

**Supplementary Table. Abnormal CMA Results in 109 Neonates with Birth Defects**

	Indication	GTG-banding Analysis	CMA	CMA Results
1	DF	DP, del(1)(p36.1p36.2)	V6	arr cgh 1p36.32p36.13(RP1-58B11->RP11-387P17)x1
2	CHD, VSD	RS visible	V6 OLIGO	arr cgh 1p36.32p36.13(RP11-22L13->RP11-387P17)x1.ish del(1)(p36.23p36.23)(RP11-92O17-)
3	DF, MCA	RS visible	V6 OLIGO	arr cgh 1p34.3p33(RP11-248I9->RP11-492I2)x3.ish dup(1)(p34.2p33)(RP11-248I9++,RP11-72N2++,RP11-492I2++)dn
4	Neuromuscular	ND	V6 OLIGO	arr cgh 1p32.1p31.3(RP11-63G10,RP11-524L19)x1.ish del(1)(p32.1p32.1)(RP11-63G10-)
5	DF, MCA,	RS visible	V6 OLIGO	arr cgh 1q21.1q24.3(RP11-337C18->RP11-44A10)x3.nuc ish 1q21.3q23.3(RP11-81P11x3,RP11-38C18x3)
6	MCA	DP, add(1)(q?42.1)	V5	arr cgh 1q42(RP5-1090A23)x1,1q44(RP11-370K11)x3dn.ish del(1)(q42.1q42.1)(RP5-1090A23-).nuc ish 1q44(RP11-370K11x3)dn
7	DF, MCA, IUGR	RS visible	V6 OLIGO	arr cgh 1q42.12q43(RP11-496N12->RP11-136B18)x1.ish del(1)(q42.3q42.3)(RP11-478H16-)
8	DF, CHD	CO, NL	V5	arr cgh 2p25.3(RP11-379N10,RP11-625N16)x3.nuc ish 2p25.3(RP11-379N10x3)
9	DF, Cleft Palate	ND	V5	arr cgh 2q22.3(RP11-89L3,RP11-90K5)x1.ish del(2)(q22.3q22.3)(RP11-89L3-,EP11-90K5-)
10	FTT	CO, NL	V6	arr cgh 2q32.1(RP11-234L7)x3.nuc ish 2q32.1(RP11-234L7x3)dn
11	MCA	CO, der(2)t(2;8)(q37.1;q24.1)	V5	arr cgh 2q37.1(RP11-690I21->RP11-491D2)x1,8q24.3(RP11-149P24->RP11-299M14)x3
12	DF	CO, del(3)(p26.3)	V6 OLIGO	arr cgh 3p26.3(RP11-385A18->RP11-33J20)x1.ish del(3)(p26.3p26.3)(RP11-392M7-)mat
13	MCA, CHD	CO, del(3)(p25)	V5	arr cgh 3p26.3p25.3(RP11-385A18->RP11-45B16)x1
14	DF	CO, add(3)(p26)	V5	arr cgh 3p26.3(RP11-385A18->RP11-392M7)x1,3p26.2p25.3(RP11-33J20->RP11-45B16)x3.ish del(3)(p26.3p26.3)(D3S4559-),dup(3)(p26.1p26.1)(RP11-194C9x3)
15	DF, Hypotonia	DP, NL	V5	arr cgh 3p26.3p25.1(RP11-385A18->RP11-271E2)x3,18p11.32p11.23(RP11-705O1->RP11-931H21)x1.ish t(3;18)(3p26.3;18p11.3)(D3S4559+,D18S552-)
16	MCA	RS visible, der(4)t(4;11)	V6 OLIGO	arr cgh 4p16.3(RP11-613L20->RP11-357G3)x1,11p15.5(RP13-46H24->RP11-38L8)x3.ish der(4)t(4;11)(p16.3;p15.5)(RP11-613L20-,RP13-870H17+)
17	CA	ND	V6 OLIGO	arr cgh 4p16.3p16.1(RP11-322F4->RP11-338K13)x3,8p23.3.p23.1(RP11-91J19->RP11-16G12)x1
