

VERIPATH LABORATORIES

Cytogenetics Laboratory
Fluorescence in situ hybridization (FISH) Tests
Constitutional Disorders

Constitutional Disorders	Cytogenetic Aberration/Locus	Probe Name	Detection Parameters
1p- Syndrome	1p36	p58	Detects deletions of terminal 1p
Ambiguous Genitalia	Xcen, Ycen	DXZ1, DYZ3	Detects sex chromosomes
Aneuploidy/Trisomy Panel	Xcen/Ycen , 13q14, 18cen, 21q22.13-q22.2,	Aneuploidy probe set	Detects aneuploidy of 13, 18, 21, X and Y
Angelman Syndrome	15q11.2q13	UBE3A	Detects deletions of UBE3A gene
Cri du chat Syndrome	5p15.2	D5S23	Detects deletions of short arm of chromosome 5
DiGeorge Syndrome	22q11.2	22q11.2 probe set	Detects deletions of 22q11.2
Male sex determination	Yp11.3	SRY	Detects presence or absence of SRY gene
Miller-Dieker Syndrome	17p13.3	LIS1	Detects deletions of LIS1 gene
Prader-Willi Syndrome	15q11.2q13	SNRPN	Detects deletions of SNRPN gene
Shprintzen Syndrome	22q11.2	22q11.2 probe set	Detects deletions of 22q11.2
Smith-Magenis Syndrome	17p11.2	SHMT1, FLII, TOP3	Detects deletions of 17p11.2
Telomere Panel	Subtelomeric Sequences	Telomere probe set	Detects cryptic rearrangements of subtelomeric sequences
Turner Syndrome	Xcen, Ycen	DXZ1, DYZ3	Detects sex chromosomes
Velocardiofacial Syndrome	22q11.2	22q11.2 probe set	Detects deletions of 22q11.2
Williams Syndrome	7q11.23	7q11.23 probe set	Detects deletions of 7q11.23
Wolf-Hirschhorn Syndrome	4p16.3	D4S96	Detects deletions of short arm of chromosome 4