

Supplemental Table 1 2008 CYCGH-01 nomenclature

2008 CYCGH-01 (n=27)

ARR 13Q11Q33.2(17943628->114126487)X3

ARR CGH 1-22(364K OLIGO)X2,X(>19K OLIGO)X1,Y(2K OLIGO)X1

ARR CGH 13(138 BAC)x3

arr cgh 13(18650699->114112478(B35))X3

ARR CGH 13(19,309,535bp->114,110,750bp)X3

arr cgh 13(30 BAC) x 3

ARR CGH 13(44 BAC)X3

ARR CGH 13(Q12.11Q34)(18601703-114077122)X3 59.48MB

arr cgh 13q11q34(17997830-114126525)x3

arr cgh 13q11qter(17.85-114.14Mb)x3

ARR CGH 13Q12.11Q34(18,601,703->114,054,857)X3

ARR CGH 13Q12.11Q34(18400395-114107637)X3

ARR CGH 13Q12.11Q34(19,309,535BP=>114,110,750BP)X3

arr cgh 13q12.11q34(chr13:19309535-114110750)x3

ARR CGH 13Q12.11Q34(OLIGONUCLEOTIDE 18601703->113132980;HG18;95541KB)x3

ARR CGH 13Q12.11Q34(RP11-187L3->RP11-569D9)X3

arr cgh 13q12.11q34(RP11-187L3->RP11-88E10)x3

ARR CGH 13q12.11q34(rp11-76K19->rp11-569D9)x3

ARR CGH 13Q12.11QTER(19,309,335->114,110,891)x3

arr cgh 13q12.11qter(RP11-301J16->RP11-569D9)x3 arr 13q11qter(rs387422->rs12874390)x3

ARR CGH 13Q12.2Q34(RP11-89J10->GS-1-L16)X3

ARR CGH 13Q12Q34(18650698-114110750 B18)X3

ARR CGH 13Q13.3QTER(RP11-186J16->GS-1-L16)X3

ARR CGH 13Q14.2Q34(48903923->114103243)X3[HG17;NCBI(B35)]

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Supplemental Table 2 2008 CYCGH-02 nomenclature

2008 CYCGH-02 (n=27)

ARR 22Q11.2Q11.2(17257787->19795835)X1

ARR CGH 22(Q11.21Q11.21)(17276972-19692296)X1 2.42MB

arr cgh 22q11.1q11.22(RP3-493F17,F5,RP11-186O8,RP11-316L10) x 1

ARR CGH 22Q11.2(17290000-19810000)X1

arr cgh 22q11.21 (chr22:17294496-19829971)x1

ARR CGH 22Q11.21(17,276,972->19,709,958)X1

ARR CGH 22Q11.21(17,294,496BP=>19,886,068BP)X1

ARR CGH 22Q11.21(17,294,496bp->19,829,971bp)X1

arr cgh 22q11.21(17.3-19.8Mb)x1

arr cgh 22q11.21(17271526->19686850(B35))x1

ARR CGH 22Q11.21(17276971-19692296 B18)X1

ARR CGH 22Q11.21(17308438->19681839)X1[HG17;NCBI(B35)]

ARR CGH 22Q11.21(17448680-19779882)X1

ARR CGH 22Q11.21(CTD-2300P14-RP11-505BIG)X1

ARR CGH 22Q11.21(CTD-2522F24->CTD-2593O4)x1

ARR CGH 22Q11.21(RP11-800B02->RP11-330P17)X1

ARR CGH 22Q11.21(RP3-493F17->RP11-316L10)X1

ARR CGH 22Q11.21Q11.21(17,294,296->19,830,112)X1
arr cgh 22q11.21q11.21(17242851-18708054)X1
arr cgh 22q11.21q11.21(CTD-2522F24->CTD-2593O4)x1 arr 22q11.21q11.21(rs5993380->cnv7555P78)x1
arr cgh 22q11.21q11.21(rp11-800b02->rp11-330p17)x1
arr cgh 22q11.21q11.21(RP11-800B02->RP11-330P17)x1
ARR CGH 22Q11.21Q11.21(RP11-800B02->RP11-330P17)X1
ARR CGH 22Q11.21Q11.21(RP3-493F17->RP11-316L10)X1

Supplemental Table3 2008 CYCGH-03 nomenclature

CYCGH-03 (n=34)

