

Identifying the cause of gonadotropin-independent precocious puberty in boys.

Male Precocious Puberty (LHCGR) Evaluation

- Precocious puberty can lead to emotional and psychosocial problems in affected children and is associated with significantly reduced adult height.¹
- Gonadotropin-independent precocious puberty in boys is a heterogenous disorder with diverse causes, such as
 - testosterone or human chorionic-gonadotropin producing tumors
 - congenital adrenal hyperplasia
 - McCune-Albright Syndrome
 - familial male-limited precocious puberty (FMPP) due to autosomal dominant gain-of-function mutations in *LHCGR*, the gene coding for the luteinizing hormone chorionic gonadotropin receptor.^{2,3}
- Identifying the underlying cause of gonadotropin-independent precocious puberty is important, since it may help determine the treatment:
 - tumors are typically excised
 - congenital adrenal hyperplasia is managed with cortisol replacement
 - FMPP and McCune-Albright Syndrome can be treated with ketoconazole or combined therapy with testolactone and spironolactone.^{4,5}

Why genetic testing?

Genetic testing for FMPP-associated mutations in *LHCGR* can allow:

- Definitive diagnosis of FMPP, helping to select the most appropriate treatment.
- Identification of asymptomatic female carriers of FMPP-associated mutations in *LHCGR* whose sons have a 50% risk of developing FMPP.
- Detection of a predisposition for FMPP in asymptomatic male infants. This would prompt increased vigilance for symptoms of FMPP and allow early intervention.

Indications for testing:

- Gonadotropin-independent precocious puberty in boys
- Family history of male-limited precocious puberty

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



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References: 1. Root AW, (2000) *Pediatr Rev* 21:10-19. 2. Shenker A, et al., (1993) *Nature* 365:652-4. 3. Kremer H, et al., (1993) *Hum Mol Genet* 2:1779-83. 4. Soriano-Guillen L, et al., (2005) *J Clin Endocrinol Metab* 90:147-51. 5. Leschek EW, et al., (1999) *J Clin Endocrinol Metab* 84:175-178.

For a brief review on Gonadotropin-Independent Precocious Puberty in Boys, please visit www.athenadiagnostics.com/DR



www.athenadiagnostics.com

Male Precocious Puberty (*LHCGR*) Evaluation

Typical Presentation:	Appearance of secondary sexual characteristics in boys age two or three
Synonyms:	Testitoxiosis Familial Male-Limited Precocious Puberty (FMPP)
Indications for Testing:	<ul style="list-style-type: none">• Gonadotropin-independent precocious puberty in boys• Family history of male-limited precocious puberty

TEST DETAILS

Test Code:	817
Test Turnaround:	14-21 days

TECHNICAL INFORMATION

Methodology:	Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region
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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 Male Precocious Puberty (*LHCGR*) Evaluation (Test #817), call Athena Diagnostics' Customer Service Representatives at:

800-394-4493 x2



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