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Research Article

Chromosomal Translocations and Couple Infertility in Cotonou

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Abstract

Background: Fertility disorders have been a significant issue affecting populations for decades. According to the World Health Organization (WHO), this issue affects approximately 17.5% of the adult population roughly one in six people worldwide. Method: This was a descriptive cross-sectional study conducted over a seven-year period at the Cotonou cytogenetics laboratory. The study aimed to highlight the impact of chromosomal translocations on couple fertility. Participants included patients found to have a chromosomal translocation via karyotyping during an infertility workup. Patients whose karyotypes revealed abnormalities other than chromosomal translocations were excluded from the study. Conventional cytogenetic techniques were employed; specifically, G-banded metaphase karyotyping was performed using lymphocyte cultures incubated at 37 ℃ for 72 hours. Results: A total of 36 cases of chromosomal translocations were identified. The genotypes involved 31 cases of Robertsonian translocations (88.9%) and 5 cases of reciprocal translocations (11.1%). Balanced Robertsonian translocations were found in 22 female patients during investigations into reproductive disorders. One case of a balanced homologous Robertsonian translocation was recorded in a patient with a history of recurrent miscarriages. Discussion: Chromosomal translocations whether balanced or unbalanced significantly impact couple fertility due to associated gamete and embryonic development abnormalities. Given this, would it not be beneficial to include karyotyping in prenuptial health screenings?

Keywords

Karyotype, Chromosome, Couple Infertility, Chromosomal Translocations, Chromosomal Abnormality, Cytogenetic Analysis

1. Introduction

Fertility disorders represent a significant issue affecting populations today. According to the WHO, approximately 48 million couples and 186 million individuals worldwide are affected by infertility, making it a public health concern. Infertility is a condition of the male or female reproductive system defined by the inability to achieve pregnancy after 12 months

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or more of regular, unprotected sexual intercourse [1]. Its etiologies are diverse, ranging from infectious and environmental causes to idiopathic ones. Genetic factors play a major role among these idiopathic causes and are the focus of this study. In most West African countries, these genetic causes are often unrecognized due to a lack of adequate technical facilities for their investigation. Chromosomal translocations are a particularly important and often overlooked category of these genetic causes. They are characterized by an exchange of genetic material between two non-homologous chromosomes. Historically, the first case of translocation observed in a patient with Down syndrome was identified in 1960; it involved chromosomes 15 and 21 [1]. There are two categories of chromosomal translocations: reciprocal translocations, which result in the formation of two derivative chromosomes but generally preserve the total amount of genetic information.

Robertsonian translocations, which lead to a loss of genetic material. This type of translocation occurs between two acrocentric chromosomes. Diagnosis relies on cytogenetic analyses specifically karyotyping as well as molecular cytogenetic techniques such as fluorescence in situ hybridization (FISH) and high-resolution microarrays [2]. As part of the investigation into fertility disorders, we performed karyotypes to study the role of chromosomal abnormalities in these conditions.

2. Material and Methods

This was a retrospective, descriptive study based on karyotype results collected over a seven-year period from May 2016 to April 2024 at the Cotonou cytogenetics laboratory cytogenetics laboratory in Cotonou.

The study population consisted of patients (male and female) referred to the Cotonou cytogenetics laboratory for karyotyping during the study period.

Patients with a chromosomal translocation identified via karyotype as part of an infertility workup were included in the study.

Patients whose karyotypes revealed abnormalities other than chromosomal translocations were excluded.

Data were gathered using individual data collection forms based on the karyotype results of patients at the Cotonou cytogenetics laboratory (Faculty of Health Sciences, Cotonou). Data analysis was performed using Excel and Epi Info (version 7.2.6.0) software.

Investigation Method

G-banded metaphase karyotyping is performed using lymphocyte cultures incubated at 37 ℃ for 72 hours. Cells are arrested in metaphase using colchicine. Following hypotonic shock with KCl, mitotic cells are fixed using a methanol/acetic acid mixture. The resulting chromosome preparations are heat-denatured (RHG banding) and Giemsa-stained. Eleven metaphases are photographed and then classified based on chromosome size, centromeric index, and banding pattern in a semi-automated manner using specialized software coupled with a camera [1].

