STAT FISH Chromosome 21

Test Code: CF21S
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. Trisomy 21 is the most common chromosomal abnormality in live born individuals. Aneuploidy of non-sex determining chromosomes increases in frequency along with increasing maternal age.

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.

Trisomy 21 or Down syndrome is a condition characterized by intellectual disability, characteristic facial features, congenital malformations and hypotonia. Trisomy 21 occurs in 1 out of every 740 newborns. Advanced maternal age increases the risk for an infant with trisomy 21. Children with trisomy 21 can have multiple malformations and intellectual disability. The intellectual impairment can vary, but typically falls within the mild to moderate range. Physical characteristics can include upslanting palpebral fissures, hypotonia, transverse palmar crease, depressed nasal bridge, brushfield spots and bilateral epicanthal folds. Ultrasound findings associated with trisomy 21 include increased nuchal translucency, cardiac defect, absent nasal bone, echogenic bowel or duodenal atresia.

In trisomy 21, there is an increased risk of congenital heart disease, leukemia, deafness, serous otitis media, Hirschsprung's disease, gastrointestinal atresias, eye disease, including cataracts and severe refractive errors, acquired hip dislocation, and thyroid disease. Adults with trisomy 21 have an increased risk of developing Alzheimer disease.

About 95% of cases of trisomy 21 result from an extra copy of chromosome 21. This extra genetic material disrupts the developmental process and causes the characteristic features of trisomy 21. Trisomy 21 is typically not inherited, but rather a random event of maternal nondisjunction in meiosis. In approximately 3% to 4% of individuals with the Down syndrome phenotype, the extra chromosomal material is the result of an unbalanced translocation between chromosome 21 and another acrocentric chromosome. Trisomy 21 caused by a translocation can be inherited and translocation carriers are at risk of having a child with trisomy 21. The remaining 1% to 2% of individuals with a Down syndrome phenotype have a mosaic form of trisomy 21 and the severity can range from mild to moderately affected.

References:

Indications

- Multiple congenital anomalies
- Dysmorphic features
- Developmental delay
- Advanced maternal age (AMA)
- Abnormal ultrasound
- Abnormal serum screen

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 chromosome 21 and only numerical abnormalities of chromosome 21 will be detected. Validation for specificity and sensitivity 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

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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)