Case: Gain_of_15q24_no_2 Folder: P245-vs02-test Date: 09-03-2009

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

No.	Label	Size	Ref. size	Size diff.	MRC size	Peak Height	Peak Width	Peak Area	Norm. Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel	Position band	Ratio	Dist. in SD	1.0 low high
4	64 -	60.92	60.65	0.27	64	164	8.4	1385	0.594	1.190	0.371	0.75		64 nt	0.50	-1.6	IIIII·
5	70 -	66.54	66.46	0.08	70	155	16.8	2603	1.117	0.913	0.264	0.80		70 nt	1.22	0.8	·II
6	76 -	72.58	72.37	0.21	76	154	20.7	3185	1.367	1.138	0.170	1.55		76 nt	1.20	1.3	·II
7	82 -	78.81	78.56	0.25	82	128	16.8	2146	0.921	0.787	0.204	0.90		82 nt	1.17	0.7	·II
Ctrl: Q-fragments					Mean	150	15.7	2330	1.000	1.007	0.252	1.00		(CV: **0.29**)	**1.07**		
8	6 a	85.17	84.95	0.22	88	1452	11.5 *	16640	0.834	0.981	0.152	1.02		6p21.3 CpG isl.	0.85	-1.0	II·
10	2 a	90.43	90.17	0.26	92	1617	10.3	16693	0.836	0.967	0.135	1.13		2q14 synt.	0.86	-1.0	I·
11	1 a	96.80	96.50	0.30	96	3039	11.4 *	34555	1.731	1.641	0.304	0.85	MV 36	1p36 CpG isl.	1.05	0.3	·I
Ctrl: D-fragments					Mean	2036	11.1	22629	1.134	1.197	0.197	1.00		(CV: **0.12**)	**0.91**		
16	Y a	114.63	114.50	0.13	118	1232	10.2	12566	0.630	0.811	0.088	1.03	13.54	Yq11	0.78	-2.1	II·
13	Y a	104.48	104.34	0.14	108	969	10.6	10254	0.514	0.566	0.065	0.97	14.10	Yq11	0.91	-0.8	I·
ctrl: Y-fragments (male ref.)					Mean	1101	10.4	11410	0.572	0.688	0.076	1.00		(CV: **0.11**)	**0.84**		
17	1 A	126.22	126.04	0.18	130	2382	11.0	26185	1.312	1.200	0.054	1.30	1.14	1p36.33	1.09	2.1	·I
25	1 A	176.58	176.49	0.09	178	1530	11.2	17078	0.856	0.942	0.074	0.75	1.75	1p36.33	0.91	-1.2	I·
23	1 A	164.65	164.53	0.12	166	2108	11.0	23266	1.166	1.181	0.073	0.95	1.95	1p36.33	0.99	-0.2	·
1p36 (1p-deletion)					Mean	2007	11.1	22176	1.111	1.108	0.067	1.00		(CV: **0.09**)	**1.01**		
39	2 B	265.00	264.89	0.11	267	1333	13.2	17530	1.009	0.948	0.040	1.36	58.30	2p16.1	1.06	1.5	·I
65	2 D	481.41	481.31	0.10	486	722	19.3	13953	0.911	1.121	0.101	0.64	61.00	2p16.1	0.81	-2.1	II·
"2p16.1 deletion syndrome"					Mean	1028	16.2	15742	0.960	1.034	0.071	1.00		(CV: **0.17**)	**0.98**		
58	3 D	416.59	416.46	0.13	418	1130	17.2	19398	1.267	1.268	0.073	0.96	198.51	3q29	1.00	0.0	·
51	3 C	354.61	354.58	0.03	359	1149	15.1	17393	0.954	1.039	0.056	1.04	198.28	3q29	0.92	-1.5	I·
"3q29 deletion syndrome"					Mean	1140	16.2	18396	1.110	1.154	0.064	1.00		(CV: **0.06**)	**0.96**		
34	4 B	230.65	230.47	0.18	232	1404	12.1	16923	0.974	0.944	0.063	0.88	1.81	4p16.3	1.03	0.5	·
61	4 D	443.27	443.03	0.24	445	749	17.7	13221	0.863	0.879	0.046	1.12	1.90	4p16.3	0.98	-0.3	·
4p16.3 Wolf-Hirschhorn region					Mean	1077	14.9	15072	0.919	0.912	0.054	1.00		(CV: **0.03**)	**1.00**		
60	5 D	435.68	435.58	0.10	436	616	17.5	10792	0.705	0.659	0.053	0.88	1.34	5p15.33	1.07	0.9	·I
41	5 C	281.49	281.39	0.10	283	1555	13.8	21409	1.174	1.115	0.071	1.12	1.40	5p15.33	1.05	0.8	·I
Cri du Chat syndrome					Mean	1086	15.6	16101	0.939	0.887	0.062	1.00		(CV: **0.01**)	**1.06**		
21	5 A	151.56	151.42	0.14	154	1955	10.4	20261	1.015	1.127	0.052	1.03	176.62	5q35.3	0.90	-2.1	I·
62	5 D	453.16	453.02	0.14	454	947	18.3	17360	1.134	1.123	0.055	0.97	176.65	5q35.3	1.01	0.2	·
Sotos syndrome					Mean	1451	14.3	18811	1.074	1.125	0.054	1.00		(CV: **0.08**)	**0.95**		
45	7 C	310.35	310.40	-0.05	310	1521	14.3	21774	1.194	1.116	0.056	0.99	73.08	7q11.23	1.07	1.4	·I
52	7 C	363.41	363.46	-0.05	364	1182	15.8	18620	1.021	1.029	0.040	1.27	73.11	7q11.23	0.99	-0.2	·
55	7 D	389.93	389.95	-0.02	391	706	16.4	11575	0.756	0.748	0.050	0.74	73.15	7q11.23	1.01	0.2	·