3. Results

A total of 36 cases of chromosomal translocations were identified in the study. The genotypes involved Robertsonian translocations in 31 cases (88.9%) and reciprocal translocations in 5 cases (11.1%). The Robertsonian translocations were found in 13 adults (mean age: 35 years) and 18 infants and children.

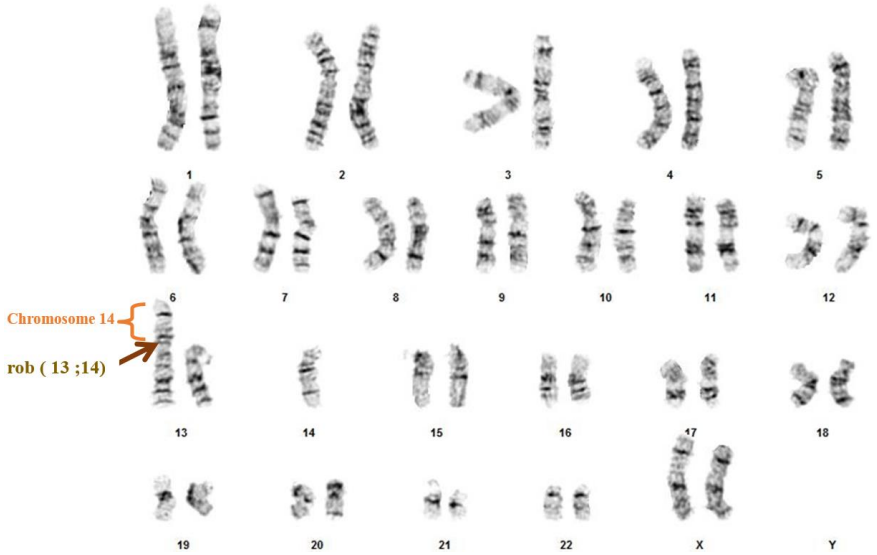

Figure 1. *Female chromosomal complement showing a structural anomaly: 45,XX,der(13;14)(p13;p13). Cytogenetics Laboratory, Faculty of Health Sciences, Cotonou, Benin.*

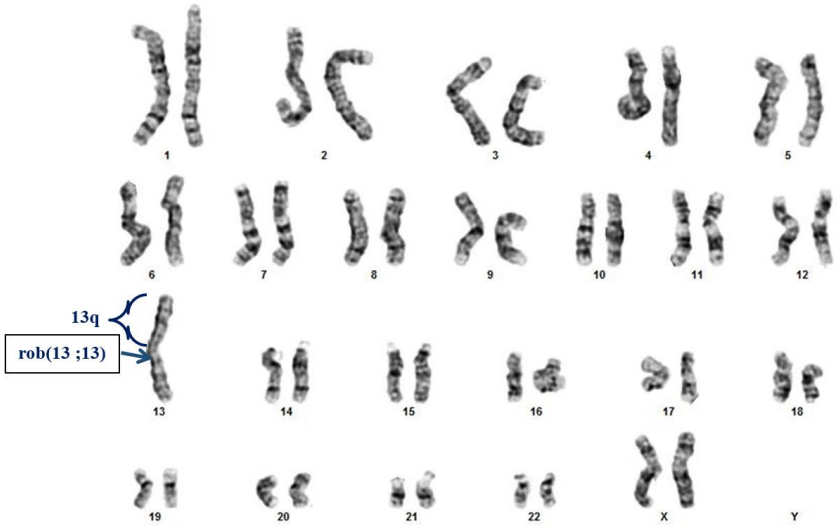

Figure 2. *Female chromosomal complement showing a structural anomaly: 45, XX, der (13;13) (q11.2; q11.2). Cytogenetics Laboratory, Faculty of Health Sciences, Cotonou, Benin.*

For reciprocal translocations, the mean age was 38 years.

The Robertsonian translocations were balanced in 22 female patients; this is illustrated by the karyotype of one patient 45, XX,der (13;14) identified during the investigation of a reproductive disorder Figure 1. In addition, nine cases of homologous Robertsonian translocation were identified, comprising one case of balanced homologous Robertsonian translocation and eight cases of unbalanced homologous Robertsonian translocation.

