

Caso Clínico/Case Report

Gravidez gemelar em mulher com Síndrome de Turner

Twin pregnancy in a woman with Turner's syndrome

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ABSTRACT

Hormone replacement therapy (HRT) is crucial for the development of normal female sexual characteristics and to prevent cardiovascular complications and osteoporosis in patients with Turner's syndrome. Fertility is extremely rare and only 2% of pregnancies are the result of spontaneous ovulation, as ovarian failure is a typical feature of the syndrome. Oocyte donation is currently seen as a treatment option for infertility in these women. The authors report the case of a 38 year-old woman with a 45, XO karyotype, submitted to ovum donation and intrauterine transfer of two embryos. A twin pregnancy was achieved with an unremarkable evolution. A cesarean section was performed near term, with the birth of two healthy boys.

Keywords: Turner syndrome; twin gestation; pregnancy; oocyte donation

INTRODUCTION

Turner's syndrome is one of the most common sex chromosome abnormalities. It is characterized by a complete or partial absence of one X chromosome and has typical phenotypic features. The most frequent chromosomal constitution is 45, XO¹. About half the patients have a mosaic chromosome complement, the most common being 45XO/46XX (15%), and 6% of the patients possess 46XXq or 46XXp deletions. Ovarian failure and amenorrhea are typical features of Turner's syndrome with 95-98% of the women being infertile due to gonadal dysgenesis²

The case of a twin pregnancy achieved after oocyte donation in a woman with Turner's syndrome is here reported.

CASE REPORT

A 38 year-old patient was referred to our institution for a pregnancy follow up. The patient suffered from gonadal dysgenesis with primary amenorrhea which motivated a karyotype analysis - a diagnosis of 45,XO Turner's syndrome was hence established. She started HRT at age of seventeen and years later a primary infertility was detected.

There were no descriptions of thyroid abnormalities, hypertension or known autoimmune diseases. The patient had some phenotypic features of Turner's syndrome, like short stature (1,49 m high), short neck, cubitus valgo, low posterior hairline and multiple pigmented nevi.

Turner's syndrome was diagnosed, revealing a 45 XO karyotype without evidence of mosaicism. The

patient became pregnant after an oocyte donation and intrauterine transfer of two embryos performed in another institution, resulting in a twin dichorionic diamniotic pregnancy. Her pregnancy follow-up in our institution started at the 12th gestational week. The twin pregnancy developed without complications with appropriate antenatal care. All ultrasound and Doppler fluxometry exams were normal, identifying a breech presentation in both fetuses at thirty five weeks. Therefore two healthy male newborns were delivered by cesarean section at thirty six weeks of pregnancy, weighting 2380 g and 2360 g. At the time of cesarean section no distinct evident ovarian tissue was found. The histological study of placenta revealed no abnormalities.

DISCUSSION

Ovarian failure is a common feature of Turner's syndrome, as there is an accelerated loss of oocytes after the 18th week of fetal life or over the first few postnatal months and years^{2,3}.

Ovaric tissue is composed of small amounts of connective tissue with follicles being absent or few and atretic; in the present case the existence of ovaries couldn't be diagnosed macroscopically. Amenorrhea and poor development of female sexual characteristics are indications for hormone replacement therapy (HRT), which contributes to the development of normal female sexual characteristics and prevents osteoporosis and cardiovascular complications.

The typical clinical features of Turner's syndrome are short stature, square appearance, webbed neck, low posterior hairline, broad chest with widely spaced nipples, cubitus valgus and multiple pigmented nevi. Patients have an elevated rate of renal and cardiovascular abnormalities. Cardiovascular anomalies occur in 10-40% of women with Turner's syndrome⁴.

Coarctation of the aorta and a bicuspid aortic valve are the most common malformations, with the dissection of aortic aneurisms being the most dangerous one with several fatal cases, mainly during pregnancy⁵. Hashimoto's thyroiditis; hypothyroidism, metabolic disorders such as glucose intolerance with mild insulin resistance are also common. As a consequence of

these abnormalities, patients with Turner's syndrome have a shorter life span than the general population. The present patient had few phenotypic characteristics of Turner's syndrome and presented no cardiovascular or renal abnormalities.

Natural pregnancies occur in at least 2% of women with Turner's syndrome². Most have a mosaic Turner's karyotype containing a 46 XX line. The rate of miscarriage, stillbirths, and abnormal babies in these pregnancies is very high^{2,6}. The reason for this poor prognosis is the diminished endometrial receptivity⁷.

In our case we couldn't find any signs of uterine hypoplasia or hypovascularization, which are frequent in women with this syndrome. Probably that is the reason why our patient supported a twin pregnancy without any sign of uterine failure.

Oocyte donation is now available as an option for infertility treatment in women with Turner's syndrome. It has been successfully used as a treatment for ovarian failure for more than two decades⁸. The implantation and pregnancy rates are similar to those cases with primary ovarian failure due to other causes². After oocyte donation the achieved pregnancies are at high risk, with an elevated miscarriage rate, which means that the cause cannot only be genetic². Additionally, there are higher rates of maternal morbidity and mortality due to hypertensive disorders, aortic rupture and dissection in oocyte donated pregnancies in women with Turner's syndrome^{5,9}.

Since a pregnancy in a Turner's syndrome patient is a high risk one, the selective transfer of one embryo should ideally be performed in order to avoid additional risks associated with multiple pregnancies¹⁰. In addition, because the implantation rate is high in these women, transfer of one embryo is acceptable and highly recommended³. This case appears to be one of the few twin pregnancies described that achieved thirty-six weeks of pregnancy without any maternal or perinatal complications. This fact contradicts the presence of a hypoplastic uterus and endometrial defect. Although there is a general consensus to avoid multiple pregnancies because of the increased risk of complications, we describe a case where the results were amazingly good.

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