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PREVALENCE OF TURNER'S SYNDROME

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ABSTRACT

Gonadal dysgenesis, 45 X condition, also known as Turner's syndrome, is the condition in which a female is missing X monosomy partially or completely. There might be a difference in the signs and symptoms seen in the affected. There are many areas of concern which are to be checked regularly like cardiovascular malformations, hearing impairment and low thyroid function. There has been no discovery of a treatment to cure this gonadal dysgenesis. However, each of the symptoms can be reduced to help the patient live a more productive life and can reach his or her almost fullest potentials in the work they do or commitment they take up further in life.

KEYWORDS:Turners syndrome,prevalence,symptoms

I. INTRODUCTION :

Ullrich-Turner's syndrome, also known commonly as Turner's syndrome, is a disorder seen in females [1] . It is the gonadal dysgenesis with abnormalities in sex chromosome [2] . It can be also said as the monosomy of X [3] . This syndrome can be characterized by cardiovascular malfunctions, aortic dilation and dissection, short stature, micrognathia, cubitus valgus, high arched palate, Modeling deformity and short metacarpals [4,5] . The most critical clinical aspects of Turner's syndrome are caused due to the congenital cardiovascular anomalies such as aortic coarctation and dissection [6] . This syndrome is found to affect 1 i every 2000 live-born girls [7].

Causes :

Available research on human embryos and gametes suggests that aneuploidy and chromosomal fragmentation occur commonly in the process of meiosis, related to the errors in development of the embryo [8, 9] . The cause for Turner's syndrome is due to the partial or complete loss of a second sex chromosome in some or all cells, with or without the cell line mosaicism [7] . This cell line mosaicism can be caused by an error which occurs in cell division in the extremely early stages of development of the unborn foetus.

Signs and Symptoms :

In Infants :Because of lymphedema, it is found that the child after birth has swollen hands and feet [10] . The combination of lymphedema with dysplastic or hypo plastic nails gives the fingers and toes a characteristic appearance like that of a sausage.

In childhood : Short stature is usually present.

In adolescents : ovarian failure, puberty delay, absence of growth spurt during adolescence are a few of the signs and symptoms which are featured during this phase of life of the patient.

Puberty : Along with the ovarian failure, there is primary or secondary amenorrhea occurring and breast development doesn't occur [10] .

Physical findings in Turner's syndrome patients :

- •Cubitus valgus
- •Lymphedema
- •Shortened fourth and fifth metatarsals and metacarpals
- •High arched palate
- •Hypoplastic or hyperconvex nails
- •Wrist Madelung deformities
- •Scoliosis
- •Hypothyroidism

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- •Cutis laxa
- •Otitis media
- •Low-set auricles
- •Cataracts

II. Diagnosis :

Diagnosis of a patient who has Turner's syndrome can be done either prenatally or postnatally.

Prenatal Diagnosis : Most cases of Turner's syndrome are discovered during amniocentesis or during the chorionic villous sampling [11] . Ultrasound is also performed to indicate the disorder by increased nuchal translucency or the presence of a cystic hygroma. Prenatal diagnosis is not sufficient as gonadal development and postnatal growth is to be checked for the final diagnosis of Turner's syndrome.

Postnatal Diagnosis : All individuals who have been suspected to suffer from Turner's syndrome must have a karyotype performed. Probing for Sry chromosome materials as the presence of Y chromosome will cause the develpoment of gonadoblastoma is essential. Diagnosis of Turner's syndrome can be considered if there is a puberty delay or a growth failure [11] . Clinical findings such as high arched palate, multiple pigmented nevi, cubits valgus, low hairline, small mandible, low set ears, short stature, hyperconvex uplifted nails, otitis media chronic problems, pubertal arrest are of help in the diagnosis of Turner's syndrome.

Management :

A patient suffering from Turner's syndrome has many areas of concern and their management is of utmost importance in the aspect of improving their lifestyle, increasing their outcome rate and reaching their fullest potential. The cure for Turner's syndrome has not been discovered yet, but there are several ways of minimizing their pain and struggle and also increasing their lifespan by few weeks.

Growth

Women with Turner's syndrome have been found to be of short stature. This is due to the deletion of the homeobox transcription factor gene, SHOX [12] . The decrease in growth rate takes place as early as one and half years of age [1] . To treat this characteristic of short stature, these girls can be administered with Growth hormone and over time, there will be a significant alteration in the height of the patient. The duration of the administration of growth hormone will be the important variable which predicts the adult height of the patient [13] .

