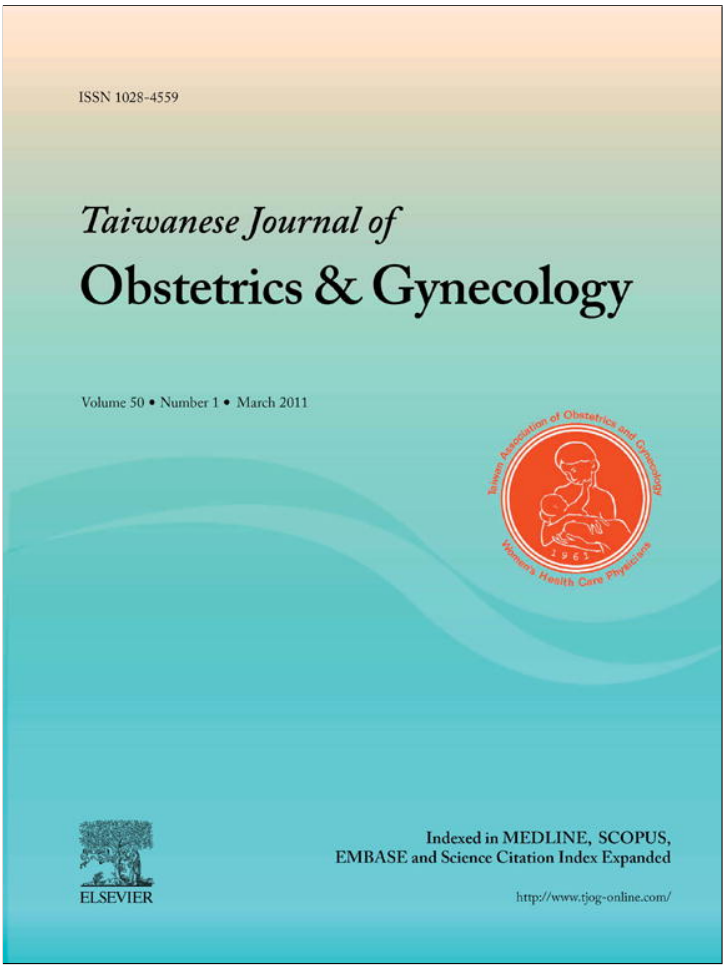

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Original Article

Unbalanced reciprocal translocations at amniocentesis

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Abstract

Objective: To present perinatal findings, modes of ascertainments, and modes of segregation in unbalanced reciprocal translocations detected at amniocentesis.

Materials and Methods: Between January 1987 and July 2010, 40 cases with unbalanced reciprocal translocations were diagnosed by amniocentesis at Mackay Memorial Hospital, Taipei, Taiwan. The 40 cases originated from 29 families; 21 families with one case, 7 families with two cases, and 1 family with five cases.

Results: Of 40 cases, 33 (82.5%) presented fetal ultrasound abnormalities and 7 (17.5%) presented no ultrasound abnormalities. Of 40 cases, 36 (90%) had a segregation mode of adjacent-1 2:2 segregation, 3 (7.5%) had a segregation mode of 3:1 segregation with tertiary trisomy, and 1 (2.5%) had a segregation mode of 3:1 segregation with tertiary monosomy. Of 29 families, 7 (24.1%) had *de novo* translocations and 22 (75.9%) had inherited translocations. In seven *de novo* cases, the main modes of ascertainments included abnormal ultrasound findings ($n = 5$) and advanced maternal age ($n = 2$). In 22 inherited families, the main modes of first ascertainment included abnormal ultrasound findings ($n = 8$), a previous aneuploid child ($n = 8$), advanced maternal age ($n = 4$), parental carrier status ($n = 1$), and abnormal maternal serum screening results ($n = 1$). Among 22 inherited families, 9 (40.9%) had a known parental carrier status, but 13 (59.1%) were unaware of parental carrier status at amniocentesis.

Conclusion: Unbalanced reciprocal translocations detected at amniocentesis are frequently associated with abnormal ultrasound findings. Prenatal diagnosis of an unbalanced translocation may incidentally detect a balanced translocation in the family. Prenatal diagnosis of fetal structural abnormalities should alert structural chromosome rearrangements and prompt cytogenetic analysis of the fetus and parents if necessary.

Keywords: Amniocentesis; Unbalanced reciprocal translocation

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Introduction

A simple reciprocal translocation is produced when there is a two-way exchange between the chromosomes resulting in the translocated segment without the centromere and the centric segment with the centromere. The rearranged chromosome is also called a derivative chromosome, or der. When the breakpoints lie within the centromere, the translocation is additionally called a whole arm translocation. When the translocation results in loss and/or increase in genetic materials, the translocation is regarded as unbalanced. When there is no loss or increase in genetic materials, the translocation is balanced. Amniocentesis may detect inherited or *de novo* reciprocal translocations with either balanced or unbalanced rearrangements. In the inherited translocation cases, the parents may have a known parental carrier status before amniocentesis or may be aware of the parental carrier status only after detection of a fetus with a chromosome aberration at amniocentesis. Jacobs et al [1] found a prevalence of 0.208% for unbalanced structural abnormalities and a prevalence of 0.017% for unbalanced reciprocal translocations at prenatal diagnosis. Here, we present our experience of prenatal diagnosis of unbalanced reciprocal translocations by amniocentesis.

