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Congenital Adrenal Hyperplasia

*CYP21A2, CYP11B1
STAR, CYP17A1
HSD3B2*



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Testing You Can Count On.

Diagnosing the cause of ambiguous genitalia, primary adrenal insufficiency, and/or endocrine hypertension in infants and children.

Congenital Adrenal Hyperplasia (CAH)¹⁻³

- CAH is a family of disorders caused by enzymatic defects in adrenal steroid biosynthesis.
- The presentation and treatment of CAH depends on the biosynthetic pathway(s) affected, the severity of the enzymatic defect, and whether gonadal steroidogenesis is also affected.
- Autosomal recessive loss-of-function mutations in any one of at least five genes have been associated with various forms of CAH:
 - 21-hydroxylase deficiency due to mutations in *CYP21A2* is the most frequent form of CAH. Elevated 17-hydroxyprogesterone levels, often first detected on a newborn screen, may be indicative of 21-hydroxylase deficiency.
 - Other forms of CAH include:
 - 11 β -hydroxylase deficiency due to mutations in *CYP11B1*.
 - 17 α -hydroxylase deficiency due to mutations in *CYP17A1*.
 - 3 β -hydroxysteroid dehydrogenase deficiency due to mutations in *HSD3B2*.
 - Lipoid CAH due to mutation in *STAR*.

Symptoms Associated with Genetic Causes of CAH

Gene	Protein	Form of CAH	External Genitalia	Glucocorticoid Activity	Mineralocorticoid Activity	Hirsutism In Women
<i>CYP21A2</i>	21-hydroxylase	Classic, salt-wasting	Ambiguous in XX	Deficient	Deficient	Yes
		Classic, simple virilizing	Ambiguous in XX	Deficient	Clinically Normal	Yes
		Non-classic	Normal	Not deficient	Not deficient	Yes
<i>CYP11B1</i>	11 β -hydroxylase	Classic	Ambiguous in XX	Clinically Normal	Excessive or Clinically Normal	Yes
		Non-classic	Normal	Clinically Normal	Clinically Normal	Yes
<i>CYP17A1</i>	17 α -hydroxylase		Female or Ambiguous in XY	Clinically Normal	Excessive or, Rarely, Clinically Normal	No
<i>HSD3B2</i>	3 β -hydroxysteroid dehydrogenase	Salt-wasting (Classic)	Ambiguous in XY, Ambiguous or Normal in XX	Deficient	Deficient	Yes
		Non-salt-wasting (Non-classic)	Ambiguous in XY, Ambiguous or Normal in XX	Clinically Normal	Clinically Normal	Probably
<i>STAR</i>	Steroid acute regulatory protein	Lipoid CAH	Female in XY	Deficient	Deficient	No

Athena Diagnostics Offers Full Sequence Analysis of the CYP21A2, CYP11B1, STAR, CYP17A1, and HSD3B2 Genes.

Why genetic testing?

Genetic testing for CAH may:

- Confirm a diagnosis of CAH.
 - Genetic testing may be more accurate than biochemical testing in mild cases of CAH or in carriers, where elevations in steroid hormone precursors can be subtle.
- Allow a differential diagnosis of the various types of CAH, helping to guide treatment.
 - Glucocorticoid replacement therapy is used for all forms of CAH.
 - Patients with the salt-wasting forms of CAH (21-hydroxylase deficiency, 3 β -hydroxysteroid dehydrogenase deficiency, or lipoid CAH) may also require mineralocorticoid replacement.
 - Estrogen replacement therapy may be indicated in phenotypically female individuals with lipoid CAH or 17 α -hydroxylase deficiency.
- Identify carriers of CAH-associated mutations, facilitating genetic counseling and the diagnosis of CAH in family members of patients.

Why Choose Athena Diagnostics?

- Athena is your single source for a wide variety of molecular diagnostics for endocrine disorders.
- Athena Diagnostics offers full sequencing for 5 genes associated with congenital adrenal hyperplasia: *CYP21A2*, *CYP11B1*, *CYP17A1*, *HSD3B2*, and *STAR*, as well as detection of the common 30kb deletion in *CYP21A2*, to help identify the cause of your patient's symptoms.
- Athena offers consultation with lab directors or genetic counselor for assistance in result report interpretation.
- Athena provides phlebotomy services through a partner hospital or home draw service to help patients gain access to testing.

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



Athena Diagnostics Genetic Testing for CAH

TEST DETAILS

CAH (CYP21A2, CYP11B1) Evaluation #879:

- Indications for Testing:**
- Ambiguous genitalia in infants
 - Premature adrenarche
 - Hirsutism and/or oligomenorrhea in women
 - Family history of congenital adrenal hyperplasia

CYP21A2 (CAH) Evaluation #880:

- Indications for Testing:**
- Elevated 17-hydroxyprogesterone levels
 - Primary Adrenal Insufficiency in infants
 - Family history of 21-hydroxylase deficiency

CYP11B1 (CAH) DNA Sequencing Test #875:

- Indications for Testing:**
- Elevated levels of deoxycorticosterone or 11-deoxycortisol
 - Ambiguous genitalia in infants with hypertension
 - Family history of 11 β -hydroxylase deficiency

CYP17A1 (CAH) DNA Sequencing Test #877:

- Indications for Testing:**
- Ambiguous genitalia in 46XY infants, after exclusion of androgen insensitivity syndrome
 - Lack of pubertal progression and primary amenorrhea in phenotypic females
 - Low-renin hypertension and/or elevated deoxycorticosterone and corticosterone levels in children with a personal or family history of ambiguous genitalia, 46XY DSD, or lack of pubertal progression and primary amenorrhea in phenotypic females
 - Family history of 17 α -hydroxylase deficiency

HSD3B2 (CAH) DNA Sequencing Test #878:

- Indications for Testing:**
- Undervirilization in 46XY or ambiguous genitalia in 46XX infants
 - Primary adrenal insufficiency in infants, after exclusion of 21-hydroxylase deficiency
 - Premature adrenarche, after exclusion of 21-hydroxylase and 11 β -hydroxylase deficiency
 - Elevated pregnenolone, 17OH-pregnenolone and/or dehydroepiandrosterone levels
 - Family history of 3 β -HSD deficiency

Lipoid CAH (STAR) Evaluation #874:

- Indications for Testing:**
- Primary adrenal insufficiency in phenotypically female infants
 - Family history of congenital lipoid adrenal hyperplasia

TECHNICAL INFORMATION AND SHIPPING CONSIDERATIONS

Methodology:	Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of gene; for CYP21A2: screening for 30kb-deletion-specific PCR product.
Test Turnaround:	14-21 days
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 genetic testing for Congenital Adrenal Hyperplasia, call Athena Diagnostics Customer Service Representatives at:

800-394-4493 x2



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