Behavioral Concern

In terms of social relationships, Turner's syndrome adolescents are more likely to fit the criteria for attention-deficit hyperactivity disorder than an adolescent girl who has no such genetic disorder or chronic illness. They are found to have fewer number of friends, poor concentration, increased anxiety and lower self esteem. Social difficulties are an area of vulnerability to patients who suffer from Turner's syndrome. To be able to treat these adolescents, there would be a need of counseling the individual and their family on the requirement of social skills and how it would be extremely useful in the social adaptation of the young patient [14] .

Musculoskeletal Defects

There is a prevalence of kyphosis and vertebral body wedging is increased in young girls suffering from Turner's syndrome. With age, there is progression in the level of kyphosis. these deformities can be found by a routine radiologic surveillance so that a treatment can be embraced to prevent or reduce the speed of the process [15] .

Cardiovascular Defects

Turner's syndrome is associated to cardiovascular defects such as bicuspid aortic valve and aortic coarctation, fetal lymphedema and aortic dissection. There is a pathogenetic connection between fetal lymphatic obstruction and defective aortic development which has been suggested by the presence of bicuspid aortic valve and aortic coarctation. Central fetal lymphedema is signified by the presence of webbing of the neck. These should be examined by magnetic resonance imaging, 24-hour ambulatory blood pressure, electrocardiogram and echocardiography.

Dermatological Concern

Turner's syndrome women who are in their late 30s and early 40s have premature wrinkling of the facial skin. Such wrinkling is seen in smokers. In patients of Turner' syndrome, it is not in relation to smoking or excessive exposure to the sun. There is a more apparent risk of keloid formation as the neck and upper chest regions are more likely to have such types of scarring. The congenital puffiness of the hands and feet take several years to be resolved [1] .

Ophthalmological Concern
Strabismus, amblyopia and ptosis are a few conditions faced by the patients inn regard to their vision. They also face visual-spatial arrangement problems.

Ear problems
The fourth most major problems associated with Turner's syndrome is hearing impairment. Majority of the women diagnosed with Turner's syndrome suffer from otitis media. A senorineural dip in hearing can be experienced at 6 years of age of the Turner's syndrome girl. These problems can be detected by audiometric and karyotype tests and also by the immunohistochemical staining of the inner ear specimens using antibodies against estrogen receptors in the fetus as well as middle aged women [16] .

Gastrointestinal
It has been discovered that there is an increased concentration of liver enzymes in patients suffering from Turner's syndrome, especially the Y-glutamyl transferase, alkaline phosphatase and alanine or aspartase aminotransferase [17] . Marked multiple focal nodular hyperplasia, nodular regenerative hyperplasia, obliterative portal venopathy, portal fibrosis, inflammatory bowel disease, non-alcoholic fatty liver disease and inflammatory infiltrates are shown in patients with Turner's syndrome.

Reproductive
95-98% of the women with Turner's syndrome are infertile due to the accelerated loss of oocytes over the first few postnatal years of months or after the 18th week of fetal life from the ovaries [18] . Oocyte donation has now changed the infertile status of women affected by Turner's syndrome [19] . Although there has been such a breakthrough in treating the case of infertility in Turner's syndrome patients by oocyte donation, the maternal mortality rate is high and are placed in high risk of death by aortic rupture or dissection during their course of pregnancy [20] .

Life Expectancy :
Due to the cardiovascular malformations, Turner's syndrome takes an important role in the three-fold overall mortality increase [21] . Most are able to lead relatively normal, healthy lives although their lifespans are reduced by 10 years than those who don't suffer this condition.

III. CONCLUSION :

Most of the patients of turner's syndrome are diagnosed at birth due to the presence of a lymphedema. The three most consistently seen features in the patient are streak gonads, utero lymphedema and short stature. The feature of short stature is invariable. It is highly impossible to predict the specific outcomes which the girl will face. The family of the patient and the patient should be counseled as there are many factors which may spoil the social interactions, confidence and self esteem of the person and stop her from achieving the maximum of her abilities. There is a need for comprehensive are in the case of patients diagnosed with Turner's syndrome from a multidisciplinary team. Although the knowledge regarding the treatment of Turner's syndrome has not been discovered yet, the knowledge on the methods of reducing the symptoms of the syndrome is vast. Care has to be taken in the administration of oestrogen. Natural oestrogens would be preferred in the administration. There will be a need for centralization of clinical care as the patient will be required to be administered over by a significant number of specialities such as otorhinology, cardiology, gynecology, gastro-enterology and ophthalmology.

