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Chromosomal Mapping Technique for Early Diagnosis of Human Genetic Diseases in Libyan populations, is it needed?

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ABSTRACT

Recently, there is growing interest in the early diagnosis of various human genetic diseases, with the emergence of the rats of several categories of genetic diseases yearly. The majority of the human genome organize into distinct structures known as chromosomes. Notably, the diagnosis of genetic disorders field clarified that chromosomes altered in their nature contributed to several human genetic diseases. Several studies indicated that the most common chromosomal aberration is aneuploidy, which is an alteration in the number of chromosomes due to an extra or missing chromosome. In line, numerical chromosomal diseases are caused by a complex combination of genetic alteration, behavioral, and environmental factors. Instances of these conditions include down syndrome, and others. Indeed, different chromosomal diseases have different phenotypes, and genotypes. However, similar features and associated diseases may overlap between those disorders, therefore, diagnosis of chromosomal diseases depending on clinical features is not accurate. Thereby, early detection of genetic diseases during (Intrauterine or neonatal life), using a suitable approach such as the chromosomal mapping technique is ideal and required. Additionally, the techniques are the cornerstone of the management of diseases associated with genetic diseases. In a society such as the Libyan population, genetic diseases impact not only physical health but also psychological and social well-being. Therefore, testing for genetic diseases is of great importance for the diagnosis, prognosis, and timely intervention of familial patients and their high-risk relatives. In this paper, our main concern is numerical chromosomal aberrations diseases. Moreover, sightsees chromosomal mapping techniques as the most suitable approach. Furthermore, clarify how our application of this technique can be the provision of appropriate that accurately detects human genetic diseases for most of the Libyan populations.

تقنية رسم الخرائط الكروموسومية للتشخيص المبكر للأمراض الوراثية لسكان ليبيا، هل نحتاج اليها ؟

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الملخص

هناك اهتمام متزايد بالتشخيص المبكر للأمراض الوراثية البشرية المختلفة. وذلك مع الازدياد الواضح لمعدلات الاصابة بالعديد من الأمراض الوراثية سنويا في الآونة الأخيرة، علميا ينتظم اغلب الجينوم البشري في تراكيب متميزة تعرف بالكروموسومات. والجدير بالذكر، مجال الاضطرابات الوراثية أوضحَ ان التغير في طبيعة

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الكروموسومات ساهم في ظهور العديد من الأمراض الوراثية البشرية،والعديد من الدراسات اشارت أن أكثر الانحرافات الكروموسومية شيوعًا هي اختلال الصيغة العددية للكروموسومات، وهو عبارة عن تغيير في عدد الكروموسومات. وذلك بفقد او اضافة كروموسوم علي العدد الثابت للنوع،وفي نفس السياق، تحدث الأمراض المتعددة الناتجة عن الاعتلالات الكروموسومية من خلال تغييرات عديدة معقدة في العوامل الجينية والسلوكية والبيئية،وتشتمل هذه الأمراض على العديد من الاعتلالات ومنها متلازمة داون وغيرها، وفي الواقع، مختلف أمراض الالاعتلات الكروموسومية لها أنماط ظاهرية وأنماط وراثية مختلفة.ولكن هناك ميزات وصفات متشابهة ومشتركة، وكذلك الأمراض الفسيولوجية المصاحبة للاعتلالات الكروموسومية قد تتداخل بين تلك الاضطرابات المرضية المتعددة. لذلك تشخيص أمراض الاعتلالات الكروموسومية الوراثية ،اعتماداًعلى الأعراض السريرية لهذه الأمراض يعتبر غير دقيق و ليس بالتشخيص الناجع.وبالتالي،الكشف المبكر عن الأمراض الوراثية في مرحلة حديثي الولادة، باستخدام نهج وطرق طبية حديثة ومناسبة مثل تقنية رسم خرائط الكروموسومات تعد هامة و مطلوبة، اضافة إلى ذلك ، فإن مثل هذه التقنيات تعتبر حديثا حجر الزاوية في إدارة و التحكم بالأمراض المرتبطة بالأمراض الوراثية المختلفة فضلا عن الكشف الدقيق للاعتلالات الكروموسومية. في مجتمعات مثل المجتمع الليبي، فإن الأمراض الوراثية لا تؤثر فقط على الصحة الجسدية للأفراد المصابين بهذا النوع من الاعتلالات، ولكن لهاأيضاًتاثير على الرفاه النفسي والاجتماعي للأفراد وذويهم. لذا ، يعد اختبار الأمراض الوراثية المبكر أمراً بالغ الاهمية للتشخيص والكشف المبكر والتدخل في الوقت المناسب لعلاج المرضى وأفراد أسرهم وخاصة الأقارب المعرضين لمخاطر عالية للاصابة بمثل هذه الاعتلالات الوراثية. في هذه الورقة ، كان التركيز الرئيسي على الأمراض المرتبطة بالانحرافات العددية للكروموسومات، علاوةً على ذلك، تسليط الضوء على تقنيات رسم الخرائط الكروموسومية كطريقة حديثة ومناسبة جدا للكشف والتشخيص المبكر، وخلال هذه الورقة العلميةايضاً تم توضح كيف أن التطبيق لهذه التقنية قد يكون نهجا حديثا و مناسبا لتوفير الجهد والمال والوقت للكشف بدقة عن الأمراض الوراثية المختلفة لمعظم السكان في ليبيا.