Materials and methods

Between January 1987 and July 2010, unbalanced reciprocal translocations were diagnosed by amniocentesis in 40 cases from 29 families at Mackay Memorial Hospital, Taipei, Taiwan because of various reasons including advanced maternal age, abnormal ultrasound findings, abnormal maternal serum screening results, a previous aneuploid child in the obstetric history or in the family, a family history of congenital anomalies or chromosome aberrations, and other reasons. Cytogenetic analyses of parental blood lymphocytes were done in all cases. The clinical data of the 40 cases are summarized in Table 1.

Results

In this study, the 40 cases of unbalanced reciprocal translocations were originated from 29 families; 21 families (Families 1–5, 7, 9, 11, 13, 15, 18–20, and 22–29) with one case, 7 families (Families 6, 8, 12, 14, 16, 17, and 21) with two cases, and 1 family (Family 10) with five cases. In the eight families with two or more than two cases, two families (Families 12 and 17) (Fig. 1) had different unbalanced reciprocal translocations, whereas the other six families had the same unbalanced reciprocal translocation in the progeny. Of these 40 cases, the mean gestational age at amniocentesis was 19.13 ± 3.90 weeks (range, 14–32 weeks) and the mean maternal age at amniocentesis was 30.55 ± 4.87 years (range, 19–42 years).

Of the 40 cases, 33 cases (82.5%) manifested fetal structural abnormalities on ultrasound, whereas the other 7 cases (17.5%) presented no ultrasound abnormalities. In the 29 families, the main modes of ascertainments included abnormal ultrasound findings ($n = 13$), a previous aneuploid child in the obstetric history or in the family ($n = 8$), advanced maternal age ($n = 6$), parental carrier status ($n = 1$), and abnormal maternal serum screening results ($n = 1$). Of these 29 families, 7 (24.1%) were associated with *de novo* translocations and 22 (75.9%) were associated with inherited translocations. In the seven *de novo* families, the main modes of ascertainments included abnormal ultrasound findings ($n = 5$) and advanced maternal age ($n = 2$). Polymorphic DNA marker analysis was applied to determine the parental origin of the *de novo* chromosome in five *de novo* cases (Cases 9, 18, 20, 27, and 28) of which three (Cases 9, 18, and 20) were of maternal origin, and two (Cases 27 and 28) were of paternal origin. In the 22 inherited families, the main modes of first ascertainment included abnormal ultrasound findings ($n = 8$), a previous aneuploid child in the obstetric history or in the family ($n = 8$), advanced maternal age ($n = 4$), parental carrier status (Case 4) ($n = 1$), and abnormal maternal serum screening results ($n = 1$). The maternal carrier status in Case 4 was identified before amniocentesis because she had a carrier sister whose carrier status was identified after prenatal diagnosis of a fetus with a balanced translocation. Among these 22 families with inherited reciprocal translocations, 9 (40.9%) had a known parental carrier status before the first amniocentesis because of a previous aneuploid child in the obstetric history or in the family ($n = 8$) or parental carrier status ($n = 1$), whereas the other 13 (59.1%) were aware of their parental carrier status only after detection of fetal aneuploidy by amniocentesis because of abnormal ultrasound findings ($n = 8$), advanced maternal age ($n = 4$), or abnormal maternal serum screening results ($n = 1$).

Of the 40 cases originated from 29 families (inherited plus *de novo*), 36 (90%) had a segregation mode of adjacent-1 2:2 segregation including one whole arm translocation (Case 1) (Fig. 2), 3 (7.5%) had a segregation mode of 3:1 segregation with tertiary trisomy (Cases 4, 5, and 19) (Fig. 4), and 1 (2.5%) had a segregation mode of 3:1 segregation with tertiary monosomy (Case 9) (Fig. 3). The translocation in Case 9 with 3:1 segregation with tertiary monosomy arose *de novo*. All the three cases (Cases 4, 5, and 19) with 3:1 segregation with tertiary trisomy had maternal inheritance of the translocation. Of the 33 cases originated from 22 inherited families, 30 (90.9%) had a segregation mode of adjacent-1 2:2 segregation, 3 (9.1%) had a segregation mode of 3:1 segregation with tertiary trisomy. For the progeny with an adjacent-1 2:2 segregating reciprocal translocation in 21 couples of 19 inherited families, the parental female carrier/male carrier ratio was 9:12. For the progeny with a 3:1 segregating reciprocal translocation in three couples of three inherited families, the parental female carrier/male carrier ratio was 3:0.