IV. REFERENCES :

[1] Sybert VP, McCauley E. Turner's syndrome. New England Journal of Medicine. 2004 Sep 16;351(12):1227-38.
[2] Sybert VP. Cardiovascular malformations and complications in Turner syndrome. Pediatrics. 1998 Jan 1;101(1):e11-.
[3] Ellison JW, Wardak Z, Young MF, Robey PG, Laig-Webster M, Chiong W. PHOG, a candidate gene for involvement in the short stature of Turner syndrome. Human molecular genetics. 1997 Aug 1;6(8):1341-7.
[4] Clement-Jones M, Schiller S, Rao E, Blaschke RJ, Zuniga A, Zeller R, Robson SC, Binder G, Glass I, Strachan T, Lindsay S. The short stature homeobox gene SHOX is involved in skeletal abnormalities in Turner syndrome. Human molecular genetics. 2000 Mar 22;9(5):695-702.
[5] Lin AE, Lippe B, Rosenfeld RG. Further delineation of aortic dilation, dissection, and rupture in patients with Turner syndrome. Pediatrics. 1998 Jul 1;102(1):e12-.

[6] Ho VB, Bakalov VK, Cooley M, Van PL, Hood MN, Burklow TR, Bondy CA. Major vascular anomalies in Turner syndrome prevalence and magnetic resonance angiographic features. Circulation. 2004 Sep 21;110(12):1694-700.
[7] Nielsen J, Wohlert M. Chromosome abnormalities found among 34910 newborn children: results from a 13-year incidence study in Århus, Denmark. Human genetics. 1991 Jan 1;87(1):81-3.
[8] Jacobs PA, Hassold TJ. The origin of numerical chromosome abnormalities. Advances in genetics. 1994 Dec;33:101-33.
[9] Lange J, Skaletsky H, van Daalen SK, Embry SL, Korver CM, Brown LG, Oates RD, Silber S, Repping S, Page DC. Isodicentric Y chromosomes and sex disorders as byproducts of homologous recombination that maintains palindromes. Cell. 2009 Sep 4;138(5):855-69.
[10] Author: Maala S Daniel, MBBS; Chief Editor: Luis O Rohena - Turner syndrome
[11] Saenger P, Wikland KA, Conway GS, Davenport M, Gravholt CH, Hintz R, Hovatta O, Hultcrantz M, Landin-Wilhelmsen K, Lin A, Lippe B. Recommendations for the Diagnosis and Management of Turner Syndrome 1. The Journal of Clinical Endocrinology & Metabolism. 2001 Jul 1;86(7):3061-9.
[12] Chen J, Wildhardt G, Zhong Z, Roeth R, Weiss B, Steinberger D, Decker J, Blum WF, Rappold G. Enhancer deletions of the SHOX gene as a frequent cause of short stature: the essential role of a 250 kb downstream regulatory domain. Journal of medical genetics. 2009 Dec 1;46(12):834-9.
[13] Plotnick L, Attie KM, Blethen SL, Sy JP. Growth hormone treatment of girls with Turner syndrome: the National Cooperative Growth Study experience. Pediatrics. 1998 Aug 1;102(Supplement 3):479-81.
[14] Mccauley E, Feuillan P, Kushner H, Ross JL. Psychosocial development in adolescents with Turner syndrome. Journal of Developmental & Behavioral Pediatrics. 2001 Dec 1;22(6):360-5.
[15] Elder DA, Roper MG, Henderson RC, Davenport ML. Kyphosis in a turner syndrome population. Pediatrics. 2002 Jun 1;109(6):e93-.
[16] Hultcrantz M. Ear and hearing problems in Turner's syndrome. Acta oto-laryngologica. 2003 Feb 1;123(2):253-7.
[17] Hjerrild BE, Mortensen KH, Gravholt CH. Turner syndrome and clinical treatment. British Medical Bulletin. 2008 Jun 1;86(1):77-93.
[18] Foudila T, Söderström-Anttila V, Hovatta O. Turner's syndrome and pregnancies after oocyte donation. Human reproduction. 1999 Feb 1;14(2):532-5.
[19] Bodri D, Vernaeve V, Figueras F, Vidal R, Guillen JJ, Coll O. Oocyte donation in patients with Turner's syndrome: a successful technique but with an accompanying high risk of hypertensive disorders during pregnancy. Human reproduction. 2006 Mar 1;21(3):829-32.
[20] Karnis MF, Zimon AE, Lalwani SI, Timmreck LS, Klipstein S, Reindollar RH. Risk of death in pregnancy achieved through oocyte donation in patients with Turner syndrome: a national survey. Fertility and sterility. 2003 Sep 30;80(3):498-501.
[21] Price WH, Clayton JF, Collyer S, De Mey R, Wilson J. Mortality ratios, life expectancy, and causes of death in patients with Turner's syndrome. Journal of Epidemiology and Community Health. 1986 Jun 1;40(2):97-102.

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