Osteogenesis Imperfecta

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OSTEO - GENESIS

Bone Disease

Genetics

IMPERFECTA

Inadequacy
What is this Disease?

- “Brittle Bone Disease”
- Genetically acquired disease
- Inadequate bone development
- Very rare: 1 in 20,000 people
- 3 Stages of development
  1. Prenatal/Neonatal
  2. Preschool/Childhood
  3. Adolescence/Adulthood
What are the Symptoms?

- Malformed Bones
- Short, small body
- Loose joints
- Blue-ish tone in the whites of the eyes
- Curved spine
- Weak teeth
- Breathing problems
- Hearing Loss
- Stunted growth
What are the Causes?

- Strictly genetic disorder that incorporates multiple genes
- Basic Defect: qualitative or quantitative lack of collagen
  - **Collagen**: The specific material that strengthens the bone
- Can be inherited from one parent or from both
Different Forms

Quantitative:
- Mutation on the COL1A gene
- Premature stop of codon
- Milder form of the disease

Qualitative:
- Chain of mutations on COL1A or COL1B gene
- Collagen formed inadequately

Type Classification:
- Total of 8 types
  - MILD: Type 1
  - MODERATE: Type 4, 5, 6
  - EXTREME: Type 2, 3, 7, 8
How do you Diagnose?

- No one specific test, series of tests performed
  - Family History
  - Medical History
  - X-Rays: bone malformation visible
  - Physical exam
  - Collagen Test: blood or skin test
- DNA testing accuracy is **60-94%**
- Prenatal mutation analysis through ultrasonographic assistance
What is the Treatment?

- NO CURE
- Treatment specific for age
- Orthotics: stabilize joints
- Walking aid, wheelchair, brace: increase mobility
- Physical therapy
- Surgery
  - Intramedullary Rod Placement
  - **Positives:** increased strength
  - **Risks:** breakage, rotational deformities, migration
- Maintain health through diet and exercise
Future Research

- **Gene Therapy**: Understand expression of the mutation
- Create an approved medication specified for the disease
- Innovative devices to replace the surgical methods
New Development: Bisphosphonates
- New drug class to increase bone density and mass

SPECIFIC CASE (Birmingham Hospital)

Background
- 9 year old patient: family history
- Recurrent fractures, low back pain, tenderness in spine
- Wheelchair bound

Testing
- Radiography signaled collapse of vertebrae
- Bone density = 0.395 g/cm²

Treatment
- Intravenously delivered bisphosphonates
- Bone density = 0.709 g/cm² in 18 months
- 44% increase in density and strength
- No adverse effects or deformity
- Wheelchair for long distances
QUESTIONS?