ARR 7Q11.23(72363697-73774409)X1 1 2.9
ARR 7Q11.23(RS1880948->cnvi0018678)x1 ARR CGH 7Q11.23(CTD-2069P19->RP11-926D5)x1 1 2.9
ARR 7Q11.23Q11.23(72297531->73780028)X1 1 2.9
ARR CGH 7Q11.23(72,210,960->73,577,926)X1 1 2.9
arr cgh 7Q11.23(72,210,964->73,577,983)x1 1 2.9
ARR CGH 7Q11.23(72,380,000-73,850,000)X1 1 2.9
ARR CGH 7Q11.23(72,404,249BP-73,771,268BP)X1 1 2.9
arr cgh 7q11.23(72,404,250bp->73,771,268bp)x1 1 2.9
ARR CGH 7Q11.23(7214506573584041)X1ARR CGH
7Q11.23(72145065>73584041)X1 1 2.9
ARR CGH 7Q11.23(72210964BP->73577983BP)X1 1 2.9
arr cgh 7q11.23(72330844-73774890)x1 1 2.9
ARR CGH 7Q11.23(72338349-73777326 B18)X1 2 5.9
arr cgh 7q11.23(72338350-73777326)x1 1 2.9
ARR CGH 7Q11.23(72395395->73676067)X1 [HG18; NCBI(B36.1)] 1 2.9
ARR CGH 7Q11.23(72404049->73771409)X1 1 2.9
ARR CGH 7Q11.23(72404248->73771269)x1 1 2.9
ARR CGH 7Q11.23(72514583-73625107)x1 1 2.9
ARR CGH 7Q11.23(B315H11>RP4-665P5)x1 1 2.9
arr cgh 7q11.23(chr7:72210964->73577983)x1 1 2.9
arr cgh 7q11.23(CTB-8H17->CTD-2529O7)x1 1 2.9
ARR CGH 7Q11.23(CTD-2069P19->RP11-926D5)X1 1 2.9
ARR CGH 7Q11.23(RP11-101D2->RP11-247L6)X1 1 2.9
ARR CGH 7Q11.23(RP11-334D20->RP4-771P4)X1 1 2.9
ARR CGH 7Q11.23(RP5-1127A24)X1 1 2.9
ARR CGH 7Q11.23Q11.23(72,210,764BP->73,578,124BP)X1 1 2.9
arr cgh 7q11.23q11.23(72321229-73905031)x1 1 2.9
arr cgh 7q11.23q11.23(oligo 72338350->73777326;hg18)X1 1 2.9
ARR CGH 7Q11.23Q11.23(RP11-101D2->RP11-247L6)X1
arr cgh 7q11.23q11.23(rp11-334D20->rp4-771p4)X1 1 2.9
ARR GGH 7911.23911.23(HG18:72382983-73777326)X1 1.4MB 1 2.9
ARR SNP 7Q11.23(rs11544045-rs1637414)X1 1 2.9
ARR SNP/CN 7Q11.23Q11.23(72361307->73780028)x1 1 2.9
DELETION 7Q11.231.44 MEGABASEKNOWN CNV BENIGN TORONTODELWTION
14Q32.330.45 MEGABASES 1 2.9

Supplemental Table 4 2008 CYCGH-04 nomenclature

CYCGH-04 (n=33)

ARR 15Q11.2Q13.1(18544068-26832495)X3
ARR 15Q11.2Q13.1(18845990->26812205)X3
ARR CGH 15Q11.2Q13.1(HG18:19109124-26199055)X3
7.01MB,22Q11.22Q11.22(HG18:20753061-21520273)X1 0.77MB
ARR CGH 15Q11.2(RP11-80H14>RP11-385H1)x3
ARR CGH 15Q11.2-15Q13.1(20418328-26199055 B18)X3
ARR CGH 15Q11.2Q12(RP11-385h1->RP11-322N14)X3
ARR CGH 15Q11.2Q13.1(18,508,603-26,761,390)X3
arr cgh 15q11.2q13.1(18579732-26735391)x3,22q11.22(20696289-21560322)x1
ARR CGH 15Q11.2Q13.1(18668296->26193908)x3,22Q11.22(20721648->21568920)x1
arr cgh 15q11.2q13.1(18668297-26997968)x3
ARR CGH 15Q11.2Q13.1(20,249,686BP->26,885,085BP)X3
ARR CGH 15Q11.2Q13.1(20,249,885->26,884,885)X3
ARR CGH 15q11.2Q13.1(20,249,886->26,884,937)x3
ARR CGH 15q11.2q13.1(20,249,886bp-->26,829,499bp)x3
ARR CGH 15Q11.2Q13.1(20,249,886BP-26,829,558BP)X3
ARR CGH 15Q11.2Q13.1(20249686->26829699)X3
ARR CGH 15Q11.2Q13.1(20249886BP->26365055BP)X3
ARR CGH 15Q11.2Q13.1(20335887>26199055)X3,22Q11.22(20747615>21514827)X1
arr cgh 15q11.2q13.1(20418329-26199055)x3 1 3.0
ARR CGH 15Q11.2Q13.1(20418329-27917571)x3
ARR CGH 15Q11.2Q13.1(22577151->26079398)X3 [HG18; NCBI(36.1)]
arr cgh 15q11.2-q13.1(chr15:20249886->26739139)x3
ARR CGH 15Q11.2Q13.1(RP11-125E1->RP11-1133P24)x3 ARR 15Q11.2Q13.1(RS2808140->RS6606925)x3
ARR CGH 15Q11.2Q13.1(RP11-125E1->RP11-1133P24)x3,22Q11.22(RP11-346I4->RP11-729G4)x1
arr CGH 15Q11.2Q13.1(RP11-385H1->RP11-322N14)X3 1
ARR CGH 15Q11.2Q13.1(RP11-385H1->RP11-322N14)X3
arr cgh 15q11.2q13.1(rp11-385hi->rp11-322n14)X3
arr cgh 15q11.2q13.1(RP11-452H4->RP11-390P7)x3
arr cgh 22q11.22q11.22(oligo 20609709->21520273;hg18)x1
ARR SNP 15Q11.2Q13.1(RS28726192-RS8040604)X3
ARR SNP/CN 15Q11.2Q13.1(18714261->26249029)x3
DUPLICATION 15Q11.2-Q13.17.09 MEGABASES DELETION 22Q11.220.6 MEGABASES SUBSET OF DGS/VFCS SYNDROM