متلازمة داون
مستشفى الأطفال بمدينة

Introduction

Heredity plays an important role in the passing of genetic material from one generation to the next. Human genome molecules contain genes that express human body how develops and their specific function. Nowadays, clearly that the majority of the human genome is organized into 23 pairs of chromosomes with distinct structures, making a total of 46 chromosomes, which vary generally in structure and also size. Hence, variations in our genomes alone or alongside the environment's multitude of factors contributed to the mechanism of Chromosomal distortion. Thereby, any chromosomal numerical or structural change can lead to medical diseases that are well known as human genetic diseases or chromosomal disorders. A growing understanding of human genetic diseases clarified that chromosomes altered in their nature correlated with several inherited disorders. Basis of clinical, pathological investigations chromosomal disorders can be classified into two main types; numerical and structural chromosomes related disorders. It is worth mentioning that any change in chromosome structure, even a small one, will lead to a specific disease, however, an altered number of chromosomes can lead to a more serious disease because a person's entire chromosome number can be severely affected. Indeed, chromosomes numerical disorders occur when there is a change in the number of chromosomes (more or fewer than 46). This numerical alteration has contributed to several diverse forms of chromosomal disorders including the most well-known numerical disorders, Down syndrome (trisomy 21), additionally to other types of numerical disorders such as trisomy 13, trisomy 18, Klinefelter syndrome as well as Turner syndrome. Recently, genetic diseases arising from chromosomal defects are widespread, worldwide in general and in Libyan society in particular, which might be attributed to the changes in the lifestyle within societies, including changing food habits where people rely heavily on fast food and canned food. Added to, many other factors, which include the lack of drug legalization, besides the spread of harmful behaviors that play a major role in chromosomal distortion, such as smoking and consuming alcohol in addition to drugs and their destructive effect on cells and their genetic material. Perhaps all these factors collectively contributed to a severe increase in human genetic diseases and their spreading capacity. Thereby, strongly need to follow the global strategy of early detection techniques for chromosomal alterations disorders.

In recent, one of the most specific techniques for early diagnosis of chromosomal alterations is genetic testing has its roots in being able to visualize whole chromosomes in a technique known as (chromosomal mapping) karyotyping. Scientists have used a staining technique that depends on a banding pattern of the chromosomes. These banding patterns make each of the chromosomes easier to identify, like a map. Each of the chromosomes has a specific structure and numbers, thereby, any deviation from the normal karyotype is recognized as a chromosome aberration.

