

## Y-Chromosome DNA Testing

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### DESCRIPTION:

- Testing for the presence of the Y chromosome is helpful in assessing patients with ambiguous genitalia, patients with mosaicism for an unidentified second sex chromosome, and prenatal samples, especially if the fetus is at risk for a sex-linked disorder. Y material is detectable even if more than 99% of analyzed cells are female.

### REASON FOR REFERRAL:

- As above
- This test detects presence of the sex-determining gene on Yp (SRY), of the q arm (alphoid) repeat region, and of the amelogenin gene in the pseudoautosomal, X-pairing region (Yp). It confirms presence of Y chromosome material but does not necessarily predict male phenotype.

### METHOD OF ANALYSIS:

- DNA from the patient is amplified by PCR and analyzed by a laser-induced fluorescence capillary electrophoresis system with internal standard. Three Y loci are analyzed as discussed above.
- Results are reported within 3 weeks of receipt of sample. Results are reported within 5 days or less of receipt of samples for "STAT" analysis. STAT analysis must be arranged in advance with the laboratory.

### REFERENCE RANGES:

- For amelogenin, a product of 106 base pairs (bp) is produced from the X chromosome, and of 112 bp from the Y. Male: 106 and 112 bp, Female: 106 bp.
- For the alphoid repeat, a product of 170 bp is produced only from the Y chromosome. Male: 170 bp, Female: No product.
- For SRY, a product of 436 bp is produced only from the Y chromosome. Male: 436 bp, Female: No product.

### SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- Prenatal testing: Please contact the laboratory.

### TEST CPT CODES:

CPT 83890 DNA extraction  
CPT 83894 DNA separation X 2  
CPT 83898 DNA amplification X 2  
CPT 83912 DNA interpretation and report

*Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.*