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# CONSTITUTIONAL FISH PROBES | APRIL 2011

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 | |
| 1p36 deletion | 1p36 | CEB108-T7-CDC2LC (p58) |
| 4p- | 4p16.3 | *WHSC1* |
| 5p- | 5p15.2 | D5S23-D5S721 |
| 15q11.2-12 duplication | 15q11.2-12 | D15S11, D15S10 |
| 22qter deletion | 22q13.3 | 22qtel (*SHANK3*) |
| Angelman | 15q11.2-12 | 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 | 15q11.2-12 | 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 |