Discussion

In this study, most unbalanced reciprocal translocations detected at amniocentesis were ascertained through abnormal ultrasound findings (44.8%, 13/29), a previous aneuploid child in the obstetric history or in the family (27.6%, 8/29), and advanced maternal age (20.7%, 6/29), but none was associated

Table 1
Clinical data for cases with unbalanced reciprocal translocations diagnosed by amniocentesis

Family/case	Indication for amniocentesis	Maternal age (yr)	Gestational age at amniocentesis (wk)	Fetal karyotype	Inheritance	Carrier status	Ultrasound abnormalities and references
1	Previous aneuploid child,[a] maternal carrier	33	16	46,XX,der(15;16)(q10;q10),+16	Maternal 46,XX,der(15;16)(q10;q10)	K	No. Ref: Chen et al [16]
2	AMA[a]	38	20	46,XY,der(3)t(2;3)(p25.3;p25)	Maternal 46,XX,t(2;3)(p25.3;p25)	UK	Single umbilical artery, short limbs. Ref: Chen et al [17]
3	AMA[a]	34	18	46,XX,der(6)t(3;6)(p23;p21.3)	*De novo*	UK	No
4	Maternal carrier status,[a,b] AMA	41	17	47,XY,+der(9)t(9;21)(q22;q22.3)	Maternal 46,XX,t(9;21)(q22;q22.3)	K	Ventriculomegaly, megacisterna magna, IUGR. Ref: Chen and Shih [18]
5	AMA[a]	39	19	47,XY,+der(21)t(12;21)(p13.3;q21)	Maternal 46,XX,t(12;21)(p13.3;q21)	UK	No. Ref: Chen et al [19]
6-1A	Previous aneuploid child,[a] paternal carrier	26	20	46,XY,der(2)t(2;3)(q37;p21)	Paternal 46,XY,t(2;3)(q37;p21)	K	HPE, cyclopia. Ref: Chen et al [20]
6-1B	The same mother as 6-1A	27	14	46,XX,der(2)t(2;3)(q37;p21)	The same as 6-1A	K	HPE, PMA. Ref: Chen et al [20]
7	Abnormal ultrasound[a]	32	32	46,XX,der(7)t(3;7)(p23;q36)	*De novo*	UK	HPE, PMA. Ref: Chen et al [20]
8-1A	Previous aneuploid child,[a] maternal carrier	25	18	46,XX,der(11)t(3;11)(q21;q23)	Maternal 46,XX,t(3;11)(q21;q23)	K	Omphalocele. Ref: Chen et al [21]
8-1B	The same mother as 8-1A	27	16	46,XY,der(11)t(3;11)(q21;q23)	The same as 8-1A	K	Omphalocele. Ref: Chen [22]
9	Abnormal ultrasound[a]	28	27	45,XX,der(4)t(4;14)(p16.3;q12),-14	*De novo* (m)	UK	IUGR, microcephaly, cardiomegaly, arrhythmia, asymmetric upper limbs. Ref: Chen et al [23]
10-1A	Previous aneuploid child,[a] maternal carrier	28	19	46,XY,der(22)t(10;22)(q24.1;p11.2)	Maternal 46,XX,t(10;22)(q24.1;p11.2)	K	Pyelectasis. Ref: Chen et al [24]
10-1B	The same mother as 10-1A	30	22	46,XX,der(22)t(10;22)(q24.1;p11.2)	The same as 10-1A	K	Pyelectasis. Ref: Chen et al [25,26]
10-2A	Maternal carrier,[a] familial translocation, sister of 10-1	26	17	46,XY,der(22)t(10;22)(q24.1;p11.2)	Maternal 46,XX,t(10;22)(q24.1;p11.2)	K	Pyelectasis.
10-2B	The same mother as 10-2A	27	18	46,XX,der(22)t(10;22)(q24.1;p11.2)	The same as 10-2A	K	No
10−3	Paternal carrier,[a] familial translocation, sister-in-law of 10-1	19	18	46,XX,der(22)t(10;22)(q24.1;p11.2)	Paternal 46,XY,t(10;22)(q24.1;p11.2)	K	Pyelectasis. Ref: Chen et al [27]
11	Abnormal ultrasound[a]	26	23	46,XX,der(7)t(3;7)(p23;q36)	Paternal 46,XY,t(3;7)(p23;q36)	UK	HPE, cyclopia. Ref: Chen et al [28]
12-1A	Abnormal ultrasound[a]	27	21	46,XY,der(18)t(18;21)(p11.3;q22.3)	Maternal 46,XX,t(18;21)(p11.3;q22.3)	UK	HPE, PMA. Ref: Chen et al [29]
12-1B	Maternal carrier,[a] the same mother as 12-1A	29	17	46,XX,der(21)t(18;21)(p11.3;q22.3)	The same as 12-1A	K	No. Ref: Chen et al [30]