Supplemental Table 5 2009 CYCGH-01 nomenclature

2009 CYCGH-01 Nomenclature (n=49)

ARR (1-22,X)X2
ARR CGH 1-22(2868BAC)X2,X(189BAC)X2,Y(22BAC)X0
ARR CGH 1-22(39,948 OLIGOS)X2,X(2,715 OLIGOS)X2,Y(364 OLIGOS)X0
ARR CGH 1-22(3900 BAC)X2,X(254 BAC)X2,Y(73 BAC)X0
ARR CGH 1-22(40,543 OLIGOS)X2,X(1,813 OLIGOS)X2,Y(138 OLIGOS)X0(HG17)
arr cgh 1-22(4327 BAC)x2,X(326 BAC)x2,Y(49 BAC)x0 [hg18;NCBI(B36.1)]
arr cgh 1-22(4952 BAC)x2,x(321 BAC)X2,Y(80 BAC)x0
ARR CGH 1-22(4991 BAC)X2,X(321 BAC)X2,Y(80 BAC)X0
ARR CGH 1-22(5108BAC)X2,X(325BAC)X2,Y(83BAC)X0

arr snp 1-22(>500,000 X 2), X(>15,000 X 2), Y (>2,000 X 0)

arr xp11.23(47,430,829-48,410,637)x3

ARR XP11.23(47,487,249-47,720,429)X3

ARR XP11.23(47481143-47743360)x3

arr Xp11.23(47500517-47666689)x3

arr(1-22,x)x2

arr(1-22,X)x2

ARR(1-22,X)x2

ARR(1-22,X)X2

arr(1-22,X)x2 normal female

normal

NORMAL:RATIO PROFILE DOES NOT SHOW SIGNIFICANT GAIN OR LOSS WE DO NOT USE ISCN 2005 FOR A NORMAL
we do not provide iscn designation for cases that are normal by a-cgh. we use an oligo array which is not included i

Supplemental Table 6 2009 CYCGH-02 nomenclature

2009 CYCGH-02 (n=51)

ARR CGH XP22.31(6,348,257BP->7,907,388BP)X0(HG17)

ARR CGH XP22.31(6,348,457-7,907,247)X0 [HG17]

arr cgh Xp22.31(6352088-->8220068)x0 [hg18;NCBI(B36.1)]

