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Study on Diagnosis of Turner Syndrome

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Abstract:

Turner's syndrome (TS) is referred to as the monosomy X, in which total or partial loss of one sex chromosome (45X) with the ratio of 1:2500 in live born infants with phenotypic females. Commonly known as Ulrich Turner Syndrome and it contain main clinical features that includes short stature, cardiac anomalies, lymphedema, primary ovarian, gonadal dysgenesis, swollen hands and feet, webbed neck and neurocognitive difficulties. Patients face various difficulties during increasing over the lifespan in given complexity of the condition. The main problem is because of the delay in diagnosis of Turner's syndrome, as only 15-30% patients are diagnosed during the first year of life. The diagnosis and care of turner syndrome was published on individuals in 1994. By the knowledge of complex etiology and detailing more about its clinical variability and difficulties conclusively allow us to develop the therapeutic and management approach of such patients. In this review paper, study about the diagnostics changes of the turner syndrome in individual.

Keywords: *Turner Syndrome, Diagnosis, Etiology*

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Introduction

Turner's syndrome, is partial or complete loss of second X-chromosome in new birth females (Ramirez and Villarreal, 2016). This syndrome is not inherited by parents, normal people contain the 46 chromosome but inherited people have 45X chromosome (Kadakol *et al.*, 2017). It is designed by Henry Turner in 1938 which included the short stature, sexual infantilism, cubitus valgus and pterygium coli (Shankar and Backelijauw, 2018). Ford *et al.* recognized the syndrome's chromosomal base in 1959 and find out that presented 45X chromosomes in patients with single X chromosome. It is occur in one every 2500-3000 live birth and it is only full monosomy which is compatible with life (Ramirez and Villarreal, 2016). Common issues was recognized in Turner syndrome that is Short stature, pubertal delay/ovarian insufficiency, cardiac and renal abnormalities, sensorineural hearing loss, ophthalmologic problems, thyroid abnormalities, metabolic syndrome, inflammatory bowel disease and neurocognitive issues (Shankar and Backelijauw, 2018). Turner syndrome with mosaicism is defined as the chromosomal abnormality may be present in some cells. This syndrome was found in 1-2% pregnant ladies and 99% have a spontaneous abortion. About 60% cases of turner syndrome that has 45X chromosomes, 5-10% cases show X chromosome anomalies which is deletion of long or short arms, isochromosomes or ring chromosomes and 6-9% cases shows a normal or structurally Y chromosomes (Iqbal, 2014).

This clinical review is focused on the latest update of diagnostic for the most common clinical concerns that is related to turner syndrome (Pinsker, 2012). It is improved the recognition of these comorbidities and their management by using recent diagnostic (Shankar and Backelijauw, 2018).

Diagnosis

Turner syndrome's diagnosis can occur at a wide range of age (Shankar and Backeljauw, 2017). In the function of clinical and hormonal findings, Diagnosis is suspected. By using a conventional Karyotype or other cytogenetic analysis, confirm the diagnosis of a subject aneuploidy. In case of 3-6% women, with turner syndrome that have a 45X/46, XY mosaic and with the risk of presenting gonadoblastoma during their lives lies between 7% to 30% (Ramirez and Villarreal, 2016).

Diagnosis was occur into two forms:

- Prenatal Diagnosis
- Postnatal Diagnosis

Prenatal Diagnosis

Prenatally, diagnosis system is increase in turner syndrome but exists the significant ascertainment partiality in that underlying reason for prenatal chromosome analysis often impacts the validity of the findings. Ultrasound findings in prenatal turner syndrome that include the nuchal translucency (result is fairly specific), cystic hygroma (ultrasound findings alone can predict turner syndrome in 30-70% cases), coarctation of the aorta and/or other left-sided heart defects, brachycephaly, malformations, renal anomalies, polyhydramnios, oligohydramnios, and growth retardation. In both (nuchal translucency and cystic hygroma) case, ultrasound finding should be seen in autosomal trisomy syndromes and finding appears in which specificity for turner syndrome depends on gestational age. Due to specific ultrasound finding, 45X fetuses are discovered, "classic" phenotypic findings are likely.

With the increasing of ultrasound finding, prenatal counseling is important because the loss of rate of spontaneous fetal for 45X fetuses. The quantity of TS occurring is as many as 3% of all the fetuses and hence later can cause around 10% of spontaneous loss of fetus including 99% of 45X embryos terminating spontaneously during the first and second trimesters.

60% of turner syndrome fetuses are electively terminated in some countries. An explanation that even with an ultrasound finding, delivery of a viable newborn is possible and many of those children go on to have an excellent quality of life included in prenatal counselling.

False positive results can occur, when a prenatal karyotype is performed for other reasons such as advanced maternal age or abnormal maternal screening tests. The fetus with 45X karyotype or loss of one chromosome in newborn phenotypic female finding then diagnosis incidentally. Fetus have not only fewer phenotypic finding but the result of karyotype can be non-specific in case of discovered mosaic karyotype.

