Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,PWDNA N	SA00058854	46	F	SA00058854
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058854	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:08				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Prader Willi/Angelman Mol Analysis					
RECEIVED: 06/07/2013 15:53 **REPORTED:** 06/10/2013 16:35					
Specimen		Blood			MCR
Specimen ID		1062121			MCR
Order Date		10 Jun 2013 09:15			MCR
Reason For Referral					MCR

Possible diagnosis of Prader-Willi (PW) or Angelman Syndrome (AS). Analyze the PW/AS critical region for alterations in the DNA methylation pattern.

Method MCR

Methylation-sensitive multiplex ligation-dependent probe amplification (MLPA) was used to test for the presence of large deletions, duplications and/or methylation defects in the Prader-Willi/Angelman syndrome (PW/AS) critical region.

Result MCR

MLPA demonstrated a normal methylation pattern. No deletions or duplications were detected. This indicates that both the maternally and paternally derived copies of the PW/AS critical region are present.

Interpretation MCR

Results suggest that this individual is unlikely to have either Prader-Willi or Angelman syndrome.

Please note that we cannot entirely rule out the diagnosis of Prader-Willi syndrome, as a small number of patients with the clinical diagnosis of Prader-Willi syndrome (approximately 2%) have alterations (e.g. point mutations or small deletions) which are not detected by this assay. Additionally, the diagnosis of Angelman syndrome is not excluded because approximately 25% of patients with the clinical diagnosis of Angelman syndrome have alterations (e.g. point mutations or small deletions) which are not detected by this assay. If not already performed, genetic testing of the UBE3A gene (UBEMS/89919 UBE3A Gene, Full Gene Analysis) may provide additional diagnostic information.

Because some chromosome abnormalities may have overlapping clinical features with Prader-Willi and Angelman syndrome, a standard chromosome study is recommended. If one has already been performed, please refer to that separate report for details.

CAUTIONS:
Test results should be interpreted in context of clinical

Performing Site Legend on Last Page of Report

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findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
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.					
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Reviewed By: Emily Christine Lauer					MCR
Release Date		10 Jun 2013 16:34			MCR

* Performing Site:

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

* Report times for Mayo performed tests are CST/CDT