Lab Dept: Anatomic Pathology

Test Name: CYP21A2 (CAH) KNOWN MUTATION

General Information

Lab Order Codes:	CYPAK
Synonyms:	Congenital Adrenal Hyperplasia Known Mutation; 21-Hydroxylase Gene Known Mutation
CPT Codes:	81403 – Molecular Pathology procedure, Level 4
Test Includes:	CYP21A2 known mutation (amplification) and CYP21A2 known mutation sequencing will always be performed. DNA extraction will always be performed at an additional charge.

Logistics

Test Indications:	Third tier confirmatory testing of positive congenital adrenal hyperplasia (CAH) newborn screens. Carrier detection of CYP21A2 mutations and genetic counseling.
Lab Testing Section:	Anatomic Pathology – Sendouts
Referred to:	Mayo Medical Laboratories (MML#: 89082)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	5 – 11 days
Special Instructions:	Include form CYP21A2 Gene Testing Patient Information Sheet with the specimen.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 mL (Minimum: 0.2 mL) blood

Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; specimens other than EDTA whole blood; frozen specimens

Interpretive

Reference Range:	An interpretive report will be provided.
Limitations:	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 s 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.
Methodology:	Polymerase Chain Reaction (PCR), DNA sequencing
References:	Mayo Medical Laboratories April 2011 Phone: 1-800-533-1710
Updates:	2/5/2013: CPT update