

Successful clinical outcome in a rare case of biparental rob(13;14) translocation following NGS-based preimplantation genetic testing for structural rearrangements

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ABSTRACT

Background: Robertsonian translocation (rob) is a structural chromosomal rearrangement resulting from the fusion of the long arms of acrocentric chromosomes. Although carriers are typically phenotypically normal, they may present with reproductive issues such as infertility and recurrent pregnancy loss due to meiotic segregation errors. This case report describes the cytogenetic findings and preimplantation genetic testing outcomes of a consanguineous couple in whom a biparental balanced rob(13;14)(q10;q10) translocation was identified.

Case presentation: A couple referred for genetic evaluation due to recurrent pregnancy loss was included. Peripheral blood karyotyping was performed in both partners, revealing a balanced rob(13;14)(q10;q10) translocation. The male partner also presented with severe oligozoospermia based on standard semen analysis. Following controlled ovarian stimulation, an IVF cycle was performed. Trophoctoderm biopsies from blastocyst-stage embryos were analyzed using next-generation sequencing (NGS)-based preimplantation genetic testing for structural rearrangements (PGT-SR). A total of 11 blastocysts were analyzed. Of these, 4 embryos (36.3%) were classified as normal or balanced, whereas 7 embryos (63.7%) demonstrated chromosomal abnormalities, including whole-chromosome aneuploidies, mosaic trisomies, and segmental imbalances. One normal/balanced embryo was selected for transfer, resulting in a successful clinical pregnancy and the subsequent birth of two healthy children.

Conclusions: Biparental rob(13;14)(q10;q10) carriage may be associated with highly complex embryonic segregation patterns and a high rate of chromosomally unbalanced embryos. NGS-based PGT-SR provides a reliable approach for identifying transferable embryos and guiding reproductive management in such rare cases.

Keywords: embryonic chromosome patterns, PGT-SR, Robertsonian translocation

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INTRODUCTION

Robertsonian translocations (robs) are among the most common structural chromosomal rearrangements observed in humans and are detected in approximately one in every 800–1,000 individuals [1,2]. These translocations result from the fusion of segments near the centromeric regions of acrocentric chromosomes (13, 14, 15, 21, and 22) and the loss of their short arms. They are typically characterized by a balanced 45-chromosome karyotype [2,3]. Since ribosomal DNA repeats are present in the short arms of acrocentric chromosomes, the loss of these regions often does not lead to a distinct clinical phenotype, and translocation carriers can gener-

ally lead a normal life [3]. However, it is known that robs may be associated with reproductive health [4].

In carriers of robs, the processes of pairing and separation of homologous chromosomes during meiosis may be altered. The chromosomes involved in the translocation form trivalent structures during meiosis, leading to different segregation patterns. Alternative segregation may result in the formation of normal or balanced gametes, whereas non-alternative segregation can lead to the formation of unbalanced gametes containing monosomy or trisomy [5,6]. This condition is associated with infertility, recurrent pregnancy loss, and the formation of chromosomally unbalanced embryos [6,7]. Additionally, certain unbalanced chromosomal combinations can cause chromosomal syndromes that may

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result in live births. In particular, robs involving chromosomes 13 and 21 are associated with an increased risk of offspring with Patau syndrome (trisomy 13) and Down syndrome (trisomy 21), respectively [8].

Rob(13;14), the most common type of robs, accounts for approximately 70% of cases in this group [3,9,10]. This type of translocation is reported as the most frequently encountered Robertsonian rearrangement in clinical cytogenetic laboratories and is particularly taken into account in genetic counseling for individuals of reproductive age [9]. Due to the history of pregnancy loss and the potential for chromosomally unbalanced embryo formation in carriers of robs, assisted reproductive techniques and genetic testing methods are frequently considered approaches [7,11].

Advances in assisted reproductive technologies have led to the widespread utilisation of preimplantation genetic testing (PGT) in cases of carriers of robs. PGT-SR enables the assessment of chromosomal imbalances in embryos during the preimplantation period [4,12]. In the contemporary era, NGS-based PGT methods have become instrumental in facilitating the high-resolution analysis of aneuploidies and structural chromosomal alterations in embryos [4,13].

