MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

CYPPS

21-Hydroxylase Gene (CYP21A2), Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00060441	SAMPLEREPORT, CYPPS ABNORMAL		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00060441	SA00060441	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		29 Jul 2013 13:00			

CYP21A2 Full Gene Analysis

CYP21A2 Full Gene Analysis

Result

MCR

Result

Result Summary: One copy of CYP21A2 has been deleted. The remaining copy of CYP21A2 harbors the known disease-causing mutation p.R356W.

The additional information provided below includes details regarding the specific genotype arrangement of CYP21 alleles for this patient, which may be important for testing of additional family members.

One copy of the active gene CYP21A2

One copy of a rearrangement creating a likely expressed (active) 5'-CYP21A2/CYP21A1P-3' hybrid

Two copies of the inactive pseudogene CYP21A1P

The following homozygous known disease-causing mutation is present in CYP21A2:

Nucleic acid, conventional nomenclature, genomic nucleotide numbering starting at ATG, omitting the common codon 9 leucine duplication: g.2108C>T Amino acid, conventional nomenclature, omitting the common codon 9 leucine duplication: p.R356W

Interpretation

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Simple Virilizing

This result is consistent with a diagnosis of congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency. The most likely phenotype is simple virilizing CAH.

Reason for Referral

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Clinical features

Patient has features suggestive of 21-hydroxylase deficient congenital adrenal hyperplasia (21-OHD CAH). Test 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.

Performing Site Legend

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

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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.

Received: 30 Jul 2013 11:07 **Reported:** 30 Jul 2013 11:47

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