

*A rapid, convenient way  
to diagnose hypophosphatemic rickets.*

# Hypophosphatemic Rickets Evaluation

## *What is Hypophosphatemic Rickets?*

**Children with hypophosphatemic rickets show renal phosphate wasting and osteomalacia, which lead to bone deformities and stunted growth.<sup>1</sup>**

- Causes of hypophosphatemic rickets include:
  - X-linked dominant loss-of-function mutations in the gene *PHEX*.<sup>2</sup>
  - autosomal dominant gain-of-function mutations in the gene *FGF23*.<sup>3</sup>
  - tumors (mostly benign) of mesenchymal origin.<sup>4</sup>
- More than 20% of genetic hypophosphatemic rickets cases are due to sporadic mutations, so patients may not have a family history.<sup>5</sup>

## *Why genetic testing?*

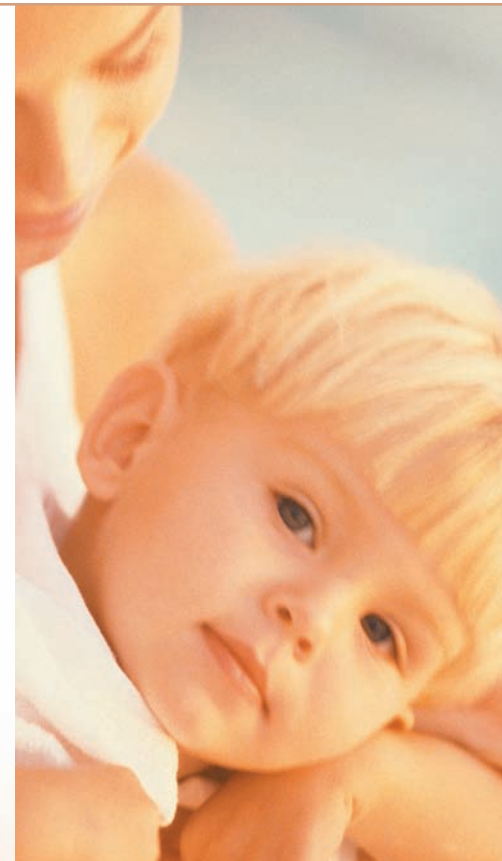
**Genetic testing for mutations in *PHEX* or *FGF23* associated with hypophosphatemic rickets may**

- Permit diagnosis of hypophosphatemic rickets based on a single blood draw in approximately 80% of cases.<sup>6</sup>
- Allow diagnosis of hypophosphatemic rickets in infants.
  - Studies show significantly improved treatment outcomes if therapy is started in infancy.<sup>7</sup>
- Help discriminate between genetic and tumor-induced hypophosphatemic rickets or osteomalacia.
- Differentiate between X-linked and autosomal dominant hypophosphatemic rickets, allowing more precise genetic counseling.
- Facilitate family testing, allowing earlier diagnosis and immediate treatment of affected family members.

## *Indications for testing:*

- Progressive bowing of legs in a child more than 18 months of age with normal vitamin D levels and marked hypophosphatemia
- Hypophosphatemia and hyperphosphaturia in the presence of inappropriately normal vitamin D levels at any age
- Family history of hypophosphatemic rickets or hypophosphatemic osteomalacia

*For complete ordering information,  
please see the reverse side.*



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 **athena diagnostics®**

*Testing You Can Count On.*

**References:** 1. Carpenter TO (1997) *Pediatric Endocrinology* 44:443-66. 2. The HYP Consortium (1995) *Nat Genet* 11:130-6. 3. ADHR Consortium (2000) *Nat Genet* 26:345-8. 4. Carpenter TO (2003) *N Engl J Med* 348:1705-8. 5. Dixon PH, et al (1998) *J Clin Endocrinol Metab* 83:3615-23. 6. Jan de Beur SM, Levine MA (2002) *J Clin Endocrinol Metab* 87:2467-73. 7. Makitie O, et al (2003) *J Clin Endocrinol Metab* 88:3591-7.

For a brief review on Hypophosphatemic Rickets, please visit [www.athenadiagnostics.com](http://www.athenadiagnostics.com).



## Hypophosphatemic Rickets Evaluation

**Typical Presentation:** Hypophosphatemia and osteomalacia; bone deformities and short stature in cases of childhood onset

**Synonyms:** Vitamin D resistant rickets

**Indications for Testing:**

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### TEST DETAILS

**Test Code:** 857

**Test Includes:** *PHEX* (Hypophosphatemic Rickets) DNA Sequencing Test, #855  
*FGF23* (Hypophosphatemic Rickets) DNA Sequencing Test, #856

**Test Turnaround:** 14-21 days

### TECHNICAL INFORMATION

**Methodology:** Polymerase Chain Reaction (PCR),  
DNA sequencing of entire protein coding region of gene

### SHIPPING CONSIDERATIONS

**Specimen Type:** Whole blood

**Volume:** 10 mL (pediatric minimum: 2 mL)

**Collection Tube:** Yellow or lavender top

**Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs

**Storage Conditions:** For short periods (until shipped) at 4°C

**Shipping Conditions:** Overnight at room temperature (specimen arrival must be less than 24 hrs after collection);  
ship Monday through Thursday only

To order the Hypophosphatemic Rickets Evaluation (Test #857), call Athena Diagnostics' Customer Service Representatives at:

**800-394-4493 x2**



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