

Constitutional FISH Testing: An Essential Tool for Genetic Analysis

Constitutional FISH (Fluorescence In Situ Hybridization) testing is a powerful technique used to detect specific chromosomal abnormalities that are present from birth and may be linked to genetic disorders, developmental delays, or congenital anomalies. By visualizing targeted regions of chromosomes with fluorescently labeled probes, FISH enables rapid and accurate identification of microdeletions, duplications, rearrangements, and other structural changes that may not be visible with standard karyotyping.

These insights can be used to advance genetic research, support assay development, and enhance understanding of chromosome biology. Empire Genomics offers a broad range of constitutional FISH probes in variety of formats, providing reliable, high-quality tools for laboratories worldwide.

Empire Genomics offers a wide range of constitutional FISH probes, each designed to study specific chromosomal regions that are important for understanding structural variation. Below are examples of some of our most widely used probes:

5P5Q FISH Probe – Cri-du-chat Region

The 5P5Q probe is designed to study deletions or duplications on the short arm (5p) and long arm (5q) of chromosome 5. These regions are often investigated in research on structural variation related to the Cri-du-chat region, making this probe a valuable tool for studying large chromosomal changes.

N25/ARSA, TBX1/ARSA, HIRA/ARSA FISH Probes – DiGeorge Region (22q11.2)

The 22q11.2 region is a well-known hotspot for chromosomal rearrangements. Our N25, TBX1, and HIRA probes each target different loci within this critical region, allowing researchers to study complex variations with precision. Together, they provide comprehensive coverage of the DiGeorge region, one of the most frequently studied areas of the human genome.

SHOX/CONX FISH Probe – SHOX Region

The SHOX gene, located in the pseudoautosomal region of the X and Y chromosomes, is a frequent focus of research into chromosome structure and growth regulation. This probe enables researchers to visualize SHOX in relation to the sex chromosomes, supporting studies of copy number changes and structural rearrangements in this unique chromosomal location.

TWIST1/ELN FISH Probe – Saethre-Chotzen & Williams-Beuren Regions

Located on chromosome 7, TWIST1 and ELN are two genes often studied in relation to structural variability across this region. This dual-target probe allows visualization of both loci simultaneously, making it a powerful tool for investigating the Saethre-Chotzen and Williams-Beuren regions, as well as broader gene-to-gene relationships.

XIST/CONX FISH Probe – X-Inactivation Locus

The XIST gene plays a central role in the process of X-chromosome inactivation, ensuring balanced gene expression between males and females. This probe targets the X-inactivation locus, giving researchers a valuable tool for studying one of the most fundamental mechanisms of chromosome biology.



Available Probes

Explore our full range of constitutional FISH probes, developed to deliver precise, reproducible results for chromosome analysis. Each probe is carefully designed and manufactured to meet the needs of advanced genetic research, giving your lab the confidence to tackle even the most complex studies.

Region Of Interest	Probe Name	SKU
Aneuploidy	Chromosome 18, X, & Y Control FISH Probe	CHR18XY-10-AGO-R
Aneuploidy	Chromosome 13/21 Version B Control FISH Probe	CHR13-CHR21-VB-10-GO-R
Angelman	UBE3A/PML FISH Probe	UBE3A-PML-20-OG-R
Cri-du-chat	5P5Q FISH Probe	5P5Q-20-GO-R
DiGeorge N25	N25/ARSA FISH Probe	N25-ARSA-20-OG-R
DiGeorge TBX1	TBX1/ARSA FISH Probe	TBX1-ARSA-20-OG-R
DiGeorge TUPLE1(HIRA)	HIRA/ARSA FISH Probe	HIRA-ARSA-20-OG-R
Kallmann	KAL1/CONX FISH Probe	KAL1-CHRX-20-OG-R
Miller-Dieker/Smith Magenis	PAFAH1B1/RAI1 FISH Probe	PAFAH1B1-RAI1-20-OG-R
Prader-Willi/Angelman	SNRPN/PML FISH Probe	SNRPN-PML-20-OG-R
Saethre-Chotzen/Williams-Beuren	TWIST1/ELN FISH Probe	TWIST-ELN-20-OG-R
SHOX	SHOX/CONX FISH Probe	SHOX-CHRX-20-OG-R
Sotos	TERT/NSD1 FISH Probe	TERT-NSD1-20-GO-R
Steroid Sulfatase	STS/CONX FISH Probe	STS-CHRX-20-OG-R
Williams-Beuren	EGFR/ELN FISH Probe	EGFR-ELN-20-GO-R
Wolf-Hirschhorn	NSD2/4Q12 FISH Probe	NSD2-4Q12-20-OG-R
X-Inactivation Locus	XIST/CONX FISH Probe	XIST-CHRX-20-OG-R

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