**Summary**

Humans have 46 chromosomes. Two of them sex: the X and Y. Women have two X chromosomes (one from the father and one from the mother). Men have one X chromosome (the mother) and another Y (his father). In the early stages of cell division, division makes a wrong part or all of the X chromosome. Most often lost is the X chromosome. The child has Turner syndrome (TS). This does not occur in children who have only one X chromosome and if needed, could not live. It is unknown what circumstances influence abnormal division. It occurs, most often chromosome lost to the father. The diagnosis of TS requires a phenotypic characteristic with full or partial absence of one X chromosome, as a result of cell division and mosaicism. The ST prevalence has 1/2 000 female births. Another third, in childhood, stunting and pubertal development of these patients. In the period of 11 years adult: Monitoring growth and development. There are associated comorbidities, to achieve adequate quality of life.

**Keywords:** Turner syndrome, short stature, chromosome X

**Introduction**

Humans have 46 chromosomes in the cells. Two of them are called sex: the X and Y. Women have two X chromosomes (one from the father and one from the mother). Men have an X chromosome (the mother) and another Y (his father). In the early stages of cell division, division makes a wrong part or all of the X chromosome is lost if the pregnancy continues, the child will have Turner syndrome (TS). This does not occur in children who have only one X chromosome and if needed, could not live. It is unknown what circumstances influence abnormal division to occur, most often chromosome lost to the father. The cause is not known. One ST diagnosis requires combining certain phenotypic characteristics with a total or partial absence of an X chromosome, as well as regulating cell line or mosaicism. It is one of the most common monosomy, prevalence of 1/2 000 1/5 000 female births. It is characterized by short stature and gonadal dysgenesis. One-third are recognized at birth by lymphedema, redundant skin or webbed neck. Another third, in childhood, stunting and pubertal development of these patients. In the period of 11 years adult: Monitoring growth and development. There are associated comorbidities, to achieve adequate quality of life.

It is associated with inflammatory bowel disease rectum, ulcerative colitis or Crohn’s disease, and colon cancer. Gastrointestinal bleeding intestinal telangiectasias. Autoimmune thyroiditis; Graves’ disease, especially if they have isochromosome X. They have a tendency to obesity and carbohydrate intolerance and type 2 increases with age, hypertriglyceridemia which is related to obesity and insulin resistance diabetes. 35% have a cardiovascular malformation: more frequent in patients bicuspid aortic valve 45 (50%), aortic coarctation (15-20%), aortic valve stenosis and hypoplastic left ventricle. X. have a higher blood pressure, to 50% may have clinical evident hypertension in adolescence.

There dilated aortic root asymptomatic to 42%, not all end in aortic dissection and rupture, with aortic valve disease, malformations of the left chambers and karyotype 45, X increases risk. Cardiovascular morbidity and mortality is increased. It must be included in monitoring cardiovascular monitoring and follow up in patients at risk. After either suspected or confirmed, apply a protocol detection, monitoring and treatment of various associated comorbidities, to achieve adequate quality of life of these patients. In the period of 11 years adult: Monitoring growth and development. The initiation of hormone replacement therapy upon detection of hypogonadism should be considered. It is important the evaluation of school and social adaptation (Figure 1).
Clinical case

Female 20 years, genetic load for DM and have product of g3 uncomplicated, mother of 23 years. Normal psychomotor development and IQ. Short stature, Without development of secondary sexual characteristics, with f 130/80 primaria. EFTA amenorrhea, c.80 x min. Height: 132cm Weight: 38kg, conjunctive with good color, epicanto, arched palate, Wide neck, short, low hairline, broad chest, breast hypertelorism, absence of secondary sexual characteristics. (Figures 2–4).

Pelvic ultrasound, uterus and left ovary small. Ovary Uterus 3.2x1x0.9 left. 1.6x1x1 absent law. TSH 9.25, T4T 8.76, 1.25 FT4, TT3 1.28, FT3 3.46 FSH 97.34. 26.82LH, Estradiol <5, Progesterone <0.03. Prolactin 6.03. Testosterone <0.025 Normal renal ultrasound Bone age 15 years. Levothyroxine and estrogen treatment is started in March 2014.

Figure 1 Multidisciplinary follow-up of turner syndrome.

Figure 2 Rhythmic heart sounds with aortic expulsivo, and 2 fixed and split noise. Karyotype 46, X.i (Xq).
Turner syndrome case presentation

Abstract

Turner syndrome is a genetic disorder characterized by the absence of one X chromosome, usually X0. It is characterized by physical features such as short stature, primary amenorrhea, and cardiovascular anomalies. Early diagnosis is crucial, as it allows for timely intervention and management of associated comorbidities. Treatment strategies include growth hormone therapy and hormone replacement therapy with estrogen and progestin.

Introduction

Turner syndrome is a genetic disorder affecting approximately 1 in 1000 live female births. It is characterized by the absence of one X chromosome, leading to clinical features such as short stature, primary amenorrhea, and cardiovascular anomalies. Early diagnosis is essential for optimal management and prognosis. This case presentation discusses a patient with Turner syndrome, highlighting the importance of early diagnosis and multidisciplinary care.

Case Presentation

A 15-year-old girl presented with short stature and primary amenorrhea. Physical examination revealed characteristics typical of Turner syndrome, including webbed neck, low posterior hairline, and shield chest. Laboratory testing confirmed the absence of one X chromosome, consistent with Turner syndrome.

Diagnostic Evaluation

Diagnostic evaluation included a comprehensive history and physical examination, genetic testing, and echocardiography. Genetic testing confirmed the diagnosis of Turner syndrome, and echocardiography demonstrated a bicuspid aortic valve.

Treatment

The patient was initiated on growth hormone therapy to promote linear growth and estrogen therapy to manage secondary sexual development and prevent osteoporosis. Multidisciplinary follow-up was scheduled to monitor for complications and adjust treatment as needed.

Discussion

Early diagnosis of Turner syndrome is crucial for optimal management and prognosis. The multidisciplinary approach to care, including genetic counseling, growth hormone therapy, and hormone replacement therapy, is essential for improving outcomes. Further research is needed to better understand the long-term effects of Turner syndrome and improve management strategies.

Conclusion

Early diagnosis of Turner syndrome is essential. It allows for timely intervention and management of associated comorbidities. A multidisciplinary approach to care, including genetic counseling, growth hormone therapy, and hormone replacement therapy, is crucial for improving outcomes. This case presentation highlights the importance of early diagnosis and multidisciplinary care in the management of Turner syndrome.

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None.

Conflict of interest

The author declares that there is no conflict of interest.

References


Figure 3 Electrocardiograma normal.

Figure 4 Ecott vaqivla bicuspid aortic valvular stenosis area light L.70 cm².

Conclusion

Early diagnosis is essential: the suspicion is based on clinical signs (physical appearance), but require cariotipo. Una after carrying out genetic diagnosis, detect associated comorbidities. Track for early diagnosis of complications and institute treatment. multidisciplinary monitoring. Full information to parents as soon as possible and the child by his parents, as requested and appropriate content to your age. Short stature, lack of secondary sexual characteristics and infertility condition the lives of patients. It is essential to optimize GH therapy and hormone replacement therapy with estrogen and progestin.8,10

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References


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