STAT FISH Chromosomes X&Y

Test Code: CFXYS
Turnaround time: 1 day - 3 days  (All abnormal findings are called out immediately.)
CPT Codes: 88271 x2, 88275 x2, 88291 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 or chromosomal microarray is required.

Sex chromosome aneuploidies include conditions such as Klinefelter syndrome (47,XXY) and Turner syndrome (45,X).

Turner syndrome (TS) occurs in females when they are missing all or part of an X chromosome. This condition occurs in 1 in 2,000 live births. Physical characteristics of Turner syndrome include webbed neck, lymphedema, short stature and shield chest. Congenital heart defects such as bicuspid aortic valve and coarctation of the aorta occur in 25-45% of individuals with Turner syndrome. Aortic dissection occurs in 1-2% of individuals with TS and is preceded by dilation. Females with TS often require hormonal therapy to progress through puberty and may experience premature ovarian failure. Often, women with TS are infertile. Other medical issues include hypertension, hearing loss, and autoimmune thyroid disease. Women and girls with TS typically have normal intelligence; however they may experience developmental delays and learning difficulties.

Klinefelter syndrome occurs in males when they have an extra X chromosome (47,XXY). This condition occurs in 1 in 500 to 1000 males. Males with Klinefelter syndrome are typically deficient in the hormone testosterone. This causes them to have reduced facial and body hair, tall stature, development of breast tissue (Gynecomastia) and small testes. Infertility occurs in 90-95% of individuals. Males with Klinefelter syndrome may have learning difficulties, especially in language tasks and verbal abilities and may have a shy sensitive personality. Males who are mosaic for Klinefelter tend to have milder manifestations of the condition. Conversely, males with variants such as 48,XXX Y tend to have a more severe phenotype.

### Indications

FISH for the X chromosome is appropriate for the following indications:

- Short stature
- Webbed neck
- Lymphedema
- Infertility
- Delayed or absent puberty
- Learning difficulties
- Coarctation of the aorta
- Cystic hygroma

FISH for the Y chromosome is appropriate for the following indications:

- Tall stature
- Absent or delayed puberty
- Infertility
- Learning difficulties
- Hypogonadism
- Gynecomastia

### 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 X and Y. Only numerical abnormalities of chromosomes X and Y 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

Specimen Requirements:
In sodium heparin (green top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

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 or chromosomal microarray is required.

**Related Tests**

- Chromosomal Microarray, EmArray Cyto (VA)
- Chromosome Analysis (CA/CB)