In the current literature, the majority of studies examining embryonic chromosome segregation patterns in rob carriers have focused on couples where only a single partner is affected [4,11]. Consequently, the reproductive challenges and unique embryonic genetic outcomes in rare cases of dual-carrier couples remain largely unmapped in the literature.

The objective of this study was to evaluate the early embryonic chromosomal distribution and meiotic segregation patterns in an extraordinarily rare clinical case where both partners synchronously carried the balanced rob(13;14) translocation. Furthermore, we aimed to present a detailed account of the high-resolution NGS-based PGT-SR processes, the resulting embryonic aneuploidy spectrum, and the clinical management strategy implemented for this structurally homozygous couple.

ETHICS APPROVAL

Approval for the research protocol was received from the Dicle University Faculty of Medicine Non-Interventional Research Ethics Committee (2020/281, date: 26 November 2020). Written informed consent was obtained from both participants prior to their inclusion in the study.

CASE PRESENTATION

Patient history and laboratory workflow

The present study focuses on a case study of a couple, comprising a 24-year-old female and a 26-year-old male, who have sought genetic counselling due to a history of recurrent pregnancy loss. Over the course of their 9-year

marriage, the couple achieved four pregnancies through in vitro fertilization (IVF), all of which resulted in spontaneous first-trimester miscarriages. Detailed medical, reproductive, and multigenerational family histories were obtained to construct a comprehensive pedigree and assess potential hereditary risk factors. Environmental factors, including smoking, alcohol consumption, substance use, and occupational exposure to hazardous chemicals, were systematically evaluated.

Semen samples collected from the male partner following a period of sexual abstinence of at least three days were analysed in accordance with the World Health Organisation's (WHO) 2021 Laboratory Guidelines for the Examination and Processing of Human Sperm [14]. Cytogenetic analysis of both partners was performed using G-banding (GTG). In routine evaluations, at least 20 metaphase spreads were examined; in cases with suspected mosaicism, the number of analyzed cells was increased up to 40. All analyses were carried out using a Zeiss optical microscope and Ikaros software, in accordance with ISCN 2020 nomenclature guidelines and previously described methods [15].

In order to ascertain the molecular basis of recurrent miscarriages in affected female patients, medical professionals elected to perform a comprehensive blood-clotting analysis, encompassing the Factor II (G20210A), Factor V Leiden, and two *MTHFR* gene alterations: C677T and A1298C.

Trophectoderm samples were obtained from blastocysts on days 5 or 6 of development. From each embryo, 5–10 trophectoderm cells were biopsied using a laser, and the entire blastocyst was subsequently vitrified.

Whole genome amplification was performed using the SurePlex™ platform (Rubicon Genomics). NGS libraries were prepared from amplified DNA and sequenced on an Illumina MiSeq platform (Illumina Inc., San Diego, CA, USA). Sequencing data were analysed using BlueFuse Multi/VeriSeq bioinformatics software.

Low-pass whole-genome sequencing generated an average coverage of approximately 0.05x, with a minimum resolution of 5 Mb for the detection of copy number variations (CNVs). Analysis included all autosomes (chromosomes 1–22) and sex chromosomes (X and Y), enabling the detection of whole-chromosome aneuploidies, segmental gains or losses, and segregation patterns associated with robs, as well as chromosomal mosaicism.

Mosaicism classification was performed according to validated laboratory criteria and PGDIS guidelines. Embryos were classified as euploid when the abnormal cell proportion was <20%, mosaic when 20–80%, and aneuploid when >80%. Only embryos reaching the blastocyst stage with sufficient DNA quality were included in the final evaluation.

