Case: Gain_of_Xq28 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
4	64 -	60.85	60.65	0.20	64	246	12.2	3000	0.772	1.190	0.371	0.75		64 nt	0.65	-1.1	IIII·
5	70 -	66.59	66.46	0.13	70	189	17.2	3246	0.836	0.913	0.264	0.80		70 nt	0.92	-0.3	I·
6	76 -	72.42	72.37	0.05	76	256	21.6	5521	1.421	1.138	0.170	1.55		76 nt	1.25	1.7	·II
7	82 -	78.59	78.56	0.03	82	229	16.5	3770	0.971	0.787	0.204	0.90		82 nt	1.23	0.9	·II
Ctrl: Q-fragments					Mean	230	16.8	3884	1.000	1.007	0.252	1.00		(CV: **0.26**)	**1.07**		
8	6 a	85.00	84.95	0.05	88	1334	13.1 *****	17425	0.844	0.981	0.152	1.02		6p21.3 CpG isl.	0.86	-0.9	I·
10	2 a	90.24	90.17	0.07	92	1762	11.3	19980	0.968	0.967	0.135	1.13		2q14 synt.	1.00	0.0	·
11	1 a	96.61	96.50	0.11	96	2674	12.4 *****	33231	1.610	1.641	0.304	0.85	MV 36	1p36 CpG isl.	0.98	-0.1	·
Ctrl: D-fragments					Mean	1923	12.3	23545	1.141	1.197	0.197	1.00		(CV: **0.08**)	**0.95**		
16	Y a	114.56	114.50	0.06	118	1588	11.0	17425	0.844	0.811	0.088	1.03	13.54	Yq11	1.04	0.4	·
13	Y a	104.36	104.34	0.02	108	1065	11.2	11923	0.578	0.566	0.065	0.97	14.10	Yq11	1.02	0.2	·
ctrl: Y-fragments (male ref.)					Mean	1327	11.1	14674	0.711	0.688	0.076	1.00		(CV: **0.01**)	**1.03**		
17	1 A	126.17	126.04	0.13	130	1929	11.6	22450	1.088	1.200	0.054	1.30	1.14	1p36.33	0.91	-2.1	I·
25	1 A	176.56	176.49	0.07	178	1567	12.5	19597	0.950	0.942	0.074	0.75	1.75	1p36.33	1.01	0.1	·
23	1 A	164.60	164.53	0.07	166	1922	11.8	22704	1.100	1.181	0.073	0.95	1.95	1p36.33	0.93	-1.1	I·
1p36 (1p-deletion)					Mean	1806	12.0	21584	1.046	1.108	0.067	1.00		(CV: **0.05**)	**0.94**		
40	2 B	264.94	264.89	0.05	267	1421	14.3	20269	1.058	0.948	0.040	1.36	58.30	2p16.1	1.12	2.7	·I
66	2 D	481.49	481.31	0.18	486	1228	20.2	24820	1.228	1.121	0.101	0.64	61.00	2p16.1	1.10	1.1	·I
"2p16.1 deletion syndrome"					Mean	1325	17.2	22545	1.143	1.034	0.071	1.00		(CV: **0.01**)	**1.11**		
59	3 D	416.48	416.46	0.02	418	1463	17.9	26233	1.298	1.268	0.073	0.96	198.51	3q29	1.02	0.4	·
52	3 C	354.59	354.58	0.01	359	1511	16.0	24179	1.174	1.039	0.056	1.04	198.28	3q29	1.13	2.4	·I
"3q29 deletion syndrome"					Mean	1487	17.0	25206	1.236	1.154	0.064	1.00		(CV: **0.07**)	**1.08**		
34	4 B	230.51	230.47	0.04	232	1300	12.6	16405	0.856	0.944	0.063	0.88	1.81	4p16.3	0.91	-1.4	I·
62	4 D	443.19	443.03	0.16	445	1019	18.5	18887	0.935	0.879	0.046	1.12	1.90	4p16.3	1.06	1.2	·I
4p16.3 Wolf-Hirschhorn region					Mean	1160	15.6	17646	0.895	0.912	0.054	1.00		(CV: **0.11**)	**0.99**		
61	5 D	435.82	435.58	0.24	436	667	18.4	12281	0.608	0.659	0.053	0.88	1.34	5p15.33	0.92	-1.0	I·
42	5 C	281.43	281.39	0.04	283	1468	15.0	22048	1.071	1.115	0.071	1.12	1.40	5p15.33	0.96	-0.6	·
Cri du Chat syndrome					Mean	1068	16.7	17165	0.839	0.887	0.062	1.00		(CV: **0.03**)	**0.94**		
21	5 A	151.46	151.42	0.04	154	2099	11.0	23028	1.116	1.127	0.052	1.03	176.62	5q35.3	0.99	-0.2	·
63	5 D	453.10	453.02	0.08	454	1312	19.1	25016	1.238	1.123	0.055	0.97	176.65	5q35.3	1.10	2.1	·I
