Case: Loss_of_1p36_no_3 Folder: P245-vs02-test Date: 09-03-2009

P245-vs02 Microdeletion syndromes (lot 1207) Female Reference: P245-vs02_t04 Ver.: 5.11

					Peak Data				Normalized Peak Area								
No.	**Label**	**Size**	Ref. size	Size diff.	MRC size	**Height**	**Width**	**Area**	Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel	band	**Ratio**	**Dist. in SD**	**1.0 low high**
4	64 -	61.03	60.65	0.38	64	175	7.9	1386	0.291	1.438	0.213	0.89		64 nt	0.20	-5.4*	|||||||·
5	70 -	66.73	66.46	0.27	70	302	18.7	5644	1.187	0.773	0.104	0.98		70 nt	1.53	4.0	·|||||
6	76 -	72.56	72.37	0.19	76	317	20.5	6484	1.363	1.055	0.125	1.12		76 nt	1.29	2.5	·|||
7	82 -	78.79	78.56	0.23	82	335	16.5	5511	1.159	0.734	0.097	1.00		82 nt	1.58	4.4*	·||||||
Ctrl: Q-fragments					Mean	282	15.9	4756	1.000	1.000	0.134	1.00		(CV: **0.53**)	**1.18**		
8	6 a	85.17	84.95	0.22	88	1984	11.9*	23538	1.153	0.834	0.103	0.95		6p21.3 CpG isl.	1.38	3.1	·||||
10	2 a	90.39	90.17	0.22	92	1884	10.4	19633	0.962	0.824	0.081	1.20		2q14 synt.	1.17	1.7	·||
11	1 a	96.71	96.50	0.21	96	3688	11.8*	43504	2.131	1.685	0.232	0.85	MV 36	1p36 CpG isl.	1.26	1.9	·|||
Ctrl: D-fragments					Mean	2519	11.4	28892	1.416	1.115	0.138	1.00		(CV: **0.09**)	**1.26**		
	Y a		114.50		118					0.811			13.54	Yq11			
	Y a		104.34		108					0.566			14.10	Yq11			
ctrl: Y-fragments (male ref.)					Mean							1.00		(CV:)			
12	1 A	126.36	126.04	0.32	130	1368	11.0	15110	0.740	1.133	0.040	1.25	1.14	1p36.33	0.65	-9.7*	|||·
20	1 A	176.62	176.49	0.13	178	958	11.9	11404	0.559	0.839	0.046	0.81	1.75	1p36.33	0.67	-6.0*	|||·
18	1 A	164.63	164.53	0.10	166	1065	11.3	11986	0.587	1.090	0.052	0.94	1.95	1p36.33	0.54	-9.7*	|||||·
1p36 (1p-deletion)					Mean	1130	11.4	12833	0.629	1.021	0.046	1.00		(CV: **0.11**)	**0.62**		
34	2 B	264.99	264.89	0.10	267	1692	13.2	22269	0.938	0.910	0.038	1.17	58.30	2p16.1	1.03	0.7	·
60	2 D	481.58	481.31	0.27	486	1063	18.7	19857	0.918	1.072	0.063	0.83	61.00	2p16.1	0.86	-2.5	|·
"2p16.1 deletion syndrome"					Mean	1378	15.9	21063	0.928	0.991	0.050	1.00		(CV: **0.13**)	**0.96**		
53	3 D	416.61	416.46	0.15	418	1757	16.7	29381	1.359	1.230	0.057	1.15	198.51	3q29	1.10	2.3	·|
46	3 C	354.63	354.58	0.05	359	1441	15.0	21649	0.897	0.959	0.061	0.85	198.28	3q29	0.94	-1.0	|·
"3q29 deletion syndrome"					Mean	1599	15.9	25515	1.128	1.094	0.059	1.00		(CV: **0.11**)	**1.03**		
29	4 B	230.61	230.47	0.14	232	1761	12.1	21339	0.898	0.896	0.058	0.88	1.81	4p16.3	1.00	0.0	·
56	4 D	443.29	443.03	0.26	445	1202	17.6	21195	0.980	0.880	0.045	1.12	1.90	4p16.3	1.11	2.2	·|
4p16.3 Wolf-Hirschhorn region					Mean	1482	14.9	21267	0.939	0.888	0.052	1.00		(CV: **0.07**)	**1.06**		
55	5 D	435.75	435.58	0.17	436	840	17.0	14314	0.662	0.655	0.037	1.02	1.34	5p15.33	1.01	0.2	·
36	5 C	281.52	281.39	0.13	283	2084	14.0	29093	1.206	1.053	0.061	0.98	1.40	5p15.33	1.14	2.5	·|
Cri du Chat syndrome					Mean	1462	15.5	21704	0.934	0.854	0.049	1.00		(CV: **0.09**)	**1.08**		
16	5 A	151.61	151.42	0.19	154	2289	10.4	23821	1.167	0.972	0.047	0.98	176.62	5q35.3	1.20	4.2*	·||
57	5 D	453.15	453.02	0.13	454	1278	17.9	22857	1.057	1.134	0.052	1.02	176.65	5q35.3	0.93	-1.5	|·
Sotos syndrome					Mean	1784	14.1	23339	1.112	1.053	0.049	1.00		(CV: **0.18**)	**1.06**		
40	7 C	310.46	310.40	0.06	310	1944	14.0	27289	1.131	1.047	0.040	1.13	73.08	7q11.23	1.08	2.1	·|
47	7 C	363.49	363.46	0.03	364	1526	15.7	24016	0.995	0.995	0.048	0.88	73.11	7q11.23	1.00	0.0	·
50	7 D	389.97	389.95	0.02	391	1125	16.1	18134	0.839	0.774	0.034	0.99	73.15	7q11.23	1.08	1.9	·|