Laboratory results and IVF outcomes

The detailed family tree and clinical reproductive history of the couple who sought genetic counselling are shown in Figure 1A. Analysis of the family tree revealed consanguinity at two distinct levels within the family structure. Firstly, a consanguineous relationship was identified between the male partner's parents (consanguinity at the parental level). Secondly, it was established that the couple themselves were first-degree cousins and shared a second-degree kinship relationship. Over the course of their nine-year marriage, due to this shared genetic background and the potential influence of chromosomal predispositions, four consecutive first-trimester spontaneous miscarriages occurred following in vitro fertilisation (IVF) cycles.

The macroscopic and microscopic semen characteristics of the male partner are presented in Table 1. Semen analysis demonstrated a sperm concentration of $0.05 \times 10^6/\text{mL}$ (reference range $12\text{--}16 \times 10^6/\text{mL}$), with a total motility of 33% (reference range 38%–42%) and a progressive motility of 23% (reference range 31%–34%). According to the reference values mentioned above, which were established by the 2021 WHO guidelines, these findings were consistent with severe oligozoospermia.

The molecular thrombophilia panel results evaluated to investigate the etiology of RPL in the female partner included the detection of four common RPL-associated mutations. The molecular analysis demonstrated that the patient was homozygous mutant (TT) for the *MTHFR* (A1298C) polymorphism. Conversely, no pathogenic variants or polymorphisms were detected in the Factor II (G20210A), Factor V Leiden, or *MTHFR* (C677T) genes, all of which exhibited a homozygous normal genotype.

Cytogenetic analysis of metaphase preparations obtained from peripheral blood lymphocytes revealed a balanced *robs* in both partners. The female partner's karyotype was determined to be $45,XX,rob(13;14)(q10;q10)$ (Figure 1B), while the male partner's karyotype was found to be $45,XY,rob(13;14)(q10;q10)$ (Figure 1C).

Given the presence of biparental balanced Robertsonian translocation carriers and severe male-factor infertility, the couple underwent an IVF cycle following controlled ovarian stimulation. A total of 23 oocytes were retrieved during the oocyte pick-up (OPU) procedure, resulting in the development of 15 embryos, numbered 1–15. Of these, 11 embryos reached the blastocyst stage and met the criteria for trophectoderm biopsy based on routine embryological assessment. These 11 embryos were subsequently included in the high-resolution NGS-based PGT-SR analysis.

NGS analysis showed that 4 of the 11 embryos (36.3%) had a normal or balanced chromosomal complement, while the remaining 7 embryos (63.7%) exhibited different types of chromosomal imbalance, including aneuploidy and mosaicism, consistent with segregation patterns observed in *robs* carriers.

The detected abnormalities included trisomy 7 and 14, monosomy 5, mosaic trisomy involving chromosomes 9 and 21, combined trisomies affecting chromosomes 5, 7, 15, 16 and X, and monosomic and trisomic segregation products of chromosomes 13 and 14. In embryo 10, a 30% mosaic segmental duplication [*dup*(13q)] involving the long arm of chromosome 13 was identified.

Table 1 summarizes the genetic findings, including mosaicism estimates for each embryo and mitochondrial DNA copy number ratios derived from bioinformatic analysis. The heatmap in Figure 2 illustrates genome-wide copy number variations, including gains, losses, and mixed alterations across all chromosomes. Based on the PGT-SR results, one normal/ balanced embryo was selected for transfer. The transfer resulted in a clinical pregnancy, and the couple subsequently had two healthy children.

DISCUSSION

In the present study, the *rob*(13;14) was detected in both male and female partners. This finding is rarely reported in the literature. Most studies on *robs* have reported that the translocation is present in only one partner [16,17,18]. When both partners carry the same translocation, the process of chromosomal segregation during meiosis becomes more complex, and the number of possible chromosomal combinations increases. This situation increases the likelihood of unbalanced gamete formation, which may result in implantation failure or early pregnancy loss [10,16]. For this reason, this case may serve as an illustrative example contributing to the understanding of embryonic chromosome distribution.

