MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: August 30, 2013
EFFECTIVE DATE: October 1, 2013

21-HYDROXYLASE GENE (CYP21A2), FULL GENE ANALYSIS
Test ID: CYPPS

USEFUL FOR:
- Third-tier confirmatory testing of positive congenital adrenal hyperplasia (CAH) newborn screens
- An adjunct to measurement of basal and adrenocorticotropic hormone stimulated 17-hydroxyprogesterone, androstenedione, and other adrenal steroid levels in the diagnosis of atypical or nonclassical cases of CAH
- Carrier detection of *CYP21A2* mutations and genetic counseling

PROFILE INFORMATION:

Test ID	Reporting Name	Available Separately	Always Performed
CYPMS	CYP21A2 Full Gene Analysis	No	Yes

TESTING ALGORITHM: When this test is ordered, *CYP21A2* full gene analysis (amplification), *CYP21A2* gene sequencing, and *CYP21A2* Deletion Detection will always be performed.

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing and Deletion Detection by Multiplex Ligation-Dependent Probe Amplification (MLPA)

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Minimum Volume: 0.2 mL
Collection Instructions: Send specimen in original tube.
Forms:
1. *CYP21A2* Gene Testing Patient Information Sheet (Supply T663) in Special Instructions
2. **New York Clients-Informed consent is required.** Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature
Whole Blood EDTA	Refrigerated (preferred)
	Ambient
	Frozen

CAUTIONS:
- Because of the complexity of the genetic structure of the *CYP21A2* locus, and the possibility that a patient's congenital adrenal hyperplasia (CAH) may be due to other gene defects, genetic testing results should be correlated carefully with clinical and biochemical data.

- This testing strategy is superior to approaches previously used, but may still miss some complex and large-scale genetic rearrangements or deletions, as well as genetic changes in far upstream or downstream gene-regulatory elements that impair *CYP21A2* gene expression. This can lead to false-negative test results.
- Rare polymorphisms in primer binding sites can lead to selective allelic drop-out, which can lead to false-negative or false-positive diagnosis.
- Patients without genetic evidence for disease-causing *CYP21A2* genetic changes may still suffer from CAH, but due to a different enzyme defect. Additional and expanded biochemical steroid profiling is, therefore, recommended if the clinical picture is strongly suggestive of CAH.

CPT CODES:

81405 – CYP21A2 Full Gene Analysis
81402 – CYP21A2 Common Variants

DAY(S) SET UP: Mondays **ANALYTIC TIME**: 5 Days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
David O'Brien, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710