

FISH Analysis

Clinical Utility:

FISH analysis is performed to evaluate a specific cytogenetic region. FISH analysis should be ordered only when A) there is clinical suspicion of: aneuploidy involving chromosomes 13, 18, 21, or the sex chromosomes; a specific microdeletion or microduplication syndrome, such as DiGeorge syndrome or Williams-Beuren syndrome; mosaicism for a specific cytogenetic condition, such as monosomy X (Turner syndrome) or B) to understand the cytogenetic mechanism of some abnormal findings on chromosomal microarray analysis such as terminal deletion and duplication or pericentromeric duplication.

FISH analysis can be performed on peripheral blood and on prenatal samples, such as amniotic fluid, chorionic villus samples (CVS), percutaneous umbilical blood samples (PUBS), or products of conception specimens.

FISH analysis on parental samples may be recommended as a follow-up when chromosomal microarray analysis on the proband reveals an abnormality suggestive of an unbalanced form of a balanced parental rearrangement. A sample from the proband is also required as a positive control and is run free of charge.

Test Methods:

FISH analysis is based on the hybridization of a fluorescently labeled probe to metaphase spreads and/or interphase nuclei prepared from cell cultures derived from amniotic fluid, chorionic villus sample, products of conception (POC), or peripheral blood. FISH probes have a specific sequence that is complementary to a particular region of the genome. The presence, absence, amplification, or translocation of the targeted region is analyzed with a fluorescent microscope.

Test Sensitivity:

FISH sensitivity depends upon the particular region being interrogated. FISH cannot detect rearrangements in genomic regions that are not specifically targeted.