Alice Rose Boyd
Born May 10, 2010 at
7:07 am
Weighed 7lbs 8 oz.
General Information

- "Osteogenesis Imperfecta" means imperfect bone formation.

- Group of rare genetic defects that affects the body’s production of collagen
  - Collagen is main protein in connective tissue

- General Characteristics:
  - Fragile or brittle bones
  - Weak muscles
  - Loose ligaments

- Four Types:
  - Types I-IV are most common
  - Recently added 4 more classifications (V-VII)
General Information

- No known cause
  - Autosomal dominant trait

- Affects 1 in 20,000-60,000 births

- Affects males & females equally

- Estimates of those affected in US are unknown
  - Best estimate is 20,000-50,000
  - No known cure
## OI Types & Characteristics:

<table>
<thead>
<tr>
<th>Type</th>
<th>Clinical Severity</th>
<th>Typical Features</th>
<th>Sclera</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Mild non-deforming</td>
<td>Normal height, mild/short stature</td>
<td>Blue</td>
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<tr>
<td>II</td>
<td>Perinatal Lethal</td>
<td>Multiple rib &amp; long bone fracture at birth</td>
<td>Blue</td>
</tr>
<tr>
<td>III</td>
<td>Severe deformity</td>
<td>Very short, triangular face, severe scoliosis</td>
<td>White (may be blue at birth)</td>
</tr>
<tr>
<td>IV</td>
<td>Moderate deformity</td>
<td>Moderately short, mild to moderate scoliosis</td>
<td>White (may be blue at birth)</td>
</tr>
</tbody>
</table>
Diagnosis:

- May have biopsy or DNA tests to determine presence of OI
  - Test detects 90% of cases
  - May have prenatal tests & counseling

- Besides tests, doctor also diagnose by:
  - Family history
  - Medical history
  - Results from a physical exam
  - X rays
Prognosis:

- Varies depending on number & severity of symptoms

- Despite numerous fractures, restricted activity, and short stature, most adults and children with OI (except Type II) lead productive and successful lives
How is OI managed?

- Caregivers education on safe handling techniques and protective positioning
- Awareness that infancy, early childhood, and pre-teen years pose to be the most challenging
- Growth and hormonal changes affect frequency of fractures
- Learn activities to avoid and how to conserve energy
- Adhere to a healthy lifestyle
Interventions:

- Physical therapy
- Care for broken bones & brittle teeth
- Pain medication
- Use of wheelchairs, braces, and other aids
Interventions:

- Physical therapy (or hydrotherapy)
  - strengthen muscles and improve mobility in a gentle manner

- Bisphosphonates (oral or intravenous)
  - increase bone mass and reduce the incidence of fracture
    - Alendronate (Merk)-oral
    - Risendronate – oral
    - Pamidronate- (Novartis)- intravenous

- Surgery
  - Insertion of metal rods in long bones to improve strength
Why we chose this topic

- Prior exposure
In patients with Osteogenesis Imperfecta, does the use of weight bearing exercise alone provide a more effective method to increase bone mass in comparison to the use of intravenous medication?
Methods:

- **Ebsco Host:**
  - Searched: physical therapy + osteogenesis imperfecta
    - Found 22 articles, but 2 were relevant to study
  - Searched: medication + osteogenesis imperfecta
    - Found 137 articles, but 2 were relevant to study

- **Google Scholar:**
  - Searched: physical therapy + osteogenesis imperfecta
    - Found 63,3000 articles, but 2 were relevant to study
  - Searched: osteogenesis imperfecta + treatment with medication
    - Found 116,000 articles, but 2 were chosen and relevant to study
Research Synthesized: Physical Therapy

- Rehabilitation of children with severe OI
- Describe common patterns of disability
- Rehabilitation intervention strategies
- The effects of a physical training:
  - exercise capacity
  - muscle force
  - subjective fatigue levels
Discussion (PT):

- Independent ambulation, but:
  - poor joint alignment
  - poor balance
  - deviated gait patterns
  - low endurance.

- All these conditions can be improved with persistent, individualized physical and occupational therapy.

- A significant improvement in aerobic capacity, muscle force and subjective fatigue was found after 3 months of training.
DISCUSSION (PT)continued

- Children with OI can participate: safely and effectively in a supervised and individual tailored training program

- A long-term benefit depends on the continuation of training sessions into adult life

- To achieve the maximum benefit, therapy should begin as soon as possible after birth
RESULTS (PT)

- Shoulder ROM improved
- LE fractures continued
- Hip flexion and abduction contractures persisted
- Improvement in Torticollis
- Highest functional achievement (Group A): few able to sit or stand w/o support
RESULTS (PT) continued

- Highest functional achievement (Group B) –
  - All ambulatory in braces
  - Two were ambulating w/o braces.
  - One child did not require assistive device

- Study from the Journal of Pediatrics:
  No fractures occurred in the intervention group but control group had 3 fractures

- Significant improvement in 3 months:
  - Aerobic capacity
  - Muscle force
  - Subjective fatigue
INTERVENTIONS (PT)

- Posture Exercises
  - Positioning
  - Soft tissue mobilizations
  - Active low back stretch
  - Standing in posterior tilt against wall

- Development exercises
  - Prone/sitting progression
  - Standing/Ambulation training

- Active ROM exercises and strengthening
  - UE/LE active resistive exercises w/weights
  - Stretching of hip abductors and Achilles tendons
  - Swimming, cycling

- Coordination exercises
Factors to Consider

- Fracture History
- Joint alignment
- Exercise tolerance
- Stamina
- Degree of muscle weakness
- Joint stiffness or laxity
Area of Future Research (Rehab)

- Focus on the perceived barriers: participate in physical exercise in unsupervised safe and effective environments.

- Focus on management of OI, should address the child’s functional needs.
Bisphosphonates:

- Mechanism of action: inhibitory affect on osteoclasts

Intravenously administered meds -
- Pamidronate- (Aredia, Novartis)- slowly infused over 4 hour period every 120 days

Orally administered meds –
- Alendronate (Fosamax, Merk)- 1 mg/kg daily
- Risendronate-

**not ideal treatment for younger pts due to**
- difficulty swallowing pills
- pills unable to be crushed b/c solution is caustic
- can cause moderate to severe gastric irritation
Results: Medication

Significant results: due to the inhibitory effect that medications have on osteoclasts

- Increased bone mineral density
  - Assessed via DXA scan
- Decreased biomarkers of bone turnover
- Moderate increases in linear bone growth
- Improved ambulation after two years of treatment
Discussion: Medication

• Although research indicates significant results from Bisphosphonates alone, treatment alongside physical therapy, occupational therapy, and surgery to correct skeletal deformities, has implications of likely being the best course of action when treating patients with osteogenesis imperfecta.

• Whyte et al 2003 published a report of a child who, after receiving very high doses of bisphosphonate over a long period of time, developed an osteoporotic-like condition. This report highlights the need for careful monitoring of children receiving these types of therapy.
REFERENCES


The Many Faces of OI
Area of Future Research (Rehab)

- Focus on the perceived barriers: participate in physical exercise in unsupervised safe and effective environments.
- Focus on management of OI, should address the child's functional needs.
Sources

- Genetics Home Reference
- Healthfinder
- MedlinePlus-Osteogenesis Imperfecta
- National Institute of Arthritis, Musculoskeletal, & Skin Diseases
- Osteogenesis Imperfecta
- The Osteogenesis Imperfecta Clinic at Kennedy Krieger Institute
- Osteogenesis Imperfecta Foundation
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