Case: Loss_of_7q11.23_no_5 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
						Peak Data			Normalized Peak Area								
3	64 -	60.63	60.65	-0.02	64	139	6.7	936	0.274	1.190	0.371	0.75		64 nt	0.23	-2.5	|||||||| ·
4	70 -	66.49	66.46	0.03	70	274	17.5	4784	1.400	0.913	0.264	0.80		70 nt	1.53	1.8	·|||||
5	76 -	72.49	72.37	0.12	76	222	20.5	4559	1.334	1.138	0.170	1.55		76 nt	1.17	1.2	·||
6	82 -	78.51	78.56	-0.05	82	187	18.1	3392	0.992	0.787	0.204	0.90		82 nt	1.26	1.0	·|||
Ctrl: Q-fragments					Mean	206	15.7	3418	1.000	1.007	0.252	1.00		(CV: **0.46**)	**1.09**		
7	6 a	85.13	84.95	0.18	88	1772	11.3 *	20111	0.867	0.981	0.152	1.02		6p21.3 CpG isl.	0.88	-0.7	| ·
9	2 a	90.32	90.17	0.15	92	1973	10.4 *	20509	0.884	0.967	0.135	1.13		2q14 synt.	0.91	-0.6	| ·
10	1 a	96.71	96.50	0.21	96	3290	11.2 *	36975	1.594	1.641	0.304	0.85	MV 36	1p36 CpG isl.	0.97	-0.2	·
Ctrl: D-fragments					Mean	2345	11.0	25865	1.115	1.197	0.197	1.00		(CV: **0.05**)	**0.92**		
14	Y a	114.59	114.50	0.09	118	1527	10.4	15865	0.684	0.811	0.088	1.03	13.54	Yq11	0.84	-1.4	|| ·
12	Y a	104.32	104.34	-0.02	108	1119	10.4	11619	0.501	0.566	0.065	0.97	14.10	Yq11	0.89	-1.0	| ·
ctrl: Y-fragments (male ref.)					Mean	1323	10.4	13742	0.593	0.688	0.076	1.00		(CV: **0.03**)	**0.86**		
15	1 A	126.30	126.04	0.26	130	2706	10.6	28581	1.232	1.200	0.054	1.30	1.14	1p36.33	1.03	0.6	·
23	1 A	176.64	176.49	0.15	178	1983	11.2	22276	0.961	0.942	0.074	0.75	1.75	1p36.33	1.02	0.2	·
21	1 A	164.61	164.53	0.08	166	2388	11.1	26416	1.139	1.181	0.073	0.95	1.95	1p36.33	0.96	-0.6	·
1p36 (1p-deletion)					Mean	2359	11.0	25758	1.111	1.108	0.067	1.00		(CV: **0.03**)	**1.01**		
37	2 B	264.94	264.89	0.05	267	1801	12.6	22742	0.993	0.948	0.040	1.36	58.30	2p16.1	1.05	1.1	·
63	2 D	481.47	481.31	0.16	486	1084	19.0	20619	1.010	1.121	0.101	0.64	61.00	2p16.1	0.90	-1.1	| ·
"2p16.1 deletion syndrome"					Mean	1443	15.8	21681	1.001	1.034	0.071	1.00		(CV: **0.10**)	**1.00**		
56	3 D	416.52	416.46	0.06	418	1631	17.3	28203	1.381	1.268	0.073	0.96	198.51	3q29	1.09	1.6	·|
49	3 C	354.50	354.58	-0.08	359	1575	14.7	23135	1.071	1.039	0.056	1.04	198.28	3q29	1.03	0.6	·
"3q29 deletion syndrome"					Mean	1603	16.0	25669	1.226	1.154	0.064	1.00		(CV: **0.04**)	**1.06**		
32	4 B	230.75	230.47	0.28	232	1919	11.9	22803	0.996	0.944	0.063	0.88	1.81	4p16.3	1.05	0.8	·|
59	4 D	443.26	443.03	0.23	445	1011	17.5	17692	0.867	0.879	0.046	1.12	1.90	4p16.3	0.99	-0.3	·
4p16.3 Wolf-Hirschhorn region					Mean	1465	14.7	20248	0.931	0.912	0.054	1.00		(CV: **0.05**)	**1.02**		
58	5 D	435.65	435.58	0.07	436	869	17.4	15110	0.740	0.659	0.053	0.88	1.34	5p15.33	1.12	1.5	·|
39	5 C	281.42	281.39	0.03	283	1901	13.7	25953	1.202	1.115	0.071	1.12	1.40	5p15.33	1.08	1.2	·|
Cri du Chat syndrome					Mean	1385	15.5	20532	0.971	0.887	0.062	1.00		(CV: **0.03**)	**1.10**		
19	5 A	151.48	151.42	0.06	154	2577	10.0	25851	1.115	1.127	0.052	1.03	176.62	5q35.3	0.99	-0.2	·
60	5 D	453.05	453.02	0.03	454	1269	17.7	22506	1.102	1.123	0.055	0.97	176.65	5q35.3	0.98	-0.4	·
Sotos syndrome					Mean	1923	13.9	24179	1.109	1.125	0.054	1.00		(CV: **0.01**)	**0.99**		
43	7 C	310.26	310.40	-0.14	310	1091	13.7	14986	0.694	1.116	0.056	0.99	73.08	7q11.23	0.62	-7.6 *	|||| ·
50	7 C	363.50	363.46	0.04	364	809	15.4	12475	0.578	1.029	0.040	1.27	73.11	7q11.23	0.56	-11.3 *	|||| ·
53	7 D	390.07	389.95	0.12	391	486	15.6	7561	0.370	0.748	0.050	0.74	73.15	7q11.23	0.50	-7.5 *	||||| ·
Williams syndrome					Mean	795	14.9	11674	0.547	0.964	0.049	1.00		(CV: **0.10**)	**0.57**		
