Osteogenesis Imperfecta

A rare, congenital, and progressive condition affecting connective tissue resulting in a widely variable clinical presentation

Presented by:

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Clinical Picture

- Bone fragility
- Osteopenia
- Varying height
- Bony deformities
- Joint laxity
- Blue sclerae
- Dentinogenesis imperfecta
- Deafness

PROGRESSIVE!

Image from https://ptrs8332011.wikispaces.com/Osteogenesis+Imperfecta
PICO Question

Does frequency and timing (age ranges) of early rehabilitation interventions affect gross motor development of pediatric patients diagnosed with type 1 Osteogenesis Imperfecta?
**Search Strategy**

- Utilize Ebscohost via BU Library
- **Databases:** Academic Search Complete, CINAHL, MEDLINE
- **Key Findings:**

<table>
<thead>
<tr>
<th>Ref #</th>
<th>Article Title (year)</th>
<th>Search Terms</th>
<th># of Results</th>
<th>Research Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Conservative versus surgical treatment of osteogenesis imperfecta: a retrospective analysis of 29 patients (2012)</td>
<td>Osteogenesis Imperfecta treatment AND surgical AND conservative</td>
<td>3</td>
<td>Level 2: Cohort Study -Outcomes research</td>
</tr>
<tr>
<td>3</td>
<td>Osteogenesis imperfecta in childhood: impairment and disability (2004)</td>
<td>Osteogenesis Imperfecta childhood impairment AND outcome</td>
<td>3</td>
<td>Level 4: Case Series -Prospective 4-year follow-up</td>
</tr>
<tr>
<td>4</td>
<td>A specialized rehabilitation approach improves mobility in children with osteogenesis imperfecta (200?)</td>
<td>Osteogenesis Imperfecta children function AND mobility AND rehabilitation</td>
<td>8</td>
<td>Level 2: Outcomes Research -Retrospective, 6-month follow-up</td>
</tr>
</tbody>
</table>
# Inclusion/Exclusion Criteria

<table>
<thead>
<tr>
<th>Type</th>
<th>Severity</th>
<th>Bony Changes</th>
<th>Sclera Color</th>
<th>Dent. Imperf.</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Non-deforming</td>
<td>Normal to mildly short</td>
<td>Blue</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Moderately deforming</td>
<td>Moderately short</td>
<td>Gray-white</td>
<td>✗</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Mild to moderate scoliosis</td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>Severely deforming</td>
<td>Very short</td>
<td>Gray</td>
<td>✗</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Triangular face</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Severe scoliosis</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Perinatally lethal</td>
<td>Multiple bony fractures @ birth</td>
<td>Dark</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Broad, long bones</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Radiographs show low bone density</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*Compare with WHO's ICF model describing the relationship between function and disability*
Treatment Options

• Surgical
• Conservative
• Medicinal – Bisphosphonates
• Therapy

• Complications?
Frequency

"On Your Feet"

<table>
<thead>
<tr>
<th>M 0</th>
<th>M 6</th>
<th>M 12</th>
</tr>
</thead>
<tbody>
<tr>
<td>1st in-patient stay</td>
<td>Home based WBV-training</td>
<td>2nd in-patient stay</td>
</tr>
<tr>
<td>13 days</td>
<td>3 months</td>
<td>6 days</td>
</tr>
</tbody>
</table>

**Daily:**
- 2 x 50 minutes for PT
- 3 x (3x3) minutes for WBV

**Weekly:**
- 3 x 40 minutes for BWSTT and RT
- 2 x 30 minutes for aquatic therapy
Interventions

1. Whole Body Vibration
2. Resistance Training
3. Isometrics
4. Body-Weight Supported Treadmill Training
5. Aquatic Therapy
6. Neurodevelopment Treatment
Conclusion(s)

• Collagen disorder cannot be treated, reversed

• Rehabilitation goals:
  – Optimal functional ability
  – Maximize level of ambulation (community walking)
  – Learn and utilize compensatory strategies for ADLs
Areas for Future Research

• Recruit and utilize larger cohorts in prospective studies dealing with:
  – Type-specific OI & associated therapy efforts
  – Experimentation of therapeutic frequencies and intensities
  – Whole body vibration approach

• Investigate anatomic basilar impression: development, effects, and treatments strategies
Any Questions?

