A NOVEL CHROMOSOMAL TRANSLOCATION IN A FEMALE WITH INFERTILITY: A CASE STUDY

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PURPOSE

Balanced reciprocal translocations (BRTs) are common chromosomal structural rearrangements formed by random de novo breakage and rejoining of two or more chromosomes. Reciprocal translocation carriers generally do not display any apparent phenotypic disease, however, approximately 1% of individuals who carry a BRT are subfertile.

The present study describes a 35-year-old female with a novel BRTs, never described before in the literature:

METHODS

A couple with one year history of primary infertility was referred to the laboratory of Genetics and Cytogenetics of Synlab-SDN (Pagani, Sa) for molecular diagnostics and chromosome analysis.

No infertility risk factors emerged from the clinical history and no drug or radiation exposure was documented. They were phenotypically normal subjects and no family history was reported. Cytogenetic analysis was carried out based on phytohaemaglutinin-stimulated peripheral blood lymphocyte cultures of the couple. Lymphocyte culturing and GTG-banding were performed following internal protocols. Karyotypes were described according to the International System for Cytogenetic Nomenclature (ISCN 2020).

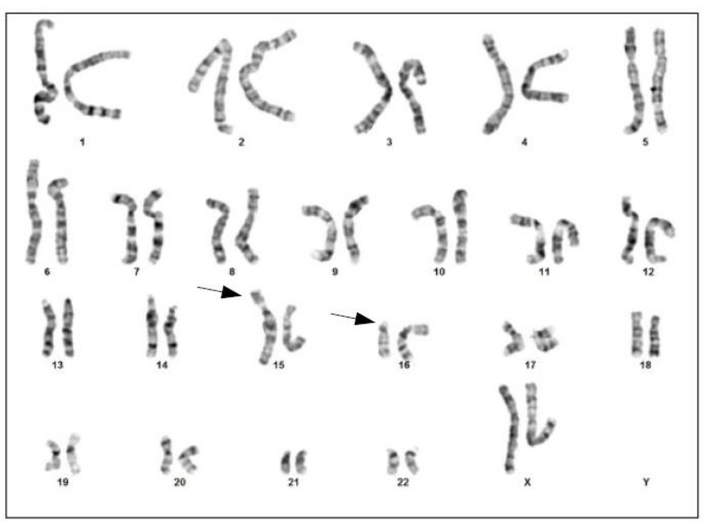

Fig.1: Patient's Karyotype: 46,XX,t(15;16)(p10,p10)

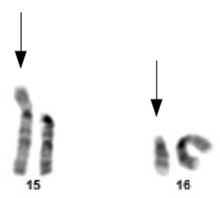

Fig. 2: Reciprocal whole-arm translocation in wich the short arms of chromosomes 15 and 16 and the long arms of these chromosomes, respectively, have been fused at the centromeres.

RESULTS

Chromosomal analysis of the couple revealed a normal 46,XY karyotype in the male, and a BRT hitherto unknown in the female partner.

Chromosomal analysis of the female revealed a novel balanced translocation between chromosomes 15 and 16 (Fig.1) indicating the karyotype to be 46,XX,t(15;16)(p10,p10). A reciprocal whole-arm translocation in which the short arm of chromosome 15 has been fused at the centromere with the long arm of chromosome 16 and consequently the p-arm of the chromosome 16 was fused to chromosome 15 (Fig,2).

CONCLUSION

To the best of our knowledge, this is the first time it is detailed the cytogenetic characterization of a patient with a karyotype 46,XX,t(15;16)(p10,p10). Genetic counselling and extension of the analysis to consanguineous was recommended.

Cytogenetic analysis is essential for all the couples with reproductive failure. The carriers of such abnormalities should be informed about birth defects risk due to de novo submicroscopic rearrangements. Adequate genetic counselling to inform them about the possibility of prenatal or preimplantation diagnosis will help the couples to make an informed reproductive decision regarding subsequent pregnancies.

References

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