18	DF, MCA	CO, del(4)(p16.2)	V6 OLIGO	arr cgh 4p16.3p16.1(RP11-613L20->RP11-136I13)x1,9q34.3(RP11-611D20->RP11-974F22)x3.ish der(4)t(4;9)(p16.3;q34.3)(RP11-613L20-,RP13-467E5+)
19	MCA	ND	V5	arr cgh 1p36.2(RP11-584P2)x3,4p16.3p15.3(GS-36P21->RP11-

20	DF, MCA, FTT	ND, dn	V6 OLIGO	34C20)x1.nuc ish 1p36.2(RP11-584P2x3),del(4)(p16.3p16.3)(RP11-203L2-) arr cgh 4q21.1q21.22(RP11-630D6->RP11-29L8)x1.ish del(4)(q21.22q21.22)(RP11-29L8-)dn arr cgh 17p13.3(RP11-235E17->RP11-48B14)x1
21	DF	RS visible, dup 4q	V6	arr cgh 4q25q26(RP11-380D23->RP11-779N9)x3.nuc ish 4q25(RP11-777N19x3)
22	DF, TOF	RS visible, dup 4q	V6	arr cgh 4q27q35.1(RP11-679C8->RP11-203L17)x3.nuc ish 4q35.1(RP11-203L17x3)
23	DF, MCA, SD	DP, NL. RS visible	V5	arr cgh 4q35.1q35.2(RP11-203L17->RP11-565A3)x1.ish del(4)(q35.1q35.2)(RP11-173M11-,RP11-565A3-)
24	DF	CO, NL	V6 OLIGO	arr cgh 4q35.2(RP11-565A3)x3,15q15.3(RP11-315A19->RP11-263I19)x1.nuc ish 4q35.2(RP11-565A3x3)dn.ish del (15)(q15.3q15.3)(RP11-263I19-)pat
25	CHD, VSD, Laryngomalacia	ND	V6 OLIGO	arr cgh 5p15.33(RP11-487N22->RP11-121L11)x1.ish del(5)(p15.33p15.33)(RP11-43F13-)
26	DF	RS visible	V6	arr cgh 5p15.33(RP11-227M19->RP11-121L11)x1
27	Craniofacial Abnl	DP, del(5)(p15.2)	V6	arr cgh 5p15.3p15.2(RP11-487N22->RP11-91M12)x1
28	DF	CO, del(5)(p13p15.33)	V5	arr cgh 5p15.3p14.2(RP11-227M19->RP11-91L13)x1
29	DF	RS visible	V6	arr cgh 5p15.33p14.2(RP11-487N22->RP11-91L13)x1
30	Ambiguous Genitalia, Premature	CO, der(5)(qter->q34::p14->qter)	V6 OLIGO	arr cgh 5p15.33p14.2(RP11-487N22->RP11-91L13)x1,5q34q35.3(RP11-109J4->RP11-80J19)x3.ish der(5)t(5;5)(p14.2;q34)(RP11-21I4+) arr cgh 16p13.2(RP11-349E19)x1.ish del(16)(p13.2p13.2)(RP11-349E19-)
31	MCA	RS visible, dn	V6 OLIGO	arr cgh 5q12.1q12.3(RP11-1A7->RP11-307L14)x1dn
32	Craniofacial Abnl Syndactyly	ND	V6	arr cgh 5q23.3(RP11-81L23)x3.nuc ish 5q23.3(RP11-81L23x3)dn
33	Hydrocephalus	CO, NL	V5	arr cgh 6p25.3(RP1-20B11->RP11-82M9)x1.ish del(6)(p25.3p25.3)(RP1-20B11-)
34	CHD, interrupted Aortic Arch	RS visible	V6 OLIGO	arr cgh 6p24.3p22.3(RP11-380L24->RP1-239K6)x1
35	No	ND	V6 OLIGO	arr cgh Xp22.32p22.31(RP11-707P20->RP11-483M24)x2.nuc ish Xp22.32(RP11-594H22x2)mat. arr cgh 6p25.3p23(RP11-812K10->RP11-127P7)x3,6q26q27(RP1-45F6->RP11-115L8)x1.ish der(6)t(6;6)(p23;q26)(RP11-299J5+,RP3-366N23-)
