Rickets
an overview: Feb 7, 2007

Dr. Ayeni & Dr. Peterson
Rickets

• Definition: Metabolic Bone Disease resulting in abnormal mineralization of the growing skeleton

• Cause: inadequate levels of calcium or phosphorous (both) for mineralization
## Causes of Osteomalacia

<table>
<thead>
<tr>
<th>Etiology</th>
<th>Ca</th>
<th>PO4</th>
<th>25-D</th>
<th>1,25-D</th>
<th>PTH</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Vitamin D abnormalities</strong></td>
<td></td>
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<tr>
<td>D-deficiency</td>
<td>↓</td>
<td>↓</td>
<td>↓</td>
<td>±↓</td>
<td>↑</td>
<td>↑ Alk Phos</td>
</tr>
<tr>
<td>Liver disease</td>
<td>↓</td>
<td>↓</td>
<td>↓</td>
<td>↓</td>
<td>N</td>
<td>↑ Alk Phos</td>
</tr>
<tr>
<td>Renal disease</td>
<td>↓</td>
<td>↑</td>
<td>N</td>
<td>↓</td>
<td>↑↑</td>
<td>↑ Alk Phos</td>
</tr>
<tr>
<td>1α hydroxylase deficiency</td>
<td>↓</td>
<td>↓</td>
<td>N</td>
<td>↓</td>
<td>↑</td>
<td>↑ Alk Phos</td>
</tr>
<tr>
<td>“vitamin D dependent rickets” type I</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Vitamin D Resistance</td>
<td>↓</td>
<td>↓</td>
<td>N</td>
<td>↑↑</td>
<td>↑</td>
<td>↑ Alk Phos</td>
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<tr>
<td>“vitamin D dependent rickets type II”</td>
<td></td>
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<tr>
<td><strong>Hypophosphatemia</strong></td>
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<tr>
<td>X-linked hypophosphatemic rickets “Vitamin D Resistant Rickets”</td>
<td>±↓</td>
<td>↓</td>
<td>N</td>
<td>N</td>
<td>±↑</td>
<td>1,25-D inappropriately normal</td>
</tr>
<tr>
<td>Autosomal dominant hypophosphatemic rickets</td>
<td>±↓</td>
<td>↓</td>
<td>N</td>
<td>N</td>
<td>±↑</td>
<td></td>
</tr>
<tr>
<td>Renal phosphate loss</td>
<td>N</td>
<td>↓</td>
<td>N</td>
<td>N</td>
<td>N</td>
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<tr>
<td>(including Fanconi’s syndrome, Dent’s disease, cadmium toxicity, heavy metal poisoning)</td>
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<tr>
<td>Excessive antacid intake</td>
<td>N</td>
<td>↓</td>
<td>N</td>
<td>±↑</td>
<td>N</td>
<td></td>
</tr>
<tr>
<td>HHRH “Hereditary hypophosphatemic rickets with hypercalciuria”</td>
<td>N</td>
<td>↓</td>
<td>N</td>
<td>↑</td>
<td>N</td>
<td>↑ urine calcium</td>
</tr>
<tr>
<td><strong>Toxicities</strong></td>
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<tr>
<td>Fluoride</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
</tr>
<tr>
<td>Etidronate</td>
<td>N</td>
<td>±↑</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td></td>
</tr>
<tr>
<td>Parenteral Aluminum</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>±↓</td>
<td>↓</td>
<td></td>
</tr>
<tr>
<td>Imatinab</td>
<td>±↓</td>
<td>↓</td>
<td>±↓</td>
<td>N</td>
<td>↑</td>
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<tr>
<td><strong>Other</strong></td>
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<tr>
<td>Hypophosphatasia</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>↓↓ Alk Phos</td>
</tr>
<tr>
<td>Acidosis</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Oncogenic osteomalacia</td>
<td>±↓</td>
<td>↓</td>
<td>N</td>
<td>↓</td>
<td>±↑</td>
<td>↑ Alk Phos</td>
</tr>
</tbody>
</table>