13	Abnormal ultrasound[a]	27	16	46,XY,der(6)t(3;6)(q22;q25.3)	Maternal 46,XX,t(3;6)(q22;q25.3)	UK	Cystic hygroma, pleural effusion. Ref: Chen [31]
14-1A	Previous aneuploid child,[a] paternal carrier[c]	26	17	46,XX,der(11)t(3;11)(p21;q23)	Paternal 46,XY,t(3;11)(p21;q23)	K	DWM. Ref: Chen et al [32]
14-1B	The same mother as 14-1A	30	16	46,XY,der(11)t(3;11)(p21;q23)	The same as 14-1A	K	HPE, PMA, pyelectasis, unilateral duplex renal system. Ref: Chen et al [33]
15	Previous aneuploid child,[a] maternal carrier	31	17	46,XY,der(12)t(9;12)(p11.2;p13.3)	Maternal 46,XX,t(9;12)(p11.2;p13.3)	K	DWM. Ref: Chen et al [34]
16-1	Previous aneuploid child,[a] paternal carrier	28	18	46,XX,der(22)t(16;22)(q12.1;q13.3)	Paternal 46,XY,t(16;22)(q12.1;q13.3)	K	Dolichocephaly. Ref: Chen et al [35]
16-2	Paternal carrier,[a] AMA, spouse of the ex-husband of 16-1	36	18	46,XX,der(22)t(16;22)(q12.1;q13.3)	The same as 16-1	K	Dolichocephaly. Ref: Chen et al [36]
17-1A	Abnormal ultrasound[a]	30	16	46,XY,der(10)t(10;18)(q25.3;q23)	Paternal 46,XY,t(10;18)(q25.3;q23)	UK	Cystic hygroma, hydrops fetalis. Ref: Chen et al [37]
17-1B	Previous aneuploid child,[a] paternal carrier, the same mother as 17-1A	31	18	46,XX,der(18)t(10;18)(q25.3;q23)	The same as 17-1A	K	Nuchal thickening, single umbilical artery, microcephaly. Ref: Chen et al [38]
18	Abnormal ultrasound[a]	28	17	46,XX,der(8)t(8;13)(p23.3;q22)	*De novo* (m)	UK	HPE, PMA, hexadactyly, hypoplastic left heart. Ref: Chen et al [39]
19	Abnormal ultrasound[a]	31	26	47,XY,+der(22)t(11;22)(q23.3;q11.2)	Maternal 46,XX,t(11;22)(q23.3;q11.2)	UK	IUGR, oligohydramnios, microcephaly, nuchal thickening. Ref: Chen et al [40]
20	Abnormal ultrasound[a]	29	17	46,XX,der(13)t(12;13)(q21.2;p13)	*De novo* (m)	UK	Ventriculomegaly, bilateral hydrothorax. Ref: Chen et al [41]
21-1A	Abnormal ultrasound[a]	33	23	46,XY,der(21)t(12;21)(q24.32;q22.2)	Maternal 46,XX,t(12;21)(q24.32;q22.2)	UK	VSD, micrognathia, rocker-bottom feet. Ref: Chen et al [42]
21-1B	Previous aneuploid child,[a] maternal carrier, AMA, the same mother as 21-1A	34	20	46,XY,der(21)t(12;21)(q24.32;q22.2)	The same as 21-1A	K	Micropenis, ventriculomegaly, right cleft lip and palate, rocker-bottom feet. Ref: Chen et al [43]
22	Abnormal ultrasound[a]	23	30	46,XY,der(4)t(4;10)(p16.1;q25.1)	Paternal 46,XY,t(4;10)(p16.1;q25.1)	UK	IUGR, single umbilical artery, prominent glabella, "lobster claw" deformities of the hands and feet. Ref: Chen et al [44]
23	Abnormal maternal serum screening[a] Abnormal ultrasound	31	18 28[d]	46,XY,der(5)t(5;14)(p13.2;q31.1)	Paternal 46,XY,t(5;14)(p13.2;q31.1)	UK	Polyhydramnios, single umbilical artery, microcephaly, corpus callosum agenesis, cerebellar hypoplasia, megacisterna magna, dilation of the third ventricle, colpocephaly, dilated right atrium, clinodactyly, short limbs. Ref: Chen et al [45]

(*continued on next page*)

Table 1 (*continued*)