36	Trisomy 18	ND	V5	arr cgh 6q27(RP3-495K2)x1,10q24q26.3(RP11-108L7->RP11-140A10)x3.nuc ish der(6)t(6;10)(q27;q26.3)

37	FTT	ND	V6 OLIGO	(RP3-495K2-,RP11-462G8+) arr cgh 7q11.23(RP11-622P13->RP4-665P5)x3.nuc ish 7q11.23(RP11-622P13x3)
38	DF	ND	V5	arr cgh 7q11.2(RP5-1127A24->RP4-665P5)x1.ish del(7)(q11.2q11.2)(RP11-622P13-)
39	CHD, Truncus Arteriosis	ND	V6	arr cgh 7q31.3(RP11-563L14)x3.nuc ish 7q31.3(RP11-563L14X3)dn
40	DF	CO, del(7)(q32q34)	V6 OLIGO	arr cgh 7q32.2q34(RP11-2E11->RP11-137O4)x1.ish del(7)(q33q33)(RP11-98D12-)
41	MCA	CO, del(7)(q33)	V6	arr cgh 7q34q36.3(RP11-137O4->RP11-789H4)x1.ish del(7)(q36.3)(GS-3K23-,RP11-452C13-)
42	CHD	CO, del(8)(p23.1p23.1)	V6 OLIGO	arr cgh 8p23.1(RP11-90J21->RP11-589N15)x1
43	CHD	ND	V6 OLIGO	arr cgh 8p23.1(RP11-90J21->RP11-589N15)x1.ish del(8)(p23.1p23.1)(RP11-252K12-)dn
44	DF, MCA	RS visible, der(8)t(8;8)	V6 OLIGO	arr cgh 8p23.3p23.1(RP11-91J19->RP11-79E11)x1,8q22.1q24.3(RP11-80P10->RP11-639O3)x3. ish der(8)t(8;8)(p23.1;q22.1)(RP11-555E9-,RP11-149P24+)
45	MCA	ND	V6 OLIGO	arr cgh 8q11.21q12.1(RP11-1134I14->RP11-107M13)x1.ish del(8)(q11.21q11.21)(RP11-113H14-)dn
46	DF, Renal Anomaly	ND	V6 OLIGO	arr cgh 8q13.3(RP11-326E22->RP11-1102P16)x1.ish del(8)(q13.3q13.3)(RP11-326E22-)
47	R/O Trisomy 13, 18, 21	CO, del(8)(q24.11q24.22)	V5	arr cgh 8q23.3q24.2(RP11-80P10->P30)x1.ish del(8)(q24.1q24.1)(P30-)
48	DF	CO, inv(9)(p11q13), der(15)(9pter->p11::9q13->p11::15q10->15qter)	V6 OLIGO	arr cgh 9p24.3p13.1 (RP11-165F24->CTD-2349C9)x3
49	Craniofacial Abnl	ND	V6 OLIGO	arr cgh 9p23(RP11-3L8,RP11-115I23)x3.nuc ish 9p23(RP11-3L8x3)
50	DF	CO, der(9)del(9)(p22.2)dup(9)(p22.2p13.3)	V6 OLIGO	arr cgh 9p24.3p22.2(RP11-165F24->RP11-163F8)x1.ish del(9)(p24.3p24.3)(RP11-165F24-) arr cgh 9p22.1p21.1(RP11-399M15->RP11-30O4)x3.nuc ish 9p22.1(RP11-399M15x3)
51	MCA	CO, NL, RS visible	V6 OLIGO	arr cgh 10q11.22q11.23(RP11-70E21,RP11-563N6)x1.ish del(10)(q11.22q11.23)(RP11-70E21-,RP11-563N6-)pat
52	DF, MCA, Ambiguous Genitalia	RS visible, del(10),t(12;15)	V5	arr cgh 10q26.1q26.3(RP11-338O1->RP11-426G8)x1