N = normal, ± means abnormality seen sometimes, Alk Phos = alkaline phosphatase.
Causes: Lovell & Winter

- Deficiency Diseases (Vit D, Ca, Phos)
- Gastrointestinal disorders
- Vitamin D - resistant rickets
- Renal Osteodystrophy
- Unusual forms
Vitamin D metabolism

7-dehydrocholesterol (Diet) → Cholecalciferol (Skin) → Bound Cholecalciferol (Blood) → 25-hydroxycholecalciferol (Liver) → 1,25-dihydroxycholecalciferol (Kidneys) → Ca Absorption (GIT)
Metabolism

Bone

\[ \text{Ca}^{2+} \text{1.0 kg} \]

\[ \text{Ca}^{2+} \text{0.5 g / Day} \]

\[ \text{Ca}^{2+} \text{9.8 g / Day} \]

\[ \text{Ca}^{2+} \text{10.0 g / Day} \]

Intestine

\[ \text{Ca}^{2+} \text{1.0 g / Day} \]

\[ \text{Ca}^{2+} \text{0.2 g / Day} \]

\[ \text{Ca}^{2+} \text{0.8 g / Day} \]

Kidney

\[ \text{Urine Ca}^{2+} \text{0.2g / Day} \]
continued
Clinical Findings

• Irritable/hypo kinetic

• Skull: prominent suture lines, softening

• Axial skeleton: pectus carinatum, prominent costochondral junction

• Small Joints: enlarged

• Long bones: bowing & fractures
Clinical Findings
Radiology

- Irregular cupping or widening of physis
- Increased height of physis
- Looser lines: pseudo fractures, transverse in complete, radiolucency on concave side of long bone (especially rib, clavicle scapula)
- Thin cortices, indistinct trabeculae
Radiology
Histology

- Zone of maturation is altered: disordered increase in zone of hypertrophy
- Poorly defined zone of calcification
- Tongues of viable cartilage not replaced by bone
- Altered blood supply (decreased)
- Poor mineralization
Differential Diagnosis (tibia vara)

- Renal Osteodystrophy
- Osteogenesis Imperfecta
- Achondroplasia
- Focal Fibro cartilaginous dysplasia
- Metaphyseal Chondrolysis
AAOS: Metabolic Work Up

- Hx + PE
- X-rays (spine pelvis, extremities)
- AP & lateral chest
- Bone scan
- **Big 10 labs**: CBC, Lytes, Bun & Cr, Glucose, Calcium, Phosphorous, Alkaline phosphatase, TSH, Thyroxine
- Serum Immuno electrophoresis, Urinalysis
Treatment

• Multidisciplinary approach
• Treating underlying cause usually addresses deformity
• Involve pediatrician, endocrinologist, gastroenterologist, nephrologist
• Residual bony deformity maybe addressed with surgery.
Treatment

• Oral phosphate, vitamin D, Calcium supplementation +/- growth hormone
• American Academy of Pediatrics 2003: 200 IU Vit D daily
• Start before 2 months of age and continue till adolescence
Orthopaedic Management

• No algorithm…numerous options!
• Including: trial of bracing, proximal tibial metaphyseal osteotomy, dome osteotomy and fixation, corrective osteotomy with intramedullary fixation, physeal stapling, osteotomy and external fixation, multiplanar fixation devices…
• Timing variable!
Retrospective series of 20 pts (11 adults)
Internal and/or external fixation
Correction of 55 segmental deformities
Complications: 18/13 (DO), 13/10 (AC)
Retrospective series of 10 pts
4.5-12 year follow up
4 good, 3 fair, 2 poor, 1 lost to Sx
Major complication: staple loosening & migration
3 initially short children with X-linked hypophosphatemic rickets treated with growth hormone

At adulthood: exceeded predicted height by 6.2 cm

But more truncal than leg length growth