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Supplemental Data

Incidence, Origin, and Predictive Model for the Detection and Clinical Management of Segmental Aneuploidies in Human Embryos

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Figure S1 - Examples of PGT-A profile plots displaying different segmental aneuploidies configurations reconstructed from five multifocal blastocyst biopsies.

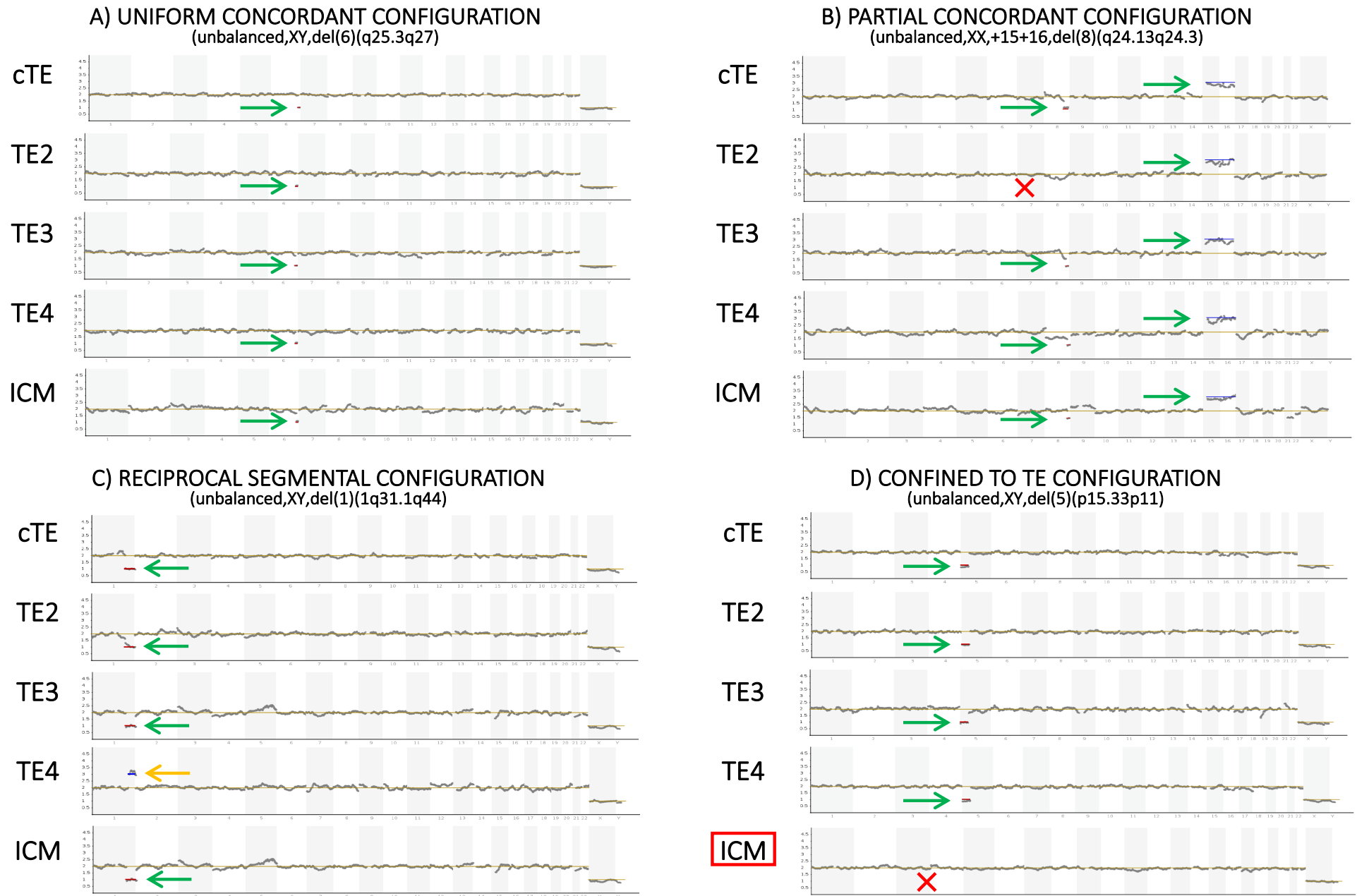


Figure S1: A) Uniform concordant configuration; **B)** Partial concordant configuration; **C)** Reciprocal segmental configuration and **D)** Confined to TE configuration. *cTE*, clinical trophoctoderm biopsy; *TE2*, second trophoctoderm biopsy; *TE3*, third trophoctoderm biopsy; *TE4*, fourth trophoctoderm biopsy; *ICM*, inner cell mass biopsy. Green arrow = confirmed aneuploidy; Yellow arrow = reciprocal aneuploidy; Red cross = unconfirmed aneuploidy.

Table S1 (A, B, C). NGS validation data

A

Barcode	Sample ID	Karyotype	Ion chef + S5 results	READS	MAPD
1	01	unbalanced,XX,+22	unbalanced,XX,+22	191,690	0.146
2	02	unbalanced,XX,+22	unbalanced,XX,+22	193,168	0.149
3	03	unbalanced,XX,+20	unbalanced,XX,+20	145,459	0.162
4	04	unbalanced,XX,+20	unbalanced,XX,+20	161,303	0.160
5	05	unbalanced,XY,+2,+21	unbalanced,XY,+2,+21	206,235	0.142
6	06	unbalanced,XY,+2,+21	unbalanced,XY,+2,+21	146,702	0.163
7	07	unbalanced,XXX	unbalanced,XXX	191,553	0.146
8	08	unbalanced,XXX	unbalanced,XXX	158,018	0.159
9	09	unbalanced,XXY	unbalanced,XXY	176,303	0.190
10	10	unbalanced,XXY	unbalanced,XXY	173,232	0.141
11	11	unbalanced,XXY	unbalanced,XXY	229,748	0.140
12	12	unbalanced,XXY	unbalanced,XXY	92,464	0.196
13	13	unbalanced,XY,+13	unbalanced,XY,+13	154,963	0.164
14	14	unbalanced,XY,+13	unbalanced,XY,+13	92,623	0.192
15	15	unbalanced,XY,+18	unbalanced,XY,+18	171,875	0.176
16	16	unbalanced,XY,+18	unbalanced,XY,+18	142,992	0.156
17	17	unbalanced,XY,+21	unbalanced,XY,+21	160,188	0.162
18	18	unbalanced,XY,+21	unbalanced,XY,+21	135,325	0.172
19	19	balanced,XX	balanced,XX	145,428	0.169
20	20	balanced,XX	balanced,XX	212,568	0.131
21	21	balanced,XY	balanced,XY	173,466	0.160
22	22	balanced,XY	balanced,XY	211,861	0.144
23	23	unbalanced,XX,+2,+21	unbalanced,XX,+2,+21	172,647	0.148
24	24	unbalanced,XX,+2,+21	unbalanced,XX,+2,+21	153,313	0.159
25	25	unbalanced,XX,+22	unbalanced,XX,+22	205,168	0.143
26	26	unbalanced,XX,+22	unbalanced,XX,+22	211,714	0.140
27	27	unbalanced,XX,+20	unbalanced,XX,+20	222,030	0.128
28	28	unbalanced,XX,+20	unbalanced,XX,+20	200,551	0.139
29	29	unbalanced,XY,+2,+21	unbalanced,XY,+2,+21	224,145	0.142
30	30	unbalanced,XY,+2,+21	unbalanced,XY,+2,+21	237,787	0.134
31	31	unbalanced,XXX	unbalanced,XXX	129,491	0.174
32	32	unbalanced,XXX	unbalanced,XXX	152,358	0.176
33	33	unbalanced,XXY	unbalanced,XXY	163,214	0.218
34	34	unbalanced,XXY	unbalanced,XXY	140,940	0.170
35	35	unbalanced,XXY	unbalanced,XXY	183,140	0.147
36	36	unbalanced,XXY	unbalanced,XXY	141,553	0.157
37	37	unbalanced,XY,+13	unbalanced,XY,+13	135,030	0.170
38	38	unbalanced,XY,+13	unbalanced,XY,+13	141,076	0.164
39	39	unbalanced,XY,+18	unbalanced,XY,+18	179,381	0.161
40	40	unbalanced,XY,+18	unbalanced,XY,+18	125,481	0.182
41	41	unbalanced,XY,+21	unbalanced,XY,+21	135,749	0.195
42	42	unbalanced,XY,+21	unbalanced,XY,+21	111,040	0.186
43	43	balanced,XX	balanced,XX	193,936	0.143
44	44	balanced,XX	balanced,XX	234,345	0.138
45	45	balanced,XY	balanced,XY	208,501	0.187
46	46	balanced,XY	balanced,XY	155,989	0.153
47	47	unbalanced,XX,+22	unbalanced,XX,+22	173,663	0.151
48	48	unbalanced,XX,+22	unbalanced,XX,+22	168,681	0.158