53	MCA	RS visible, der(10)t(10;18)	V6 OLIGO	arr cgh 10q26.13q26.3(RP11-338O1->RP11-108K14)x1,18q23(RP11-162A12->RP11-89N1)x3.ish der(10)t(10;18)(q26.13;q23)(RP11-462G8-,RP11-451L19+)
54	DF, MCA	ND	V5	arr cgh 11p15.5p11.2(RP11-706A13->RP13-46H24)x3.nuc ish

55	CHD, Coarctation of Aorta	RS visible	V6	11p15.5(RP5-998N23x3)
56	DF, MCA	ND	OLIGO	arr cgh 11q14.2q25(RP11-720D4->RP11-419F8)x3.
57	GI Abnl	RS visible, der(12)	V6	arr cgh 11q23.2q25(RP11-64D24->RP11-469N6)x3,13q12.11q12.3(RP11-301J16->RP11-90M5)x3.
58	DF	RS visible, der(15)t(12;15)	OLIGO	ish der(13)t(11;13)(q23.2;q12.3)(RP11-861E23+,RP11-90M5+)
59	Trisomy 13	CO, +13	V6	arr cgh 12q21.2(RP11-734E19->RP11-8P16)x1.ish
60	DF, Polydactyly, Cataracts	DP, +13	OLIGO	der(12)del(12)(q21.2q21.2)inv(12)(q13.2q23.3)(RP11-8P16-)
61	DF	RS confirm	V6	arr cgh 12q24.31qter(RP11-197N18->RP11-386I8)x3.nuc ish
62	Trigonocephaly	CO, der(13)t(13;13)(p11.1;q14.1)	V6	der(15)t(12;15)(q24.31;p11.2)(RP11-669N7+)
63	Dandy Walker Syndrome	DP, add(13)(q12)	V6	arr cgh 13(47 BAC)x3
64	DF, Macrocephaly, Club Feet	CO,	V6	arr cgh 13(47 BAC)x3
65	Cyanotic Episode	der(14;14)(q10;q10)[3]/46,XX[38]	OLIGO	arr cgh 13q14.2q34(RP11-305D15->RP11-569D9)x3
66	Trisomy 13, 18, 21	CO, del(15)(q11.2q13)	V6	arr cgh 13q14.3q33.2(RP11-768P11->RP11-166E2)x1
67	DF, Cryptorchidism, R/O Trisomy 13/18	ND	V6	arr cgh 14(RP11-98N22->RP11-47P23)x3.nuc ish 14q32.2(RP11-187K17x3)[84]/14q32.2(RP11-187K17x2)[116]
68	DF, MCA	ND	OLIGO	arr cgh 14q11.2q21.1(RP11-76P17->RP11-588D7)x1.ish
69	RUBINSTEIN TAYBI	ND	V6	der(18)t(14;18)(q11.2;p11.3)(RP11-260G13-)
70	Trisomy 18	CO, +18	OLIGO	arr cgh 15q11.2q12(RP11-289D12)->RP11-345N11)x1.ish
71	DF, Possible Trisomy 18	CO, +18	V6	del(15)(q11.2q11.2)(5e9-)
72	Goldenhar Syndrome	CO, +18	V6	arr cgh 15q11.2q12(RP11-289D12-->RP11-345N11)x1
73	Heterotaxy	DP, NL	V6	arr cgh 15q11.2q12(RP11-484P15->RP11-345N11)x1.ish
74	Trisomy 21	DP, +21	OLIGO	del(15)(q11.2q12)(5e9-)
75	DF, Trisomy 21, Noonan's	CO, +21	V6	arr cgh 16p13.3(RP11-75P12,RP11-95J11)x1.ish
76	Trisomy 21	CO, +21	OLIGO	del(16)(p13.3p13.3)(RT100-,RT166-)
			V6	arr cgh 18(44 BAC)x3
			V6	arr cgh 18(44 BAC)x3
			OLIGO	arr cgh 18(44 BAC)x3
			V6	arr cgh 19p13.3(RP11-52O9)x3,nuc ish 19p13.3(RP11-52O9x2)dn
