Prenatal Rapid Screen by FISH for 13, 18, 21, X and Y

**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. Aneuploidy of the non-sex chromosomes increases in frequency as maternal age increases.

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 required.

**Indications**

- FISH for aneuploidy is appropriate for the following indications:
  - Parental concern
  - Abnormal ultrasound
  - Abnormal maternal serum screen
  - Increased nuchal translucency or nuchal fold
  - Advanced maternal age
  - Family history

**Methodology**

Interphase FISH is performed on direct amniotic fluid and chorionic villi 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**

Submit only 1 of the following specimen types

**Type: Amniotic Fluid**

Specimen Requirements:

Collect 20-30 ml of amniotic fluid (discard the first 1-2 ml). Place in sterile conical centrifuge tubes.

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Type: Chorionic Villi**

Specimen Requirements:

Collect 15-30 mg of chorionic villi using sterile technique. Place in sterile tube(s) with EGL transport media or other sterile culture media.

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Special Instructions**

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

**Related Tests**

- Prenatal Chromosomal Microarray, EmArray Cyto (CMPRE)
- Prenatal Chromosome Analysis (CV/AD)