

Patient Name SAMPLEREP, PWDNA A	Patient ID SA00058855	Age 46	Gender F	Order # SA00058855
Ordering Phys CLIENT, CLIENT				DOB 06/10/1966
Client Order # SA00058855	Account Information C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			Report Notes
Collected 06/06/2013 00:00				
Printed 06/11/2013 16:09				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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Prader Willi/Angelman Mol Analysis
RECEIVED: 06/07/2013 15:53 **REPORTED:** 06/10/2013 16:39

Specimen Blood MCR

Specimen ID 1062122 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 an abnormal methylation pattern. No deletions or duplications were detected. The abnormal methylation pattern suggests an absence of the maternally derived copy of the PW/AS critical region.

Interpretation MCR

These results are consistent with the diagnosis of Angelman syndrome.

Please note that MLPA does not distinguish between paternal uniparental disomy (UPD) and the presence of an imprinting mutation. Additional studies involving molecular genetic analysis are required to distinguish between these possibilities.

CAUTIONS:

Test results should be interpreted in context of clinical 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.

Performing Site Legend on Last Page of Report

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Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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Reviewed By:					MCR
Emily Christine Lauer					
Release Date		10 Jun 2013 16:37			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREPORT,PWDNA A	Collection Date and Time 06/06/2013 00:00	Report Status Final
Page 2 of 2		** End of Report **

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