

Child With Suspected Rickets

Suggestive history and physical findings	Initial laboratory and/or radiologic work-up can include:	When to refer	Items useful for consultation	Additional information
<p>Symptoms/Signs: Short stature, bowed legs Delay in walking, muscle pain, possible seizures if low calcium</p> <p>Dietary history: exclusive breast fed infant with no vitamin D supplementation, low calcium intake</p> <p>Physical findings: Broadening of wrists/ankles, rachitic rosary, prominent forehead, bowlegs, Harrison's groove</p> <p>Alopecia is seen in the rare hereditary vitamin D resistant rickets due to mutation in vitamin D receptor</p> <p>Differential Diagnosis</p>	<p>Blood tests:</p> <ul style="list-style-type: none"> • Total and ionized calcium • Albumin • Phosphorus • Magnesium • Alkaline phosphatase • Intact PTH • 25-OH Vitamin D • 1, 25-dihydroxy-vitamin D <p>X-ray:</p> <ul style="list-style-type: none"> • X-ray of wrists of knees 	<p>Urgent: if seizures present, check calcium level and admit for IV calcium if needed</p> <p>Routine: Ideally the child should be seen by a specialist soon after the diagnosis is suspected and before treatment is begun so an accurate diagnosis can be made.</p> <p>Prompt diagnosis and treatment is indicated to prevent compromise of adult stature in rare inherited causes of rickets.</p> <p>Find a Pediatric Endocrinologist</p>	<p>Previous growth data/growth charts</p> <p>Pertinent medical records</p> <p>Recent laboratory and radiologic studies</p>	<p>Additional Information</p> <p>Vitamin D Deficiency and Rickets: A Guide for Families</p> <p>References</p>

Differential Diagnosis:

Optimal mineralization of bone requires optimal concentration of calcium, phosphorous, and vitamin D. Deficiency of any of these three can result in rickets. The main causes of rickets can be differentiated to acquired and hereditary causes.

- Acquired causes:
 - Nutritional vitamin D deficiency: This is most often seen in the setting of an exclusive breast fed infant who is not receiving any vitamin D supplementation. Breast milk is a poor source of vitamin D. AAP recommends supplementing all breast fed infants with 400IU of vitamin D daily. Dark colored skin limits the production of vitamin D by the skin. Similarly, high altitude or cold temperatures will limit body's production of endogenous vitamin D from UVB radiation due to limited sun exposure.
 - Liver disease: Severe liver disease limits the production of 25-hydroxyvitamin D which is substrate for the active 1, 25-dihydroxyvitamin D.
 - Kidney disease: Severe kidney disease limits the activity of 1 α -hydroxylase enzyme which converts 25-hydroxyvitamin D to active 1, 25-dihydroxyvitamin D.
- Hereditary causes:
 - 1 α -hydroxylase deficiency: Mutation in the 1 α -hydroxylase gene leading to defective or absent 1 α -hydroxylase leads to rickets which responds to calcitriol treatment.
 - Hereditary vitamin D resistant rickets: This rare cause of rickets is caused by mutation in vitamin D receptor. This is an autosomal recessive condition which is very rare with only around 100 cases reported worldwide. Children with this condition can have alopecia.
 - Hereditary hypophosphatemic rickets. In this condition, there is a defect in phosphorous reabsorption in the kidney. This can be isolated to phosphorous reabsorption or can be a part of generalized renal tubular reabsorption disorder. Most cases of isolated phosphorus reabsorption disorder are inherited in an X-linked dominant fashion but autosomal recessive and autosomal dominant inheritance has also been described. Children with hypophosphatemic rickets do not typically have hypocalcemia. Treatment consists of supplementation with phosphorous and calcitriol.

Additional Information:

- Rickets is defective mineralization of growing bone.
- Classic radiologic features of rickets are: fraying, splaying and widening of the metaphysis.
- Biochemically, rickets is characterized by elevated alkaline phosphatase.

Treatment: Therapy depends on etiology. Prompt work up and treatment is recommended to prevent compromise of adult stature.

Suggested References and Additional Reading:

- Elder CJ, Bishop NJ. Rickets. Lancet, 2014; 383: 1665-76.
- Misra M, et al. Vitamin D deficiency in children and its management: review of current knowledge and recommendations. 2008; 122: 398-417.

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