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A Case Report of Turner Syndrome and Literature Review

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Abstract: Turner syndrome (TS), also known as congenital dysplasia of ovary syndrome, is the most common female chromosomal abnormality, and the incidence of live birth girls is 1/2 000 ~ 1/2 500. The main clinical manifestations of Turner syndrome are spinal trunk abnormality, growth retardation, sexual dysplasia and special physical characteristics, and abnormal sex hormone, abnormal growth hormone, dyslipidemia, etc. This article summarized and analyzed the clinical characteristics and test results of a child with Turner syndrome, combined with literature review, in order to improve the understanding of Turner syndrome and clinical diagnosis and treatment.

Keywords: Turner Syndrome; Clinical Features; Sex Hormones; Lipids.

1. Introduction

Turner syndrome (TS), also known as congenital dysplasia of ovary syndrome, is the most common female chromosomal abnormality, with an incidence of 1/2,000 ~ 1/2,500 live born girls [1]. It is caused by the complete or partial deletion of an X chromosome in all or part of somatic cells, or other structural abnormalities of the X chromosome, and is one of the common human chromosome diseases. The main clinical manifestations of Turner syndrome are growth retardation, sexual dysplasia and special physical features. This article summarized and analyzed the clinical characteristics and test results of a child with Turner syndrome, combined with literature review, in order to improve the understanding of Turner syndrome and clinical diagnosis and treatment.

2. Clinical Data

Patient, male, 13 years old, chronic course. History of present disease: More than 3 years ago, the patient's family found that the patient had unequal shoulder height, manifested as right-side elevation, left lower limb lameness, and no abnormal hair or pigmentation spots on the back skin. The patient was diagnosed with "scoliosis" in another hospital, and no special treatment was given. Two years ago, the patient's family found that the right back was raised and showed progressive aggravation, walking about 1000 meters, the lumbar acid discomfort, can continue to walk after sitting down and rest, have been reviewed in another hospital for three times, found that the scoliosis slowly aggravated, suggested surgery treatment, but limited to local medical conditions, the patient had concerns about surgery, no treatment. Now for further surgical treatment, outpatient "scoliosis" into my department. The patient had no special discomfort such as low fever, night sweats, fatigue and poor appetite in the recent period. Her spirit, sleep and diet were good, her bowel movements were normal, and her weight did not increase or decrease significantly.

Previous history: Normal health condition, denied hypertension, diabetes, coronary heart disease. Denied the history of hepatitis, tuberculosis and other infectious diseases, at the age of 3 due to "congenital dislocation of the right hip" surgery treatment; In late 2020, he was diagnosed with tethered cord syndrome in the Children's Hospital Affiliated to Soochow University, and underwent surgery in early 2021, with no significant lumbar acid distress after surgery. Deny the history of trauma, blood transfusion, food and drug allergies. Vaccination history is unknown.

Specialist physical examination: Height 145.8cm, (-2SD) BMI 22.1kg/m2, finger spacing 146cm, no neck webbing, no hairline, male juvenile genitalia, Adam's apple protrudation was not obvious, beard, pubic hair, armpit hair did not grow. Short stature, breast development, secondary sexual characteristics did not develop.

Imaging examination: No obvious abnormalities were found in the thyroid gland and pituitary gland. The bone age of the left wrist was consistent with the bone age of a 14-year-old boy. Double adrenal ultrasonography showed hypoechoic nodules in the right adrenal area. Ultrasonography in bilateral mammary glands showed no obvious abnormalities in the general structure, blood flow and function of the heart, and the physical examination showed breast development and no development of secondary sexual characteristics.

According to the test results, coagulation, electrolyte, kidney function, uric acid, lipid, glucose, liver function, eight items before surgery and stool routine were basically no special, alpha-fetoprotein was 6.540ng/mL; Carcinoembryonic antigen :1.110ng/mL; Adrenocorticotropin (ACTH)17.22pmol/L↑; Cortisol (COR) 486.30nmol/L; Insulin-like growth factor (IGF-1)111.00ng/mL; Parathyroid hormone (PTH) 2.96pmol/L; Six elements in whole blood: no obvious abnormality; 25 hydroxyvitamin D:25 hydroxyvitamin D(Vit-D)14.7ng/mL↓, 25 hydroxyvitamin D3(Vit-D3)14.7ng/mL, 25 hydroxyvitamin D2(VitD-2)<2.2ng/mL. Six sex hormones: six sex hormones: estradiol <18.35pmol/L, progesterone 1.630nmolL, luteinizing hormone <0.10IU/L, testosterone 0.322nmol/L. Estradiol, luteinizing hormone, testosterone decreased, progesterone increased, supplementary diagnosis: genetic metabolic disease to be eliminated, it is recommended to improve genetic testing. The sex hormone results are shown in Table 1. The growth hormone stimulation test of the child showed that the peak value of growth hormone was less than 10ng/ml. Combined with the medical history and physical examination, the supplementary diagnosis was: growth hormone deficiency. The results of growth hormone

stimulation test are shown in Table 2. There was no abnormality in blood lipid.

