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

**Type:** Whole Blood

**Specimen Requirements:**

In EDTA (purple top) tube:  
- Infants (2 years): 3-5 ml  
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.