arr cgh Xp22.31(6463137-8109132)x0

ARR CGH XP22.31(6561154->7992120 B18)X0

ARR CGH XP22.31(6561154-7992120 B18)X0

ARR CGH XP22.31(RP11-294K6->CTD-2369L20)x0

ARR CGH XP22.31(RP11-294K6->CTD-2369L20)X0

ARR CGH XP22.31(RP11-483M24->RP11-274G7)X0

ARR CGH XP22.31(RP11-483M24->RP11-60P14)X0

arr cgh Xp22.31(RP11-483M24->RP11-60P14)X1,Xp22.12(RP3-488H23)X3

ARR CGH XP22.31XP22.31(RP11-48324->RP11-274G7)X1

arr snp Xp22.31 (rs6654819-rs4830389) X 0

arr Xp22.3(6,947,719-7,922,174)x0

arr Xp22.31(6,410,891-7,841,856)x0 1

ARR XP22.31(6,410,891-8,075,976)X0

arr Xp22.31(6,459,644-8,086,161)x0

arr Xp22.31(6,465,771-8,084,046)x0

ARR XP22.31(6,465,815-8,084,744)X1

arr Xp22.31(6,467,203-7,992,261)x1

ARR XP22.31(6,467,402-8,091,811)X0

arr Xp22.31(6,472,947-8,039,507)x0

ARR XP22.31(6,472,947-8,039,507)X0

arr xp22.31(6,476,806-8,075,294)x0

ARR XP22.31(6,477,005-8,091,811)X1

ARR XP22.31(6,477,902-8,095,645)X0

ARR XP22.31(6,487,037-8,091,950)X0

ARR XP22.31(6,487,237-8,075,154)X1

arr Xp22.31(6,487,238-8,091,751)x0

ARR XP22.31(6,498,521-8,057,652)X0

arr xp22.31(6,498,721-8,057,452)x1

ARR XP22.31(6,498,721-8,057,511)X0
ARR XP22.31(6,498,721-8,057,511)x1
arr xp22.31(6,498,750-8,057,482)x0
arr Xp22.31(6,513,905-7,805,167)x0
ARR XP22.31(6,561,155-7,992,120)X0
arr Xp22.31(6,647,544-8,049,828)x0
ARR XP22.31(6.42-8.06)X1
arr Xp22.31(6458457-8086494)x0
ARR XP22.31(6461783-8075153)x0
ARR XP22.31(6466759-8079297)X0
arr Xp22.31(6477902-8095645)x0
ARR XP22.31(6477902-8095645)x0
ARR XP22.31(6477902-8095645)X1
ARR XP22.31(6498521-8010791)X0
arr xp22.31p22.31(6561155-7992120)x0
XP22.31DEL 6410891-7841856
ARR CGH P15.33P15.31(3369579->7379441 B18)x3 1 1.8
ARR CGH 5P15.33(75148-2945279 B18)X1,5P15.31P15.33(3369579-7679441B18)X3
ARR CGH 5P15.33(CTD-2065F11->RP11-289J13)X1,5P15.33P15.31(RP11-437F24->CTD-2339L17)x3
ARR CGH 5P15.33(RP11-323M9->RP11-572E2)X1,5P15.33P15.31(RP11-20K9->RP11-109F5)X3
arr cgh 5p15.33(RP11-44H14->RP11-572E2)x1,5p15.32->5p15.31(RP11-20K9->RP11-114M17)x3
ARR(1-22)X2,(XY)X1 1 1.8
ARRCGH5p15.33(273228-2945420)X1 ARRCGH5p13.3(3214629-32185621)X3
ARRCGH14q32.33(106216526-106251281)X0 ARRCGH14q32.32-q32.33(105016641-105277086)X3
CHR5P15.33 93489-2945420DELETION CRI DU CHAT 1 1.8
arr 17p13.3(2,043,600-2,535,405)x1
ARR 17P13.3(2,043,600-2,535,405)X1
ARR 17P13.3(2,043,600-2,535,405)X1 [HG18]
arr 17p13.3(2,043,629-2,535,375)x1
arr 17p13.3(2,094,659-2,570,897)x1
arr 17p13.3(2,104,679-2,541,288)x1
arr 17p13.3(2,123,023-2,668,225)x1
arr 17p13.3(2020340-2575920)x1
ARR 17P13.3(2020340-2577460)X1
ARR 17P13.3(2028421-2558481)X1
ARR 17P13.3(2043600-2535405)X1
arr 17p13.3(Chr17:2043600-2535405)x1
arr 17p13.3p13.3(1,844,305-2,652,089)X1
arr 17p13.3p13.3(2,003,397-2,570,939)x1
arr 17p13.3p13.3(2104679-2549373)x1
arr cgh 1-22(5108BAC)x2,X(325BAC)x2
ARR CGH 17P13.3(2,043,400BP->2,535,546BP)X1
arr cgh 17p13.3(2000813-2586017)x1
ARR CGH 17P13.3(2022855->2549373 B18)x1
arr cgh 17p13.3(2022855->2570879)x1 [hg18;NCBI(B36.1)]
ARR CGH 17P13.3(2104678-2549373 B18)X1
ARR CGH 17P13.3(CTD-2350B15->CTD-3225D9)x1

ARR CGH 17P13.3(RP11-297M11->RP11-582F10)X1
ARRCGH14q 32.33(105402801-105845858)X1 ARRCGH17p13.3(2022656-2549514)X1
CHR17P13.3 2104479-2541439DELETION MILLER-DIEKER