The karyotype analysis result of peripheral blood chromosome examination was 45, and X was consistent with the karyotype of Turner syndrome, so it was diagnosed as Turner syndrome.

Table 1. Sex hormone results

Test item	result	reference interval unit
Estradiol	<18.35 ↓	94.8-223 (pmol/L)
Progesterone	1.630 ↑	<0.474(nmol/L)
Follicle stimulating hormone	2.51	1.5-12.4(IU/L)
Luteinizing hormone	<0.10 ↓	1.7-8.6(IU/L)
Prolactin	210.70	86.0-324.0(ulU/ml)
Testosterone	0.322↓	9.9-27.8(nmol/L)

Table 2. Results of growth hormone stimulation test

Time of administration	Growth hormone (ng/mL)	Growth hormone (ng/mL)
	The amino	acid L-dopa
Before medication	0.04	0.04
30 minutes of medication	0.81	2.37
60 minutes of medication	0.28	6.20
90 minutes of medication	0.14	3.43

3. Discussion

The characteristic signs of Turner syndrome are short stature, gonadal dysplasia and neck webbing. The growth and development of this child has been delayed for many years, which is rare in the spinal bone disease department but common in the endocrinology department. It needs to be distinguished from the following diseases in clinical practice: 1) endocrine diseases, such as hypothyroidism, growth hormone deficiency, precocious puberty, Cushing's syndrome in children, congenital adrenal hyperplasia, etc. 2) Metabolic diseases, including carbohydrate metabolic diseases such as mucopolysaccharidosis and amino acid metabolic diseases; 3) Chronic systemic diseases, such as chronic diseases of respiratory, circulatory, digestive and blood systems; 4) chondrodysplasia; 5) Nutritional disorders, such as undernutrition or deficiency, malabsorption syndrome, etc.; 6) neurological disorders, such as brain hypoplasia or retardation; 7) Delayed puberty. The patient was short in stature, physical examination showed breast development, abnormal sex hormone, growth hormone deficiency, imaging examination, peripheral blood chromosome karyotype analysis and a series of examinations confirmed Turner syndrome.

The diagnosis of Turner syndrome mainly relies on clinical features and chromosome karyotype detection in peripheral blood. Currently, a variety of abnormal chromosome karyotypes have been found, mainly including chromosome haplotype 45, XO accounting for about 50%, chimeric body type and X chromosome structural abnormalities [2]. The child in this study is a typical 45, XO haplotype. Due to the different purposes of detection and detection specimens, chromosome karyotype analysis can be roughly divided into peripheral blood chromosome karyotype analysis, neonatal umbilical cord blood chromosome karyotype analysis, amniotic fluid chromosome karyotype analysis, villus chromosome karyotype analysis, bone marrow blood chromosome karyotype analysis, pleuroabdominal fluid chromosome karyotype analysis. This child was diagnosed by peripheral blood chromosome karyotype analysis. Peripheral blood chromosome karyotype analysis and gene detection are the gold standard for the diagnosis of Turner syndrome.

In this study, the patient is a 45 XO chromosome haplotype with a male clinical phenotype, which is extremely rare. Most 45 XO patients present with Turner syndrome in females, but there are very few reports of 45 XO males. A small number of patients with TS have Y chromosome material present or have chromosomal markers (derived from fragments of the X or Y chromosomes) that cause the phenotype to be male. This child may have Y chromosome material, which requires further chromosome microarray analysis and SRY gene detection. However, because the child did not continue to be treated in our hospital, no further tests were conducted. There is a strong correlation between virilization of TS children and gonad blastoma and Y chromosome material.

The main clinical manifestations of the children in this study were: short stature, left lower limb limping, trunk slightly tilted to the left, shoulders unequal height, scoliosis, breast development, etc. The main features of the patients are usually short stature, short mandible and retraction, large ears and low position, short neck, low posterior hairline, undulating neck, widening of breast spacing, breast and uterine dysplasia, ovaries without follicles, primary amenorrhea, and sterility. Cubitus valgus, some mild intellectual impairment. Short stature is the main clinical presentation in TS patients. In addition to short stature, TS patients are often accompanied by gonadal dysplasia, and most TS patients require estrogen replacement therapy to induce puberty and maintain subsequent normal uterine development. In this child, physical examination showed breast development, abnormal sex hormone results, decreased estradiol, luteinizing hormone, testosterone, and elevated progesterone. Abnormal growth hormone stimulation test indicated that the child was deficient in growth hormone. Abnormal sex hormone and growth hormone in this patient may be due to the haplotype of the 45 XO chromosome and the lack of an X sex chromosome, resulting in abnormal sex hormone and hormone in the child. Because there are genes regulating gonad development on the X sex chromosome, the abnormal development of gonad will lead to abnormal clinical

manifestations in the child. As the patient in this paper was not treated in our hospital, the effect of follow-up treatment could not be tracked.

