain)
  - Improvement in pulmonary function
  - No reoperations / readmissions / complications

Important to recognize symptoms or progression of disease early – less morbidity / better outcomes!
Thank you

www.nemours.org/spinecenter

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