EGL Genetics | Eurofins Clinical Diagnostics
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STAT FISH Chromosome 13

Test Code: CF13S
Turnaround time: 1 day - 3 days (All abnormal findings are called out immediately.)
CPT Codes: 88291 x1, 88271 x2, 88275 x2

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 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.

Concurrent G-banded chromosome analysis or chromosomal microarray is required.

Trisomy 13 or Patau syndrome is a disorder characterized by major congenital malformations, such as those of the heart, brain, kidney, and spinal cord. Trisomy 13 occurs approximately in 1 out of every 16,000 newborns. Advanced maternal age increases the risk for an infant with trisomy 13. Individuals with trisomy 13 have severe intellectual disabilities. Physical characteristics may include cleft palate, holoprosencephaly, microcephaly, polydactyly, and rocker bottom feet. Infants with trisomy 13 often have such severe medical complications that many die within the first days or weeks of life. The median survival for children with this disorder is 7 days. Five to 10% of children with trisomy 13 live past year one of life; however, those that do will have severe anomalies which usually leads to their death. Ultrasound findings for trisomy 13 include cleft palate, polydactyly and rocker bottom feet.

Most cases of trisomy 13 result from an extra copy of chromosome 13. This extra genetic material disrupts the development process and causes the characteristic features of trisomy 13. In individuals with a mosaic form of trisomy 13, the severity can range from normal to severely affected. Trisomy 13 is typically not inherited, but rather a random event of nondisjunction in meiosis. Trisomy 13 caused by a translocation can be inherited and translocation carriers are at risk of having a child with trisomy 13.

References:

- www.ghr.nlm.nih.gov/condition/trisomy-13
- Smith, D.W. (2006) Trisomy 13 Syndrome. Smith's Recognizable Patterns of Human Malformation. Philadelphia, Elsevier Saunders, 6th Edit. pp. 18-21.

Indications

- Multiple congenital anomalies
- Dysmorphic features
- IUGR
- Advanced maternal age (AMA)
- Abnormal ultrasound

Methodology

Interphase FISH analysis 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 13 and only numerical abnormalities of chromosome 13 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.

Special Instructions

Concurrent G-banded chromosome analysis or chromosomal microarray is required.

Related Tests

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