Cytogenetic and Statistical Study on Klinefilter’S Syndrome in Baghdad

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Abstract
This study was carried out to determine the percentage and chromosomal karyotyping in patients with klinfelter’s syndrome in Baghdad. One hundred cases collected from Infertility and I.F.V Center in Baghdad & Yarmok Hospital during the period (2006-2009) to determine the frequency of Klinefilter’s syndrome. In I.V.F Center hundred infertile male studied Klinefilter form 1.5% percentage of infertile men and statistical study in Yarmok hospital illustrated Klinefilter form 5.2% of patients admitted in situation for mental deficiencies .The statistical study conclusion that Klinefilter’s syndrome varies according to the cases kind Chromosomal analysis & seminal examine for cases with Klinfelter’s syndrome according to the clinical feature and symptoms which related to Klinefilter, small testes (<10mm.), breast enlargement, reduced facial and body hair were studied karyotype. The Chromosomal study illustrated first karyotype of three cases abnormal number for sex chromosome (two X chromosome) 47, XXY and one case with mosaic karyotype 46, XY / 47, XXY . General seminal examination illustrated aizoospermia for all cases .The Cytogenetic study conclusion that percentages of cases with only abnormal metaphase 75% while the case with mosaic metaphase 25%.

Introduction
Klinefilter syndrome is disorder that affects only in males. Klinefilter syndrome is much rarer, occurring in 1: 50,000 or fewer male birth. Males with this condition typically have small testes (<10) that do not produce enough testosterone, which is the hormone that directs male sexual development before birth and during puberty [1, 2]. A shortage of testosterone during puberty can lead to breast enlargement (gynecomstia), reduced facial and body hair, children and adults with Klinefilter syndrome tend to be taller than other males in their age. Compared with other men, adult males with Klinefilter syndrome have an increased risk of developing breast cancer and a chronic inflammatory disease called systemic lupus erythematosus. Their chance of developing these disorders is similar to that of normal adult females. Boys with Klinefilter syndrome may have learning disabilities and difficulty with speech and language development. They tend to be quiet, sensitive, and personality characteristics vary among males with this condition [3, 4, 5, 6]. Klinefilter’s syndrome is characterized by an abnormal number of X chromosomes. Klinefilter is not an inherited disease, but occurs at random. It’s usually the result of an error that occurs during cell division during the formation of egg or sperm cells division called nondisjunction Normal males have one X chromosome and one of Y chromosome (46, XY), but in Klinefilter the male has an extra copy of the X chromosome 47, XXY figure 1 [1, 2, 3, 5]. Mosaic is the male has two type of cell normal and abnormal karyotype 46, XY/47, XXY & also not inherits as a random event during cell division early in fetal development. As a result, some of the body's cells have one X chromosome and one Y chromosome (46, XY), and other cells have an extra copy of the X chromosome (47, XXY) [5, 7]. Most men with this syndrome are unable have father children (infertility) due to their low sperm count a few individual who believe themselves to be XXY males may actually be XY/XXY mosaic these males may have normal cell in the testes in great enough , the individual should be able to be father [1, 8, 9]. It most often diagnosed in adulthood using a karyotype, an analysis of the patient's chromosomes taken from a blood sample, or during a woman's pregnancy by taken sample from the amniotic fluid that surrounds the fetus (amniocentesis)and from the placenta (chorionic villous sampling (CVS) [2,4,11,12].
Fig. (1) Karyotype of a young man with Klinefelter syndrome showing an extra X chromosome 47, XXY.

Material & Methods

100 infertile men collected from infertility centre and I.V.F in Baghdad were selected according to the following criteria.

- Male with infertility.
- Having abnormalities in morphology and count of sperm in seminal fluid examination.
- Clinical feature of Klinefelter’s syndrome.

General Semen Exam:
1- Seminal fluid samples were collected.
2- Incubated the seminal fluid at 5-15 minute in 37 °C.
3- One drop of sample on slide examined by microscope [13].

Cytogenetic Methods:
1- Collected (5ml) of venous peripheral blood.
2- Added (0.3ml) or 3 drops of blood in (5-7ml) culture media PRMI 1640.added 0.1 of PHA (phytohaemagglutinin) incubate 3 days (71hours) in 37 °C .
3- Added colchicines and incubate one hour.
4- A hypotonic solution KCL 10 ml added to tube culture 20 minutes in 37 °C in order to swells the cells and separates the individual chromosomes.
5- Harvesting & Fixation treatment with acetic acid methanol 1:3. The cells are spread on micro slides an air dried slide preparation [4].

Result of Cytogenetic Study:
Selected four from 100 case with Klinefelter’s syndrome have symptoms like small testes (<10mm.) breast enlargement, reduced facial and body air, General seminal examination illustrated azoosperma for all cases. Three of X chromosome of the lymphocyte in blood culture with karyotype 47,XXY and one case with mosaic (two cell line first group cell abnormal in the number of chromosome X two copies of chromosome) 47,XXY karyotype Fig.(3) and second group cell normal karyotype 46,XY as show in Fig.(4). The Table (1) illustrated karyotype of four cases and the percentage of each karyotype.

