## Y-Chromosome: Microdeletion Analysis

<table>
<thead>
<tr>
<th>Test Code: <strong>YD</strong></th>
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<tbody>
<tr>
<td>Turnaround time: <strong>2 weeks</strong></td>
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<tr>
<td>CPT Codes: <strong>81403 x1</strong></td>
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</tbody>
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### 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) or ACD (yellow 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.