In a society such as the Libyan population, chromosomal aberrations and diseases impact not only physical health but also psychological and social well-being. Therefore, testing for early diagnosis of genetic diseases is of great importance for the diagnosis and potentially directing therapeutic intervention. Besides timely intervention of familial patients and their high-risk relatives. Moreover, early diagnosis can also be useful in terms of gaining access to relevant support whether this is financial, social, or practical. Nowadays, early molecular diagnosis of chromosomal aberrations is an urgent necessity, not a luxury that can be dispensed in a society such as the Libyan society. Thus, the present paper focuses on such chromosomal numerical disorders. Additionally, aimed to identify important early diagnostic strategies for numerical chromosomal disorders. Besides, clarify the potential of chromosomal mapping (Karyotyping Technique) as the more suitable approach, that is currently used in clinical settings to identify several types of chromosomal alterations disorders.

Methodology

In this study, quantitative methods were utilized. Data were gathered from Benghazi Pediatric Hospital at time points during 2005-2020. This method is particularly useful in studying, determining, and gaining a detailed distribution and spread of human genetic diseases, particularly chromosomal numerical alteration among the population of Benghazi city. This method is also for identifying and obtaining further in-depth information on the categories of human genetic

diseases and additionally to determine measures of the most common chromosomal numerical alteration disorders in Benghazi city. The criteria were considered when selecting Benghazi Pediatric Hospital that the sample participants in this study were pediatric age group.

Result and Discussion

A genetic disease is any disease caused by an aberration in the genetic material of an individual. Most genetic diseases are the direct result of a mutation in the human genome that contributes to diseases that have a complex pattern of inheritance. Chromosomal aberrations are either numerical (concerning the addition or subtraction of an entire chromosome or set of chromosomes) or structural alterations. Some people inherit genetic disorders from their parents, the acquired changes or mutations in others can occur either randomly or due to some environmental exposure. Genetically determined diseases include those resulting from chromosomal aberrations that typically occur due to a problem with the cell genome.

Concurrently, several investigations have confirmed that numerical aberrations represent a noteworthy proportion of chromosomal variations found in humans. The chromosomal aberrations may occur in both division meiosis and mitosis and therefore present in all cells or mosaic models [30]. Medically, an early genetic disorders diagnosis would be ideal to help in early intervention and treatment, which will improve the outcome of the patient. Genetic disorders impact not only physical health but also the affected individuals, family, and society, as many are associated with a severe and permanent disability. Hence, the ultimate goal of this study is to use suitable techniques for early diagnosis to identify individuals who are at risk of appearance of chromosomal aberrations disorders. Thus at this point, advanced criteria for the screening of hereditary disorders, which include some review of personal and family histories, as well as genetic testing (Karyotyping Technique) are the most significant strategies, which help in early intervention or, if possible prevent the development of genetics disease [19]. This study focuses on the diagnosis of chromosome numerical disorders. The study had several stages and phases. The data was collected from the Paediatric Hospital in Benghazi for a timeline from 2005 to 2020 to detect the incidence of human genetic disease (chromosomal numerical alteration) in Benghazi city. In addition to measuring the highest disorder category among all the provided chromosomal abnormalities.

Figure 1, shows out of 2257 cases, trisomy 21 documented the highest disorder were counted for 2176 among the provided diseases. The 2nd was trisomy 18 with 51 cases, while the 3rd was Trisomy 13 with 36 cases, and lastly Turner syndrome with about 12 cases. Although Klinefelter syndrome is included in our study categories, however, the data collected from Benghazi Pediatric Hospital displays zero cases.

