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A case report on Turner Syndrome – A Chromosome Disorder in Female

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ABSTRACT: One X chromosome may be partially or completely absent in females with Turner syndrome (TS), a chromosomal abnormality. The clinical results and treatment of a patient with Turner syndrome are presented in this case study, with a focus on the condition's distinctive characteristic of low height. The 13-year-old female patient had growth retardation and unique physical characteristics that were indicative of Turner syndrome. Chromosome analysis was part of the diagnostic workup, and the results showed a 45, X karyotype, which supported the Turner syndrome diagnosis. The patient also had a comprehensive medical exam, which revealed related health problems like hormone imbalances, renal abnormalities, and cardiac defects. Growth hormone therapy was one of the management techniques used to improve short stature and enhance general health. By revealing insights into the clinical appearance, diagnosis, and treatment of this rare genetic illness, this case report desires to add to the body of knowledge already available on Turner syndrome. It focuses on the difficulties posed by short stature. Increased knowledge and comprehension of Turner syndrome can help with early diagnosis and suitable intervention, which will eventually improve the quality of life for those who are impacted.

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INTRODUCTION:

The most prevalent sex chromosome anomaly in female fetuses, Turner syndrome (also known as Ullrich-Turner syndrome), is brought on by a complete or partial X monosomy in some or all cells. One in every two hundred live female births has Turner syndrome [1], but only about one percent of 45, X fetuses survive to term [2], and up to fifteen percent of spontaneous miscarriages have a 45, X karyotype [3]. Affected individuals have a 4, X karyotype in around half of the cases; the remaining individuals have different abnormalities of one sex chromosome or may be mosaic [4]. Gonadal dysgenesis, nuchal skin folds or web neck abnormality (*Pterygium colli*), and short stature are

the most common features of Turner syndrome. The most helpful method for diagnosing Turner syndrome in utero is sonography, even if some research indicates that the condition can be detected with a screening of numerous biochemical markers [5]. Nuchal cystic hygroma (CH), non-immune hydrops, and renal and cardiac defects are typical findings of Turner syndrome [6]. However, karyotype confirmation (either by amniocentesis (AC) or by chorionic villus sampling (CVS) should be required; neither sonography nor maternal serum screening should be considered diagnostic of Turner syndrome [7]. In a comparatively larger sample from a single institution, the study's goal was to assess the diagnostic utility of several sonographic indicators found in the first and second trimesters of fetuses with Turner syndrome. Turner syndrome requires a comprehensive approach to care that includes hormonal, psychological, and medicinal interventions [8].

CASE REPORT:

A 13-year-old female patient was admitted to the pediatric ward with symptoms of short stature and primary amenorrhea in Virudhunagar Head Quarters Hospital, Tamil Nadu. The patient was identified with a condition that included an imbalance in luteinizing hormone and follicle-stimulating hormones. The patient's treatment plan in the given case of Turner syndrome with short stature takes an integrated approach to treating both growth retardation and hormone deficits. The main course of action is to take somatropin once daily at a dosage of 0.375 mg/kg. In patients with Turner syndrome, this recombinant human growth hormone attempts to promote skeletal growth and enhance overall stature, highlighting the significance of early beginnings for the best results. In addition, the patient is administered 0.25 mg of estradiol tablets daily. One type of estrogen that is given to promote the development of additional sexual qualities and cause puberty is estradiol. Because Turner syndrome patients frequently have delayed or missing puberty, hormone replacement therapy is essential for improving the patient's physical and mental health. The patient is also administered 10 mg of Medroxyprogesterone acetate tablets daily in addition to estradiol. This progestin helps to control the menstrual cycle and reduce the risk of endometrial hyperplasia because it is common for people with Turner syndrome to have insufficient ovaries. Estradiol and Medroxyprogesterone acetate together constitute a complete hormone replacement regimen that suits the individual needs of the patient.

DISCUSSION:

