MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, 86374	L3MRNG9166109	18	F	G9166109

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
03/08/11 09:09 A	03/08/11 09:09 A	03/09/11 10:21 A	DATE OF BIRTH: 7/23/1992

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

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

Known 45,X Mosaicism Reflex, FISH — **REPORTED: 03/08/11 09:14 A**

Specimen	**Blood**	**MCR**
Specimen ID	**1053704**	**MCR**
Order Date	**08 Mar 2011 09:10**	**MCR**

Method — **MCR**

FISH for sex chromosome mosaicism using probes for centromeres of X (DXZ1) and Y (DYZ3).

Reason For Referral	**amenorrhea**	**MCR**

Results — **MCR**

nuc ish(DXZ1x1)[353/500]/(DXZ1,DYZ3)x1[147/500]
Of 500 interphase nuclei, 70.6% had one X signal and 29.4% had an X and a Y signal. The normal cutoff is <1.0% and <0.6% for presence of a second X signal or the presence of a Y signal, respectively.

Interpretation — **MCR**

The cytogenetic findings are consistent with a diagnosis of Turner syndrome. The routine chromosome analysis result revealed a non-mosaic 45,X karyotype. Some studies suggest that everyone with Turner syndrome has chromosomal mosaicism. Since some have 46,XY mosaicism, or a structural abnormality of the Y, we scored 500 interphase cells for X and Y chromosome "signals". 353 cells had 1 X signal, and 147 cells had X and Y signals. In a nonmosaic 45,X we would expect fewer than 1.0% of cells to have both X and Y signals. The data therefore suggest that this individual is a mosaic with a minor 46,XY cell population or some cells with a structurally abnormal Y chromosome. A genetic consultation may be of benefit.
DISCLAIMER: This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN. It has not been

** Perform Site Legend on last page of report*

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cleared or approved by the U.S. Food and Drug
Administration. This FISH assay does not rule out other
chromosome abnormalities.

Consultant	**Heather Lynn Owen**	**MCR**
Report Date	**08 Mar 2011 09:12**	**MCR**

Analyte Specific Reagent
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determined by Laboratory Medicine and Pathology, Mayo
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*** 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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