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

Physician: Doctor, Example

Patient: Patient, Example

DOB	5/19/1983
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

X-Chromosome Inactivation Analysis

ARUP test code 2006352

X-Chromosome Inactivation Specimen: Whole Blood

X-Chromosome Inactivation Interpretation: **Indeterminate** *

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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

Result: XCI Ratio 75:25

Interpretation: Indeterminate. The XCI ratio determined by methylation analysis of the androgen receptor (AR) gene locus is of uncertain clinical significance. An XCI ratio greater than 85:15 in an XX female supports non-random, skewed XCI in the sample type tested. XCI ratios less than 75:25 suggest normal, random XCI in the sample type tested. The XCI ratio detected in this sample falls between these ranges; therefore, caution should be used when interpreting this result. Please see the background information included in this report for assay limitations, including standard deviation of this assay.

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 nonrandom or "skewed" pattern of X-chromosome inactivation (XCI). The pattern of XCI may vary among tissue types. XCI ratios of 50:50 to 74:26 suggest random XCI, ratios greater than 85:15 suggest nonrandom XCI, and ratios from 75:25 to 85:15 should be interpreted with caution.
Cause: Nonrandom XCI may result by chance or from secondary cell selection in females who are heterozygous for X-chromosome rearrangements, carriers of pathogenic variants 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 10-15 percent of females 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 with a standard deviation 0.08 for XCI ratios of 50:50-79:50; 0.05 for XCI ratios 80:20 or greater. 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, pathogenic variants in X-linked genes or neoplastic disease. If a nonrandom 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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
X-Chromosome Inactivation Specimen	23-345-401364	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
X-Chromosome Inactivation Interpretation	23-345-401364	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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