

Supplemental Table 1s. Analysis of the 18 loci investigated

Near telomere	Selective Tiling version of probe available	Region	Size	Probe Coverage (bp)	Non-RM Bases (bp)	Pct Masked	Num Gaps	Median Gap Size (kb)	Max Gap Size (bp)	Pct Probe Coverage across whole region	Pct Probe Coverage across only Non-RM region
yes		chr10:135142720-135266554	123,834	26,750	40,282	67%	43	405	16661	22%	66%
	yes	chr15:029818104-030297359	479,255	144,050	200,065	58%	263	354	14652	30%	72%
yes	yes	chr16:000142049-000170588	28,539	15,850	18,426	35%	22	290	1834	56%	86%
yes	yes	chr16:000155524-000171101	15,577	10,350	11,640	25%	12	152	1702	66%	89%
	yes	chr17:003490013-003535283	45,270	24,850	29,054	36%	33	361	2028	55%	86%
		chr19:033058446-033091539	33,093	17,050	18,372	44%	15	379	9516	52%	93%
		chr2:110219652-110316643	96,991	31,550	39,227	60%	49	306	10462	33%	80%
yes		chr2:242646152-242723252	77,100	8,050	45,320	41%	49	369	3864	10%	18%
yes	yes	chr4:000041213-000103724	62,511	6,450	22,028	65%	36	477	7482	10%	29%
yes		chr4:001955027-001978148	23,121	8,550	9,053	61%	5	660	1221	37%	94%
yes		chr5:000074949-000191587	116,638	40,150	59,620	49%	58	226	11887	34%	67%
yes		chr5:000124704-000167026	42,322	29,750	32,078	24%	23	178	3371	70%	93%
yes		chr6:000174615-000284943	110,328	55,350	63,183	43%	80	244.5	8350	50%	88%
	yes	chr6:026292020-026298726	6,706	3,550	4,582	32%	6	318	611	53%	77%
		chr8:116712325-116750490	38,165	28,750	31,615	17%	27	102	2396	75%	91%
yes		chr9:000210053-000285546	75,493	30,650	36,950	51%	65	309	6708	41%	83%
		chr9:138270360-138349890	79,530	34,350	36,865	54%	25	289	30523	43%	93%
	yes	chrX:139312266-139343233	30,967	6,750	7,677	75%	8	1010	12349	22%	88%

RM = Repeat Masked

Supplemental Table 2. Results for Simple Tiling Probes for 18 Loci

Locus	Region size (kb)	Tile size (kb)	I	Sp	Se	Rep	Values
Xq27.1*	31	6.8	1	-----	-----	-----	1
2q13	97	32	1	5	5	5	20
2q37.3*	77	8.0	4	5	5	5	19
4p16.3*	63	6.5	2	5	5	5	17
4p16.3	23	8.6	4	5	5	5	19
5p15.3	117	40	5	5	5	5	20
5p15.3	42	30	5	5	5	5	20
6p22.2	6.7	3.6	1	-----	-----	-----	1
6p25.3**	110	55	5	1	5	5	16
8q23.3	38	29	3	5	5	5	18
9p24.3	76	31	5	5	5	5	20
9q34.3	80	34	5	5	5	5	20
10q23	124	27	5	5	5	5	20
15q13.3*	479	144	5	5	5	5	20
16p13.3*	29	16	4	5	5	5	19
16p13.3*	16	10	4	5	5	5	19
17p13.3*	45	25	5	5	5	5	20
19q12	33	17	4	5	5	5	19

I = Intensity; Sp = Specificity; Se = Sensitivity; Rep = Reproducibility

***Selective Tiling version of probe available **This locus represents a known polymorphic site and was shown to hybridize to two separate regions.**

Supplemental Table 3: Results for Selective Tiling Probes in Seven Loci

Locus	Region size (kb)	Tile Size (kb)	I	Sp	Se	Rep	Value
6p22.2	6.7	3.6	2	4	4	5	15
15q13.3	479	144	5	5	5	5	20
16p13.3	29	16	5	5	5	5	20
16p13.3	16	10	5	5	5	5	20
17p13.3	45	25	5	5	5	5	20
4p16.3	63	8.6	3	5	5	5	18
Xq27.1	31	6.8	3	5	5	1	14

I = Intensity; Sp = Specificity; Se = Sensitivity; Rep = Reproducibility

Supplemental Table 4. Oligonucleotide-based FISH confirmed aCGH findings in 100% of samples tested

Locus	Array Findings	FISH Findings
2q13	Deletion	Deletion
2q37.3	Deletion	Deletion
4p16.3	Duplication	Duplication
4p16.3	Deletion	Deletion
5p15.3	Duplication	Duplication
5p13.3	Duplication	Duplication
6p25.3	Duplication	Duplication
9p24.3	Duplication	Duplication
9q34.3	Duplication	Duplication
10q26.3	Duplication	Duplication
15q13.3	Duplication	Duplication