

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

Patient ID SA00060444	Patient Name SAMPLEREPOR, CYCMS ABNORMAL	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00060444	Client Order Number SA00060444	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 29 Jul 2013 13:00		

CYP21A2 Full Gene Analysis, Prenatal

<p>Result MCR</p> <p>Result</p> <p>Result Summary: Homozygous for the known disease-causing mutation(s): p.P453S .</p> <p>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.</p> <p>Two copies of the active gene CYP21A2</p> <p>Two copies of the inactive pseudogene CYP21A1P</p> <p>The following homozygous known disease-causing mutation is present in CYP21A2:</p> <p>Nucleic acid, conventional nomenclature, genomic nucleotide numbering starting at ATG, omitting the common codon 9 leucine duplication: g.2578C>T Amino acid, conventional nomenclature, omitting the common codon 9 leucine duplication: p.P453S</p> <p>Please note, per request of the ordering provider, maternal cell contamination studies were not performed for this prenatal testing.</p> <p>Interpretation MCR</p> <p>Non-classical CAH</p> <p>This result is consistent with a diagnosis of congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency. The most likely phenotype is non-classical CAH.</p>	<p>Reason for Referral MCR</p> <p>Confirmed Fam Hist</p> <p>Fetus has a confirmed 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.</p> <p>Method MCR</p> <p>Sequencing and MLPA</p> <p>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.</p> <p>Comment MCR</p> <p>GC consult</p> <p>A genetic consultation may be of benefit.</p>
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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



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Reviewed By

MCR

LYNETTE HANSEN

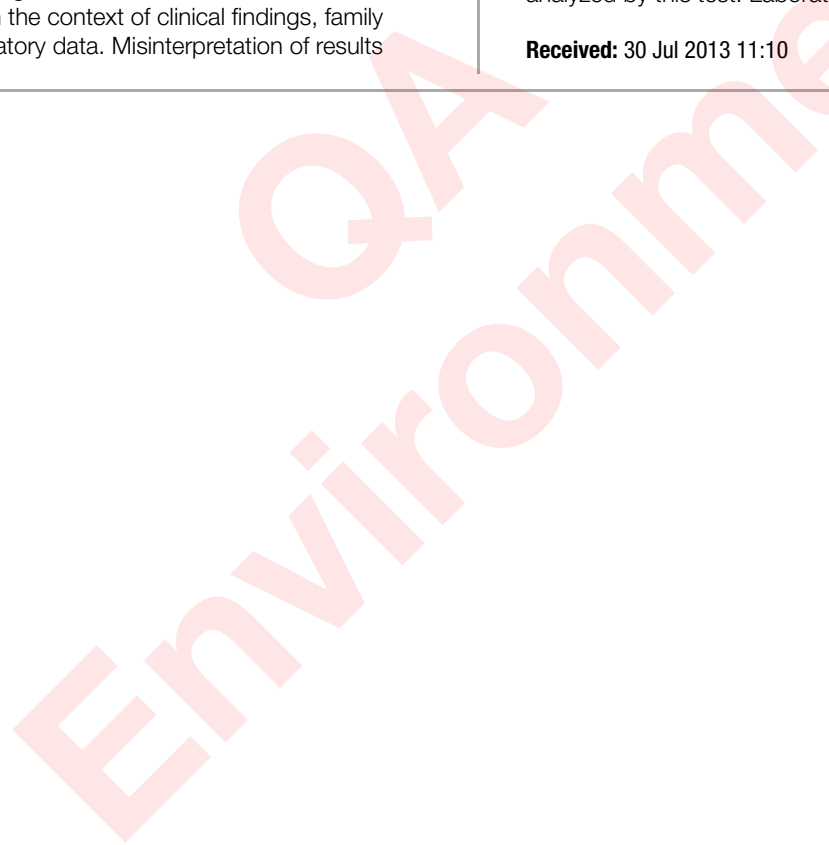
ADDITIONAL INFORMATION

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.

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