STAT FISH Chromosomes 13, 18, 21, X, and Y

Test Code: CFRSB

Turnaround time: 1 day - 3 days. ((All abnormal findings are called out immediately.))

CPT Codes: 88271 x1, 88291 x1, 88275 x1

Condition Description

Chromosome disorders due to non-disjunction of chromosomes 13, 18, 21, X, and Y together comprise the majority of the microscopically detectable chromosome disorders.

Analysis by Fluorescence In Situ Hybridization (FISH) allows for the most rapid detection of the most common chromosome disorders. Results can typically be reported in 24-48 hours from the time of receipt.

Concurrent G-banded chromosome analysis with or without chromosomal microarray is recommended.

Indications

FISH for aneuploidy is appropriate for the following indications:

- Multiple congenital anomalies
- Dysmorphic features
- Developmental delay
- Advanced maternal age (AMA)
- Abnormal ultrasound
- Abnormal serum screen
- Parental concern
- Increased nuchal translucency or nuchal fold
- Family history

Methodology

Interphase FISH is performed on uncultured peripheral blood samples using commercially available probes.

Detection

FISH is very sensitive in the detection of aneuploidy. This probe set is specific to chromosomes 13, 18, 21, X, and Y. Only numerical abnormalities of these chromosomes will be detected. Validation for specificity and sensitivity are performed on each probe. Control probes are present in all probe sets.

Specimen Requirements

Type: Whole Blood (EDTA and Sodium Heparin)

Specimen Requirements:

- Sodium Heparin and EDTA
- Infants (Children <2 years): 3-5 ml in both tubes
- Older Children & Adults: 7-10 ml in both tubes

Specimen Collection and Shipping:

- Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

- Chromosomal Microarray, EmArray Cyto (test code VA)
- Chromosomal SNP array (test code CMSNP)
- Chromosome Analysis (test codes CA, CB, MM)