ARUP LABORATORIES

500 Chipeta Way | Salt Lake City, UT 84108-1221
Phone: (800) 522-2787 | Fax: (801) 583-2712
www.aruplab.com | www.arupconsult.com

CONSTITUTIONAL FISH PROBES | AUGUST 2013

FISH assays only analyze the DNA sequence corresponding to the FISH probe for gain or loss of sequence and, in the case of metaphase FISH, indicate the cytogenetic location of that sequence. Therefore, a normal result does not rule out alterations elsewhere in the genome, small deletions or duplications within the sequence complementary to the probe, point mutations, or other mutational mechanisms yet unknown. Therefore, FISH should be considered as an adjunct to other genomic analyses. In addition, for assays in which FISH has identified an abnormality, routine chromosome analysis or other molecular genetic analyses may be necessary to determine the mutational mechanism accounting for the FISH result.

Recommended use:

1. To screen for microdeletions and microduplications associated with known syndromes.
2. For the study of individuals who have previously had a normal G-banded karyotype but have an unexplained abnormal phenotype that is suspicious for one of the following disorders.

Please note: Some probes listed below may be recommended by the laboratory to follow up on abnormal chromosome studies and are not associated with specific genetic syndromes.

Since probes on a genomic microarray specifically target subtelomeric and pericentromeric sites and locations of known microdeletion syndromes, abnormalities at any of these regions can be detected in a single analysis; it may be more efficient to order a microarray analysis instead of several different FISH probes.

AVAILABLE PROBES

SYNDROMES		
SUSPECTED DIAGNOSIS	**PROBE TARGET**	**GENE(S)/UNIQUE SEQUENCE**
Aneuploidy, common	13/18/21/X/Y	
4p-	4p16.3	*WHSC1*
5p-	5p15.2	D5S23-D5S721
15q11.2-13 duplication	15q11.2-13	D15S11, D15S10
22qter deletion	22q13.3	22qtel (*SHANK3*)
Angelman	15q11.2-13	D15S10
Cri-du-chat	5p15.2	D5S23-D5S721
DiGeorge	22q11.2	TUPLE-1 (HIRA)
Kallman	Xp22.3	*KAL1*
Male detection (*SRY*)	Yp11.3	*SRY*
Miller-Dieker (Lisencephaly)	17p13.3	*LIS1*
Phelan McDermid	22q13.3	22qtel (*SHANK3*)
Prader-Willi	15q11.2-13	D15S10
SHOX	Xp22.3	*SHOX*
Smith-Magenis	17p11.2	*SHMT1-TOP3-FL11-LLGL1*
SRY	Yp11.3	*SRY*
Steroid sulfatase deficiency (*STS*)	Xp22.3	*STS*
Velocardiofacial (VCF)	22q11.2	TUPLE-1 (HIRA)
Williams (elastin)	7q11.23	*ELN-LIMK1*-D7S613
Wolf-Hirschhorn	4p16.3	*WHSC1*

MISCELLANEOUS (Please contact the lab prior to ordering.)		
SUSPECTED DIAGNOSIS	**PROBE TARGET**	**GENE(S)/UNIQUE SEQUENCE**
Acrocentric p-arm		NOR regions of all acrocentric chromosomes
X centromere	Xcen	DXZ1
X inactivation locus	Xq13	*XIST*
Y centromere	Ycen	DYZ1
Yp11.3	Yp11.3	*SRY*
Yq12	Yq12	DYZ1-YsatIII