Table (1)
The percentage of karyotype Klinefelter’s syndrome.

<table>
<thead>
<tr>
<th>No.</th>
<th>Karyotype</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>46 , XXY</td>
<td>75</td>
</tr>
<tr>
<td>2</td>
<td>46 , XXY</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>46 , XXY</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>46, XY /47, XXY</td>
<td>25</td>
</tr>
</tbody>
</table>

Fig.(2) Metaphase of abnormal cell 47,XXY.
Discussion

Cytogenetic study illustrated the abnormal in chromosome number in three cases which have all clinical feature of Klinefelter their karyotype 47,XXY the percentage 75% of these karyotype lower than the percentage which recorded by [1,9] and one case with tow cell line mosaic 25% some cells with normal karyotype 46, XY and other cell line with 47,XXY the percentage is high than percentage recorded by [1,9]. The percentage of Klinefelter syndrome more than mosaic. Normal male inherit one X chromosome and Y chromosome the karyotype of normal male 46, XY. Single Y chromosome is sufficient to produce maleness while it's absent is necessary for femaleness. Male abnormalities are the result of irregular number of ether the X, or the Y chromosome or both. In Klinefelter syndrome the extra X chromosome lead to some female clinical features like enlarge breast no hair in body and face. Have two X chromosome 47,XXY and lead to infertility male [6,7,10,11,12]. Statistical study has been made to know percentage of Klinefelter syndrome compare with other syndrome Hundred cases studied with infertility.

We found the Klinefelter’s syndrome 5% when compared infertility male The percentage is over than the world percentage 1% [1,5,6]. The percentage of Klinefelter syndrome between compare with other mental deficiencies (2-9) and the average 5% illustrated in Table (2) for years 2006 till 2009, in the year 2006 the number of patients was low because of bad security circumstances in Baghdad, but the percentage started to raise after that year that may refer to improving of security situation The percentage 5.2% is higher than world percentage 1%[9,10]. The study conclusion that number and Percentage of Klinefelter's syndrome varies according to different population.

References


Statistically study

This study statistical in Yarmok hospital for those patients who submitted the hospital for medical diagnosis in years from 2006-2009. The study illustrated that percentage of Klinefelter syndrome average 5.2% comparinwith other mental retardation Table (2). The study illested hundreded infertility males addmited inI.V.Center 15 of them with klinefelter's syndrom. The percentage of Klinefelter syndrome(1.50%).

<table>
<thead>
<tr>
<th>Year</th>
<th>Number of other genetic syndrome</th>
<th>Number of Klinefelter syndrome</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>2006</td>
<td>47</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>2007</td>
<td>22</td>
<td>2</td>
<td>9</td>
</tr>
<tr>
<td>2008</td>
<td>33</td>
<td>2</td>
<td>6</td>
</tr>
<tr>
<td>2009</td>
<td>46</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Average = 5.2</td>
</tr>
</tbody>
</table>

Table (2)
Percentage of Klinefelter syndrome among cases with deficiency during the period from (2006 – 2009).

Fig (3) Metaphase of abnormal cell mosaic case 47, XXY.

Fig (4) Metaphase of normal cell 46, XY mosaic.
أجري تحليل وراثة خلوية على أربعة مصابين تم اختيارهم من العينة اعتمادًا على بعض الصفات السريرية التي يمتلكها المصابين بمرضية متلازمة كلاينفالتير مثل صغر الحضيض، حجم الأذن، ورم الشعر في العين. وجد أن 3 من حالات هيئة كروموسومية غير طبيعية 47,XXY وحالة واحدة مزائبة تمثل نوعين من الخلايا مجموعة خلايا مماثلة هيئة كروموسومية طبيعية 46,XY ومجموعة أخرى لها هيئة كروموسومية غير طبيعية 47,XXY. عند اجراء الفحص المختبري للسائل المنوي، كانت نتيجة الفحص عدم وجود الحيامن في جميع الحالات. يمكن الاستنتاج أن نسبة الحالات التي أعطت فحصًا وراثيًا خلويًا غير طبيعي للفحص الاستوائي لجميع الحالات نسبة (75%) في حين كانت نسبة (25%) للحالات التي لها فحص وراثيًا خلويًا نوع مزائبة. وأن جميع الحالات التي تم دراستها كانت تعاني من الأعمق.

الخلاصة

أجريت دراسة إحصائية في مركز العقم وأطفال الأذلباب في بغداد مستشفى البرموك لتحديد نسبة المصابين بمتلازمة كلاينفالتير. وجد أن الكلاينفالتير بشكل 1.5% من حالات عدم الرجال وعند إجراء دراسة إحصائية مماثلة في مستشفى البرموك. وجد أن نسبة المصابين بمتلازمة الكلاينفالتير 5.2% مقارنة مع الحالات المصابة بالإمساك العصبي والراثية. يمكن الاستنتاج أن نسبة الإصابة بالمتلازمة تختلف اعتمادًا حسب نوع الحالات المرضية.