From an epidemiological and population genetics perspective, the simultaneous presence of the same *rob*(13;14)(q10;q10) structural rearrangement in both parents is a very rare occurrence. Although *robs* can occur sporadically in the general population, cases where both parents carry the same translocation are mostly reported in geographically isolated communities or families with a high degree of consanguinity. In this case, the detection of the same translocation in both spouses suggests that it may be related to the second-degree consanguinity between the couple. Pedigree analysis points to the presence of a balanced translocation that may have arisen in a common ancestor and could have been transmitted across different family lines over generations, eventually recombining in this couple. This scenario may be consistent with a possible founder effect. It is thought that such structural similarities, based on a common ancestor, may increase complexity in meiotic segregation processes and, as a result, may pave the way for the formation of nullisomic, disomic and mosaic chromosomal configurations during embryonic development. However, the clini-



Table 1. Chromosomal abnormalities detected in the 11 embryos following NGS-based PGT-SR analysis

Embryo numbers correspond to the original embryo numbering (Embryos 1–15), while only the 11 embryos selected for trophectoderm biopsy and genetic analysis are presented. The mosaicism rate represents the estimated proportion of cells with chromosomal abnormalities within each embryo. Abbreviations: NGS- next generation sequencing, PGT-SR- preimplantation genetic testing for structural rearrangements.

