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Case report

47, XYY Syndrome and its Association with Male Infertility: Case Report

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Abstract

47, XYY syndrome is one of the most common sex chromosomal anomaly found in humans after Klinfelter syndrome (47, XXY). It is frequently associated with infertility in males. This syndrome has an extra Y chromosome (XYY) due to non-disjunction of the Y chromosome in paternal meiotic II. The presence of an extra Y chromosome causes hormonal disbalance in the gonads that responsible for abnormal function of human chorionic gonadotropin. In our case of infertile men with severe oligozoospermia that also confirm by conventional cytogenetic analysis of the peripheral blood lymphocytes revealed the constitutional karyotype of 47, XYY. This report is likely to be helpful for counselling and early management of such infertile males.

Introduction

47, XYY syndrome is the most common sex chromosomal anomaly observed in men, which is frequently associated with infertility. In general population 47, XYY syndrome occurring in approximately 1 out of 1,000 live male births, but more frequently found in infertile males [1]. Generally there are no phenotypic abnormalities in 47, XYY syndrome, but many reports showed that the XYY boys are more risky for behavioural problems, mild learning disability, delayed speech and language development and tall stature [2]. Men with 47, XYY syndrome can have endocrine dysfunction, variable sperm count ranging from normal to oligozoospermia [3]. Many studies showed that most persons with 47, XYY have some problems related to behavioural, learning disability, delayed speech or language development [4]. There are various extents of spermatogenic failures, but males are usually sterile. In 47, XYY syndrome males cause hormonal disbalance in the gonadal environment which affects the normal function of human chorionic gonadotropin [5]. During the present investigation, we have reported one case of infertility among men with 47, XYY karyotype.

Case presentation

The case was a 35 year old married male came to hospital with complaint of infertility. There was no family history with similar complaints. Physical examination revealed that the person was phonotypicaly a normal male with a height of 168 cm and a weight of 64.5 kg. Hormonal levels demonstrated that FSH and LH were increased to 29.7 and 18.3 mlU/ml, respectively and a very low Testosterone level (1.2 ng/ml). Seminal analysis result showed that he is having severe oligozoospermia (sperm concentrations less than 1 million sperm/ml).

Discussion

There is no need to emphasize that infertility in human reproduction period continues to be a major cause of concern. Sex chromosomal anomalies related to infertility in men were studied in the present investigation. The present study was undertaken to identify the genetic cause responsible for infertility in men. Cytogenetic analysis with GTC banding techniques of infertile men revealed sex chromosomal anomalies. In this case had increased FSH and LH levels. The testosterone level was low. Abnormal levels of hormone may be associated with infertility in males. Many studies showed Luteinizing hormone, Follicular stimulating hormone is performed to assess the reproductive endocrinological axis and thus effective spermatogenesis [5]. Other studies have also shown the correlation between FSH, LH and Testosterone with infertility in men [6]. FSH levels are mainly associated with the number of spermatogonia when these cells are absent, FSH values are usually increased. Inclusion of extra sex chromosome (47, XYY) was observed in the present study. It is in conformity with an earlier report by Faeza et al. and Ratcliffe et al. [6,7].

The abnormalities in present case may due to the presence of an extra sex chromosome, CFTR gene mutation and Y chromosome microdeletion. A few cases of Y chromosome structural rearrangements involved failure of pairing the X and Y chromosomes. These include a dicentric Y chromosome and pericentric inversion of the Y chromosome [8,9]. A gain of extra sex chromosomes resulting in numerical changes may be due to non disjunction of chromosome during gametogenesis. Due to non-disjunction there is a possibility that a particular pair of chromosomes is transmitted to a gamete or it may be lost. There can be error in the gametogenesis due to the accumulation of mutations which might have taken place during the life span of an individual. There is a need to identify the specific loci on the chromosome involved in different types of chromosomal anomalies. Spectral karyotyping, NOR banding and molecular techniques should be undertaken to identify infertility in males. In a nutshell, cytogenetic studies, particularly on the sex chromosome with special reference to Y chromosome in males are required to be explored in Indian population group where the problem of infertility is often encountered. Such studied are more important for management and genetic counselling. The cytogenetic analysis is recommended for all infertile subjects, which will be very useful for genetic counselling.

Conclusions Men with 47,XYY syndrome have a diverse spectrum of clinical presentation. Because of the heterogeneous phenotype and potential lack of symptoms, diagnosis may be difficult, especially if fertility is not compromised. However, in our patient and in our review of the literature, it appears that many men with 47,XYY syndrome will likely have decreased fertility potential. These patients may ultimately require assisted reproductive techniques in order to achieve pregnancy. Genetic evaluation is recommended prior to proceeding. We recommend careful screening of these patients and referral to primary physicians for long-term follow-up given the increased incidence of associated comorbidities

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