Williams syndrome					Mean	1136	15.5	17323	0.990	0.964	0.049	1.00		(CV: **0.04**)	**1.02**		
56	8 D	399.90	399.69	0.21	401	701	16.7	11722	0.766	0.776	0.036	1.06	116.75	8q24.12	0.99	-0.3	·
59	8 D	424.66	424.33	0.33	427	860	17.5	15053	0.983	0.895	0.047	0.94	117.73	8q24.11	1.10	1.9	·I
Langer-Giedion syndrome					Mean	781	17.1	13388	0.874	0.835	0.042	1.00		(CV: **0.08**)	**1.04**		
46	9 C	319.32	319.40	-0.08	319	1649	14.2	23419	1.284	1.292	0.072	1.05	100.95	9q22.33	0.99	-0.1	·
57	9 D	408.07	408.03	0.04	409	1166	16.9	19699	1.287	1.119	0.069	0.95	100.95	9q22.33	1.15	2.4	·I
"9q22.3 deletion syndrome"					Mean	1408	15.5	21559	1.285	1.206	0.070	1.00		(CV: **0.10**)	**1.07**		
18	10 A	132.79	132.56	0.23	136	1689	10.8	18282	0.916	0.948	0.043	1.16	8.14	10p	0.97	-0.7	·
50	10 C	349.23	349.23	0.00	349	1205	14.8	17860	0.979	0.979	0.060	0.84	10.59	10p15.1	1.00	0.0	·
DiGeorge region 2 (10p)					Mean	1447	12.8	18071	0.948	0.963	0.052	1.00		(CV: **0.02**)	**0.98**		
32	11 B	218.77	218.68	0.09	220	1765	11.8	20871	1.202	1.246	0.061	1.00	31.78	11p13	0.96	-0.7	·
WAGR syndrome					Mean	1765	11.8	20871	1.202	1.246	0.061	1.00		(CV:)	**0.96**		
31	15 B	213.51	213.54	-0.03	214	1864	11.9	22180	1.277	1.226	0.047	1.34	21.48	15q11.2	1.04	1.1	·
36	15 B	245.28	245.06	0.22	247	1275	12.8	16333	0.940	0.962	0.058	0.86	22.65	15q12	0.98	-0.4	·
42	15 C	289.84	289.75	0.09	292	1373	13.5	18564	1.018	1.018	0.061	0.86	22.76	15q12	1.00	0.0	·
22	15 A	158.14	158.00	0.14	160	1690	11.2	18933	0.949	0.930	0.051	0.94	23.17	15q12	1.02	0.4	·
Prader-Willi / Angelman					Mean	1551	12.4	19003	1.046	1.034	0.054	1.00		(CV: **0.03**)	**1.01**		
27	15 A	189.79	189.75	0.04	190	3453	11.4	39306	1.969	1.299	0.130	0.78	72.50	15q24.1	1.52	5.2 *	·IIIII
47	15 C	326.40	326.43	-0.03	325	1224	14.7	18045	0.989	0.771	0.049	1.22	72.80	15q24.1	1.28	4.4 *	·III
"15q24 deletion syndrome"					Mean	2339	13.1	28676	1.479	1.035	0.090	1.00		(CV: **0.12**)	**1.37**		
24	16 A	170.95	170.91	0.04	172	1788	10.8	19356	0.970	1.018	0.063	1.00	3.87	16p13.3	0.95	-0.8	·
Rubinstein-Taybi syndrome					Mean	1788	10.8	19356	0.970	1.018	0.063	1.00		(CV:)	**0.95**		
19	17 A	139.18	138.97	0.21	142	2122	11.2	23753	1.190	1.248	0.053	1.27	2.51	17p13.3	0.95	-1.1	·
35	17 B	236.25	236.22	0.03	238	1329	12.2	16165	0.931	0.900	0.067	0.73	2.52	17p13.3	1.03	0.5	·
Miller-Dieker region					Mean	1726	11.7	19959	1.060	1.074	0.060	1.00		(CV: **0.06**)	**0.98**		
63	17 D	463.36	463.07	0.29	465	1118	18.5	20698	1.352	1.331	0.092	0.83	17.53	17p11.2-##	1.02	0.2	·
40	17 B	271.96	271.90	0.06	274	1165	13.2	15387	0.886	0.940	0.043	1.25	17.83	17p11.2	0.94	-1.3	I·
44	17 C	302.92	302.90	0.02	303	1053	13.9	14625	0.802	0.839	0.053	0.91	18.08	17p11.2	0.96	-0.7	·
Smith-Magenis syndrome					Mean	1112	15.2	16903	1.013	1.037	0.063	1.00		(CV: **0.04**)	**0.97**		
38	17 B	258.44	258.33	0.11	260	845	13.3	11208	0.645	0.621	0.038	0.88	26.56	17q11.2	1.04	0.6	·
48	17 C	334.44	334.45	-0.01	335	1416	14.8	20990	1.151	1.118	0.053	1.12	26.58	17q11.2	1.03	0.6	·
NF1 microdeletion syndrome					Mean	1131	14.0	16099	0.898	0.870	0.046	1.00		(CV: **0.01**)	**1.03**		
64	17 D	470.69	470.58	0.11	472	601	18.8	11271	0.736	0.719	0.047	0.70	41.26	17q21.31	1.02	0.4	·
33	17 B	224.99	224.79	0.20	226	2003	12.2	24483	1.409	1.325	0.050	1.21	41.44	17q21.31	1.06	1.7	·I
49	17 C	341.64	341.78	-0.14	342	1120	15.5	17377	0.953	0.932	0.039	1.09	41.45	17q21.31	1.02	0.5	·
"17q21.31 microdeletion"					Mean	1241	15.5	17710	1.033	0.992	0.046	1.00		(CV: **0.02**)	**1.04**		