Summary of validation results	
Concordant diagnosis per sample	100%, n=48/48;(95%CI=93.41-98.24)
Concordant diagnosis per chromosome	100%, n=1104/1104;95%CI=99.75-99.95
Sensitivity per chromosome	100.0% (n=46/46;95%CI=92.29-100.00)
Specificity per chromosome	100% (n=1058/1058;95%CI=99.65-100.00)

A: Validation data of whole chromosome aneuploid detection from cell lines with known whole chromosome alterations.

B

Barcode	Sample ID	Karyotype TE1	Length	Karyotype TE2	READS	MAPD
1	01	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	328,338	0.195
2	02	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XY,del(5)(p15.33p15.32)	303,345	0.138
3	03	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	259,805	0.156
4	04	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12.2p11.23)	418,440	0.114
5	05	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	260,634	0.146
6	06	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XX,del(5)(p15.33p15.32)	171,997	0.149
7	07	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	128,956	0.176
8	08	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12.2p11.23)	190,785	0.126
9	09	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	159,202	0.116
10	10	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XY,del(5)(p15.33p15.32)	356,937	0.126
11	11	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	198,055	0.126
12	12	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12p11.2)	102,049	0.12
13	13	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	328,338	0.195
14	14	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XY,del(5)(p15.33p15.32)	303,345	0.138
15	15	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	259,805	0.156
16	16	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12.2p11.23)	418,440	0.114
17	17	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	260,634	0.146
18	18	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XX,del(5)(p15.33p15.32)	171,997	0.149
19	19	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	128,956	0.176
20	20	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12.2p11.23)	190,785	0.126
21	21	unbalanced,XY,del(15)(q11.2q13)	7.8 Mb	unbalanced,XY,del(15)(q11.2q13)	159,202	0.116
22	22	unbalanced,XX,del(5)(p15.3)pat.ish	5.8Mb	unbalanced,XY,del(5)(p15.33p15.32)	356,937	0.126
23	23	unbalanced,XY,del(5)(p15.2p14)	6.1 Mb	unbalanced,XY,del(5)(p15.31p14.3)	198,055	0.126
24	24	unbalanced,XY,del(20)(p12p11.2)	5.8 Mb	unbalanced,XY,del(20)(p12p11.2)	102,049	0.12

Summary of validation results	
Concordant diagnosis per sample	100%, n=24/24;(95%CI=85.75-100.00)
Concordant diagnosis per chromosome	100%, n=552/552;(95%CI=99.33-100.00)
Sensitivity per chromosome	100%, n=24/24;(95%CI=85.75-100.00)
Specificity per chromosome	100%, n=528/528;(95%CI=99.30-100.00)

B: Validation data of segmental aneuploid detection from cell lines with known sub-chromosomal alterations.

