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PATIENT REPORT

Patient Age/Sex: 29 years Female

Specimen Collected: 29-Oct-25 14:22

X-Chromosome Inactiviation Analysis **|Received: 29-Oct-25 14:22** **Report/Verified: 30-Oct-25 11:22**

Procedure	Result	Units	Reference Interval
X-Chromosome Inactivation Specimen	Whole Blood		
X-Chromosome Inactivation Interpretation	Random [f1] [i1]		

Result Footnote

f1: X-Chromosome Inactivation Interpretation

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

Result: Random XCI Ratio 66:34

Interpretation: A random XCI pattern was detected by methylation analysis of the androgen receptor (AR) gene locus. An XCI ratio of less than 75:25 in an XX female does not support non-random XCI in the sample type tested.

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]

Test Information

i1: X-Chromosome Inactivation Interpretation

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

Test Information

i1: X-Chromosome Inactivation Interpretation

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.