			V5	arr cgh 21(16 BAC)x3
			V5	arr cgh 21(16 BAC)x3
			V6	arr cgh 21(19 BAC)x3

77	DF, Possible Trisomy 21	CO, +21	V6	arr cgh 21(19 BAC)x3
78	DF, Cleft Palate, Trisomy 21	CO, +21	V6	arr cgh 21(19 BAC)x3.ish 21q22.3(DSCRx3)
79	Trisomy 21	CO, +21	V6	arr cgh 21(19 BAC)x3
80	DF	CO, +21	OLIGO V6	arr cgh 21(19 BAC)x3
81	DF, MCA	CO, +21	OLIGO V6	arr cgh 21(19 BAC)x3
82	DF, MCA, CHD		OLIGO V6	arr cgh 22(20 BAC)x3.ish 22q11.2(RP11-296H20x3),22q13.1(RP11-69B8x3)
83	MCA	DP(prenatal), NL	V5	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(D22S75-)
84	MCA	CO, NL	V5	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(D22S75-)
85	CHD	CO, NL	V5	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(RP11-186O8-)
86	DF	ND	V5	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(RP11-186O8-)
87	CHD	CO, del(22)(q11.21q11.23)	V5	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(RP11-316L10x1)
88	CHD	DP, del 22q	V6	arr cgh 3p26.3(RP11-306H5)x3.nuc ish 3p26.3(RP11-306H5x3) arr cgh 13q12.12(RP11-760M1)x3.nuc ish 13q12.12(RP11-760M1x3) arr cgh 22q11.21(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.21q11.21)(N25-)
89	CHD	ND	V6	arr cgh 22q11.2(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.2q11.2)(F5-,D22S75-)
90	CHD, Truncus Arteriosus	CD, del(22)(q11.21q11.23)	V6	arr cgh 22q11.21(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.21q11.21)(N25-)
91	CA	ND	V6	arr cgh 22q11.21(RP11-186O8->RP11-165F18)x1.ish del(22)(q11.21q11.21)(RP11-186O8-)
92	CHD	CO, NL	V6	arr cgh 22q11.21(RP11-243A4->RP11-165F18)x1.ish del(22)(q11.21q11.21)(N25-)
93	CHD	ND	OLIGO V6	arr cgh 22q11.21(RP11-243A4->RP11-165F18)x1.ish del(22)(q11.21q11.21)(RP11-316L10-)
94	CHD, Coarctation of the Aorta	ND	OLIGO V6	arr cgh 22q11.21(RP11-243A4->RP11-165F18)x1.ish del(22)(q11.21q11.21)(RP11-316L10-)
95	DF,CHD	ND	OLIGO V6	arr cgh 22q11.21(RP11-316L10,RP11-165F18)x1.ish

96	DF, Turner Syndrome	CO, 45,X	OLIGO V6	del(22)(q11.21q11.21)(RP11-316L10-) arr cgh X(111 BAC)x1
97	DF	CO, NL	OLIGO V6	arr cgh Yp11.2(RP11-115H13->RP11-418M8)x0, 4q35.1(RP11-173M11)x3.ish del(Y)(p11.2p11.2)(RP11-418M8-).nuc ish 4q35.1(RP11-173M11x3)