C

Bar code	Sample ID	Abnormal parental Karyotype	First biopsy result	READS	MAPD	Second biopsy result results	READS	MAPD
1	01	46,XX,t(7;18)(p14;q21.3)	unbalanced,XX,del(7)(p22.3p14.3), dup(18)(q21.32q23)	113,812	0.185	unbalanced,XX,del(7)(p22.3p14.3), dup(18)(q21.32q23)	202,171	0.131
2	02	46,XY,t(1;16)(p34;p13.3)	unbalanced,XY,del(1)(p36.33p34.3)	252,551	0.156	unbalanced,XY,del(1)(p36.33p34.3)	255,934	0.15
3	03	46,XY,t(8;22)(q24;q11)	unbalanced,XY, del(8)(q24.13q24.3), dup(22q11.1q13.33)	237,043	0.161	unbalanced,XY, del(8)(q24.13q24.3), dup(22q11.1q13.33)	203,842	0.145
4	04	46,XX,t(1;4)(q21p14)	unbalanced,XX,dup(1)(q23.3q44), del(4)(p16.3p15.2)	302,629	0.137	unbalanced,XX,dup(1)(q23.3q44), del(4)(p16.3p15.2)	263,016	0.14
5	05	46,XY,t(7;18)(q11.1;q11.1)	unbalanced,XX,dup(7)(p22.3p11.1), del(18)(p11.32p11.21)	213,837	0.159	unbalanced,XX,dup(7)(p22.3p11.1), del(18)(p11.32p11.21)	275,063	0.146
6	06	46,XY,t(5;10)(p13;q11.2)	unbalanced,XX,del(5)(p13.3q35.3), dup(10)(p15.3q11.23)	263,529	0.163	unbalanced,XX,del(5)(p13.3q35.3), dup(10)(p15.3q11.23)	349,467	0.15
7	07	46,XX,t(6;8)(q21;q24.1)	unbalanced,XY,dup(6)(q21q27)	137,716	0.155	unbalanced,XY,dup(6)(q21q27)	206,620	0.165
8	08	46,XX,t(11;18)(p11.2;q21.1)	unbalanced,XX,del(11)(p15.5p12), dup(18)(q21.32q23)	389,210	0.136	unbalanced,XX,del(11)(p15.5p12), dup(18)(q21.32q23)	255,902	0.137
9	09	46,XY,t(4;7)(q27;p15)	unbalanced,XX,del(4)(q27q35.2), dup(7)(p22.3p14.3)	333,194	0.141	unbalanced,XX,del(4)(q27q35.2), dup(7)(p22.3p14.3)	265,908	0.137
10	10	46,XX,t(2;13)(q33;q12)	unbalanced,XX,dup(2)(p25.3q35) del(2)(q35q37.3),del(13)(q11q14.1), dup(13)(q14.11q34)	255,555	0.153	unbalanced,XX,dup(2)(p25.3q35) del(2)(q35q37.3),del(13)(q11q14.1), dup(13)(q14.11q34)	234,603	0.154

C: Validation data of know meiotic segmental aneuploidies detected with PGT-SR in trophoctoderm rebiopsies.

Table S2: Complete overview of blastocyst profiles from aneuploid and euploid cTE samples.

