Patient ID	Patient Name		Birth Date	Gender	Age
SA00001255	SAMPLEREPDMGLM, VLD20150727A0005		2008-01-01	M	7
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00001255	SA00001255	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		26 Jul 2015 14:30			

Uniparental Disomy

Result Summary
MCR

POSITIVE FOR UPD 15

Result
MCR

The informative markers D15S128, D15S1012, D15S153, D15S131, D15S205, D15S127, and D15S120 demonstrated that
the proband has inherited only maternal alleles for these markers (paternal alleles were not detected). Please see attached pedigree for results.

Interpretation
[1] MCR

Results are consistent with maternal heterodisomy/isodisomy for chromosome 15 and a diagnosis of Prader-Willi syndrome.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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.

Rare polymorphisms exist that could lead to false-negative or false-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 Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Reason for Referral
MCR

Evaluate for uniparental disomy (UPD) for chromosome 15.

Specimen
MCR

WB Whole Blood

Method
MCR

A PCR-based analysis on the DNA from the proband and parents was used to test for the presence of uniparental disomy (UPD) of chromosome 15 in the proband. The following microsatellite markers were used: D15S128, D15S1002, D15S165, D15S1007, D15S1012, D15S978, D15S153, D15S131, D15S205, D15S127, D15S130, and D15S120.

Released By
MCR

EMILY LAUER

Received: 28 Jul 2015 08:22 **Reported:** 29 Jul 2015 13:56

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292