

## Multiplex Trisomy / Sex Chromosome Screen

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### DESCRIPTION:

- This is a DNA test to examine tetranucleotide repeat polymorphisms for evidence of trisomy for selected chromosomes, 13, 18, and 21, and also detects the sex chromosomes (X and Y).
- This test does not replace G-banded karyotype analysis or fluorescence in situ hybridization (FISH) for the detection of trisomy. It is available in selected situations after consultation with the laboratory director.

### REASONS FOR REFERRAL:

- Rapid diagnosis of trisomy in a newborn.
- Rapid diagnosis of trisomy for a fetus.
- Diagnosis of trisomy and gender in a fetal loss with nonviable tissue (unable to karyotype).

### METHOD OF ANALYSIS:

- Size polymorphisms of tetranucleotide repeat alleles are determined by multiplex PCR amplification using fluorescently labeled specific primers. Typically, three loci per selected chromosome are amplified in the patient. Allele sizes and peak areas are determined by a high resolution laser-induced fluorescence capillary electrophoresis system with internal standard.
- 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:

- The loci should show a 2:1 peak area ratio of one allele to the other if two alleles are observed, or three alleles should be found to confirm trisomy. At least 2 of the three loci should be informative.

### 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.
- Fetal tissue: fresh, fixed, frozen accepted, with parental bloods if necessary.

### TEST CPT CODES:

CPT 83890 DNA extraction  
CPT 83894 DNA separation X 3  
CPT 83901 DNA amplification multiplex X 3  
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.*