Family/case	Indication for amniocentesis	Maternal age (yr)	Gestational age at amniocentesis (wk)	Fetal karyotype	Inheritance	Carrier status	Ultrasound abnormalities and references
24	Previous aneuploid child,[a] paternal carrier, AMA	35	18	46,XX,der(2)t(2;15)(q37.3;q24.3)	Paternal 46,XY,t(2;15)(q37.3;q24.3)	K	No. Ref: Chen et al [46]
25	AMA[a] Abnormal ultrasound	37	16 21[d]	46,XY,der(13)t(10;13)(q25.1;q34)	Paternal 46,XY,t(10;13)(q25.1;q34)	UK	Pyelectasis. Ref: Chen et al [47]
26	Abnormal ultrasound[a]	31	20	46,XY,der(22)t(16;22)(p12.2;q13.31)	Paternal 46,XY,t(16;22)(p12.2;q13.31)	UK	Fetal ascites, ventriculomegaly. Ref: Chen et al [48]
27	AMA	42	18	46,XY,der(13)t(7;13)(p15.3;q33.3)	*De novo* (p)	UK	Microcephaly, DWM, nuchal edema, TGA. Ref: Chen et al [49]
28	Abnormal ultrasound[a]	31	20 22[d]	46,XX,der(1)t(1;20)(p36.23;p12.1)	*De novo* (p)	UK	VSD, ventriculomegaly, midface hypoplasia. Ref: Chen et al [50]
29	AMA	36	14	46,XX,der(11)t(7;11)(q22;p13)	Paternal 46,XY,t(7;11)(q22;p13)	UK	No

[a] Main mode of ascertainment; [b] Translocation carrier status identified because of a balanced translocation fetus conceived by her sister; [c] Translocation carrier status identified because of an aneuploid child conceived by her sister-in-law; [d] Referred to confirmation and repeat amniocentesis.

AMA = advanced maternal age; DWM = Dandy-Walker malformation; HPE = holoprosencephaly; IUGR = intrauterine growth restriction; K = known at amniocentesis; (m) = *de novo* structural reorganization is of maternal origin; (p) = *de novo* structural reorganization is of paternal origin; PMA = premaxillary agenesis; Ref = reference; TGA = transposition of great arteries; UK = unknown at amniocentesis; VSD = ventricular septal defects; wk = week; yr = year.