54	8 D	399.90	399.69	0.21	401	964	16.5	15893	0.778	0.776	0.036	1.06	116.75	8q24.12	1.00	0.1	·
57	8 D	424.71	424.33	0.38	427	1015	17.6	17874	0.875	0.895	0.047	0.94	117.73	8q24.11	0.98	-0.4	·
Langer-Giedion syndrome					Mean	990	17.0	16884	0.827	0.835	0.042	1.00		(CV: **0.02**)	**0.99**		
44	9 C	319.17	319.40	-0.23	319	2083	13.9	28964	1.341	1.292	0.072	1.05	100.95	9q22.33	1.04	0.7	·
55	9 D	408.38	408.03	0.35	409	1529	16.9	25775	1.262	1.119	0.069	0.95	100.95	9q22.33	1.13	2.1	·|
"9q22.3 deletion syndrome"					Mean	1806	15.4	27370	1.302	1.206	0.070	1.00		(CV: **0.06**)	**1.08**		
16	10 A	132.92	132.56	0.36	136	2138	10.8	23108	0.996	0.948	0.043	1.16	8.14	10p	1.05	1.1	·|
48	10 C	349.21	349.23	-0.02	349	1687	14.8	24988	1.157	0.979	0.060	0.84	10.59	10p15.1	1.18	3.0	·||
DiGeorge region 2 (10p)					Mean	1913	12.8	24048	1.077	0.963	0.052	1.00		(CV: **0.08**)	**1.11**		
30	11 B	218.80	218.68	0.12	220	2727	11.7	31933	1.394	1.246	0.061	1.00	31.78	11p13	1.12	2.4	·|
WAGR syndrome					Mean	2727	11.7	31933	1.394	1.246	0.061	1.00		(CV:)	**1.12**		
29	15 B	213.60	213.54	0.06	214	2340	11.7	27464	1.199	1.226	0.047	1.34	21.48	15q11.2	0.98	-0.6	·
34	15 B	245.38	245.06	0.32	247	1679	12.6	21214	0.926	0.962	0.058	0.86	22.65	15q12	0.96	-0.6	·
40	15 C	289.80	289.75	0.05	292	1878	13.5	25276	1.171	1.018	0.061	0.86	22.76	15q12	1.15	2.5	·|
20	15 A	158.12	158.00	0.12	160	1937	11.0	21321	0.919	0.930	0.051	0.94	23.17	15q12	0.99	-0.2	·
Prader-Willi / Angelman					Mean	1959	12.2	23819	1.054	1.034	0.054	1.00		(CV: **0.08**)	**1.01**		
25	15 A	189.88	189.75	0.13	190	2958	11.1	32732	1.411	1.299	0.130	0.78	72.50	15q24.1	1.09	0.9	·|
45	15 C	326.19	326.43	-0.24	325	1237	14.4	17752	0.822	0.771	0.049	1.22	72.80	15q24.1	1.07	1.0	·|
"15q24 deletion syndrome"					Mean	2098	12.7	25242	1.117	1.035	0.090	1.00		(CV: **0.01**)	**1.07**		
22	16 A	170.96	170.91	0.05	172	2210	10.9	24182	1.043	1.018	0.063	1.00	3.87	16p13.3	1.02	0.4	·
Rubinstein-Taybi syndrome					Mean	2210	10.9	24182	1.043	1.018	0.063	1.00		(CV:)	**1.02**		
17	17 A	139.18	138.97	0.21	142	2761	11.1	30742	1.326	1.248	0.053	1.27	2.51	17p13.3	1.06	1.5	·|
33	17 B	236.30	236.22	0.08	238	1739	12.0	20905	0.913	0.900	0.067	0.73	2.52	17p13.3	1.01	0.2	·
Miller-Dieker region					Mean	2250	11.6	25824	1.119	1.074	0.060	1.00		(CV: **0.03**)	**1.04**		
61	17 D	463.26	463.07	0.19	465	1515	18.3	27664	1.355	1.331	0.092	0.83	17.53	17p11.2-##	1.02	0.3	·
38	17 B	271.93	271.90	0.03	274	1698	13.0	22010	0.961	0.940	0.043	1.25	17.83	17p11.2	1.02	0.5	·
42	17 C	302.82	302.90	-0.08	303	1478	13.8	20346	0.942	0.839	0.053	0.91	18.08	17p11.2	1.12	2.0	·|
Smith-Magenis syndrome					Mean	1564	15.0	23340	1.086	1.037	0.063	1.00		(CV: **0.06**)	**1.05**		
36	17 B	258.44	258.33	0.11	260	1075	12.7	13625	0.595	0.621	0.038	0.88	26.56	17q11.2	0.96	-0.7	·
46	17 C	334.29	334.45	-0.16	335	1862	14.8	27541	1.275	1.118	0.053	1.12	26.58	17q11.2	1.14	2.9	·|
NF1 microdeletion syndrome					Mean	1469	13.7	20583	0.935	0.870	0.046	1.00		(CV: **0.12**)	**1.06**		
62	17 D	470.73	470.58	0.15	472	873	18.3	15987	0.783	0.719	0.047	0.70	41.26	17q21.31	1.09	1.4	·|
31	17 B	225.06	224.79	0.27	226	2544	11.9	30210	1.319	1.325	0.050	1.21	41.44	17q21.31	1.00	-0.1	·
47	17 C	341.67	341.78	-0.11	342	1333	14.8	19702	0.912	0.932	0.039	1.09	41.45	17q21.31	0.98	-0.5	·
"17q21.31 microdeletion"					Mean	1583	15.0	21966	1.005	0.992	0.046	1.00		(CV: **0.05**)	**1.01**		