Sotos syndrome					Mean	1706	15.0	24022	1.177	1.125	0.054	1.00		(CV: **0.08**)	**1.04**		
46	7 C	310.43	310.40	0.03	310	1619	15.2	24553	1.192	1.116	0.056	0.99	73.08	7q11.23	1.07	1.4	·I
53	7 C	363.44	363.46	-0.02	364	1194	16.1	19249	0.935	1.029	0.040	1.27	73.11	7q11.23	0.91	-2.4	I·
56	7 D	389.85	389.95	-0.10	391	863	17.4	15002	0.742	0.748	0.050	0.74	73.15	7q11.23	0.99	-0.1	·
Williams syndrome					Mean	1225	16.2	19601	0.957	0.964	0.049	1.00		(CV: **0.09**)	**0.98**		
57	8 D	399.62	399.69	-0.07	401	928	17.8	16560	0.820	0.776	0.036	1.06	116.75	8q24.12	1.06	1.2	·I
60	8 D	424.51	424.33	0.18	427	1118	18.5	20685	1.024	0.895	0.047	0.94	117.73	8q24.11	1.14	2.7	·I
Langer-Giedion syndrome					Mean	1023	18.2	18623	0.922	0.835	0.042	1.00		(CV: **0.06**)	**1.10**		
47	9 C	319.47	319.40	0.07	319	1910	15.2	29103	1.413	1.292	0.072	1.05	100.95	9q22.33	1.09	1.7	·I
58	9 D	408.12	408.03	0.09	409	1366	18.0	24635	1.219	1.119	0.069	0.95	100.95	9q22.33	1.09	1.5	·I
"9q22.3 deletion syndrome"					Mean	1638	16.6	26869	1.316	1.206	0.070	1.00		(CV: **0.00**)	**1.09**		
18	10 A	132.73	132.56	0.17	136	1532	11.9	18178	0.881	0.948	0.043	1.16	8.14	10p	0.93	-1.6	I·
51	10 C	349.25	349.23	0.02	349	1359	15.7	21320	1.035	0.979	0.060	0.84	10.59	10p15.1	1.06	0.9	·I
DiGeorge region 2 (10p)					Mean	1446	13.8	19749	0.958	0.963	0.052	1.00		(CV: **0.09**)	**0.98**		
32	11 B	218.73	218.68	0.05	220	1796	12.4	22318	1.164	1.246	0.061	1.00	31.78	11p13	0.93	-1.3	I·
WAGR syndrome					Mean	1796	12.4	22318	1.164	1.246	0.061	1.00		(CV:)	**0.93**		
31	15 B	213.56	213.54	0.02	214	2027	12.5	25344	1.322	1.226	0.047	1.34	21.48	15q11.2	1.08	2.1	·I
36	15 B	245.09	245.06	0.03	247	1327	13.2	17518	0.914	0.962	0.058	0.86	22.65	15q12	0.95	-0.8	·
43	15 C	289.73	289.75	-0.02	292	1512	14.8	22337	1.085	1.018	0.061	0.86	22.76	15q12	1.07	1.1	·I
22	15 A	158.07	158.00	0.07	160	1761	11.8	20757	1.006	0.930	0.051	0.94	23.17	15q12	1.08	1.5	·I
Prader-Willi / Angelman					Mean	1657	13.1	21489	1.082	1.034	0.054	1.00		(CV: **0.06**)	**1.05**		
27	15 A	189.77	189.75	0.02	190	1894	12.0	22778	1.104	1.299	0.130	0.78	72.50	15q24.1	0.85	-1.5	II·
48	15 C	326.53	326.43	0.10	325	1270	15.8	20128	0.977	0.771	0.049	1.22	72.80	15q24.1	1.27	4.2 *****	·III
"15q24 deletion syndrome"					Mean	1582	13.9	21453	1.041	1.035	0.090	1.00		(CV: **0.26**)	**1.10**		
24	16 A	171.00	170.91	0.09	172	1635	12.0	19653	0.952	1.018	0.063	1.00	3.87	16p13.3	0.94	-1.0	I·
Rubinstein-Taybi syndrome					Mean	1635	12.0	19653	0.952	1.018	0.063	1.00		(CV:)	**0.94**		
19	17 A	139.09	138.97	0.12	142	2056	12.1	24778	1.201	1.248	0.053	1.27	2.51	17p13.3	0.96	-0.9	·
35	17 B	236.29	236.22	0.07	238	1269	13.5	17081	0.891	0.900	0.067	0.73	2.52	17p13.3	0.99	-0.1	·
Miller-Dieker region					Mean	1663	12.8	20930	1.046	1.074	0.060	1.00		(CV: **0.02**)	**0.97**		
64	17 D	463.30	463.07	0.23	465	1431	19.4	27750	1.373	1.331	0.092	0.83	17.53	17p11.2-##	1.03	0.5	·
41	17 B	271.96	271.90	0.06	274	1256	14.1	17732	0.925	0.940	0.043	1.25	17.83	17p11.2	0.98	-0.3	·
