MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, FSMS	L3MRNG9150547	34Y	F	G9150547

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
05/25/10 10:39 A	05/25/10 10:39 A	06/15/10 01:59 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

VALIDATION TESTING

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

17p11.2 Deletion/Duplication, FISH

Specimen	**Blood**	**MCR**
Specimen ID	**1050919**	**MCR**
Order Date	**25 May 2010 11:14**	**MCR**

Reason for Referral **MCR**

r/o deletion/duplication of 17p11.2

Method **MCR**

FISH for Smith Magenis and Potocki-Lupski syndromes was performed using a probe for the RAI1 critical region (SMS, which includes RP11-1149K20) at 17p11.2 and a control locus (RP11-47G8) at 17q12.

Result **ish 17p11.2(SMSx2)** **MCR**

nuc ish(SMSx2)
SMS hybridized to each chromosome 17 in ten metaphase cells. Of 200 interphase nuclei, 0% had three SMS signals. The normal cutoff is <8% for duplication of SMS.

Interpretation **MCR**

The result is within normal limits. No deletion or duplication of the RAI1 critical region (SMS) for Smith-Magenis and Potocki-Lupski syndromes at 17p11.2 was observed. A genetic consultation may be of benefit.
A full chromosome analysis (test #8696) was not performed in this laboratory, but may be of benefit if clinically warranted and it may be possible to perform this test on cells that we have stored from this specimen.
An array CGH study (test #88898) may be of benefit to detect clinically significant gains or losses of chromosomal material not visible by this FISH test.
For more information regarding further testing, please call 800-533-1710.
DISCLAIMER: This test was developed and its performance

** Perform Site Legend on last page of report*

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TESTING, FSMS	Final	05/25/10 10:39 A

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characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN. It has not been cleared or approved by the U.S. Food and Drug Administration. This FISH assay does not rule out other chromosome abnormalities.

Consultant MCR
Rhett Patrick Ketterling MD

Released Date 01 Jun 2010 11:12 MCR

*** PERFORMING SITE**

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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TESTING, FSMS	Final	05/25/10 10:39 A