Case: Loss_of_7q11.23_no_5 Folder: P245-vs02-test Date: 09-03-2009

P245-vs02 Microdeletion syndromes (lot 1207) Male 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**
26	22 A	195.36	195.29	0.07	196	1776	10.9	19338	0.834	0.890	0.065	0.83	17.89	22q11.21	0.94	-0.9	| ·
28	22 B	208.17	207.99	0.18	208	2211	11.5	25331	1.106	1.109	0.061	1.11	18.09	22q11.21	1.00	-0.1	·
51	22 D	371.52	371.47	0.05	373	1276	16.7	21291	1.043	0.920	0.053	1.06	19.57	22q11.21	1.13	2.3	· |
22q11.21 (DiGeorge)					Mean	1754	13.0	21987	0.994	0.973	0.060	1.00	(CV: **0.10**)		**1.03**		
35	22 B	252.15	251.96	0.19	253	2188	12.5	27391	1.196	1.321	0.100	1.05	49.49	22q13.33-##	0.91	-1.2	| ·
52	22 D	382.28	382.09	0.19	382	1740	16.6	28940	1.417	1.442	0.120	0.95	49.50	22q13.33-#	0.98	-0.2	·
22q13 (Phelan-McDermid)					Mean	1964	14.6	28166	1.307	1.382	0.110	1.00	(CV: **0.06**)		**0.94**		
41	X C	295.48	295.46	0.02	297	1351	13.1	17699	0.820	0.751	0.048	1.00	32.29	Xp21.2	1.09	1.4	· |
Chromosome X control probe					Mean	1351	13.1	17699	0.820	0.751	0.048	1.00	(CV:)		**1.09**		
27	X B	199.75	199.64	0.11	202	896	10.0	8926	0.390	0.457	0.047	0.99	153.02	Xq28	0.85	-1.4	| ·
18	X A	147.04	146.82	0.22	148	1108	8.9	9887	0.426	0.538	0.065	0.84	152.95	Xq28	0.79	-1.7	|| ·
24	X A	183.22	183.16	0.06	184	1226	11.1	13550	0.584	0.678	0.060	1.16	152.94	Xq28-#	0.86	-1.6	| ·
Xq28 (RETT / MECP2)					Mean	1077	10.0	10788	0.467	0.558	0.057	1.00	(CV: **0.04**)		**0.84**		
Mean values				0.10		1676	13.6	21969	**0.999**	1.000	0.060	**4**			0.99	Total of all except	
Standard deviations				0.14		(Coef. of variance:		0.277)	0.272	0.224					0.14	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	6.79
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.23
Mean height of first probes AB	> 450 (800)	2024
Mean height of last probes CD	> 280 (500)	1342
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.51
Mean group CV of weighted ratio	<0.20 (0.15)	0.06
3 uinidentified peak areas / 54 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.

Male Reference

Abn. peaks: 7q11.23 7q11.23 7q11.23

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 217 (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)