

Y-Chromosome: SRY Analysis

Test Code: DY

Turnaround time: 2 weeks

CPT Codes: 81403 x1

Condition Description

Indications

This test is performed on patients who require a very quick analysis to determine presence/absence of Y-chromosome material, such as infants born with ambiguous genitalia, X-linked disorders, or cases of tissue mosaicism. PCR-based analysis, with male and female controls, is utilized to determine the presence/absence of the Y-chromosome. Results include which regions of the Y-chromosome were analyzed and which regions were detected.

Methodology

PCR-based amplification for a portion of the SRY gene, uses primers specific for the centromere, DYZ3, and specific for the q arm, Y3.4. Male and female controls are used in combination to determine the presence/absence of the Y-chromosome. Results include which regions of the Y-chromosome were analyzed and which regions were detected or absent. Point mutations or rearrangements are not detected by this analysis.

Detection

Over 99% of Y-chromosomes will be detected by this assay.

Reference Range

Qualitative assay.

Specimen Requirements

Submit only 1 of the following specimen types

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.