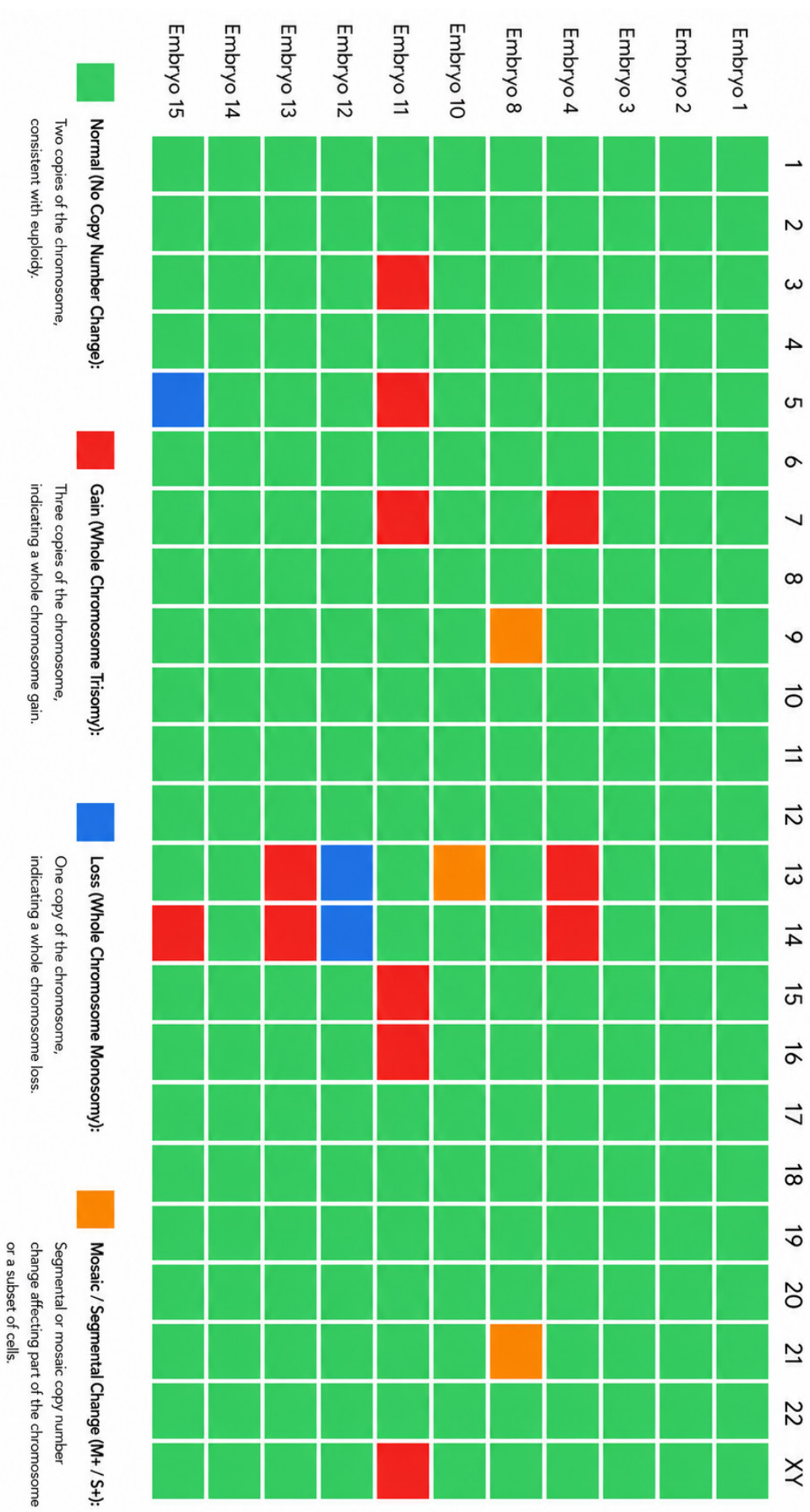


Figure 2. Heatmap representation of chromosomal copy-number profiles in the 11 embryos analyzed by NGS-based PGT-SR

Embryo numbers correspond to the original embryo numbering (Embryos 1–15), while only the 11 embryos selected for trophectoderm biopsy and genetic analysis are displayed. Four embryos (Embryos 1, 2, 3, and 14) exhibited a normal or balanced chromosomal complement, whereas the remaining seven embryos showed whole-chromosome gains, losses, mosaic abnormalities, or segmental copy-number alterations.

cal impact of such chromosomal arrangements may vary between cases, and their relationship with implantation outcomes and early pregnancy loss remains variable. In this context, preimplantation genetic testing provides an important opportunity to evaluate the embryonic consequences of these complex meiotic segregation patterns and to characterize the chromosomal constitution of embryos generated from such unions.

NGS-based PGT-SR analysis of the embryos identified both normal or balanced chromosomal complements and a range of chromosomal abnormalities, including mosaic trisomy as well as segmental chromosomal gains and losses. Similar findings have been reported in previous PGT-SR studies involving translocation carriers.

As also reported by Munné et al., a considerable proportion of embryos from translocation carrier couples present chromosomal imbalances [19]. These observations support the known association between structural chromosomal rearrangements and altered embryonic chromosomal segregation patterns.

The presence of the translocation in both maternal and paternal gametes has been demonstrated to contribute to elevated levels of unbalanced chromosomal profiles at the embryonic level.

From a molecular perspective, robs are thought to arise due to the susceptibility of ribosomal DNA regions and repeat sequences in the short arms of chromosomes 13 and 14 to erroneous recombination. Misalignment and recombination events in these regions can facilitate translocation formation and lead to chromosomal segregation errors during gametogenesis [10,20]. The mosaic trisomies and partial chromosomal gains or losses observed in our study support the complexity of meiotic segregation in translocation carriers.

Another mechanism discussed in the literature is the interchromosomal effect (ICE), which refers to the influence of a translocation on the segregation of chromosomes not directly involved, thereby increasing the risk of aneuploidy in other chromosomes [21,22]. Machev et al. reported increased autosomal and gonosomal disomy rates in sperm from male robs carriers using FISH analysis [23]. In the present case, aneuploidy and mosaicism were also observed in chromosomes not directly involved in the translocation. However, given the descriptive nature of this single case report, no conclusion regarding the presence of an interchromosomal effect can be drawn.

Recent studies have shown that embryonic chromosome segregation in carriers of robs may not always be fully explained by classical meiotic models. In a comprehensive analysis conducted by Benn and colleagues, it was reported that more complex chromosomal patterns, beyond the classical alternative and adjacent seg-

regation models, may emerge in embryos derived from translocation carriers [24]. In this study, various chromosomal distributions were identified, including combinations where trisomy and monosomy co-occurred, higher-than-expected monosomy rates, and rare 3:0 segregation patterns. Researchers have suggested that this situation may be associated with mechanisms such as early chromatid separation, anaphase delay, reverse segregation, or early mitotic errors [24]. The mosaic chromosomal structures observed in our study and the aneuploidies in chromosomes not directly involved in the translocation suggest that such complex segregation mechanisms may play a role in the early stages of embryonic development.