Supplemental Table 7 2009 CYCGH-03 nomenclature

2009 CYCGH-03 (n=57)

AAR 5p15.33(131,945-2,915,164)X1,5p15.33p15.31(3,018,786-7,709,666)X3
ARR 5P13.33(75,149-2,915,024)X1,5P15.33P15.31(3,110,571-7,709,526)X3
ARR 5P15.33(0-3,010,253)X1,5P15.33P15.31(3,010,253-7,787,633)X3
arr 5p15.33(0-3,029,252)x1;5p15.33-p15.31(3,029,252-7,766,035)x3
ARR 5P15.33(1,867,332-2,982,621)X1,5P15.33P15.31(2,991,109-7,707,075)X3
arr 5p15.33(106,403-2,945,279)x1
arr 5p15.33(107,954-2,985,297)x1,5p15.33p15.31(3,022,962-7,719,959)x3
arr 5p15.33(108,467-3,000,715)x1,5p15.33p15.31(3,011,154-7,731,472)x3
ARR 5p15.33(108,467-3,000,655)X1,5p15.33p15.31(3,011,154-7,731,472)X3
ARR 5P15.33(124903-2982004)X1,5P15.33P15.31(3016687-7729377)X3
arr 5p15.33(132,145-2,915,025)x1,5p15.33p15.31(3,018,986-7,709,527)x3 1 1.8
ARR 5P15.33(132,145-2,915,025)x1,5P15.33p15.31(3,018,986-7,709,527)x3
ARR 5P15.33(132,145-2,915,025)X1,5P15.33P15.31(3,018,986-7,709,527)X3
arr 5p15.33(234837-2796864)x1,5p15.33p15.31(3256245-7591116)x3
arr 5p15.33(387,034-2,827,801)x1,5p15.33p15.31(3,010,724-7,150,950)x3
arr 5p15.33(62,592-2,795,763)x1,5p15.33p15.31(3,473,138-7,549,486)x3
ARR 5P15.33(68,520-2,992,031)X1,5P15.33P15.31(2,994,346-7,728,886)X3
arr 5p15.33(68,520-2,992,032)x1,5p15.33p15.31(2,994,346-7,731,445)x3
arr 5p15.33(68520-2992031)x1,arr 5p15.33p15.31(3017690-7731444)x3
ARR 5P15.33(68520-3002644)X1,5P15.33P15.31(3002898-7731444)x3
arr 5p15.33(69,497-2,964,508)x1,5p15.33p15.31(3,098,972-7,683,516)x3
ARR 5P15.33(73,747-2,987,319)x1,5P15.33P15.31(2,992,706-7,728,709)x3
ARR 5P15.33(74,949-2,915,165)X1,5P15.33P15.31(3,110,371-7,709,667)X3
ARR 5P15.33(75,149-2,914,965)X1,5P15.33P15.31(3,110,571-7,709,467)X3
arr 5p15.33(75,149-2,915,024)x1,5p15.33p15.31(3,110,571-7,709,526)x3
ARR 5P15.33(75,149-2,915,024)X1,5P15.33P15.31(3,110,571-7,709,526)X3
arr 5p15.33(75,149-2,981,945)x1;arr 5p15.33-p15.31(3,018,987-7,724,046)X3
arr 5p15.33(75,178-2,914,994)x1,5p15.33p15.31(3,110,600-7,709,496)x3
ARR 5P15.33(75149-2915024)X1,5P15.31P15.33(3110571-7709526)X3
ARR 5P15.33(78,942-2,982,145)X1,5P15.33P15.31(3,018,787-7,724,246)X3
arr 5p15.33(80,564-2,987,319)x1,5p15.33p15.31(2,992,706-7,728,709)X3 1 1.8
ARR 5P15.33(88,779-2,990,136)x1,5P15.33P15.31(3,013,778-7,728,886)x3
arr 5p15.33(91,139-2,986,362)X1,5p15.33p15.31(3,008,932-7,717,530)X3
arr 5p15.33(92,607-2,982,145)x1
arr 5p15.33(Chr5:75149-2915024)x1,5p15.33p15.31(Chr5:3110571-7709526)x3
arr 5p15.33p15.33(146,896-2,872,658)X1,5p15.33p15.31(3,172,080-7,675,958)X3
arr 5p15.33p15.33(148243-2945279)x1,15p15.33p15.31(3203418-7679441)x3
arr 5p15.33p15.33(68,520-2,990,114)x1,5p15.33p15.31(2,990,136-7,728,886)x3
ARR 5PTERP15.31(PTER-2,982,145)X1,5P15.31P13.2(3,018,787-7,748,330)x3
ARR 5PTERP15.33(75,149-2,915,024)X1,5P15.33P15.31(3,110,571-7,709,526)X3 [HG18]
arr cgh 5p15.33(132145->2915025)x1,5p15.33p15.31(3018986->7709527)x3[hg18;NCBI(B36.1)]

arr cgh 5p15.33(67744-3022962)x1 arr cgh 5p15.31p15.33(2985239-7758207)x3
 ARR CGH 5P15.33(74,949BP->2,915,165BP)X1,5P15.33P15.31(3,110,371BP->7,709,667BP)X3
 ARR CGH 5P15.33(75148->2945279 B18)x1
 ARR CGH P15.33P15.31(3369579->7379441 B18)x3 1 1.8
 ARR CGH 5P15.33(75148-2945279 B18)X1,5P15.31P15.33(3369579-7679441B18)X3
 ARR CGH 5P15.33(CTD-2065F11->RP11-289J13)X1,5P15.33P15.31(RP11-437F24->CTD-2339L17)x3
 ARR CGH 5P15.33(RP11-323M9->RP11-572E2)X1,5P15.33P15.31(RP11-20K9->RP11-109F5)X3
 arr cgh 5p15.33(RP11-44H14->RP11-572E2)x1,5p15.32->5p15.31(RP11-20K9->RP11-114M17)x3
 ARR(1-22)X2,(XY)X1 1 1.8
 ARRCGH5p15.33(273228-2945420)X1 ARRCGH5p13.3(3214629-32185621)X3
 ARRCGH14q32.33(106216526-106251281)X0 ARRCGH14q32.32-q32.33(105016641-105277086)X3
 CHR5P15.33 93489-2945420DELETION CRI DU CHAT 1 1.8