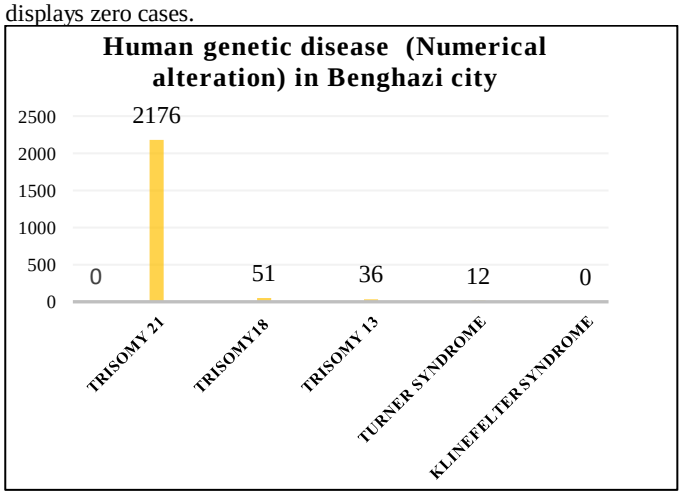

Fig 1: Shows that there is a sharp rise in the cases number of trisomy 21 with grows by more than 2000 cases. The data reveals that trisomy 21 is a high disorder account among the provided chromosomal alteration disorders. While 2nd disorder was trisomy 18 with drop cases numbers around 51 cases, 3rd is Trisomy 13 with 36 cases, Turner syndrome reported a low case with about 12 cases, lastly, the number of in Klinefelter syndrome shows level off by zero point cases.

Indeed, Turner syndrome and Klinefelter syndrome are common chromosomal abnormalities, however, they demonstrated the least number, which might be due to missed diagnosis cases as turner syndrome and some Klinefelter syndrome individuals' expressions of normal mental status, and late complications compared to other chromosomal numerical disorders. Therefore, accurate and early genetic diagnosis is essential to determine and detect any chromosomal abnormalities in early life.

Figure 2, demonstrates that Trisomy 21 starting from 2005 until 2020 where the prevalence of Down syndrome was less than a hundred cases per year until 2010, at which there was a slight increase in the number of cases reported by 133 cases, however in 2011 a drop in the number of reported cases was noted, which might be due to lack of collected data as during the mentioned time there was February revolution and war as a consequence reaching to the hospital and providing data was risky. Moreover, our idea is supported due to the increase of the data on Down syndrome cases in 2012. Whereas reaching a peak of reported cases in 2013 with 202 cases, in Benghazi city population, which is considered a bit high.

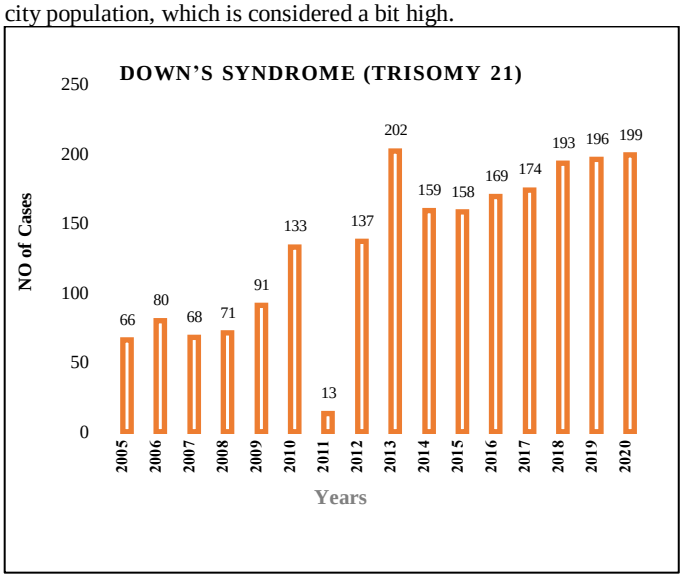

Fig. 2: Shows the incidence of Trisomy 21 starting from 2005 until 2020. The graph shows that there has been an increase in cases numbers of Down syndrome. The number of cases reached a peak in 2013, which a reported 202 cases of DS. The data can be seen that DS cases remain steady during the last three years of study.

Worth mentioning these data of admitted cases with chromosomal number alteration to Benghazi Pediatric Hospital due to coexisting diseases, in previously clinically diagnosed individuals such as dysentery, cardiovascular disease, respiratory infection (etc…). Consequently, the accurate numbers of chromosomal alterations disorders cases in general and down syndrome specifically are not yet accurately measured.