The case of balanced homologous Robertsonian translocation involved the fusion of the long arms of both chromosome 13s at band q11.2. This case was identified in a 35-year-old female patient with a history of recurrent miscarriages. This chromosomal anomaly, shown in Figure 2, is associated with a 45, XX, der(13;13)(q11.2;q11.2) karyotype. The cases of unbalanced homologous Robertsonian translocations identified in this study involved chromosome 21. These translocations led to the formation of isochromosomes 21, resulting in trisomy 21 in eight children.

4. Discussion

Karyotyping was ordered for adults in 25% of cases. This test is typically requested to investigate the underlying cause of fertility issues or recurrent miscarriages. Its primary purpose is to detect balanced chromosomal rearrangements, particularly balanced translocations. It is generally ordered as part of a fertility workup or following recurrent miscarriages typically after two or three pregnancy losses [3].

Two forms of balanced Robertsonian translocations der(13;14) and der(13;13) resulting from the fusion of two acrocentric chromosomes were discovered in women during the investigation of reproductive disorders. The derivative chromosome (denoted "der") is composed of the long arms of the original chromosomes, with the short arms being lost [4]. Karyotyping was performed on these patients due to reproductive difficulties, specifically six recurrent miscarriages in one case and five pregnancies that ceased at eight weeks of gestation in the other. Carriers of reciprocal translocations generally exhibit a normal phenotype, unless the translocation breakpoint causes a disruption of a gene [5]. However, in most cases, these individuals face a high risk of producing unbalanced gametes; this explains both fertility issues such as infertility and recurrent miscarriages and congenital malformations in pregnancies that reach term [6, 7]. Additionally, two cases of reciprocal chromosomal translocations were identified in male patients, involving chromosomes 2 and 5, and chromosomes 2 and 15 [t(2;5) and t(2;15)], respectively.

These patients sought medical attention for reproductive difficulties linked to altered sperm morphology, specifically teratozoospermia. According to the literature, carriers of a balanced Robertsonian translocation generally do not display clinical features but are at risk for infertility, recurrent pregnancy loss, and the birth of children with congenital anomalies, as well as intellectual and developmental disabilities [8].

Indeed, the present study revealed eight cases of unbalanced homologous Robertsonian translocations in children. These children had trisomy 21 due to an unbalanced Robertsonian translocation.

Trisomy 21 resulting from a translocation between chromosomes 14 and 21 [9]. Most cases of trisomy 21 caused by a translocation between chromosomes 14 and 21 designated 46,der(14;21)+21 result from the inheritance of a balanced Robertsonian translocation, 45,der(14;21), from one parent [10]. Typically, the carrier parent has a normal phenotype but faces an increased risk of passing on an unbalanced chromosomal combination to their offspring due to the formation of trivalents during meiosis [11].

Approximately 30% to 35% of the gametes produced by this parent are abnormal. Fertilization of an oocyte or sperm

carrying the der(14;21) by a normal gamete results in trisomy 21 in the child [11]. Hence the need to offer genetic counseling to affected families to assess recurrence risks and inform parents about prenatal diagnostic options.

5. Conclusion

Chromosomal translocations, whether balanced or unbalanced, significantly impact a couple's fertility. They are responsible not only for conception difficulties and recurrent miscarriages linked to gamete and embryonic development abnormalities but also for the occurrence of malformations and even, at times, malignancies. Given this, would it not be beneficial to include karyotyping in pre-marital screening?

6. Recommendations

Include karyotyping in premarital screening.

Abbreviations

FISH	Fluorescence in Situ Hybridization
WHO	World Health Organization
KCl	Potassium Chloride
der	Derivative Chromosome
rob	Robertsonian Translocation

Author Contributions

Tuo-Benie Wako-Tianwa Alice: Writing – original draft
Goulai Bi You Bazago Etienne: Formal Analysis
Diessongo Nodouma Safiatou: Data curation
Coulibaly Quidana Desiree: Conceptualization
Azonbakin Simon: Supervision, Validation
Yao Gnangoran Victor: Supervision
Laleye Anatole: Supervision

Conflicts of Interest

The authors declare no conflict of interest.

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