Supplemental Table 8 2009 CYCGH-04 nomenclature

2009 CYCGH-04 (n=57)

arr 17p13.3(1,872,529-2,652,130)x1
 arr 17p13.3(1990643-2531046)x1
 ARR 17P13.3(2,006,377-2,580,339)X1
 arr 17p13.3(2,013,578-2,594,109)x1
 ARR 17P13.3(2,014,547-2,577,460)X1
 arr 17p13.3(2,014,547-2,584,136)x1
 arr 17p13.3(2,015,100-2,566,365)x1
 ARR 17P13.3(2,017,757-2,555,396)X1
 arr 17p13.3(2,019,699-2,571,657)X1
 ARR 17P13.3(2,020,340-2,570,939)x1
 arr 17p13.3(2,020,442-2,555,744)x1
 ARR 17P13.3(2,020,442-2,560,922)X1
 ARR 17P13.3(2,022,855-2,570,879)x1
 arr 17p13.3(2,022,856-2,566,165)x1
 arr 17p13.3(2,023,102-2,571,657)X1
 ARR 17P13.3(2,028,220-2,571,018)X1
 ARR 17P13.3(2,028,221-2,566,365)X1
 arr 17p13.3(2,028,420-2,570,879)x1
 ARR 17P13.3(2,028,420-2,570,879)X1
 ARR 17P13.3(2,038,515-2,525,398)X1
 arr 17p13.3(2,042,015-2,541,288)x1
 ARR 17P13.3(2,042,462-2,571,657)x1
 ARR 17P13.3(2,043,400-2,535,546)X1
 ARR 17P13.3(2,043,400-2,566,365)X1
 ARR 17P13.3(2,043,600-2,534,404)X1
 arr 17p13.3(2,043,600-2,535,405)x1
 ARR 17P13.3(2,043,600-2,535,405)X1
 ARR 17P13.3(2,043,600-2,535,405)X1 [HG18]
 arr 17p13.3(2,043,629-2,535,375)x1
 arr 17p13.3(2,094,659-2,570,897)x1
 arr 17p13.3(2,104,679-2,541,288)x1
 arr 17p13.3(2,123,023-2,668,225)x1

arr 17p13.3(2020340-2575920)x1
 ARR 17P13.3(2020340-2577460)X1
 ARR 17P13.3(2028421-2558481)X1
 ARR 17P13.3(2043600-2535405)X1
 arr 17p13.3(Chr17:2043600-2535405)x1
 arr 17p13.3p13.3(1,844,305-2,652,089)X1
 arr 17p13.3p13.3(2,003,397-2,570,939)x1
 arr 17p13.3p13.3(2104679-2549373)x1
 arr cgh 1-22(5108BAC)x2,X(325BAC)x2
 ARR CGH 17P13.3(2,043,400BP->2,535,546BP)X1
 arr cgh 17p13.3(2000813-2586017)x1
 ARR CGH 17P13.3(2022855->2549373 B18)x1
 arr cgh 17p13.3(2022855->2570879)x1 [hg18;NCBI(B36.1)]
 ARR CGH 17P13.3(2104678-2549373 B18)X1
 ARR CGH 17P13.3(CTD-2350B15->CTD-3225D9)x1
 ARR CGH 17P13.3(RP11-297M11->RP11-582F10)X1
 ARRCGH14q 32.33(105402801-105845858)X1 ARRCGH17p13.3(2022656-2549514)X1
 CHR17P13.3 2104479-2541439DELETION MILLER-DIEKER
 ARRCGH5p15.33(273228-2945420)X1 ARRCGH5p13.3(3214629-32185621)X3
 ARRCGH14q32.33(106216526-106251281)X0 ARRCGH14q32.32-q32.33(105016641-105277086)X3
 CHR5P15.33 93489-2945420DELETION CRI DU CHAT 1 1.8

Supplemental Table 9 2010 CYCGH-01 nomenclature
CYCGH-01 (n=72)