Williams syndrome					Mean	1532	15.3	23146	0.988	0.938	0.041	1.00		(CV: **0.04**)	**1.06**		
51	8 D	399.81	399.69	0.12	401	872	16.5	14385	0.665	0.753	0.051	0.88	116.75	8q24.12	0.88	-1.7	|·
54	8 D	424.62	424.33	0.29	427	1107	17.5	19361	0.895	0.897	0.048	1.12	117.73	8q24.11	1.00	0.0	·
Langer-Giedion syndrome					Mean	990	17.0	16873	0.780	0.825	0.049	1.00		(CV: **0.08**)	**0.95**		
41	9 C	319.46	319.40	0.06	319	2017	14.4	29056	1.204	1.194	0.050	0.92	100.95	9q22.33	1.01	0.2	·
52	9 D	408.28	408.03	0.25	409	1449	16.6	24053	1.112	1.115	0.040	1.08	100.95	9q22.33	1.00	-0.1	·
"9q22.3 deletion syndrome"					Mean	1733	15.5	26555	1.158	1.155	0.045	1.00		(CV: **0.01**)	**1.00**		
13	10 A	132.86	132.56	0.30	136	1824	11.4	20812	1.020	0.895	0.048	0.90	8.14	10p	1.14	2.6	·|
45	10 C	349.34	349.23	0.11	349	1334	15.1	20105	0.833	0.938	0.041	1.10	10.59	10p15.1	0.89	-2.6	|·
DiGeorge region 2 (10p)					Mean	1579	13.2	20459	0.926	0.917	0.044	1.00		(CV: **0.18**)	**1.00**		
27	11 B	218.82	218.68	0.14	220	2375	11.9	28267	1.190	1.184	0.062	1.00	31.78	11p13	1.01	0.1	·
WAGR syndrome					Mean	2375	11.9	28267	1.190	1.184	0.062	1.00		(CV:)	**1.01**		
26	15 B	213.63	213.54	0.09	214	2492	11.9	29533	1.243	1.162	0.060	0.90	21.48	15q11.2	1.07	1.3	·|
31	15 B	245.24	245.06	0.18	247	1537	12.4	19125	0.805	0.909	0.047	0.90	22.65	15q12	0.89	-2.2	|·
37	15 C	289.79	289.75	0.04	292	1662	13.5	22510	0.933	0.932	0.035	1.25	22.76	15q12	1.00	0.0	·
17	15 A	158.17	158.00	0.17	160	1737	11.5	19959	0.978	0.838	0.041	0.95	23.17	15q12	1.17	3.4	·||
Prader-Willi / Angelman					Mean	1857	12.3	22782	0.990	0.960	0.046	1.00		(CV: **0.11**)	**1.03**		
22	15 A	189.90	189.75	0.15	190	2371	11.5	27205	1.333	1.228	0.057	1.07	72.50	15q24.1	1.09	1.8	·|
42	15 C	326.43	326.43	0.00	325	1172	14.7	17286	0.716	0.753	0.040	0.93	72.80	15q24.1	0.95	-0.9	·
"15q24 deletion syndrome"					Mean	1772	13.1	22246	1.025	0.990	0.049	1.00		(CV: **0.09**)	**1.02**		
19	16 A	171.04	170.91	0.13	172	1840	11.3	20882	1.023	0.921	0.053	1.00	3.87	16p13.3	1.11	1.9	·|
Rubinstein-Taybi syndrome					Mean	1840	11.3	20882	1.023	0.921	0.053	1.00		(CV:)	**1.11**		
14	17 A	139.26	138.97	0.29	142	2304	11.5	26517	1.299	1.138	0.031	1.42	2.51	17p13.3	1.14	5.2*	·|
30	17 B	236.30	236.22	0.08	238	1750	12.5	21929	0.923	0.859	0.058	0.58	2.52	17p13.3	1.08	1.1	·|
Miller-Dieker region					Mean	2027	12.0	24223	1.111	0.999	0.044	1.00		(CV: **0.04**)	**1.12**		
58	17 D	463.41	463.07	0.34	465	1846	18.3	33713	1.559	1.385	0.097	0.75	17.53	17p11.2-##	1.13	1.8	·|
35	17 B	272.00	271.90	0.10	274	1616	13.2	21302	0.897	0.934	0.038	1.30	17.83	17p11.2	0.96	-1.0	·
39	17 C	302.94	302.90	0.04	303	1379	13.8	19015	0.788	0.796	0.044	0.95	18.08	17p11.2	0.99	-0.2	·
Smith-Magenis syndrome					Mean	1614	15.1	24677	1.081	1.038	0.060	1.00		(CV: **0.08**)	**1.01**		
33	17 B	258.37	258.33	0.04	260	1097	13.4	14693	0.619	0.578	0.041	0.94	26.56	17q11.2	1.07	1.0	·|
43	17 C	334.60	334.45	0.15	335	1756	14.5	25518	1.058	1.044	0.065	1.06	26.58	17q11.2	1.01	0.2	·
NF1 microdeletion syndrome					Mean	1427	14.0	20106	0.838	0.811	0.053	1.00		(CV: **0.04**)	**1.04**		
59	17 D	470.80	470.58	0.22	472	845	18.3	15481	0.716	0.728	0.046	0.78	41.26	17q21.31	0.98	-0.3	·
28	17 B	224.94	224.79	0.15	226	2453	12.3	30206	1.272	1.273	0.049	1.26	41.44	17q21.31	1.00	0.0	·
44	17 C	341.84	341.78	0.06	342	1349	15.2	20558	0.852	0.896	0.046	0.95	41.45	17q21.31	0.95	-1.0	·
"17q21.31 microdeletion"					Mean	1549	15.3	22082	0.947	0.966	0.047	1.00		(CV: **0.03**)	**0.98**		

Case: Loss_of_1p36_no_3 Folder: P245-vs02-test Date: 09-03-2009

P245-vs02 Microdeletion syndromes (lot 1207) Female Reference: P245-vs02_t04 Ver.: 5.11

No.	Label	Size	Ref. size	Size diff.	MRC size	Height	Width	Area	Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel	band	Ratio	Dist. in SD	1.0 low high
23	22 A	195.36	195.29	0.07	196	1756	10.9	19061	0.934	0.767	0.034	1.08	17.89	22q11.21	1.22	5.0*	·‖
25	22 B	208.14	207.99	0.15	208	1997	11.9	23826	1.003	1.070	0.053	0.96	18.09	22q11.21	0.94	-1.3	| ·
48	22 D	371.52	371.47	0.05	373	1136	16.7	18958	0.877	0.942	0.047	0.96	19.57	22q11.21	0.93	-1.4	| ·
22q11.21 (DiGeorge)					Mean	1630	13.2	20615	0.938	0.926	0.044	1.00	(CV: **0.16**)		**1.04**		
32	22 B	252.04	251.96	0.08	253	2359	13.1	30949	1.303	1.330	0.090	0.75	49.49	22q13.33-##	0.98	-0.3	·
49	22 D	382.30	382.09	0.21	382	1680	16.8	28159	1.302	1.436	0.059	1.25	49.50	22q13.33-#	0.91	-2.3	| ·
22q13 (Phelan-McDermid)					Mean	2020	14.9	29554	1.303	1.383	0.075	1.00	(CV: **0.05**)		**0.93**		
38	X C	295.55	295.46	0.09	297	2356	13.6	32118	1.331	1.392	0.054	1.00	32.29	Xp21.2	0.96	-1.1	·
Chromosome X control probe					Mean	2356	13.6	32118	1.331	1.392	0.054	1.00	(CV:)		**0.96**		
24	X B	199.76	199.64	0.12	202	1824	11.1	20301	0.855	0.895	0.045	1.00	153.02	Xq28	0.95	-0.9	·
15	X A	147.05	146.82	0.23	148	1998	10.2	20298	0.994	0.964	0.056	0.86	152.95	Xq28	1.03	0.5	·
21	X A	183.23	183.16	0.07	184	2330	11.5	26762	1.311	1.214	0.053	1.14	152.94	Xq28-#	1.08	1.8	·|
Xq28 (RETT / MECP2)					Mean	2051	10.9	22454	1.053	1.025	0.051	1.00	(CV: **0.06**)		**1.02**		
Mean values			0.14			1653	13.9	22360	**0.995**	1.000	0.050	**4**			1.00	Total of all except	
Standard deviations			0.09		(Coef. of variance:			0.239)	0.232	0.196					0.13	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	4.29
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.64
Mean height of first probes AB	> 450 (800)	1866
Mean height of last probes CD	> 280 (500)	1449
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.29
Mean group CV of weighted ratio	<0.20 (0.15)	0.09
1 uinidentified peak area / 52 peak areas	< (0.02)	0.00

Individual peaks having normalized area > 4.0 SD from the ref. mean and ratio <0.65 or >1.3 indicate 'abnormal' probe area.

Female Reference

Abn. peaks: 1p36.33

An "*" marks: Size Diff.>0.5, Peak Height>7000, unexpected peak width, and "Dist. in SD">4.0.
Ratio group mean and coefficient of variance (CV) are weighted by the ref. weights
Labels A,B,... define normalization groups; a,b,... labeled probes do not contribute to normalization.
Mean Rox height is 146 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.08

("#" marked probes are often low when CpG-D-fragments are low)