Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CYPPS NORMAL	SA00060440	47	F	SA00060440
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00060440	**Account Information**			**Report Notes**
Collected 07/29/2013 13:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 08/30/2013 13:48				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CYP21A2 Full Gene Analysis					
RECEIVED: 07/30/2013 11:06 **REPORTED:** 07/30/2013 11:47					
CYP21A2 Full Gene Analysis					
Reason for Referral		Not provided			MCR
Reason for referral was not provided by the requesting physician.					
Method		Sequencing and MLPA			MCR
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.					
Result		Negative			MCR
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		Negative			MCR
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%).					
Comment		GC consult			MCR
A genetic consultation may be of benefit.					
Reviewed By		LYNETTE HANSEN			MCR

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CYPPS NORMAL	SA00060440	47	F	SA00060440
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00060440	**Account Information**			**Report Notes**
Collected 07/29/2013 13:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 08/30/2013 13:48				

Test	Flag	Results	Unit	Reference Value	Perform Site*

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.

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.