Embryo ID	Embryo grade	Patient Age	ICM biopsy	Clinical TE biopsy	Segmental length (Mb)	TE2	TE3	TE4
C01	5AB	25	unbalanced,XX,-4	unbalanced,XX,del(4)(p16.3q31.1)	141.21-49.82	unbalanced,XX,del(4)(q31.1q35.2)	unbalanced,XX,del(4)(q31.1q35.2)	unbalanced,XX,-4
C02	5AA	28	unbalanced,XY,dup(9)(p24.3q22.2)	unbalanced,XY,dup(9)(p24.3q22.2)	92.33	unbalanced,XY,dup(9)(p24.3q22.2)	unbalanced,XY,dup(9)(p24.3q22.2)	unbalanced,XY,dup(9)(p24.3q22.2)
C03	5AB	33	unbalanced,XX,del(9)(p24.3p13.1)	unbalanced,XX,del(9)(p24.3p13.1)	39.6	unbalanced,XX,del(9)(p24.3p13.1)	unbalanced,XX,del(9)(p24.3p13.1)	unbalanced,XX,del(9)(p24.3p13.1)
C04	5BB	39	unbalanced,XY,-17,dup(20)(p13p11.1)	unbalanced,XY,-17,dup(20)(p13p11.1)	26.260	unbalanced,XY,-17,dup(20)(p13p11.1)	unbalanced,XY,-17,dup(20)(p13p11.1)	unbalanced,XY,-17,dup(20)(p13p11.1)
C05	5BB	32	unbalanced,XY,del(3)(q11.1q29)	unbalanced,XY,del(3)(q11.1q29)	104.46	unbalanced,XY,del(3)(q11.1q29)	unbalanced,XY,del(3)(q11.1q29)	unbalanced,XY,del(3)(q11.1q29)
C06	5AA	32	unbalanced,XY,del(2)(p25.3p11.2)	unbalanced,XY,del(2)(p25.3p11.2)	89.62	unbalanced,XY,del(2)(p25.3p11.2)	unbalanced,XY,del(2)(p25.3p11.2)	unbalanced,XY,del(2)(p25.3p11.2)
C07	5AA	32	unbalanced,XY,dup(2)(q11.1q37.3)	unbalanced,XY,dup(2)(q11.1q37.3)	147.78	unbalanced,XY,dup(2)(q11.1q37.3)	unbalanced,XY,dup(2)(q11.1q37.3)	unbalanced,XY,dup(2)(q11.1q37.3)
C08	5AA	24	unbalanced,XY,del(6)(q25.3q27)	unbalanced,XY,del(6)(q25.3q27)	12.59	unbalanced,XY,del(6)(q25.3q27)	unbalanced,XY,del(6)(q25.3q27)	unbalanced,XY,del(6)(q25.3q27)
C09	5BB	40	unbalanced,XX,del(4)(p16.3p13)	unbalanced,XX,del(4)(p16.3p13)	41.55	unbalanced,XX,del(4)(p16.3p13)	unbalanced,XX,del(4)(p16.3p13)	unbalanced,XX,del(4)(p16.3p13)
C010	5BB	23	unbalanced,XY,del(8)(p23.3p11.1)	unbalanced,XY,del(8)(p23.3p11.1)	43.78	unbalanced,XY,del(8)(p23.3p11.1)	unbalanced,XY,del(8)(p23.3p11.1)	unbalanced,XY,del(8)(p23.3p11.1)
C011	5BC	33	unbalanced,XY,del(4)(q27q35.2)	unbalanced,XY,del(4)(q27q35.2)	69.75	unbalanced,XY,del(4)(q27q35.2)	unbalanced,XY,del(4)(q27q35.2)	unbalanced,XY,del(4)(q27q35.2)
C012	5BB	37	unbalanced,XX,del(9)(p24.3p12)	unbalanced,XX,del(9)(p24.3p12)	42.60	unbalanced,XX,del(9)(p24.3p12)	unbalanced,XX,del(9)(p24.3p12)	unbalanced,XX,del(9)(p24.3p12)
C013	5BB	26	unbalanced,XX,del(3)(p26.3p25.1)	unbalanced,XX,del(3)(p26.3p25.1)	16.023	unbalanced,XX,del(3)(p26.3p25.1)	unbalanced,XX,del(3)(p26.3p25.1)	unbalanced,XX,del(3)(p26.3p25.1)
C014	6AB	40	unbalanced,XY,+15,del(8)(q22.3q24.3)	unbalanced,XY,+15,del(8)(q22.3q24.3)	40.59	unbalanced,XY,+15,del(8)(q22.3q24.3)	unbalanced,XY,+15,del(8)(q22.3q24.3)	unbalanced,XY,+15,del(8)(q22.3q24.3)