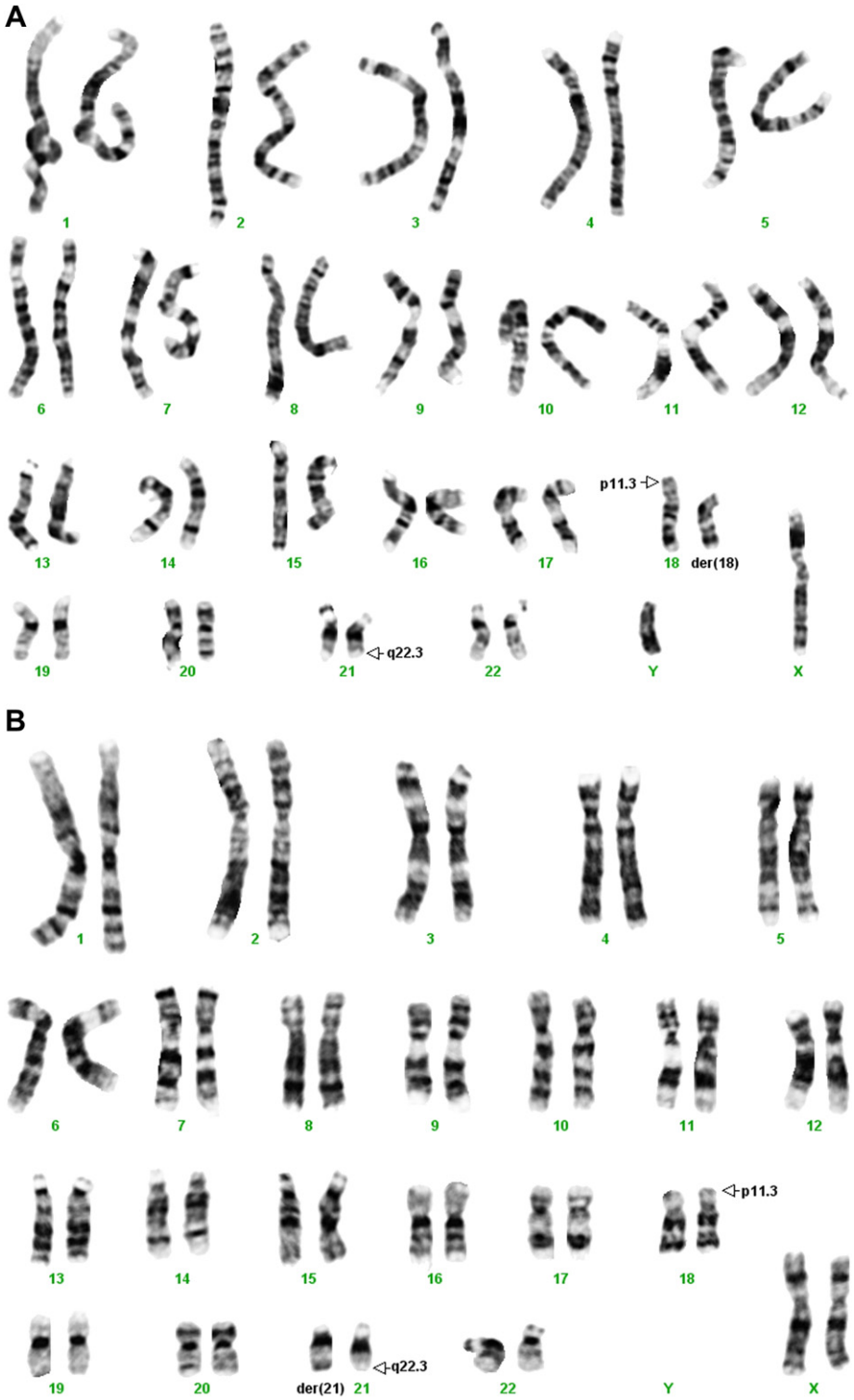

Fig. 1. A family (Family 12) with two different unbalanced reciprocal translocations (Cases 12-1A and 12-1B) of adjacent-1 2:2 segregation. (A) Case 12-1A with a karyotype of 46,XY,der(18)t(18;21)(p11.3;q22.3). (B) Case 12-1B with a karyotype of 46,XX,der(21)t(18;21)(p11.3;q22.3).

with a carrier couple ascertained through two or more miscarriages. This finding is in accordance with the observation reported by Franssen et al [2] that inherited unbalanced structural chromosome abnormalities at prenatal diagnosis are rarely ascertained through recurrent miscarriage. In contrast, inherited balanced reciprocal translocations at amniocentesis are ascertained through recurrent miscarriage as often as through a previous aneuploid child [3]. It has been reported that the carrier couples ascertained through a previous aneuploid child are at a higher risk of unbalanced viable offspring than those ascertained through miscarriages [4–7]. Franssen et al [2] found that the main modes of ascertainment at

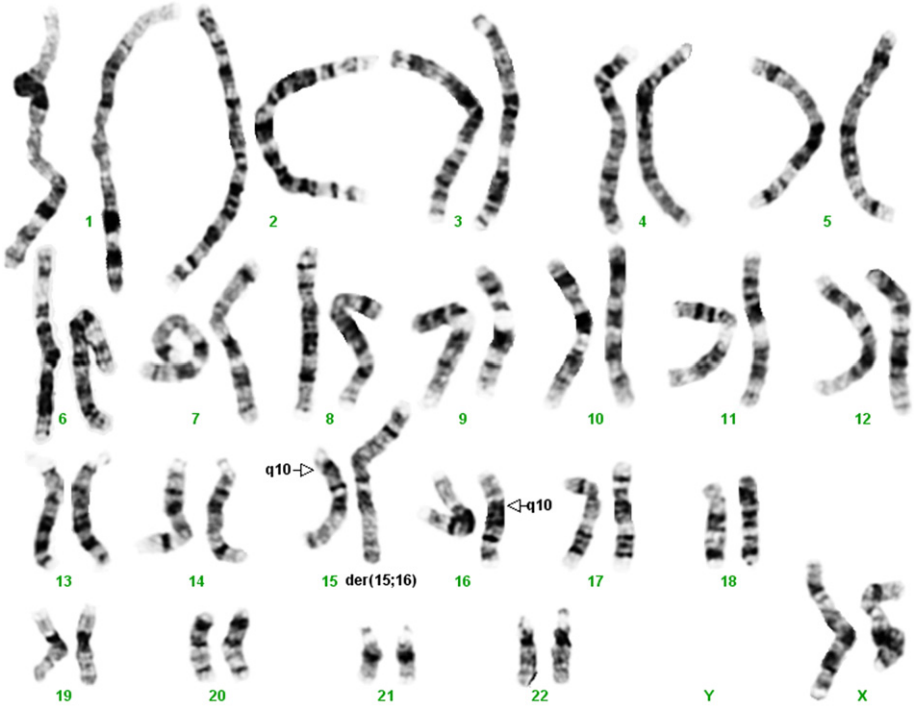

Fig. 2. A case (Case 1) with a whole arm unbalanced reciprocal translocation of adjacent-1 2:2 segregation and a karyotype of 46,XX,der(15;16)(q10;q10),+16.

prenatal diagnosis in cases with inherited unbalanced structural chromosomal abnormalities were a previous child with an unbalanced karyotype (48.2%, 27/56), congenital abnormalities at ultrasound examination (19.6%, 11/56), and advanced maternal age (8.9%, 5/56). Our study shows that the mode of ascertainment through abnormal ultrasound findings is as often as that through a previous aneuploid child in the obstetric history or in the family at amniocentesis in cases with inherited unbalanced reciprocal translocations. This implies that, in addition to the family history of a previous aneuploid child, prenatal ultrasound plays a very important role in the prenatal diagnosis of unbalanced reciprocal translocations. In