Diagnostic information is received by the high-resolution ultrasound and fetal echocardiography. Fetuses with turner syndrome are detected by using the maternal biomarker or maternal plasma DNA sequencing but is still preliminary stages (Pinsker, 2012).

Postnatal Diagnosis

During infancy, Lymphedema is the most common disease that is seen in 97% cases while short stature is most commonly leads to evaluation during childhood

and adolescence that is seen in 82% cases. Y-chromosomal material may be present in 5% individual which have a marker chromosome that is revealed by the analysis of karyotype. Current guidelines advocate screening for Y material if signs of virilization develop or a marker chromosome has already been identified, because the risk of developing gonadoblastoma with Y material present ranges from 5–30% in recent studies.

Earlier age of diagnosis, delay in the diagnosis of turner syndrome is shown by retrospective analyses, averaging 5 years after patients had fallen below the 5th percentile in height to time of diagnosis. 20% patient are diagnosed after the age of 12 years and important questions are arise that how to diagnosis of turner syndrome earlier. Earlier diagnosis, if diagnosis could be done non-invasively or as part of newborn screening that should be allow for detection of cardiovascular and renal anomalies which is unidentified until the time is diagnosed and facilitate for treatment of growth failure.

The value of high through put pyrosequencing of buccal swabs for turner syndrome is the recent advances testing. By using pyrosequencing, quantitate relative allele strength, readily detect loss of an entire X-chromosome or mosaicism with up to 97% sensitivity in this testing. This technology is very useful for non-invasive screening for turner syndrome **(Pinsker, 2012)**.

Review of Literature

Hjerrild, Mortensen and Gravholt (2008), in this paper, patients are needed to comprehensive care from a multidisciplinary team whose are suffer from turner syndrome and team done the best practice from outpatient clinic with special importance on turner syndrome. Related to turner syndrome knowledge is limited, this syndrome is infrequently seen by clinicians and patients that have a range of questions related to syndrome.

Pinker (2012), up to 30% of cases of turner syndrome are diagnosed prenatally which is showed a normal karyotype at delivery that is done by cytogenetic registry. This study is complicated by the fact that mosaicism is common finding in chorionic villus sampling or amniocentesis. In this review paper, improvement in the diagnosis of growth failure, cardiac disease and ovarian failure for the care of women with turner syndrome.

Iqbal (2014), stated that multidisciplinary care involving newborn screening, regular cardiovascular examination, GH and estrogen supplements with appropriate pubertal development had changed the scenario, with the hope of giving them a normal quality of life.

Bondy (2014), using newer genetic screening tests for the diagnosis of postnatal turner syndrome that requires further validation, with the 20-30 cell karyotype remaining the gold standard.

Ramirez and Villarreal (2016), turner syndrome is a chromosomal syndrome, in which partial or complete loss of second sex chromosome in newborn female which may cause a sub-diagnosis of its cases. It is most important that know the clinical and genetic aspects which will lead to a timely detection of cases and proper management of co-morbidities of genetic condition.

Yang (2017), stated that marked advances in diagnosis and first treatment of diagnosis in 1939 by Henry Turner. For diagnosis, feasibility of molecular technique needs to be further established. In young adults with turner syndrome, improve quality of life by development of normal height attainment and age appropriate pubertal.

Culen *et al.* (2017), in this review paper, growing the upcoming or expected difficulties with turner syndrome. The aim of this review, provided the treatment of girls with turner syndrome through the help of psychologist or other healthcare providers with challenges. It is conducted that a multidisciplinary standard of care based on a well-defined screening would greatly improve the long term outcomes of patients with turner syndrome.

Kadakol (2017), clinical and structural changes in chromosomes with turner syndrome like Y chromosome mosaicism. Type of genetic disorders is the essential study of chromosome, which will turn in help for diagnosis.

Shankar and Backeljauw (2018), turner syndrome's guidelines are based on expert consensus and evidence for optimal hormone replacement throughout the age spectrum in turner syndrome is still evolving. Patient's health and quality of life related to turner syndrome are improved by adequate support and medical care.

Conclusion

Turner syndrome is a mutation due to which many diseases are occur such as short stature, webbed neck, pubertal delay/ ovarian insufficiency, cardiac and renal abnormalities, sensorineural hearing loss, ophthalmologic problems, thyroid abnormalities etc. This syndrome is improved by giving the clinical and medical health care of patients by best practice of medical and psychologist. Postnatal turner syndrome (a type of turner syndrome) is diagnose by using the

screening test, it is the recent technology. It is conducted with a multidisciplinary standard of care based on a well-defined screening, would greatly improve the long term outcomes of patients with turner syndrome.

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