Case: Gain_of_15q24_no_2 Folder: P245-vs02-test Date: 09-03-2009

P245-vs02 Microdeletion syndromes (lot 1207) Male 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
28	22 A	195.31	195.29	0.02	196	1531	11.2	17192	0.861	0.890	0.065	0.83	17.89	22q11.21	0.97	-0.4	·
30	22 B	208.12	207.99	0.13	208	1717	11.7	20125	1.159	1.109	0.061	1.11	18.09	22q11.21	1.04	0.8	·
53	22 D	371.43	371.47	-0.04	373	896	17.2	15413	1.007	0.920	0.053	1.06	19.57	22q11.21	1.09	1.6	·|
22q11.21 (DiGeorge)					Mean	1381	13.4	17577	1.009	0.973	0.060	1.00	(CV: **0.06**)		**1.04**		
37	22 B	252.10	251.96	0.14	253	1770	13.1	23118	1.331	1.321	0.100	1.05	49.49	22q13.33-##	1.01	0.1	·
54	22 D	382.20	382.09	0.11	382	1354	16.7	22654	1.480	1.442	0.120	0.95	49.50	22q13.33-#	1.03	0.3	·
22q13 (Phelan-McDermid)					Mean	1562	14.9	22886	1.405	1.382	0.110	1.00	(CV: **0.01**)		**1.02**		
43	X C	295.50	295.46	0.04	297	965	13.4	12902	0.707	0.751	0.048	1.00	32.29	Xp21.2	0.94	-0.9	|·
Chromosome X control probe					Mean	965	13.4	12902	0.707	0.751	0.048	1.00	(CV:)		**0.94**		
29	X B	199.75	199.64	0.11	202	770	10.5	8054	0.464	0.457	0.047	0.99	153.02	Xq28	1.01	0.1	·
20	X A	147.02	146.82	0.20	148	1008	9.6	9700	0.486	0.538	0.065	0.84	152.95	Xq28	0.90	-0.8	|·
26	X A	183.19	183.16	0.03	184	947	11.3	10725	0.537	0.678	0.060	1.16	152.94	Xq28-#	0.79	-2.4	||·
Xq28 (RETT / MECP2)					Mean	908	10.5	9493	0.496	0.558	0.057	1.00	(CV: **0.13**)		**0.90**		
Mean values				0.09		1356	13.9	18004	**1.019**	1.000	0.060	**4**			1.02	Total of all except	
Standard deviations				0.10		(Coef. of variance:		0.294)	0.274	0.224					0.10	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	8.57
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.28
Mean height of first probes AB	> 450 (800)	1643
Mean height of last probes CD	> 280 (500)	1079
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.52
Mean group CV of weighted ratio	<0.20 (0.15)	0.07
4 uinidentified peak areas / 54 peak areas	< (0.02)	0.01

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

Male Reference

Abn. peaks: 15q24.1

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 166 (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)