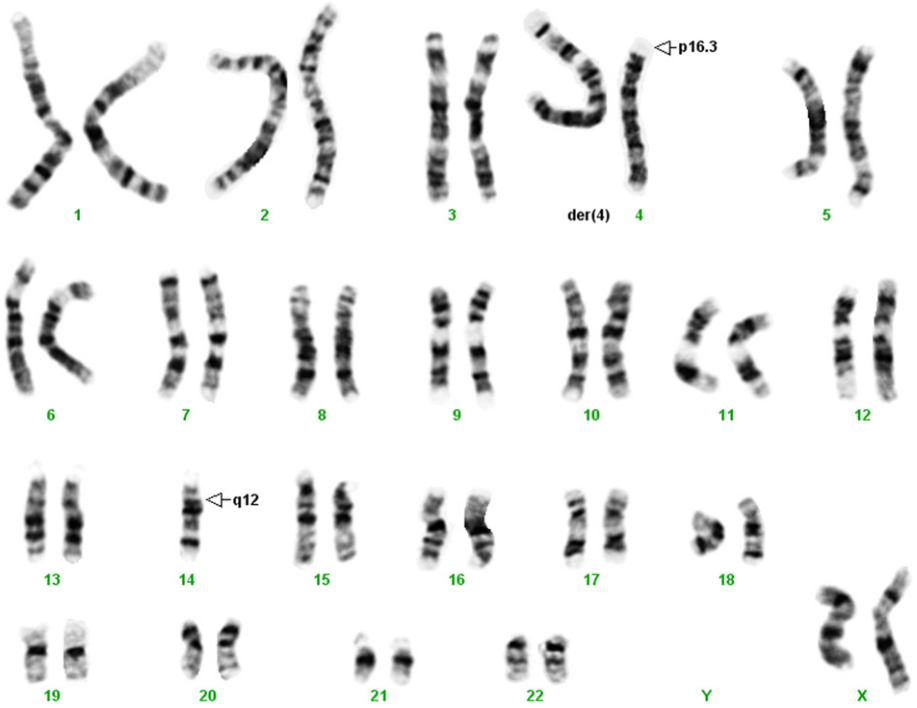

Fig. 3. A case (Case 9) with an unbalanced reciprocal translocation of 3:1 segregation with tertiary monosomy and a karyotype of 45,XX,der(4)t(4;14)(p16.3;q12),-14.

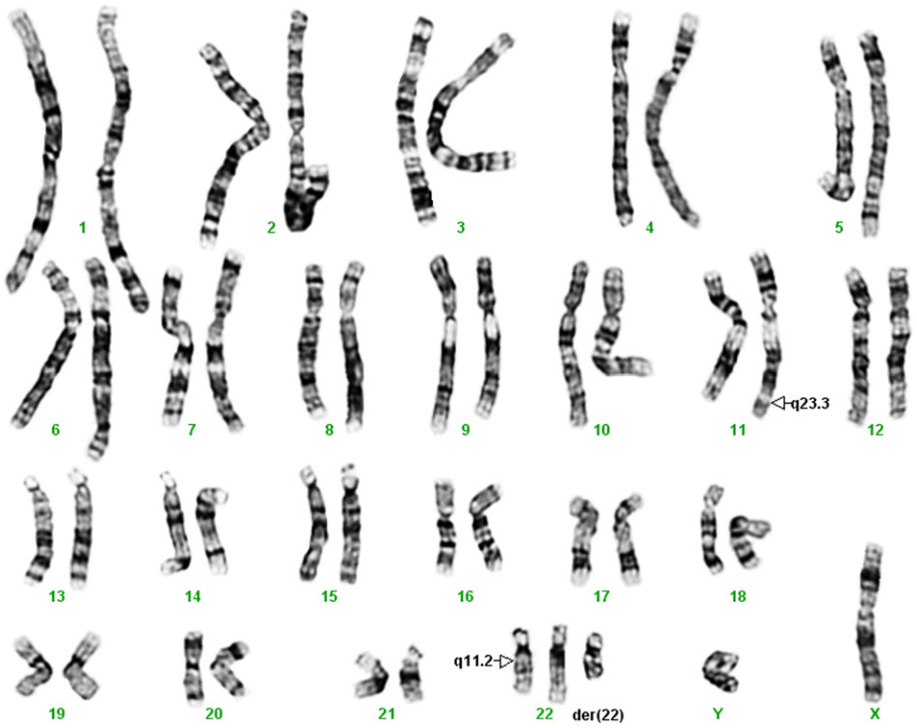

Fig. 4. A case (Case 19) with an unbalanced reciprocal translocation of 3:1 segregation with tertiary trisomy and a karyotype of 47,XY,+der(22)t(11;22)(q23.3;q11.2).

fact, we observed that more than 80% of the fetuses with unbalanced reciprocal translocations were associated with sonographically detectable structural abnormalities.

