Cytogenetic and Statistical Study on Klinefilter´S Syndrome in Baghdad

Suad Gazi Jafer[*], Zeina Tarek Saleh[**] and Abtesam Hadi Mutlake[**]
[*]Department of Biology , Collage of Science, Karbala university.
[**]Department of Biotechnology, Collage of Science, Al-Nahreen University.

Aabstract

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(tow X chromosome) 47, XXY and one case with mosaic karyotape 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]. Klinfilter`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 karyotape 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,].

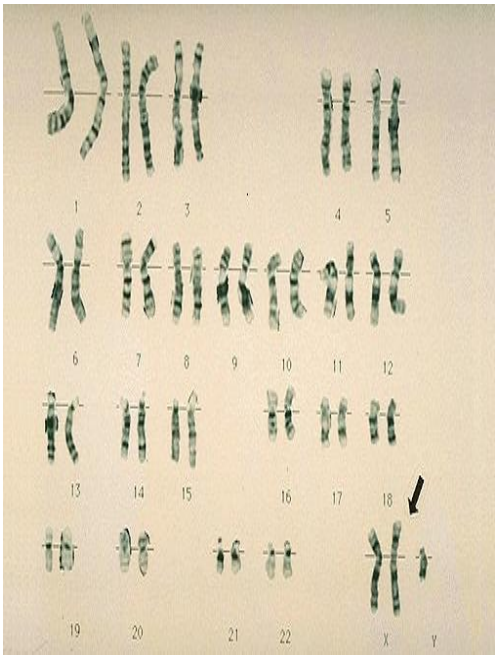

Fig. (1) Karyotype of a young man with Klinefilter syndrome showing an extra X chromosome 47, XXY.

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 Klinefilter`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 slid 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 Klinefilter`s syndrome have symptoms like small testes (<10mm.) breast enlargement, reduced facial and body air, General seminal examination illustrated azoospermia 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,XXYkaryotype 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.

No.	*Karyotype*	*%*
1	**46 , XXY**	**75**
2	**46 , XXY**	
3	**46 , XXY**	
4	**46, XY /47, XXY**	**25**

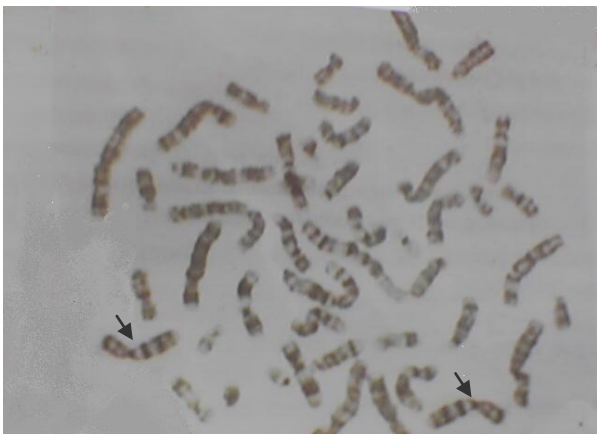

Fig.(2) Metaphase of abnormal cell 47,XXY.

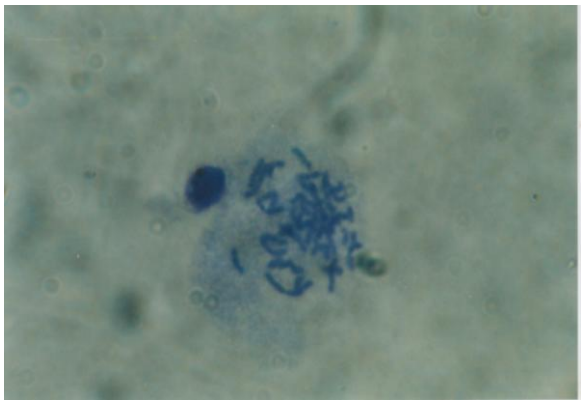

Fig (3) Metaphase of abnormal cell mosaic case 47, XXY.

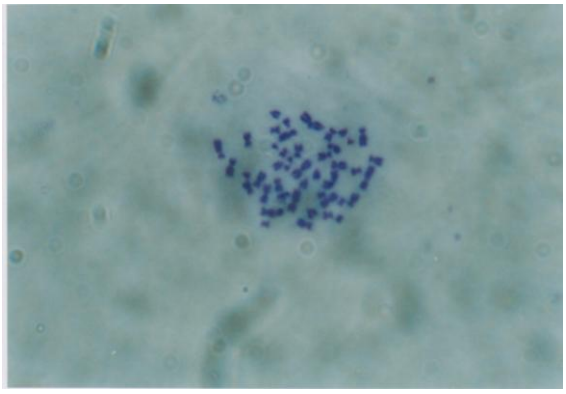

Fig (4) Metaphase of normal cell 46, XY mosaic.

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 Klinefilter syndrome avearage 5.2% comparinwith other mental retradation Table (2). The study illestated handerd infertility males addmited inI.V.Fcenter 15 of them with klinfelter`s syndrom. The percentage of Klinefilter syndrome(1.50%).

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

Year	*Number of other genetic syndrome*	*Number of Klinefilter syndrome*	*%*
2006	47	1	2
2007	22	2	9
2008	33	2	6
2009	46	2	4
			Average = 5.2

Discussion

Cytogenetic study illustrated the abnormal in chromosome number in three cases which have all clinical feature of Klinefilter 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 Klinefilter 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 Klinefilter 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 Klinefilter syndrome compare with other syndrome Hundred cases studied with infertility.

We found the Klinefilter`s syndrome. 5% when compared infertility male The percentage is over than the world percentage 1% [1,5,6].The percentage of Klinefilter 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 Klinefilter's syndrome varies according to different population.

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أجري تحليل وراثة خلوية على أربعة مصابين تم اختيارها من العينة اعتمادا على بعض الصفات ألسريريه التي يمتلكها المصابين بمرضى متلازمة كلاينفيلتير مثل صغر الخصى(ملم>10)، كبر حجم الأثداء ، عدم ظهورالشعر في الوجه والجسم وجد أن 3 من حالات له هيئة كروموسومية غير طبيعية 47,XXY وحالة واحدة موزائيك تمتلك نوعين من الخلايا مجموعة خلايا تمتلك هيئة كروموسومية طبيعية 46,XY ومجموعة أخرى لها هيئة كروموسومية غير طبيعية 47,XXY. عند أجراء الفحص ألمختبري للسائل المنوي كانت نتيجة الفحص عدم وجود الحيامن لجميع الحالات. يمكن الاستنتاج أن نسبة الحلات التي أعطت فحصاً وراثياً خلوي غير طبيعي للطور ألاستوائي لجميع الخلايا نسبة (75%) في حين كانت النسبة (25%) للحلات التي لها فحص وراثيا خلوياً نوع موزائك. وأن جميع الحالات التي تم دراستها كانت تعاني من العقم.

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الخلاصة

أجريت دراسة إحصائية في مركز العقم وأطفال الأنابيب في بغداد ومستشفى اليرموك لتحديد نسبة المصابين بمتلازمة كلاينفلتر. وجد أن الكلاينفلتر يشكل 1,5% من حالات عقم الرجال وعند إجراء دراسة إحصائية مماثلة في مستشفى اليرموك .وجد ان نسبة المصابين بمتلازمة الكلاينفلتر 5.2% مقارنتاً مع الحلات المصابة بالا مرض الولادية الوراثية يمكن الاستنتاج أن نسبة الإصابة بالمتلازمة يختلف اعتمادا حسب نوع الحالات المرضية.