Some studies have shown that the incidence of lipid metabolism disorders and dyslipidemia in TS patients is increased, mainly as shown by the increased TG level in Turner syndrome patients aged 11-15 years compared with age-matched control group, and the incidence of high TG and critical high TG are also increased [4]. Foreign studies have found that the total cholesterol (TC) of 11–15-year-old TS patients without any drug treatment was higher than that of the age-matched normal control group, and the cholesterol (TC) in TS patients was significantly affected by age, body mass index, body mass index, and body mass index. BMI) is associated [3]. The possible relationship between dyslipidemia in TS patients and insufficient estrogen level and chromosome karyotype in TS patients during adolescence has been prospectively studied [4]. Is dyslipidemia in Turner syndrome due to insufficient estrogen secretion, or is it the result of abnormal sex chromosome karyotype? In order to determine the mechanism for further study, Ostberg et al., Van et al., selected women with premature ovarian failure as the control group in order to exclude the influence of estrogen on lipids, and found that TG was still elevated in TS patients, and Turner syndrome patients had a complete or partial deletion of an X chromosome gene quantity in all or part of somatic cells. It is speculated that in addition to estrogen effects, the number of genes lost or defective on the X chromosome or functional changes in gene imprinting may affect lipid metabolism. However, the blood lipid of the child in this study was not abnormal, which may be affected by diet, etc. It was not possible to determine whether the child was undergoing hormone therapy, and the blood lipid was not continuously monitored, only four blood lipid tests were performed once, and eight blood lipid tests were not performed.

To sum up, Turner syndrome is rare among inpatients in the spine osteopathy department, which reminds doctors that congenital chromosomal abnormalities, a rare type of disease, must not be missed for children with clinical spinal malformations, such as intelligence abnormalities, secondary gland dysplasia, growth retardation or other malformations. Turner syndrome patients can be treated with hormone replacement therapy, if necessary, the purpose of treatment is to improve adult height, promote the development of secondary sexual characteristics, and prevent multiple complications. If early detection, early diagnosis and early intervention can improve the clinical phenotype of patients and improve the subsequent quality of life.

References

[1] Yan Chun, Wang Muti. Pediatric Endocrinology [M]. Beijing: People's Medical Publishing House, 2006:348-355.

[2] Yesilkaya E, Bereket A, Darendeliler F, Bas F, et al. Turner syndrome and associated problems in Turkish children: a multicenter study [J]. J Clin Res Pediatr Endocrinol. 2015 Mar; 7 (1): 27-36.

[3] Ross JL, Feuillan P, Long LM, et al. Lipid abnor-malities in Turner syndrome [J]. J Pediatr, 1995,126 (2): 242-245.

[4] Huang Siqi, Chen Hongshan, Du Minlian et al. Analysis of dyslipidemia and related factors in Chinese children and adolescents with Turner syndrome [J]. Journal of Sun Yat-sen University (Med Edition),2018,39(03):420-426. (in Chinese)

[5] Yan Kunli. Analysis of chromosome karyotype distribution and lipid level in children with Turner syndrome [J/OL]. Zhejiang University, 2015: 1-17. Yan KL. The study on the chromosomal karyotypes distribbution and lipid concentrations in children with Turner syndrome [J/OL]. Zhejiang Univ, 2015:1-17.

[6] Qin Shuang, Luo Songping, Ju Rui. China expert consensus Turner syndrome (2022 edition) [J]. Chinese journal of practical gynecology and obstetrics, 2022, 38 (4): 424-433. The DOI: 10.19538 / j. f k2022040111.

[7] Zheng X M, Liu G L.45, abnormal sex differentiation induced by XO [J]. Chinese Journal of Practical Internal Medicine, 2004 (11):649-651. (in Chinese)

[8] LI P, CHENG F, XIU L. Height outcome of the recombinant human growth hormone treatment in Turner syndrome: a meta-analysis[J]. Endocr Connect, 2018, 7(4): 573-583.

[9] CARPINI S, CARVALHO A B, de LEMOS-MARINI S H V, et al. FSH may be a useful tool to allow early diagnosis of Turner syndrome[J]. BMC Endocr Disord, 2018, 18: 1186-1194.

[10] SHANKAR R K, BACKELJAUW P F. Current best practice in the management of Turner syndrome[J]. Ther Adv Endocrinol Metab, 2018, 9(1): 33-40.