The above-aforementioned context may be explained how the presence of an early and clear diagnosis of chromosomal aberrations disorders is of great importance for several reasons, confirming the suspected diagnosis to clarify risks in affected individuals, and directing therapeutic intervention. Moreover, accurate diagnosis can also be useful in support terms whether this is financial or social, or, if possible, preventing the development of genetic disorders in Libyan society. Hence, significant strategies, chromosomal mapping techniques in early newborn life for any doubtful case, and an accurate statistical system together and provide more reliable information about genetic aberrations disorders in the Libyan population are urgently required.

Interestingly, the present data has displayed a dramatic increase in Down syndrome cases, partially in the last decade. Several factors play a role in increasing the risk of chromosomal aberrations disorders on top of that maternal age, family history as well as war

remnants, in addition, other factors need to be investigated and studied thoroughly. Structural and numeric chromosomal abnormalities occur in approximately 0.6% of live births and often result in malformations, dysmorphism, and/or developmental disabilities. Most aneuploidies are a consequence of improper segregation of the chromosome pairs during meiosis [1], [22].

In the field of chromosomal disorders, it is found that aneuploidies of autosomal chromosomal are more tolerated from the sex chromosome alterations [22]. The most common numeric disorder is down syndrome, or trisomy 21, which occurs in nearly 1/800 of live newborns, which defect with enormous medical issues. On the other hand, trisomy 13 and 18 are considered less common disorders. While, trisomy 8 and 9, rarely occur [1].

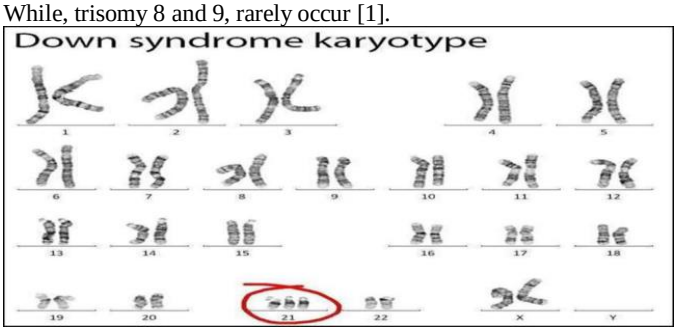

Fig 3: Reveals that there have been a marked extra copy of chr-21, chromosomes mapping techniques (Karyotyping) of Down syndrome (Trisomy 21).

Generally, down syndrome (DS) or trisomy 21 is caused by variation of whole or part of chromosome 21 in all or some cells of the body, which lead to a high dosage of gene expression of the trisomic genes (Figure.3). Although down syndrome (DS) is the most predominant chromosomal numerical aberrations worldwide that cause of intellectual disabilities. However, in the middle east region, there are no accurate investigations of the DS population [18].

Concurrently, several studies have explained that DS has high genetic complexity and phenotype variability. Moreover, large numbers of DS patients in different populations of the world run into several extra health issues. Several clinical conditions associated with DS affect the multisystem. These medical problems include congenital heart defects, hematopoietic diseases, and early-onset Alzheimer's disease. Additionally, certain mental retardation is close related to Down syndrome. In early infancy, gastrointestinal anomalies, weak neuromuscular tone, and hypotonia are often issues that have a strong possibility implication for down syndrome.

Further, several studies prove that the Down syndrome population has specific characteristics of facial and physical features, which include dysmorphic features of the head, neck, and airways, and audio-vestibular and visual impairment. A systematic study has revealed that down syndrome, is also disposed to several other GI disorders like gastroesophageal reflux (GERD), chronic constipation, and intermittent diarrhea, a celiac disease. As well as Hirschprung disease (HD), which is reported in about 12% of all cases of (HD) [2].