C015	5CB	22	unbalanced,XY,del(5)(p15.33p14.1)	unbalanced,XY,del(5)(p15.33p14.1)	27.86	unbalanced,XY,del(5)(p15.33p14.1)	unbalanced,XY,del(5)(p15.33p14.1)	unbalanced,XY,del(5)(p15.33p14.1)
C016	3BB	26	unbalanced,XY,del(18)(q12.1q23)	unbalanced,XY,del(18)(q12.1q23)	45.68	unbalanced,XY,del(18)(q12.1q23)	unbalanced,XY,del(18)(q12.1q23)	unbalanced,XY,del(18)(q12.1q23)
C017	4AA	39	unbalanced,XY,+16,del(11)(p15.5p12)	unbalanced,XY,+16,del(11)(p15.5p12)	40.80	unbalanced,XY,+16,del(11)(p15.5p12)	unbalanced,XY,+16,del(11)(p15.5p12)	unbalanced,XY,+16,del(11)(p15.5p12)
C018	4BB	43	unbalanced,XX,+1,-4,-6,+22	unbalanced,XX,+1,-6,+22,del(4)(q25q35.2)	81.71	unbalanced,XX,+1,+22	unbalanced,XX,+1,+22	unbalanced,XX,+1,-4,-6,+22
C020	5AB	33	unbalanced,XX,dup(9)(p12q34.3)	unbalanced,XX,dup(9)(p12q34.3)	99.74	unbalanced,XX,dup(9)(p12q34.3)	unbalanced,XX,dup(9)(p12q34.3)	balanced,XX
C021	5AA	39	unbalanced,XX,+15,+16,del(8)(q24.13q24.3)	unbalanced,XX,+15,+16,del(8)(q24.13q24.3)	20.57	unbalanced,XX,+15,+16	unbalanced,XX,+15,+16,del(8)(q24.13q24.3)	unbalanced,XX,+15,+16,del(8)(q24.13q24.3)
C022	5AA	31	unbalanced,XX,del(7)(q11.23q36.3)	unbalanced,XX,del(7)(q11.23q36.3)	86.537	balanced,XX	unbalanced,XX,del(7)(q11.23q36.3)	unbalanced,XX,del(7)(q11.23q36.3)
C023	5AB	21	unbalanced,XY,dup(17)(q12q25.3)	unbalanced,XY,dup(17)(q12q25.3)	46.46	balanced,XY	unbalanced,XY,dup(17)(q12q25.3)	balanced,XY
C024	5BB	29	unbalanced,XX,del(3)(q13.11q29)	unbalanced,XX,del(3)(q13.11q29)	92.40	balanced,XX	unbalanced,XX,del(3)(q13.11q29)	balanced,XX
C025	5AA	39	unbalanced,XX,+15,+16,dup(8)(p23.3q24.13)	unbalanced,XX,+15,+16,dup(8)(p23.3q24.13)	124.74	unbalanced,XX,+15,+16	unbalanced,XX,+15,+16	unbalanced,XX,+15,+16
C026	5AB	26	unbalanced,XY,del(1)(q31.1q44)	unbalanced,XY,del(1)(q31.1q44)	59.10-44.99-104.09	unbalanced,XY,del(1)(q31.1q44)	unbalanced,XY,del(1)(q21.1q44)	unbalanced,XY,dup(1)(q32.1q44)
C027	5BB	39	unbalanced,XX,del(7)(q22.3q36.3)	unbalanced,XX,del(7)(p22.3p11.1)	50.64-57.77-97.16	unbalanced,XX,del(7)(q31.1q36.3)	unbalanced,XX,del(7)(q11.21q36.3)	unbalanced,XX,-7
C029	5CB	26	unbalanced,XX,del(4)(q27q35.2)	unbalanced,XX,del(4)(q27q35.2)	117.29-69.754-113.30	unbalanced,XX,del(4)(q27q35.2)	unbalanced,XX,del(4)(p16.3q25)	unbalanced,XX,-4
C030	5AB	29	balanced,XY	unbalanced,XY,del(5)(p15.33p11)	46.40	unbalanced,XY,del(5)(p15.33p11)	unbalanced,XY,del(5)(p15.33p11)	unbalanced,XY,del(5)(p15.33p11)
C031	5BB	37	balanced,XX	unbalanced,XX,del(1)(p36.33p36.11)	25.12	unbalanced,XX,del(1)(p36.33p36.11)	unbalanced,XX,del(1)(p36.33p36.11)	unbalanced,XX,del(1)(p36.33p36.11)
