Constitutional Disorders (Microdeletion Syndrome) FISH Probes

Chromosome	Locus / Loci	Related Syndrome or Condition
4	4p16.1 / 4p11-q11	Wolf-Hirschhorn Syndrome
5	5q31 / 5p15.2	Cri du Chat
10	10q23 / 10p11.1-q11.1	PTEN
13	13q14	Retinoblastoma region (~10% are deletions)
13	13q14	Trisomy 13
15	15q11-13 / 15q22 / 15p11.2	Prader-Willi / Angelman Syndrome
17	17p11.2 / 17q21	Smith-Magenis
17	17p13.3 / 17q21.1	Miller-Dieker
18	18p11.1q11.1	Trisomy 18
21	21q22.13-q22.2	Trisomy 21
22	22q11.2 / 22q13	VCF / DiGeorge Region
X	Xp22.3 / Xp11.1-q11.1	X-linked Ichthyosis / Steroid Sulfatase Deficiency
X	Xp22.3 / Xp11.1-q11.1	Kallman Syndrome
X/Y	Yp11.3 / Xp11.1q11.1	Sex determination / Ambiguous genitalia
X/Y (Centromeres)	Xp11.1q11.1 / Yp11.1q11.1	Sex determination / Ambiguous genitalia
Multiple	13, 18, 21, X, Y	Prenatal FISH for trisomy and sex chromosome anomalies

Please contact the lab at 513-636-4474 or labgeneticcounselors@cchmc.org with additional questions related to FISH testing and/or FISH probe availability.

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changing the outcome together

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