In addition to the appearance of higher frequency of other medical disorders such as various types of leukemia disorders. Whereas, the patients with DS demonstrate a unique spectrum of malignancies, which include leukemia as well as solid tumors [3]. In the line, studies discussed that DS patients have a high relative risk of leukemia about 10– 20 fold, with a growing risk of 2% at 5 years old and 2.7% by age of 30 years old [9]. Moreover, ear, nose, and throat problems are frequently prescribed as relatively common medical issues in down syndrome patients. Because of the structural abnormalities in the inner ears thereby, sensorineural hearing loss has also been related to the down syndrome population [15]. In addition to that, the DS population is commonly associated with thyroid gland dysfunction. It is now understood that Hypothyroidism can be congenital or acquired at any time during the life of DS patients. The evidence from the newborn screening program in New York has reported a high frequency of congenital hypothyroidism in DS syndrome newborns as compared to the others [3].

The following part moves on to the second most common autosomal trisomy syndrome, which is trisomy 18 syndrome. The condition is also termed Edwards syndrome, which triggers the presence of an extra chromosome 18, either full, mosaic trisomy, or partial trisomy 18q (Figure. 4). Data about the trisomy 18 syndrome gives an account of the frequency of live-born is estimated as 1/6,000-1/8,000. Additionally, several studies suggest an association between the prevalence of trisomy 18 rises and the growing maternal age. The recurrence risk for a family with a child with full trisomy 18 is about 1% [5].

Fig.4: Shows clearly that there is an extra copy of chr-18, chromosomes mapping techniques (Karyotyping) of Edwards syndrome (Trisomy 18).

Recent developments in the field of chromosomal aberrations disorders have provided important information on the clinical features that are allied with Trisomy 18 patients. These are involving psychomotor and growth retardation, microcephaly, microphthalmia also severe from malformed ears, micrognathia or retrognathia, microstomia, and distinctively clenched fingers, as well as other congenital malformations.

In term of Patau syndrome is the least widely and most severe of the viable autosomal trisomies. It is now understood that median survival is less than 3 days. Patau syndrome is triggered by an additional copy of chromosome number 13 (Figure. 5). Recent research has revealed that the frequency of Patau syndrome is around 1 case per 8,000-12,000 live births [30]. The evidence of Dysmorphic features in Patau syndrome cases is similar to other chromosomal abnormalities disorders. However, there is a difference in the clinical presentation which include cleft lip, cleft palate, polydactyly, microcephaly, and microphthalmia. In addition to various types of hernias and neural tube defects [25]. The most common chronic disease linked with Patau syndrome is cardiac disorders, whereas 80% of those who were very clearly demonstrated effective cardiac defects. Moreover, capillary hemangiomas and polycystic kidneys, and other renal defects have been established in Patau syndrome patients. Notably, previous research has established that the most common causes of death in Patau syndrome are cardiopulmonary arrest estimated (at 69%), congenital heart disease (13%), besides pneumonia by (4%) [16].

In this case, Turner syndrome is characterized by sex chromosomes (x) through altering the partial or complete absence of one X chromosome (45, X karyotype) (Figure.6). Estimates of Turner syndrome occurs in one out of 2,500 to 3,000 live female births; however, many more 45, X conceptuses do not survive, whereas post the first trimester. Turner syndrome has contributed to causes 10% of all first-trimester miscarriages.

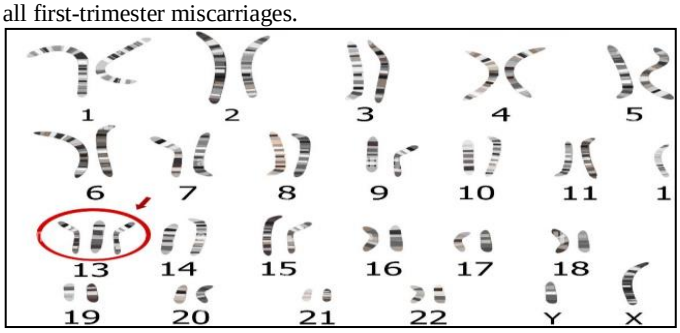

Fig. 5: Shows that there has been an extra copy of Chr-13, chromosomes mapping techniques (Karyotyping) of Patau syndrome (Trisomy 13).