45	17 C	302.98	302.90	0.08	303	948	14.8	14051	0.682	0.839	0.053	0.91	18.08	17p11.2	0.81	-3.0	II·
Smith-Magenis syndrome					Mean	1212	16.1	19844	0.994	1.037	0.063	1.00		(CV: **0.12**)	**0.95**		
39	17 B	258.40	258.33	0.07	260	897	13.8	12338	0.644	0.621	0.038	0.88	26.56	17q11.2	1.04	0.6	·
49	17 C	334.53	334.45	0.08	335	1574	15.8	24841	1.206	1.118	0.053	1.12	26.58	17q11.2	1.08	1.6	·I
NF1 microdeletion syndrome					Mean	1236	14.8	18590	0.925	0.870	0.046	1.00		(CV: **0.03**)	**1.06**		
65	17 D	470.78	470.58	0.20	472	741	19.2	14198	0.703	0.719	0.047	0.70	41.26	17q21.31	0.98	-0.3	·
33	17 B	224.86	224.79	0.07	226	1882	13.3	24938	1.301	1.325	0.050	1.21	41.44	17q21.31	0.98	-0.5	·
50	17 C	341.86	341.78	0.08	342	1229	16.1	19753	0.959	0.932	0.039	1.09	41.45	17q21.31	1.03	0.7	·
"17q21.31 microdeletion"					Mean	1284	16.2	19630	0.988	0.992	0.046	1.00		(CV: **0.03**)	**1.00**		

Case: Gain_of_Xq28 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 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
28	22 A	195.29	195.29	0.00	196	1319	11.4	14979	0.726	0.890	0.065	0.83	17.89	22q11.21	0.82	-2.5	‖ ·
30	22 B	208.08	207.99	0.09	208	1573	12.6	19878	1.037	1.109	0.061	1.11	18.09	22q11.21	0.93	-1.2	| ·
54	22 D	371.38	371.47	-0.09	373	1058	17.7	18722	0.927	0.920	0.053	1.06	19.57	22q11.21	1.01	0.1	·
22q11.21 (DiGeorge)					Mean	1317	13.9	17860	0.897	0.973	0.060	1.00	(CV: **0.10**)		**0.93**		
38	22 B	251.98	251.96	0.02	253	1931	14.1	27144	1.416	1.321	0.100	1.05	49.49	22q13.33-##	1.07	0.9	·|
55	22 D	382.18	382.09	0.09	382	1611	17.7	28571	1.414	1.442	0.120	0.95	49.50	22q13.33-#	0.98	-0.2	·
22q13 (Phelan-McDermid)					Mean	1771	15.9	27858	1.415	1.382	0.110	1.00	(CV: **0.06**)		**1.03**		
44	X C	295.48	295.46	0.02	297	1092	14.3	15591	0.757	0.751	0.048	1.00	32.29	Xp21.2	1.01	0.1	·
Chromosome X control probe					Mean	1092	14.3	15591	0.757	0.751	0.048	1.00	(CV:)		**1.01**		
29	X B	199.76	199.64	0.12	202	1528	12.0	18372	0.959	0.457	0.047	0.99	153.02	Xq28	2.10	10.6*	·|||||||||
20	X A	146.91	146.82	0.09	148	1944	10.7	20796	1.008	0.538	0.065	0.84	152.95	Xq28	1.87	7.2*	·||||||||
26	X A	183.17	183.16	0.01	184	2227	12.6	27983	1.356	0.678	0.060	1.16	152.94	Xq28-#	2.00	11.3*	·|||||||||
Xq28 (RETT / MECP2)					Mean	1900	11.8	22384	1.108	0.558	0.057	1.00	(CV: **0.05**)		**2.00**		
Mean values				0.07		1461	14.8	20970	**1.041**	1.000	0.060	**4**			1.07	Total of all except	
Standard deviations				0.07		(Coef. of variance:		0.205)	0.212	0.224					0.25	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	5.31
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.23
Mean height of first probes AB	> 450 (800)	1671
Mean height of last probes CD	> 280 (500)	1260
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.33
Mean group CV of weighted ratio	<0.20 (0.15)	0.07
5 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: Xq28 Xq28-# Xq28

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 215 (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)