Benn et al. suggested that the sex of the parent carrying the translocation may play a role in determining chromosomal distribution in embryos [24]. They proposed that inherent biological differences between male and female gametogenesis could influence the likelihood of producing normal/balanced or unbalanced chromosomal combinations. This observation is supported by previous studies showing that recombination patterns and chromosome segregation mechanisms differ between spermatogenesis and oogenesis.

PGT-SR studies have shown that problems with proper chromosome segregation frequently occur in embryos obtained from couples who are carriers of robs. Alfarawati et al. found that many embryos from translocation carrier couples carried chromosomal numerical or structural abnormalities, emphasizing the importance of comprehensive genetic screening in improving embryo selection and reproductive outcomes [25]. In a more extensive study conducted by Tan et al., it was reported that the proportion of blastocysts capable of genetic transfer was slightly higher in male surrogates than in female surrogates (31.7% versus 34%) [26]. This finding suggests the potential for biological differences in the formation processes of sperm and egg cells to influence chromosome distribution in embryos.

In their study, Kaddouri-Kaddouri et al. reported that there may be marked and statistically significant impairments in sperm parameters in men who are carriers of rob. In contrast, it has been reported that the response to controlled ovarian stimulation may be largely preserved in female RT carriers [27]. These findings appear to be generally consistent with the observations obtained in the present study and suggest that cytogenetic analysis and karyotype examination may be an important component in the assessment of the aetiology of infertility, particularly in male carriers.

In the literature, it has been reported that the rates of unbalanced chromosomal distribution in embryos from translocation carrier couples are higher (above 50%) in

cleavage-stage biopsies and lower in blastocyst biopsies; this suggests that chromosomal imbalances may decrease in the later stages of embryo development [28]. The embryo distribution obtained in our study is consistent with these ranges and supports the clinical value of PGT-SR in couples carrying translocations. Furthermore, in studies employing comprehensive chromosome screening, high rates of chromosomal abnormalities have been reported in embryos from couples carrying translocations, and it has been emphasized that the selection of suitable embryos via PGT-SR may improve clinical pregnancy outcomes [29,30]. In the present case, transfer of a normal/balanced embryo resulted in a clinical pregnancy, and the couple subsequently had two healthy children, further supporting the potential clinical utility of PGT-SR in the management of rare biparental rob(13;14) carriers.

CONCLUSIONS

This report describes the cytogenetic and preimplantation genetic testing findings of a rare case in which both partners carried a balanced rob(13;14)(q10;q10) translocation. NGS-based PGT-SR enabled the characterization of chromosomal abnormalities and the identification of normal/balanced embryos for transfer. The findings from this case may contribute to the limited literature on biparental rob carriers and provide additional information regarding embryo chromosomal segregation patterns in such couples.

ABBREVIATIONS

DNA – deoxyribonucleic acid

FISH – fluorescence in situ hybridization

ICE – interchromosomal effect

IVF – in vitro fertilisation

NGS – next-generation sequencing

PGT – preimplantation genetic testing

PGT-SR – preimplantation genetic testing for structural rearrangements

Rob(s) – Robertsonian translocation(s)

WHO – World Health Organization

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AUTHORS' CONTRIBUTION

İY – writing (original draft), analysis, data management

EA – writing (original draft), analysis, data management

GGE – Writing (original draft), analysis, data management

MB – writing (review & editing), data visualization

DO – writing (review & editing), cytogenetic analysis

SiK – writing (original draft), writing (review & editing)

ST – writing (original draft), writing (review & editing)

MB – investigation, formal analysis

Mİ – investigation, formal analysis

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All authors have read and approved the final version of the manuscript.

CONFLICT OF INTEREST

None to declare.

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