46,XY,arr 8p23.3p23.1(162,852-7,890,986)x1,8p23.1p21.1(12,625,316-27,438,262)x3
 8P DEL P23.3-23.1ARR 183050-704079689 DUP P23.1-21.1ARR 12664411-27439877
 arr 8p23.1p23.3(151472-7322035)x1,8p21.1p23.1(11879251-27487115)x3
 arr 8p23.3(154,984-7,356,405)x1, 8p23.1p22(12,595,527-27,449,950)x3
 ARR 8P23.3P23.1(103,565-7,213,835)X1,8P23.1P21.1(12,466,379-27,450,457)X3
 arr 8p23.3-p23.1(0-6,715,256)x1;8p23.1-p21.2(12,571,349-27,375,402)x3
 ARR 8P23.3P23.1(0-7,938,904)X1,8P23.1P21.1(12,256,860-27,463,426)X3
 arr 8p23.3p23.1(103,386-7,027,251)x1,8p23.1p21.1(12,072,394-27,452,125)x3
 ARR 8P23.3P23.1(103,565-7,158,495)X1,8P23.1P21.1(12,415,742-27,452,125)X3
 arr 8p23.3p23.1(103,565-7,202,009)x1,8p23.1p21.1(12,071,704-27,461,090)x3
 arr 8p23.3p23.1(103,565-7,213,834)x1,8p23.1(12,071,704-27,448,983)x3
 ARR 8P23.3P23.1(113,277-6,897,267)x1,8P23.1P21.1(12,313,630-27,452,722)X3
 arr 8p23.3p23.1(133,196-6,396,170)x1,8p22p21.2p12,687,920-27,462,612)x3
 arr 8p23.3p23.1(146,703-6,998,207)x1,8p23.1p21.1(12,574,653-27,429,397)x3
 ARR 8P23.3P23.1(151,473X2,166,475-6,901,461X1,6,929,719X2),
 8P23.1P21.1(12,602,169X2,12,627,659-27,431,497X3,27,458,283X2)
 ARR 8P23.3P23.1(166,452-7,139,225)x1,8P23.1P21.1(12,535,243-27,439,736)x3
 arr 8p23.3p23.1(166,614-7,697,225)x1,8p23.1p21.1(12,627,430-27,447,517)x3
 Arr 8p23.3-p23.1(166452-6913276)x1; arr 8p23.1-p21.1(12600375-27447317)x3
 ARR 8P23.3P23.1(166814-6926706 B18)X1,8P23.1P21.1(12600375-27447376 B18)X3
 arr 8p23.3-p23.1(1-6867773)x1 8p23.1-p21.1(12077338-27417713)x3
 arr 8p23.3p23.1(176,279-6,901,486)x1,8p23.1p21.2(12,627,630-27,375,773)x3