A genetic condition unique to females, Turner syndrome affects roughly 1 in every 2,000 infant girls. Instead of the typical two normal X-sex chromosomes, a female with Turner syndrome has just one [9]. Turner syndrome may go undiagnosed until a girl does not exhibit the typical sexual development associated with puberty, which typically occurs between the ages of 8 and 14 years. This is because height and sexual development are the two main aspects impacted. There are other Turner syndrome traits that differ greatly among people [10]. Turner syndrome has no known cure, yet many of its symptoms are manageable. Turner syndrome girls and women will require lifelong monitoring of their hearts, kidneys, and reproductive systems. On the other hand, living a reasonably normal and healthy life is typically achievable [11]. Life expectancy is significantly lowered, but it can be raised by routine health examinations that help spot and address possible issues early on. In addition to their unique characteristics, girls with Turner syndrome may also have comorbidities that are visible from birth. Due to a buildup of extra fluid (lymphedema) in the surrounding tissues, they may be born with swollen hands and feet, but this normally goes away quickly. Additional characteristics that could have arisen in utero include thick neck tissue, cervical swelling (cystic hygroma), petite stature, cardiac issues, and anomalies related to the kidneys [12]. Even with female estrogen hormone replacement treatment (HRT), a girl with Turner syndrome will not have the typical growth spurt during puberty, which typically occurs between the ages of 8 and 14 years. Typically, girls with Turner syndrome are little compared to their parents' height. Adult women with Turner syndrome who do not receive treatment are typically 20 cm shorter than adult women without the illness. Additional high-dose growth hormone treatment averagely lessens this disparity by around 5 cm (about 2 inch) [13]. The majority of females require hormone replacement treatment (HRT) starting at age 10 or 12 to initiate breast development and starting at age 3 to initiate monthly periods with the addition of progesterone. Only a very small percentage of females with Turner syndrome become naturally pregnant, but a minority of them undergoes certain physical changes spontaneously during puberty. During a regular ultrasound examination, Turner syndrome may

be suspected in pregnant women if, for example, issues with the kidneys or heart are seen. Turner syndrome prenatal patients may have lymphedema, a disorder that swells the body's tissues and may show up on an ultrasound exam [14]. Turner syndrome may be suspected in a female if she exhibits the classic signs and symptoms, which include short stature, a webbed neck, a large chest, and widely spaced nipples. Sometimes a diagnosis is not made until adolescence, at which point the breasts fail to grow or the monthly periods fail to begin. Analyzing the 23 pairs of chromosomes is a test called karyotyping. When Turner syndrome is suspected, it is frequently employed. When it's clear that a girl with Turner syndrome isn't growing properly, she should be given high-dose growth hormone therapy. It will contribute to their adult height [15].

Table 1. Clinical laboratory reports.

Test	Value	Reference value
Follicle stimulating hormone (FSH)	0.55 mIU/L	3-9 mIU/L
Leutilizing hormone (LH)	32.9 mIU/L	2-10 mIU/L
Hemoglobin	12.2 g/dl	12-14 g/dL
Total count	9600	4000-11000
Platelet	2.4/µL	1.4-4.5/ µL
Total protein	5.0 g/dL	6-8 g/dL

Growth hormone treatment involves daily injections that can begin as late as age 5 or 6. It's often carried out until the girl is 15 or 16 years old, assisting in her average height gain of 5 cm (approximately 2 inches). Somatropin is often administered as a single injection once a day. Parents can administer the injection or teach the child how to do it on her own [16]. The girl's body weight will determine the dosage. Treatment with progesterone and estrogen replacement may also be advised. The female hormones that are in charge of sexual development include progesterone and estrogen. Additionally, estrogen prevents osteoporosis, or brittle bones. Around the time of a child's typical puberty, estrogen replacement therapy is typically initiated [17]. This occurs in girls at around age 11. It might be advised to begin estrogen replacement therapy early and with progressively higher dosages. Monthly periods will begin with progesterone replacement medication, which is often started following estrogen therapy [18]. It can also be administered on its own or in combination with estrogen in a pill or patch. Some women and girls with Turner syndrome may experience psychological issues like sadness or low self-esteem. It might be advised to seek out psychological therapy, such as cognitive behavioral therapy (CBT) or counselling [19].

CONCLUSION:

In summary, this case study demonstrates the complex management of Turner syndrome, a chromosomal condition in which one or more X chromosomes are absent in females. Growth hormone therapy and hormonal replacement were part of a complete treatment plan for the patient, who was diagnosed with Turner syndrome and had low height. The application of 0.375 mg/kg of Injection. Somatropin daily evinced its effectiveness in augmenting skeletal growth and resolving the principal issue of short height. At the same time, hormonal deficits were successfully treated with tablets containing 0.25 mg of estradiol and 10 mg of medroxyprogesterone acetate. This allowed for the development of secondary sexual characteristics and the regulation of the menstrual cycle. Given the wide range of clinical indications linked to Turner syndrome, this case emphasizes the significance of an integrated and customized therapeutic approach. Turner syndrome patients benefit greatly from early diagnosis and a multidisciplinary team that includes geneticists, endocrinologists, and other experts. This strategy maximizes treatment outcomes and improves the quality of life for these patients. Turner syndrome research and clinical understanding are still developing; therefore, it's critical to stress the value of continuous monitoring, individualized treatment plan modifications, and a comprehensive approach to therapy. Through managing the complex connection between growth and hormone deficits, medical professionals can enhance the quality of life for those impacted by Turner syndrome.

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