Indeed, little is known about Turner syndrome and it is not clear what major risk factors that associated with this alteration. Despite the importance of maternal age in increased rates of chromosomal disorders such as Down syndrome and other chromosomal

abnormalities, however, there remains a paucity of evidence on Turner syndrome. Data from several studies suggest that recurrence of Turner syndrome in subsequent pregnancies is rare [23], [26].

Nevertheless, not all genes from the second chromosome are inactivated in Turner syndrome. Some genes escape X-inactivation through a process initiated by the X-inactivation-specific transcript (XIST). The loss of these non-inactivated X genes causes the phenotypic manifestations characteristic of Turner syndrome [26].

The existing body of research on X chromosomes suggests that all females with Turner syndrome are infertile [17]. Clinical manifestation of Turner syndrome varies and may be subtle, however, they usually include short stature, a broad chest with widely spaced nipples, cubitus valgus, congenital lymphedema, and a lack of spontaneous pubertal development because of ovarian sex hormone insufficiency [12], [29].

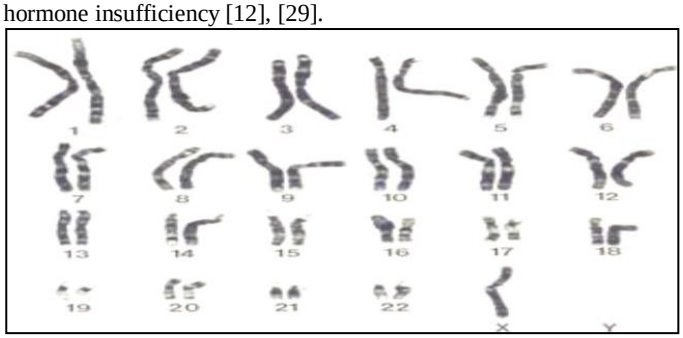

Fig. 6: Shows that there has been monosomy alteration in the sex chromosome (45, X). Chromosome mapping techniques (Karyotyping) of Turner syndrome.

Moreover, Turner syndrome individuals are at risk of congenital heart defects, besides an increased risk of renal defect, and sensorineural hearing loss. Further, it has previously been observed that Turner syndrome patients sever from osteoporosis, obesity, diabetes, and also atherogenic lipid profile [13]. Although Turner syndrome individuals typically have normal intelligence, however, may suffer from some serious problems with nonverbal, social, and psychomotor skills [6], [21], [28].

Another important aspect of chromosomal aberrations disorders is Klinefelter syndrome. The term Klinefelter syndrome is generally understood to mean alterations in the number of sex chromosomes, thereby, it is seeming to be the most common sex chromosome disorder (Figure.7). Klinefelter syndrome is a chromosomal alter condition that occurs as a result of the presence of an extra X chromosome in male cells, this could affect the physical and intellectual development of the Klinefelter syndrome individual. Unfortunately, the rated frequency of Klinefelter syndrome affects around 1 in 650 male newborns [20]. The evidence shows that most often, Klinefelter individuals have the normal X and Y chromosomes, but one extra X chromosome is also present, with a total number of 47 chromosomes (47, XXY). This condition runs individuals with Klinefelter syndrome to have an extra copy of multiple genes on the X chromosome [10], [27].

Recent research has revealed that Klinefelter syndrome is not inherited; the addition of an extra X chromosome occurs during the formation of reproductive cells. Mosaic Klinefelter syndrome (46, XY/47, XXY) is also not inherited. It seems possible that these results are due to a random error of cell division in early fetal development [20], Accordingly, it is becoming extremely mistake to ignore the existence of early and accurate diagnosis of chromosomal aberrations disorders, and sex chromosome alteration in special.

