Table SI. Details of copy number variation cases covered by Prenatal BoBs™ assay.

Case no.	Karyotype	Prenatal BoBs™ assay				
		Chromosome involved (number of probes in target region)	Number of probes exceeds the threshold S-F/S-M	≥n-1 probes exceed the threshold	NMR, F/M (threshold)	Exceeds the fixed threshold
1	47,XX,+21	Chr21 (5)	5/5	Yes	1.30/1.32 (1.15)	Yes
2	47,XX,+21	Chr21 (5)	5/5	Yes	1.26/1.24 (1.12)	No
3	47,XY,+21	Chr21 (5)	5/5	Yes	1.29/1.32 (1.13)	Yes
4	47,XY,+21	Chr21 (5)	4/5	Yes	1.18/1.23 (1.12)	No
5	47,XX,+21	Chr21 (5)	3/4	No	1.20/1.25 (1.13)	No
6	47,XX,+21	Chr21 (5)	5/4	Yes	1.29/1.24 (1.12)	No
7	47,XY,+21	Chr21(5)	5/4	Yes	1.30/1.26 (1.12)	Yes
8	47,XX,+21	Chr21 (5)	4/5	Yes	1.24/1.26 (1.11)	No
9	47,XY,+21	Chr21 (5)	5/5	Yes	1.30/1.33 (1.13)	Yes
10	47,XY,+21	Chr21 (5)	5/5	Yes	1.33/1.26 (1.12)	Yes
11	47,XX,+21	Chr21 (5)	5/5	Yes	1.31/1.30 (1.12)	Yes
12	47,XY,+21	Chr21 (5)	4/4	Yes	1.23/1.25 (1.16)	No
13	47,XX,+21	Chr21 (5)	5/5	Yes	1.30/1.29 (1.11)	Yes
14	46,XY,+21, der(21;21)(q10;q10)	Chr21 (5)	5/5	Yes	1.29/1.25 (1.12)	No
15	45,XY,-21	Chr21 (5)	5/5	Yes	0.68/0.67 (0.88)	Yes
16	47,XY,+18	Chr18 (5)	5/5	Yes	1.35/1.42 (1.13)	Yes
17	47,XY,+18	Chr18 (5)	5/5	Yes	1.34/1.36 (1.12)	Yes
18	47,XX,+18	Chr18 (5)	5/5	Yes	1.29/1.36 (1.18)	Yes
19	47,XY,+13	Chr13 (5)	4/5	Yes	1.22/1.28 (1.13)	No
20	47,XY,+13	Chr13 (5)	4/4	Yes	1.29/1.35 (1.13)	Yes
21	47,XX,+13	Chr13 (5)	5/5	Yes	1.22/1.28 (1.12)	No
22	47,XXY	ChrX (5)	5	Yes	1.47 (1.09)	Yes
23	47,XXY	ChrX (5)	5	Yes	1.58 (1.11)	Yes
24	47,XXY	ChrX (5)	5	Yes	1.56 (1.10)	Yes
25	47,XXY	ChrX (5)	5	Yes	1.51 (1.14)	Yes
26	47,XXY	ChrX (5)	5	Yes	1.66 (1.11)	Yes
27	47,XXX	ChrX (5)	5	Yes	1.31 (1.13)	Yes
28	47,XYY	ChrY (5)	5	Yes	1.66 (1.19)	Yes
29	47,XYY	ChrY (5)	5	Yes	1.81 (1.21)	Yes
30	45,X	ChrX (5)	5	Yes	0.71 (0.80)	Yes
31	mos 45,X[63]/47,XXX[23]	ChrX (5)	5	Yes	0.88 (0.91)	No
32	mos 46,XY[45]/47,XXY[8]	ChrX (5)	3	No	1.12 (1.10)	No
33	mos 45,X[37]/46,XX[20]	ChrX (5)	5	Yes	0.81 (0.89)	No
34	mos 45,X[19]/46,XX[31]	ChrX (5)	5	Yes	0.82 (0.88)	No
35	mos 45,X[6]/46,XY[69]	ChrY (5)	5	Yes	0.71 (0.85)	Yes
36	mos 45,X[8]/46,X, psu dic(Y)(q12)[52]	ChrY (5)	5	Yes	0.50 (0.85)	Yes
37	mos 45,X[54]/46,XY[7]	ChrY (5)	5	Yes	0.50 (0.81)	Yes
38	46,XY[a]	ChrX (5)	5	Yes	1.31 (1.12)	Yes
		ChrY (5)	5	Yes	0.54 (0.82)	Yes
39	46,XY	Chr15 (PWS,7)	7/7	Yes	1.34/1.32 (1.17)	Yes
40	46,XY	Chr15 (PWS,7)	7/7	Yes	0.67/0.70 (0.86)	Yes
41	46,XY	Chr5 (CDC,8)	8/8	Yes	0.62/0.62 (0.86)	Yes
42	mos 48,XN,+5, +12[4]/46,XN[57]	Chr5 (CDC,8)	2/1	No	0.97/1.00 (1.09)	No
43	46,XX	Chr8 (LGS,7)	3/3	No	1.17/1.17 (1.12)	No
					1.37/1.40[(C1-3)b]	Yes
44	46,XY	Chr8 (LGS,7)	2/2	No	1.18/1.18 (1.14)	No
					1.45/1.44[(C5-6)b]	Yes
45	46,XY	Chr17 (MDS,6)	6/6	Yes	0.65/0.65 (0.89)	Yes
46	46,XX	Chr22 (DGS,4)	4/4	Yes	1.28/1.28 (1.14)	No
47	46,XY	Chr22 (DGS,4)	4/4	Yes	1.28/1.30 (1.10)	Yes
48	46,XY	Chr22 (DGS,4)	4/4	Yes	0.66/0.68 (0.89)	Yes

Table SI. Continued.

Case no.	Karyotype	Prenatal BoBs™ assay				
		Chromosome involved (number of probes in target region)	Number of probes exceeds the threshold S-F/S-M	≥n-1 probes exceed the threshold	NMR, F/M (threshold)	Exceeds the fixed threshold
49	46,XY	Chr10 (DGS,4)	4/4	Yes	0.66/0.66 (0.88)	Yes
50	46,XY,del(4)(p16.1)	Chr4 (WHS,5)	5/5	Yes	0.76/0.75 (0.89)	Yes
51	46,XY,del(4)(p16.1)	Chr4 (WHS,5)	5/5	Yes	0.73/0.72 (0.88)	Yes
52	46,XY	Chr17 (SMS,4)	2/2	No	0.79/0.76 (0.87)	Yes
					0.59/0.57[(C1-2)b]	Yes

[a]Case of maternal cell contamination with normal karyotype after culture. [b]The MNR of the deviation probes (serial number of deviation probes). MNR, mean normalized ratio; S-F/F-M, sample-to-female reference/sample-to-male reference; der, Derivative chromosome; mos, Mosaic; PWS, Prader-Willi syndrome; LGS, Langer-Giedion syndrome; MDS, Miller-Dieker syndrome; WHS, Wolf-Hirschhorn syndrome; CDC, Cri du Chat syndrome; DGS, DiGeorge syndrome; SMS, Smith-Magenis syndrome; Chr, chromosome; BoBs, BACs-on-Beads.