FISH Analysis

Clinical utility of this service:
FISH analysis is performed to evaluate a specific cytogenetic region. FISH analysis should be ordered:

- When there is clinical suspicion of:
  - 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)
  - Aneuploidy
- To understand the cytogenetic mechanism of some abnormal findings on chromosomal microarray analysis such as the presence of both terminal deletion and duplication or pericentromeric duplication or other aberration requiring further clarification. (For FISH analysis on the proband and/or the proband’s parents as a follow-up to a microarray performed by GeneDx, please see FISH Follow-up Analysis.)

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.

Reason for Referral:
- For Prenatal Diagnostic Samples: Fetal abnormalities detected by ultrasound, abnormal maternal serum screening test result, noninvasive prenatal screening, advanced maternal age. GeneDx is validated to perform FISH analysis on prenatal/products of conception samples for the DiGeorge Syndrome/ Velocardiofacial Syndrome (DGS/VCFS) region.
- For Postnatal Diagnostic Samples: Suspicion for aneuploidy, mosaic aneuploidy, or a specific microdeletion or microduplication syndrome. GeneDx is validated to perform FISH analysis on cultured peripheral blood for aneuploidy involving chromosomes 13, 18, 21, and the sex chromosomes, and the following regions: DiGeorge Syndrome/Velocardiofacial Syndrome (DGS/VCFS)
  - Williams-Beuren syndrome
  - Smith-Magenis Syndrome (SMS)
  - Miller-Dieker Syndrome (MDS)
  - Kallmann syndrome
  - SRY
  - Steroid Sulfatase Deficiency (STS)
  - Wolf Hirschhorn Syndrome (WHS)
  - Prader-Willi/Angelman Syndrome
- For Parental Follow-up of a child with an abnormal chromosome or microarray analysis suggestive of an unbalanced form of a parental chromosomal rearrangement. (For parental FISH analysis as a follow-up to a microarray performed by GeneDx, please see FISH Follow-up Analysis.)
- For any other indication: Please call GeneDx and ask to speak with one of our array genetic counselors

Test method:
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.

**Specimen Requirements and Shipping/Handling:**
- *Blood:* A single tube with 1-5 mL whole blood in sodium heparin (green top). Ship overnight at ambient temperature, using a cool pack in hot weather. Although not optimal, specimens may be refrigerated for 3 days prior to shipping.

**Required Forms:**
Prenatal or Cytogenetic Sample Submission (requisition) form – complete all pages, including Payment Options or Institutional Billing Instructions

**Turn-Around Time:** 2 weeks
For test codes, prices, and CPT codes please refer to the Prenatal Testing or Cytogenetics page on our website: [www.genedx.com](http://www.genedx.com)