C032	5BB	32	balanced,XY	unbalanced,XY,dup(4)(q13.3q35.2)	115.59-93.66	unbalanced,XY,del(4)(q22.3q35.2)	balanced,XY	unbalanced,XY,-4
C033	6BB	26	balanced,XX	unbalanced,XX,dup(9)(q21.11q34.3)	70.32	unbalanced,XX,dup(9)(q21.11q34.3)	balanced,XX	unbalanced,XX,dup(9)(q21.11q34.3)
C034	5AA	30	balanced,XY	unbalanced,XY,del(12)(q21.31q24.33)	52.28	unbalanced,XY,del(12)(q21.31q24.33)	unbalanced,XY,del(12)(q21.31q24.33)	balanced,XY
C035	5AA	37	balanced,XX	unbalanced,XX,dup(7)(p22.3p14.1)	40.07	balanced,XX	balanced,XX	balanced,XX
C036	5BB	29	balanced,XX	unbalanced,XX,dup(4)(q21.22q35.2)	107.62	balanced,XX	balanced,XX	balanced,XX
C037	5BB	29	balanced,XX	unbalanced,XX,dup(8)(p23.3p22.3)	14.11	balanced,XX	balanced,XX	balanced,XX
C039	5AB	37	unbalanced,XX,+21	unbalanced,XX,+21,del(4)(q32.1q35.2)	33.88	unbalanced,XX,+21	unbalanced,XX,+21	unbalanced,XX,+21
C040	5AB	21	balanced,XY	unbalanced,XY,dup(X)(q24q28)	34.86	balanced,XY	balanced,XY	balanced,XY
C041	5AA	28	balanced,XX	unbalanced,XX,dup(4)(q34.3q35.2)	11.96	balanced,XX	balanced,XX	balanced,XX
C042	5BB	29	balanced,XX	unbalanced,XX,dup(11)(q13.3q25)	65.17	balanced,XX	balanced,XX	balanced,XX
C043	5AB	29	balanced,XY	unbalanced,XY,dup(1)(p36.33p36.13)	16.60	balanced,XY	balanced,XY	balanced,XY
C044	5BB	45	unbalanced,X0,-9,+12,-14	unbalanced,X0,-9,+12,-14,dup(6)(p25.3p22.3)	22.01	unbalanced,X0,-9,+12,-14	unbalanced,X0,-9,+12,-14	unbalanced,X0,-9,+12,-14
C045	5AB	39	balanced,XX	unbalanced,XX,del(8)(q11.23q24.3)	90.53	balanced,XX	balanced,XX	balanced,XX
C046	5AB	25	unbalanced,XY,+2	unbalanced,XY,+2,del(1)(p36.33p36.13)	16.60	unbalanced,XY,+2	unbalanced,XY,+2	unbalanced,XY,+2
C047	5AA	22	balanced,XY	unbalanced,XY,dup(9)(p24.3p2)	13.88	balanced,XY	balanced,XY	balanced,XY
C048	5AA	22	balanced,XY	unbalanced,XY,del(10)(q25.1q26.3)	27.76	balanced,XY	balanced,XY	balanced,XY
C049	5AB	26	balanced,XX	unbalanced,XX,dup(16)(q11.2q24.3)	43.91	balanced,XX	balanced,XX	balanced,XX
C050	3BB	26	balanced,XY	unbalanced,XY,dup(9)(q21.11q34.3)	70.31	balanced,XY	balanced,XY	balanced,XY
C051	5AA	36	balanced,XX	unbalanced,XX,dup(16)(q11.2q24.3)	43.90	balanced,XX	balanced,XX	balanced,XX
C052	6BC	21	balanced,XY	unbalanced,XY,dup(17)(q22q25.3)	30.65	balanced,XY	balanced,XY	balanced,XY
C053	5BB	32	balanced,XY	unbalanced,XY,dup(3)(p26.3p11.1)	90.44	balanced,XY	balanced,XY	balanced,XY
C054	5BB	35	unbalanced,XX,-22	unbalanced,XX,-22,dup(6)(q14.1q27)	94.30	unbalanced,XX,-22	unbalanced,XX,-22	unbalanced,XX,-22
C055	5BB	29	balanced,XX	balanced,XX,dup(10)(q22.2q26.3)	59.94	balanced,XX	balanced,XX	balanced,XX

