

FISH Follow-up Analysis

Clinical Utility:

FISH Follow-up analysis is performed to evaluate a specific cytogenetic region as a follow-up to chromosomal microarray analysis performed by GeneDx.

FISH Follow-up should be ordered:

On a proband to understand the cytogenetic structure of an abnormal finding detected by chromosomal microarray analysis at GeneDx, such as duplicated material that may represent a ring chromosome or located elsewhere.

On parents when chromosomal microarray analysis on the proband at GeneDx reveals an abnormality suggestive of an unbalanced form of a balanced parental rearrangement. A sample from the proband is also required as a charged positive control.

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.

For FISH testing that is not a follow-up to chromosomal microarray analysis at GeneDx, including FISH for a specific microdeletion or microduplication syndrome, mosaicism for a specific cytogenetic condition, or determination of the cytogenetic structure or mechanism of an abnormal finding detected by chromosomal microarray by an outside lab, please see “FISH Analysis” (<http://www.genedx.com/test-catalog/available-tests/fish-analysis/>).

Test Methods:

FISH analysis is based on the hybridization of a fluorescently labeled probe to metaphase spreads 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.