Y-Chromosome: Microdeletion Analysis

**Test Code:** YD  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81403 x1

### Condition Description

### Indications

This test is performed on patients with azoospermia or oligospermia, as 3-30% of these men will have microdeletions in the Y-chromosome. The type of deletion present can determine the cause of infertility in some males, and have prognostic value in determining intervention. There are a total of four AZoospermic Factor (AZF) regions on the Y-chromosome: AZFa, AZFb, AZFc, and AZFd. These deletions in the Yq11 region are mostly undetectable by cytogenetic analysis.

Most cases of Y-chromosome microdeletions are new mutations. If transmitted, any child receiving the Y-chromosome will have the microdeletion. In other words, all sons will be affected.

### Methodology

Molecular analysis can detect the presence/absence of these deletions by PCR (using eighteen sequence tagged sites).

### Detection

Over 99% of AZFa, AZFb, AZFc, and AZFd 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™ 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.