In this study, 24.1% (7/29) of the families had *de novo* fetal aneuploidy, and of the seven *de novo* cases, six (85.7%) manifested abnormal ultrasound findings. In a study of structural rearrangements detected at prenatal diagnosis, Hume et al [8] found that 37.6% (65/173) of the cases arose *de novo*, and 45% of the *de novo* cases manifested abnormal ultrasound findings. In case of a *de novo* fetal unbalanced reciprocal translocation, characterization of the nature of the aberrant chromosome will require molecular cytogenetic technologies such as spectral karyotyping, fluorescence *in situ* hybridization, and array-based comparative genomic hybridization. Quantitative fluorescent polymerase chain reaction (QF-PCR) using polymorphic DNA markers can additionally determine the parental origin of the aberrant chromosome. The acquired molecular results are very useful in genetic counseling. Structural reorganizations are usually familial (80%), but may arise *de novo* (20%) [9]. In a study of 32 cases with *de novo* structural chromosome rearrangements including deletions, duplications, translocations, and ring chromosomes, Olson and Magenis [10] reported that 84.4% (27/32) of the cases were paternal in origin. However, we did not observe such a preferential paternal origin in the cases with *de novo* unbalanced reciprocal translocations. In our study, among five *de novo* cases with QF-PCR analysis, three were of maternal origin and two were of paternal origin.

Prenatal diagnosis of an unbalanced translocation may incidentally detect a balanced translocation in the family. Chen et al [3] reported that in the 61 families with an inherited reciprocal translocation detected at amniocentesis, 67.2% (41/61) were unaware of their parental carrier status at amniocentesis. Chen et al [11] previously reported that in the 16 families with an inherited acrocentric rearrangement involving chromosomes other than Chromosome 21, 87.5% (14/16) were unaware of their parental carrier status at amniocentesis. Chen et al [12] also reported that in the six families with an inherited heterologous acrocentric rearrangement involving Chromosome 21, 50% (3/6) were unaware of their parental carrier status at amniocentesis. In this study, we found that in the 22 families with an inherited reciprocal translocation, 59.1% (13/22) were unaware of their parental carrier status at amniocentesis.

Balanced reciprocal translocations are the most frequent chromosome rearrangements in humans, occurring in 0.16–0.20% (from 1/625 to 1/500) of live births [1,13,14]. The carriers of a balanced reciprocal translocation are usually phenotypically normal because of a balanced complement of genes. However, because of the segregation modes of 2:2 alternate, 2:2 adjacent-1, 2:2 adjacent-2, 3:1 with tertiary trisomy or monosomy, 3:1 with interchange trisomy or monosomy, and 4:0 with double trisomy or monosomy, a balanced reciprocal translocation carrier can produce 32 different gametes, only two of which would result in a normal complement or a balanced rearrangement by the 2:2 alternate rearrangement [15]. Our study shows that the conceptuses of 2:2 adjacent-1 segregation and 3:1 segregation can be viable at amniocentesis with the former accounting for about 90% and the later accounting for 10% of the fetuses with unbalanced reciprocal translocations

detected by amniocentesis. Our study also shows that in the adjacent-1 2:2 segregating reciprocal translocation, the parental male carriers have the same risk of unbalanced progeny as the female carriers, indicating that there is little effect of adjacent-1 2:2 segregation on the fertility of the male carriers, and that in the 3:1 segregating reciprocal translocation, the parental male carriers have a lower risk of unbalanced progeny than the female carriers, indicating that there is a great effect of 3:1 segregation on the fertility of the male carriers. This observation is in accordance with the results reported by Daniel et al [5].

This study demonstrates that the same unbalanced rearrangement with similar recurrent congenital malformations in the consecutive pregnancies is not unusual in familial unbalanced reciprocal translocations at amniocentesis. For instances, in Family 6, recurrent holoprosencephaly was noted in two sib fetuses with partial monosomy 2q37→qter and partial trisomy 3p21→pter; in Family 8, recurrent omphalocele was noted in two sib fetuses with partial monosomy 11q23→qter and partial trisomy 3q21→qter; in Family 10, recurrent pyelectasis was noted in four fetuses conceived by three women with partial trisomy 10q24.1→qter and partial monosomy 22p11.2→pter; in Family 14, recurrent brain anomaly was noted in two sib fetuses with partial monosomy 11q23→qter and partial trisomy 3p21→pter; and in Family 16, recurrent dolichocephaly was noted in two sib fetuses with partial trisomy 16q12.1→qter and partial monosomy 22q13.3→qter.

In summary, we have presented the results of prenatal diagnostic examinations for unbalanced reciprocal translocations using amniocentesis. Unbalanced reciprocal translocations detected at amniocentesis are frequently associated with abnormal ultrasound findings and prenatal diagnosis of an unbalanced translocation may incidentally detect a balanced translocation in the family. We suggest that prenatal diagnosis of fetal structural abnormalities should alert structural chromosome rearrangements and prompt cytogenetic analysis of the fetus and parents if necessary.

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