MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

CYCMS

21-Hydroxylase Gene (CYP21A2), Full Gene Analysis, Prenatal

Patient ID	Patient Name		Birth Date	Gender	Age
SA00060443	SAMPLEREPORT, CYCMS NORMAL		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00060443	SA00060443	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		29 Jul 2013 13:00			

CYP21A2 Full Gene Analysis,Prenatal

Result

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Negative

There is no evidence of disease-causing mutations or large rearrangements between the CYP21A2 gene and CYP21A1P pseudogene. MLPA analysis indicates two copies of the CYP21A2 gene are present.

Interpretation

MCR

Negative

There is no genetic evidence for congenital adrenal hyperplasia due to CYP21A2 deficiency. There is no genetic evidence for carrier status for CYP21A2 deficiency. Rarely, some individuals who have a diagnosis of 21-OHD CAH and involvement of the CYP21A2 gene may have mutations or deletions that are not identified by the described testing methodology. In addition, some individuals with a CAH phenotype may have mutations in genes other than CYP21A2 (<10%).

Reason for Referral

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Reported Fam History

Fetus has a reported family history of 21-hydroxylase deficient congenital adrenal hyperplasia (21-OHD CAH). Test prenatal specimen for the presence of mutation(s) within the CYP21A2 gene and large rearrangements between CYP21A2 and the CYP21A1P pseudogene.

Method

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Sequencing and MLPA

Four sets of primer pairs amplify all possible recombinant forms of CYP21A2, CYP21A1P (the pseudogene), and their hybrids 5'-CYP21A2/CYP21A1P-3' and 5'-CYP21A1P/CYP21A2–3' via PCR to determine whether there are large rearrangements between the gene and pseudogene. Fluorescent DNA sequence analysis is then performed on all exons of the active form of CYP21A2 and any presumed active hybrid to test for the presence of sequencing mutations (GenBank accession number: NM_000500.5). If necessary, further analysis may be performed on non-expressed copies of CYP21A2 or hybrids to gain insight into possible rearrangements. In addition, multiplex ligation-dependent probe amplification (MLPA) was performed to determine exact copy numbers of the active gene (CYP21A2), its inactive pseudogene (CYP21A1P), and any rearrangements. However, this technology cannot determine the cis/trans status (cis=same chromosome, trans=opposite chromosomes) of the identified normal gene(s) and rearrangement(s). Family studies of blood relatives might assist in determination of the cis/trans status.

Comment

MCR

GC consult

A genetic consultation may be of benefit.

Reviewed By

MCR

LYNETTE HANSEN

ADDITIONAL INFORMATION

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. A list of common polymorphisms identified for this patient is available from the Endocrine Laboratory upon request. Rarely, individuals may have a mutation or deletion in the gene(s) tested that is not identified by the described testing methodology. In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test. Laboratory developed test.

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Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905