98	DF, MCA	ND	V6	mos arr cgh 12p13.33p11.21(RP11-598F7->RP11-8P13)x3.ish i(12)(p13.3p13.3)(RP11-407G6++)[5]/12p13.3(RP11-407G6x2)[5]nuc ish 12cen(cep12x3),12p13.3(RP11-407G6x4)[35]/12cen(cep12x2),12p13.3(RP11-407G6x2)[15]
99	DF, MCA	DP, 45,X	V6 OLIGO	mos arr cgh 12p13.33p11.21(RP11-598F7->RP11-8P13)x3nuc ish 12p13.32(RP11-429A20x2),12p13.33(RP11-359B12x2)[173]/12p13.32(RP11-429A20x6),12p13.33(RP11-359B12x6)[27]
100	RDS	ND	V6 OLIGO	mos arr cgh 12p13.33p11.21(RP11-598F7->RP11-8P13)x3.ish i(12)(p13.2q11)(RP11-656E20++,D12Z3+)[3/50].nuc ish (RP11-656E20x4,D12Z3x3)[37/200]
101	DF	CO, NL	V5	mos trisomy 9.nuc ish 9cen(CEP 9x2)[139]/9cen(CEP 9x3)[57]
102	DF	ND	V6	mos trisomy 9
103	MCA	CO, 47,XY,+inv dup(15)(q13)[39]/46,XY[1]	V6 OLIGO	arr cgh 15q11.2q13.3(RP11-289D12->RP11-348B17)x3.nuc ish 15q11.2(RP11-125E1x4)[91]/15q11.2(RP11-125E1x2)[9]
104	DF, FTT	CO, Mos +18	V6 OLIGO	mos arr cgh 18(44 BAC)x3.ish 18(RP11-78H1x3,D18Z1x3)[26]/i(18)(p11.32q11.1)(RP11-78H1++D18Z1+)[4]
105	IUGR	CO, 47,XX,+18[16]/46,XX[4]	V6 OLIGO	mos arr cgh 18p11.32q23(RP11-705O1->RP11-89N1)x3.nuc ish 18p11.32(RP11-607C2x3),18p11.22(RP11-752I5x3)[89/100]
106	MCA	RS visible, mos inv dup 22	V6 OLIGO	mos arr cgh 22q11.1q11.21(RP11-48E19->RP11-47L18)x3.nuc ish 22q11.21(RP11-91O6x3,RP11-165F18x3)[195]/22q11.21(RP11-91O6x4,RP11-165F18x4)[55]/22q11.21(RP11-91O6x2,RP11-165F18x2)[50]
107	Hypoplastic Lt. Heart	CO, 46,X,i(Y)(q10)[17]/45,X[3]	V5	mos Turner arr cgh X(66 BAC)x1,Y(9 BAC)x1
108	Ambiguous Genitalia	CO, 45,X[26]/46,X,indic(Y)(q11.2)[24]	V6 OLIGO	mos Turner arr cgh Y(RP11-112L19->RP11-223K9)x0
109	Ambiguous Genitalia	RS visible	V6 OLIGO	mos Turner arr cgh Yp11.31q11.22(RP11-400O10->RP11-223K9)x1.nuc ish Xcen(DXZ1x1),Ycen(DYZ3x0)[88]/Xcen(DXZ1x1),Ycen(DYZ3x1)[12]

CHD, congenital heart disease; DF, dysmorphic features; MCA, multiple congenital anomalies;

RDS, respiratory distress syndrome; FTT, failure to thrive; GI, gastrointestinal; IUGR, intrauterine growth retardation; CDH, congenital diaphragmatic hernia

NL, normal; Abnl, abnormal; DP, done previously; CO, concurrent; ND, not done; RS, retrospective; Mos, mosaicism; mar, marker chromosome; dn, *de novo*; inv, inversion; dup, duplication; del, deletion; der, derivative; add, additional material