ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	5/12/1979
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

X-Chromosome Inactivation Analysis

ARUP test code 2006352

X-Chromosome Inactivation Specimen	Whole Blood
X-Chromosome Inactivation Interpretation	Normal

Indication for Testing: Assess pattern of X-chromosome inactivation (XCI).

Result: Random XCI Ratio 66:34

Interpretation: A normal, random XCI pattern was detected by methylation analysis of the androgen receptor (AR) gene locus. An XCI ratio of less than 80:20 in an XX female does not support skewed XCI in the sample type tested.

Recommendation: Medical management should rely on clinical findings and family history.

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: X-Chromosome Inactivation Analysis

CHARACTERISTICS: Females usually have two copies of the X-chromosome, one of which becomes randomly inactivated early in embryonic development in a process known as lyonization. If either the paternally or maternally derived X-chromosome is preferentially inactivated, this results in a non-random or skewed pattern of X-chromosome inactivation (XCI). The pattern of XCI may vary among tissue types. XCI ratios of 50:50 to 79:21 are considered random, ratios of 80:20 to 89:11 are moderately skewed and ratios of 90:10 to 100:0 are highly skewed.
CAUSE: Non-random XCI may result by chance or from secondary cell selection in women who are heterozygous for X-chromosome rearrangements, carriers of mutations in X-linked genes, or affected with neoplastic disease.
GENE TESTED: The androgen receptor (AR) gene on the X chromosome.
CLINICAL SENSITIVITY: Approximately 90 percent. An estimated 10 percent of women have skewed X-inactivation by chance. However, skewed XCI may be seen more frequently with increasing age.
METHODOLOGY: Methylation-sensitive restriction digest followed by PCR and fragment analysis.
LIMITATIONS: Testing is limited to XX females only. This assay will be uninformative in up to 20 percent of females due to homozygosity for the polymorphic AR gene locus analyzed. XCI patterns may differ among tissues; therefore, the XCI ratio reported is for the tissue type tested. Although this test will detect the methylation status of the X-chromosomes, it will not determine if the X inactivation pattern is associated with rearrangements of the X chromosome, mutations in X-linked genes or neoplastic disease. If a non-random XCI pattern is present, the parent of origin of the active X cannot be determined without testing parental samples. XCI ratios should not be used to predict prognosis for female carriers of X-linked disorders as variable expressivity may result due to other genetic or environmental modifiers. Because the level of XCI may differ in prenatal specimens and whole blood, this test is not recommended for prenatal diagnosis. Diagnostic errors can occur due to rare sequence variations.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
X-Chromosome Inactivation Specimen	17-073-122274	3/14/2017 4:37:00 PM	3/15/2017 4:29:15 PM	3/24/2017 5:04:00 PM
X-Chromosome Inactivation Interpretation	17-073-122274	3/14/2017 4:37:00 PM	3/15/2017 4:29:15 PM	3/24/2017 5:04:00 PM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical