Case: Gain_of_17p13.3_no_2 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
3	64 -	60.90	60.65	0.25	64	156	7.6	1190	0.488	1.438	0.213	0.89	64 nt		0.34	-4.5 *	·|||||||
4	70 -	66.64	66.46	0.18	70	200	17.1	3412	1.398	0.773	0.104	0.98	70 nt		1.81	6.0 *	·||||||||
5	76 -	72.50	72.37	0.13	76	189	15.9	3001	1.229	1.055	0.125	1.12	76 nt		1.17	1.4	·||
6	82 -	78.81	78.56	0.25	82	127	17.0	2161	0.885	0.734	0.097	1.00	82 nt		1.21	1.6	·||
Ctrl: Q-fragments					Mean	168	14.4	2441	1.000	1.000	0.134	1.00	(CV: **0.51**)		**1.15**		
7	6 a	85.18	84.95	0.23	88	1379	11.8 *	16268	0.803	0.834	0.103	0.95	6p21.3	CpG isl.	0.96	-0.3	·
9	2 a	90.35	90.17	0.18	92	1565	10.7 *	16673	0.823	0.824	0.081	1.20	2q14	synt.	1.00	0.0	·
10	1 a	96.80	96.50	0.30	96	2860	11.7 *	33382	1.648	1.685	0.232	0.85	MV 36 1p36	CpG isl.	0.98	-0.2	·
Ctrl: D-fragments					Mean	1935	11.4	22108	1.092	1.115	0.138	1.00	(CV: **0.02**)		**0.98**		
	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:)				
11	1 A	126.23	126.04	0.19	130	2052	11.2	22913	1.131	1.133	0.040	1.25	1.14	1p36.33	1.00	0.0	·
19	1 A	176.64	176.49	0.15	178	1496	11.8	17589	0.869	0.839	0.046	0.81	1.75	1p36.33	1.04	0.6	·
17	1 A	164.61	164.53	0.08	166	1932	11.4	21999	1.086	1.090	0.052	0.94	1.95	1p36.33	1.00	-0.1	·
1p36 (1p-deletion)					Mean	1827	11.4	20834	1.029	1.021	0.046	1.00	(CV: **0.02**)		**1.01**		
33	2 B	264.99	264.89	0.10	267	1250	14.0	17529	0.912	0.910	0.038	1.17	58.30	2p16.1	1.00	0.0	·
59	2 D	481.70	481.31	0.39	486	811	21.0	17030	1.051	1.072	0.063	0.83	61.00	2p16.1	0.98	-0.3	·
"2p16.1 deletion syndrome"					Mean	1031	17.5	17280	0.981	0.991	0.050	1.00	(CV: **0.01**)		**0.99**		
52	3 D	416.76	416.46	0.30	418	1148	18.7	21510	1.327	1.230	0.057	1.15	198.51	3q29	1.08	1.7	·|
45	3 C	354.70	354.58	0.12	359	1180	16.0	18822	0.987	0.959	0.061	0.85	198.28	3q29	1.03	0.5	·
"3q29 deletion syndrome"					Mean	1164	17.3	20166	1.157	1.094	0.059	1.00	(CV: **0.03**)		**1.06**		
28	4 B	230.74	230.47	0.27	232	1357	12.6	17137	0.891	0.896	0.058	0.88	1.81	4p16.3	1.00	-0.1	·
55	4 D	443.49	443.03	0.46	445	771	20.1	15476	0.955	0.880	0.045	1.12	1.90	4p16.3	1.09	1.7	·|
4p16.3 Wolf-Hirschhorn region					Mean	1064	16.4	16307	0.923	0.888	0.052	1.00	(CV: **0.06**)		**1.05**		
54	5 D	435.81	435.58	0.23	436	590	19.2	11317	0.698	0.655	0.037	1.02	1.34	5p15.33	1.07	1.2	·|
35	5 C	281.40	281.39	0.01	283	1360	14.7	19992	1.049	1.053	0.061	0.98	1.40	5p15.33	1.00	-0.1	·
Cri du Chat syndrome					Mean	975	16.9	15655	0.874	0.854	0.049	1.00	(CV: **0.05**)		**1.03**		
15	5 A	151.48	151.42	0.06	154	1956	10.2	20035	0.989	0.972	0.047	0.98	176.62	5q35.3	1.02	0.4	·
56	5 D	453.26	453.02	0.24	454	996	19.9	19869	1.226	1.134	0.052	1.02	176.65	5q35.3	1.08	1.8	·|
Sotos syndrome					Mean	1476	15.1	19952	1.108	1.053	0.049	1.00	(CV: **0.04**)		**1.05**		
39	7 C	310.08	310.40	-0.32	310	1349	15.4	20745	1.088	1.047	0.040	1.13	73.08	7q11.23	1.04	1.0	·
46	7 C	363.72	363.46	0.26	364	1282	17.1	21956	1.152	0.995	0.048	0.88	73.11	7q11.23	1.16	3.2	·||
49	7 D	390.27	389.95	0.32	391	762	17.9	13633	0.841	0.774	0.034	0.99	73.15	7q11.23	1.09	2.0	·|
Williams syndrome					Mean	1131	16.8	18778	1.027	0.938	0.041	1.00	(CV: **0.05**)		**1.09**		
50	8 D	400.19	399.69	0.50	401	652	18.0	11756	0.726	0.753	0.051	0.88	116.75	8q24.12	0.96	-0.5	·
53	8 D	424.82	424.33	0.49	427	731	19.6	14309	0.883	0.897	0.048	1.12	117.73	8q24.11	0.98	-0.3	·
Langer-Giedion syndrome					Mean	692	18.8	13033	0.804	0.825	0.049	1.00	(CV: **0.01**)		**0.98**		
40	9 C	319.04	319.40	-0.36	319	1635	15.5	25358	1.330	1.194	0.050	0.92	100.95	9q22.33	1.11	2.7	·|
51	9 D	408.45	408.03	0.42	409	1086	18.4	20020	1.236	1.115	0.040	1.08	100.95	9q22.33	1.11	3.0	·|
"9q22.3 deletion syndrome"					Mean	1361	17.0	22689	1.283	1.155	0.045	1.00	(CV: **0.00**)		**1.11**		
12	10 A	132.76	132.56	0.20	136	1530	11.1	16943	0.837	0.895	0.048	0.90	8.14	10p	0.93	-1.2	|·
44	10 C	349.38	349.23	0.15	349	1073	16.0	17211	0.903	0.938	0.041	1.10	10.59	10p15.1	0.96	-0.9	·
DiGeorge region 2 (10p)					Mean	1302	13.6	17077	0.870	0.917	0.044	1.00	(CV: **0.02**)		**0.95**		
26	11 B	218.83	218.68	0.15	220	1834	12.5	22838	1.188	1.184	0.062	1.00	31.78	11p13	1.00	0.1	·
WAGR syndrome					Mean	1834	12.5	22838	1.188	1.184	0.062	1.00	(CV:)		**1.00**		
25	15 B	213.65	213.54	0.11	214	1948	12.3	23947	1.245	1.162	0.060	0.90	21.48	15q11.2	1.07	1.4	·|
30	15 B	245.32	245.06	0.26	247	1324	13.5	17856	0.929	0.909	0.047	0.90	22.65	15q12	1.02	0.4	·
36	15 C	289.75	289.75	0.00	292	1265	14.4	18279	0.959	0.932	0.035	1.25	22.76	15q12	1.03	0.8	·
16	15 A	158.13	158.00	0.13	160	1785	11.4	20290	1.002	0.838	0.041	0.95	23.17	15q12	1.20	4.0	·||
Prader-Willi / Angelman					Mean	1581	12.9	20093	1.034	0.960	0.046	1.00	(CV: **0.07**)		**1.08**		
21	15 A	189.88	189.75	0.13	190	2057	11.7	24157	1.193	1.228	0.057	1.07	72.50	15q24.1	0.97	-0.6	·
41	15 C	326.06	326.43	-0.37	325	904	15.3	13832	0.726	0.753	0.040	0.93	72.80	15q24.1	0.96	-0.7	·
"15q24 deletion syndrome"					Mean	1481	13.5	18995	0.959	0.990	0.049	1.00	(CV: **0.01**)		**0.97**		
18	16 A	170.97	170.91	0.06	172	1536	11.6	17754	0.877	0.921	0.053	1.00	3.87	16p13.3	0.95	-0.8	·
Rubinstein-Taybi syndrome					Mean	1536	11.6	17754	0.877	0.921	0.053	1.00	(CV:)		**0.95**		
13	17 A	139.09	138.97	0.12	142	3046	11.6	35347	1.745	1.138	0.031	1.42	2.51	17p13.3	1.53	19.5 *	·|||||
29	17 B	236.27	236.22	0.05	238	2009	13.1	26364	1.371	0.859	0.058	0.58	2.52	17p13.3	1.60	8.9 *	·||||||
Miller-Dieker region					Mean	2528	12.4	30856	1.558	0.999	0.044	1.00	(CV: **0.03**)		**1.55**		
57	17 D	463.49	463.07	0.42	465	1048	20.0	20970	1.294	1.385	0.097	0.75	17.53	17p11.2-##	0.93	-0.9	|·
34	17 B	271.96	271.90	0.06	274	1254	14.1	17690	0.920	0.934	0.038	1.30	17.83	17p11.2	0.99	-0.4	·
38	17 C	302.79	302.90	-0.11	303	1074	14.9	16000	0.839	0.796	0.044	0.95	18.08	17p11.2	1.05	1.0	·|
Smith-Magenis syndrome					Mean	1125	16.3	18220	1.018	1.038	0.060	1.00	(CV: **0.06**)		**0.99**		
32	17 B	258.34	258.33	0.01	260	814	14.1	11449	0.595	0.578	0.041	0.94	26.56	17q11.2	1.03	0.4	·
42	17 C	334.27	334.45	-0.18	335	1319	15.9	20988	1.101	1.044	0.065	1.06	26.58	17q11.2	1.05	0.9	·|
NF1 microdeletion syndrome					Mean	1067	15.0	16219	0.848	0.811	0.053	1.00	(CV: **0.02**)		**1.04**		
58	17 D	470.94	470.58	0.36	472	565	19.9	11216	0.692	0.728	0.046	0.78	41.26	17q21.31	0.95	-0.8	·
27	17 B	225.07	224.79	0.28	226	1815	12.9	23425	1.218	1.273	0.049	1.26	41.44	17q21.31	0.96	-1.1	·
43	17 C	341.61	341.78	-0.17	342	975	16.4	15979	0.838	0.896	0.046	0.95	41.45	17q21.31	0.94	-1.3	|·
"17q21.31 microdeletion"					Mean	1118	16.4	16873	0.916	0.966	0.047	1.00	(CV: **0.01**)		**0.95**		

Case: Gain_of_17p13.3_no_2 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	Peak Data: Height	Width	Area	Normalized Peak Area: Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel	band	Ratio	Dist. in SD	1.0 low high
22	22 A	195.36	195.29	0.07	196	1330	11.2	14955	0.738	0.767	0.034	1.08	17.89	22q11.21	0.96	-0.8	·
24	22 B	208.23	207.99	0.24	208	1656	12.4	20503	1.066	1.070	0.053	0.96	18.09	22q11.21	1.00	-0.1	·
47	22 D	371.91	371.47	0.44	373	890	18.4	16359	1.010	0.942	0.047	0.96	19.57	22q11.21	1.07	1.5	·|
22q11.21 (DiGeorge)					Mean	1292	14.0	17272	0.938	0.926	0.044	1.00	(CV: **0.06**)		**1.01**		
31	22 B	252.16	251.96	0.20	253	1692	13.6	23076	1.200	1.330	0.090	0.75	49.49	22q13.33-##	0.90	-1.4	|·
48	22 D	382.54	382.09	0.45	382	1295	18.4	23778	1.467	1.436	0.059	1.25	49.50	22q13.33-#	1.02	0.5	·
22q13 (Phelan-McDermid)					Mean	1494	16.0	23427	1.334	1.383	0.075	1.00	(CV: **0.08**)		**0.98**		
37	X C	295.41	295.46	-0.05	297	1852	14.4	26744	1.403	1.392	0.054	1.00	32.29	Xp21.2	1.01	0.2	·
Chromosome X control probe					Mean	1852	14.4	26744	1.403	1.392	0.054	1.00	(CV:)		**1.01**		
23	X B	199.83	199.64	0.19	202	1422	11.3	16135	0.839	0.895	0.045	1.00	153.02	Xq28	0.94	-1.3	|·
14	X A	147.02	146.82	0.20	148	1838	9.8	18082	0.893	0.964	0.056	0.86	152.95	Xq28	0.93	-1.3	|·
20	X A	183.23	183.16	0.07	184	1744	11.8	20552	1.015	1.214	0.053	1.14	152.94	Xq28-#	0.84	-3.7	‖·
Xq28 (RETT / MECP2)					Mean	1668	11.0	18256	0.916	1.025	0.051	1.00	(CV: **0.06**)		**0.90**		
Mean values				0.15		1373	14.8	19219	**1.031**	1.000	0.050	**4**			1.03	Total of all except	
Standard deviations				0.21		(Coef. of variance:		0.237)	0.230	0.196					0.13	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**	
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	8.30	
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.23	
Mean height of first probes AB	> 450 (800)	1695	
Mean height of last probes CD	> 280 (500)	1065	
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.59	**high**
Mean group CV of weighted ratio	<0.20 (0.15)	0.04	
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: 17p13.3 17p13.3

1 quality warning!

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 122 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.12

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