UPMC
LIFECHANGINGMEDICINE

PITTSBURGH CYTOGENETICS LABORATORY

NON-ONCOLOGY FISH PROBE LIST

PROBE FOR MICRODELETION AND DUPLICATION		
SYNDROMES	**REGIONS**	**PROBES**
1p36 microdeletion	1p36	p58
Cri-du-Chat syndrome	5p15.2/5q31	D5S23, D5S721/EGR1
DiGeorge/Velo-Cardio-Facial syndrome	22q11.2	TUPLE1 (HIRA)
DiGeorge II	10p14	Di-George II/SE10
Kallmann syndrome	Xp22.3	KAL/XCEP
Miller-Dieker syndrome/Lissencephaly	17p13.3	LSI LIS1/LSI RARA
Phelan-McDermid syndrome	22q13.3	SHANK3
Prader-Willi/Angelman Region	15q11.2	SNRPN/15CEP (D15Z1)/PML; D15S10/15CEP (D15Z1)/PML;LSI D15S11/15CEP (D15Z1)
SHOX1 deletion	Xp22.33;Yp11.3	SHOX
Smith-Magenis syndrome	17p11.2/17q21.1	SMS Region/RARA
Sotos syndrome deletion	5q35	NSD1
Steroid Sulfatase deficiency	Xp22.3/XCEP	STS/CEPX
SRY	Yp11.3	SRY
Wolf-Hirschhorn syndrome	4p16.3	WHS/4CEP
Williams syndrome deletion	7q11.23/7q31	ELN/D7S486, D7S522

AneuVysion PROBE		
SYNDROMES	**REGIONS**	**PROBE REGION**
Sex chromosomes aneuploidy Trisomy 18	Xp11.1-q11.1/ Yp11.1-q11.1/18p11.1-q11.1	DXZ1/ DYZ3/ D18Z1
Trisomy 13/ Trisomy 21	13q14/ 21q22.1-q22.2	RB1/D21S259, D21S341, D21S342 (21q22.13-q22.2)

CHROMOSOME ENUMERATION PROBES		
Chromosome 1 (1CEP)	Chromosome 9 (9CEP)	Chromosome 16 (16CEP)
Chromosome 2 (2CEP)	Chromosome 10 (10CEP)	Chromosome 17 (17CEP)
Chromosome 3 (3CEP)	Chromosome 11 (11CEP)	Chromosome 17 (18CEP)
Chromosome 4 (4CEP)	Chromosome 12 (12CEP)	Chromosome 20(20CEP)
Chromosome 6 (6CEP)	Chromosome 13/21 (13/21 alpha sat)	Chromosome 21/13 (alpha sat)
Chromosome 7 (7CEP)	Chromosome 14/22 (14/22 alpha sat)	Chromosome 22/14 (alpha sat)
Chromosome 8 (8CEP)	Chromosome 15 (alpha sat)	Chromosome X (XCEP)
Chromosome Y (YCEP)	Chromosome Y heterochromatic region (DYZ1)	

SUBTELOMERIC PROBES	
The 41 TelVysion probes are specific to:	p and q subtelomeres of chromosomes 1-12 and 16-20 q subtelomeres of the acrocentric chromosomes (13, 14, 15, 21, and 22) Xp/Yp and Xq/Yq pseudo-autosomal region subtelomeres

CUSTOM PROBES
Custom probes prepared from human genome BAC library

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