

Identify children at high risk for end organ damage from hypertension.

Endocrine Hypertension (HSD11B2) Evaluation

What is Apparent Mineralocorticoid Excess (AME)?

- AME is characterized by early-onset hypertension, hypokalemia, and metabolic alkalosis in the presence of low plasma renin activity and low serum aldosterone and often leads to nephropathy, retinopathy, neuropathy, cardiovascular disease, or even death at a young age.¹
- AME can be effectively treated, allowing prevention or even reversion of end-organ damage.¹
- AME is caused by autosomal recessive loss-of-function mutations in the gene *HSD11B2*, which codes for the enzyme 11- β -hydroxysteroid dehydrogenase type 2.² Reduced activity of this enzyme allows cortisol to activate the mineralocorticoid receptor and thus increase renal sodium re-absorption in a renin-angiotensin independent manner.¹

Why genetic testing?

Genetic testing for mutations in *HSD11B2* associated with AME

- Permits fast and accurate diagnosis of AME without the need for 24-hour urine samples.
- Allows timely initiation of targeted therapy with spironolactone, which can prevent or even reverse potentially fatal end-organ damage.¹
- Can detect carriers of AME, who may be at increased risk for late-onset hypertension.^{3,4}

Indications for testing:

- Persistent low-renin hypertension in young children
- Family history of AME

For complete ordering information, please see the reverse side.

References: 1. Dave-Sharma S et al (1998) *J Clin Endocrinol Metab* 83:2244-54. 2. Mune T et al. (1995) *Nat Genet* 10(4):394-9. 3. Lavery GG et al (2003) *Hypertension* 42:123-9. 4. Li A et al. (1997) *J Hypertens* 15:1397-402.

For a brief review on Endocrine Hypertension, please visit www.athendiagnosics.com/DR



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Testing You Can Count On.

Endocrine Hypertension (*HSD11B2*) Evaluation *Apparent Mineralocorticoid Excess*

- Typical Presentation:** Persistent low-renin hypertension with associated hypokalemia
- Indications for Testing:**
- Persistent low-renin hypertension in young children
 - Family history of AME

TEST DETAILS

- Test Code:** 881
- Test Turnaround:** 14-21 days

TECHNICAL INFORMATION

- Methodology:** Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of gene
- Patents:** United States Patent No. 5,883,240

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

www.athenadiagnostics.com

Call Athena Diagnostics' Customer Service Representatives to order the Endocrine Hypertension (*HSD11B2*) Evaluation (Test #881) at:

800-394-4493 x2



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