arr 8p23.3p23.1(176,477-6,895,593)x1,8p23.1p21.1(12,652,676-27,427,313)x3
 arr 8p23.3p23.1(177,860-6,780,946)x1,8p23.1p21.1(12,264,428-27,447,517)x3
 ARR 8P23.3P23.1(180,5,986,630)X1,8P23.1P21.2(12,604,206-27,380,739)X3
 arr 8p23.3P23.1(181,330-6,867,914)x1,8P23.1P21.1(12,285,264-27,417,854)x3
 ARR 8P23.3P23.1(181,330-6,867,914)X1,8P23.1P21.1(12,630,584-27,417,854)X3
 ARR 8P23.3P23.1(181,530-6,867,773)x1,8P23.1P21.1(12,630,784-27,487,174)x3
 arr 8p23.3p23.1(181,560-8,117,301)x1,8p23.1p21.1(12,630,814-27,417,684)x3
 ARR 8P23.3P23.1(181,605-7,918,726)X1,8P23.1P21.1(12,630,903-27,417,662)x3
 arr 8p23.3p23.1(181530-6867773)x1,8p23.1p21.1(12630784-27487174)x3
 arr 8p23.3p23.1(181530-7290647)x1,8p23.1p21.1(12285464-27431526)x3
 arr 8p23.3p23.1(181560-6867744)x1,8p23.1p21.1(12630814-27417684)x3
 ARR 8P23.3P23.1(184,417-6,867,914)x1,8p23.1p21.1(12,630,584-27,417,854)X3
 ARR 8P23.3P23.1(184,417-6,867,914)X1,8P23.1P21.1(12,630,584-27,417,854)X3
 ARR 8P23.3P23.1(184,417-6,867,914)X1,ARR 8P23.1P21.1(12,630,584-27,417,854)X3
 ARR 8P23.3P23.1(184,617-7,256,229)X1,8P23.1P21.1(12,630,784-27,417,654)X3
 arr 8p23.3p23.1(184617-6860848)x1,8p23.1p21.1(12624473-27447376)x3
 arr 8p23.3p23.1(192,262-6,907,722)x1,8p23.1p21.1(11,319,611-27,555,823)x3
 arr 8p23.3p23.1(192,262-6,907,722)x1,8p23.1p21.1(12,627,495-27,444,693)x3
 arr 8P23.3P23.1(192,262-6,907,722)X1,8P23.1P21.1(12,627,495-27,444,693)X3
 ARR 8P23.3P23.1(192,262-6,907,722)X1,8P23.1P21.1(12,627,495-27,444,693)X3
 arr 8p23.3p23.1(192,262-6,907,722)x1,8p23.1p21.1(12,627,495-27,444,693)x3 Abnormal Male
 arr 8p23.3p23.1(192,262-6,907,722)x1,8p23.1p21.1(12,627,495-27,454,079)x3
 arr 8p23.3p23.1(192,262-8,168,114)x1,8p23.1p21.1(11,895,815-27,470,160)x3
 arr 8p23.3p23.1(192,262-8,179,723)x1,8p23.1p21.1(12,627,495-27,444,693)x3 [hg18;NCBI(B36.1)]
 arr 8p23.3p23.1(202,704-6,867,774)x1,8p23.1p21.1(12,630,783-27,417,714)x3
 ARR 8P23.3P23.1(218688-6907722)X1,8P23.1P21.1(12627495-27489075)X3
 ARR 8P23.3P23.1(355,060-6,962,020)X1,8P22P21.2(12,792,036-27,363,402)X3
 ARR 8P23.3P23.1(36,386-7,158,495)X1,8P23.1P21.1(12,071,704-27,448,983)X3
 arr 8p23.3p23.1(36,386-7,212,767)x1,8p23.1p21.1(12,483,107-27,450,457)x3
 arr 8P23.3P23.1(36,386-7,242,508)X1,8P23.1P21.1(12,071,704-27,448,983)X3
 arr 8p23.3p23.1(36386-7213701)x1,8p23.1p21.1(12487414-27450456)x3
 ARR 8P23.3P23.1(36386-7213834)X1,8P23.1P21.1(12599114-27450456)X3
 ARR 8P23.3P23.1(52,041-6,928,066)x1,8P23.1P21.1(12,570,457-27,450,267)x3
 ARR 8p23.3p23.1(52,041-6,930,491)x1,8p23.1p21.1(12,595,527-27,448,983)x3
 ARR 8P23.3P23.1(63,832-6,901,461)X1,8P23.1P21.1(12,602,169-27,741,612)X3
 ARR 8P23.3P23.1(63,832-7,821,343)X1,8P23.1P21.1(12,276,208-27,431,497)X3
 ARR 8P23.3P23.1(181,530-6,867,773)X1,8P23.1P21.1(12,630,784-27,417,713)X3 [HG18]
 arr 8pterp23.1(154,984-6,994,825)X1, 8p23.1p21.1(12,595,527-27,452,125)X3
 ARR 8PTERP23.1(1-6,949,430)x1,8P22P21.1(12,836,407-27,419,319)x3
 arr 8pterp23.1(pter-8,117,471)x1 arr 8p23.1p21.1(12,600,175-27,447,517)x3 Sex: male
 arr(1-22)x2,(XY)x1
 ARR.8P23.1P23.3(181530-6867773)X1,8P21.1P23.1(12630784-27417713)X3
 ARR8P23.3P23.1(184,647-6,901,461)X1,8P23.1P21.1(12,602,169-27,458,283)X3
 arr8p23.3p23.1(591,043-7,204,451)X1,8p23.3p23.1(12,276,640-27,448,984)X3
 ARR8P23.3P23.1(OBP-6954184BP)X1,8P23.1P21.1(12181423BP-27452414BP)X3
 27,417,854x3,27,487,115x2) (hg18)

Supplemental Table 10 2010 CYCGH-02 nomenclature

CYCGH-02 (n=72)

11P15.5-15.4 DUP1916931-33953071.48MBP DUP OF BWSMICRODUP

46,XY,arr Yq11.223q11.23(24,130,810-26,656,320)x2

arr (1-22)x2(XY)x1

arr (1-22)x2, (XY)x1

arr (1-22)x2,(xy)x1

arr (1-22)x2,(XY)x1

arr (1-22)X2,(XY)X1

ARR (1-22)X2,(XY)X1

arr 8p11.1(43,591,775-43,848,453)x1

arr 8p23.3p23.1(166,452-6,901,436)x1,8p23.1p21.1(12,489,697-27,467,648)x3

arr Yq11.223(23274521-23697630)x3,Yq11.223q11.23(25351719-26905403)x3

arr(1-22)x2, (xy)x1

arr(1-22)x2,(xy)x1

arr(1-22)x2,(XY)x1

arr(1-22)X2,(XY)X1

ARR(1-22)x2,(XY)x1

ARR(1-22)x2,(XY)X1

ARR(-122)X2,(XY)X1

ARR(1-22)X2,(XY)X1

arr(1-22)x2,(xy)x1 (hg18)

arr(1-22)x2,(XY)x1 [hg18;NCBI(B36.1)]

arr(1-22)x2,(XY)x1 normal male

arr(1-22)x2,(XY)x1 Normal Male