C056	5AB	29	balanced,XX	balanced,XX,dup(X)[q21.1q28]	76.18	balanced,XX	balanced,XX	balanced,XX
C057	6BB	36	balanced,XY	balanced,XY		balanced,XY	unbalanced,XY,+8	balanced,XY
C058	5AA	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C059	5AA	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C060	5BA	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C061	5AA	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C062	5BB	36	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C063	5AB	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C064	5AA	23	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C065	5AA	31	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C066	5AB	36	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C067	4BB	31	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C068	5AB	36	balanced,XX	balanced,XX	60.51	balanced,XX	unbalanced,XX,dup(6)(6q21q27)	unbalanced,XX,del(6)(6q21q27)
C069	5AA	38	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C070	4AA	26	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C072	4BB	25	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C073	4AA	25	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C074	5BB	34	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C075	5AA	34	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C076	5BC	33	balanced,XX	balanced,XX		balanced,XX	balanced,XX	unbalanced,XX,+14
C077	5BC	33	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C077	5AB	33	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C079	5AB	34	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C080	5AB	34	balanced,XY	balanced,XY		balanced,XY	balanced,XY	balanced,XY
C081	5AB	34	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX
C082	5AB	34	balanced,XX	balanced,XX		balanced,XX	balanced,XX	balanced,XX