Typically, some people with features of Klinefelter syndrome have an additional X chromosome in only some of their cells while other cells have a normal set of male sex chromosomes. Rarely, other cells may have extra chromosome aberrations, in these individuals, the condition can broadly be defined as mosaic Klinefelter syndrome (46, XY/47, XXY). Data from several studies suggest that fewer than 10% of individuals with Klinefelter syndrome are believed that have the mosaic model [24]. A further definition of variants Klinefelter syndrome is given by several conditions resulting from the presence of more than one extra sex chromosome in each cell. These conditions have become distinguished by 48, XXXY syndrome, 48, XXYY syndrome, besides 49, XXXXY syndrome [10]. Several effects result when individuals undergo Klinefelter syndrome. These are including taller than the average additionally infertile; however, the signs and symptoms of Klinefelter syndrome vary among boys and men with this condition. In some cases, the features of the condition are so mild that the condition is not diagnosed until puberty or adulthood. Unfortunately, up to 75% of affected men and boys with Klinefelter syndrome are never diagnosed [10].

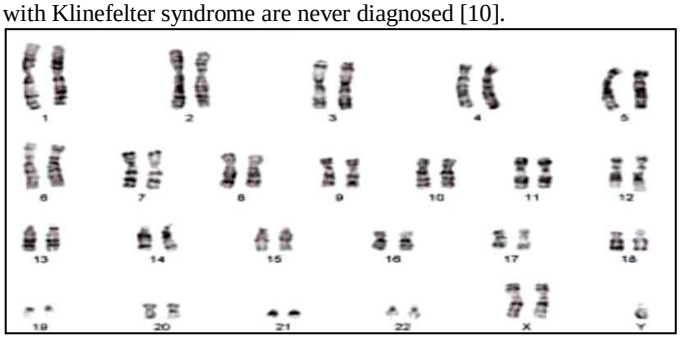

Fig. 7: Shows clearly that there is an extra copy of sex Chromosomes (X- Chr-). Chromosome mapping techniques (Karyotyping) of Klinefelter syndrome.

In line, the present study data are in accordance with the previous context where the data collected from Benghazi Pediatric Hospital displays zero cases of Klinefelter syndrome. Although Klinefelter syndrome included a high-frequency rate in comparison to other chromosomal alteration disorders such as Down syndrome. Hence, the present study offers some important insights into addressing the issue of early diagnosis of chromosomal aberrations disorders in the Libyan population that is strongly needed. Previous studies explained that the small testes and decline of hormone production are a major influence on Klinefelter syndrome individual's infertility.

Moreover, lack of treatment and shortage of testosterone can lead to several medical issues such as delayed or incomplete puberty, breast enlargement (gynecomastia), decreased muscle mass, and decreased bone density [11], [27]. Furthermore, Klinefelter syndrome can be associated with learning disabilities and several issues with speech and language development. However, the features of variants Klinefelter syndrome disorders tend to be more severe than those of Klinefelter syndrome and affect more parts of the body [11].

As discussed above, chromosomal alterations disorders are associated with many compacted human diseases. Currently, several techniques have been developed to detect different variations in numbers and subtle rearrangements of the chromosomes. Recent advances in the mapping and banding techniques are the most reliable and valid techniques in the chromosomal alterations field. They allowed for identifying any alteration in the chromosomal numbers, cryptic or submicroscopic imbalances, which gain a detailed understanding of subtle rearrangements that affected one or a few chromosome bands. Thus obtain further in-depth information on the categories of chromosomal alterations disorders.

In Conclusion

Numerous chromosomal alterations must be early detected and diagnosed in the intrauterine or neonate period. In a society such as the Libyan population, genetic diseases impact not only physical health but also psychological and social well-being. Therefore, testing for genetic diseases is of great importance for the diagnosis, prognosis, and timely intervention of familial patients and their high-risk relatives. At this point, the present work offers some important insights into compacted diseases that more contributed to the chromosomal numerical disorders. Furthermore, this study opens the doors to the extent of the danger and the spread of chromosomal alteration disorders within Libyan society. Thereby, strongly need to follow the global strategy of early diagnosis techniques by using specific techniques with a high percentage of accuracy such as chromosomal mapping (Karyotyping) and other advanced molecular techniques.

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