Table S3: Logistic regression analysis using confirmation occurrence as independent variable to assess the co-variates involved in segmental aneuploidy confirmation following cTE biopsy diagnosis.

	B	S.E.	df	Sig.	Exp(B)	95% C.I. for EXP(B)	
						Lower	Upper
center(1)	-.445	.612	1	.468	.641	.193	2.127
morpho			3	.804			
morpho(good)	.021	.784	1	.978	1.021	.220	4.745
morpho(average)	-.256	.798	1	.748	.774	.162	3.702
morpho(poor)	.275	.753	1	.715	1.317	.301	5.766
dayOfBiopsy			2	.991			
dayOfBiopsy(6)	.061	.831	1	.941	1.063	.208	5.420
dayOfBiopsy(7)	.098	.763	1	.898	1.103	.247	4.919
FemaleAge	.050	.042	1	.236	1.051	.968	1.142
sperm(1)	-.409	.470	1	.385	.664	.264	1.670
Male Age	-.012	.028	1	.676	.988	.936	1.044
Confirmed in second TE	3.65	.912	1	.000	38.719	6.485	231.169
Segmental Size	.012	.006	1	.047	1.028	1.000	1.056
Constant	-1.442	2.136	1	.500	.003	.027	

	N	Mean	Std. Deviation	95% Confidence Interval for Mean		Minimum	Maximum
				Lower Bound	Upper Bound		
				not confirmed	52		
confirmed	56	60.4070	33.87579	51.3350	69.4790	12.59	147.78
Total	108	53.9107	34.71073	47.2895	60.5320	.00	147.78

Table S4: Demographic data of the study population

Number of couples producing blastocysts with segmental aneuploidies, n	53
Maternal age, mean \pm SD (min-max)	38 \pm 3 (31-44) yr
Duration of infertility, mean \pm SD (min-max)	3 \pm 2 (1-10) yr
Cause of Infertility, %	Idiopathic, 54% Tubal factor, 7% Endometriosis, 11% Endocrine-ovulatory, 4% Male factor, 24%
Basal Follicle Stimulating Hormone, mean \pm SD (min-max)	7 \pm 4 (3-11) IU/l
Basal Anti-Mullerian Hormone, mean \pm SD (min-max)	2 \pm 1 (0.5-5) ng/ml
Body Mass Index	20 \pm 3 (16-23)
Paternal age, mean \pm SD (min-max)	40 \pm 5 (33-53) yr
Sperm factor, %	Normozoospermic, 51% Moderate male factor, 25% Severe male factor, 24%