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Successful Twin Pregnancy in A Woman with 46,XY Complete Gonadal Dysgenesis (Swyer Syndrome): A Rare Case Report

Dr. Shweta Patki[1], Dr. Aditi Kunte[2], Dr. Surhud Patki[3], Dr. Satish Manohar Patki[4], Dr Ujjwala Patki[5], Dr Shriswaroop Shriprakash Kulkarni[6]

[1,2,3,4,5] Consultant Obstetrician and Gynecologist, Patki Hospital
[6] Anesthesia, Patki Hospital

Abstract

46,XY complete gonadal dysgenesis, also known as Swyer syndrome, is a rare disorder of sex development characterized by female phenotype, presence of Müllerian structures, and non-functional streak gonads. These patients typically present with primary amenorrhea and require assisted reproductive techniques using donor gametes to achieve pregnancy. Reports of twin pregnancies in such women are exceedingly rare.

We report a 32-year-old phenotypic female with primary amenorrhea who was found to have a 46,XY karyotype. Pelvic imaging revealed a normally formed uterus and vagina with streak gonads. Diagnostic hysteroscopy confirmed a normal uterine cavity. Pregnancy was achieved through assisted reproduction using donor gametes. A twin intrauterine pregnancy was diagnosed at 6 weeks scan. Cervical length was found to be on the lower side at NT scan and prophylactic cervical cerclage was performed. At 24 weeks of gestation, the patient developed pregnancy-induced hypertension, which was managed successfully with oral labetalol. The pregnancy progressed without major complications, and an elective cesarean section was performed at 37 weeks of gestation due to twin pregnancy with breech presentation of the first twin. Two healthy female neonates were delivered.

This case highlights the reproductive potential of women with 46,XY gonadal dysgenesis and emphasizes the importance of meticulous antenatal surveillance in achieving favorable outcomes, even in twin gestations.

Keywords: Swyer syndrome, 46 XY gonadal dysgenesis, twin pregnancy, donor gametes, primary amenorrhea.

1. Introduction

46,XY complete gonadal dysgenesis (Swyer syndrome) is a rare disorder of sex development resulting from failure of testicular differentiation in a genetically male individual. Due to absence of anti-Müllerian hormone and testosterone, Müllerian structures persist, external genitalia develop along female lines, and the gonads remain as non-functional streaks. Patients usually present with primary amenorrhea and absence of spontaneous puberty.

With advances in assisted reproductive technology, pregnancy has become possible in these women using donor gametes. However, most reported pregnancies are singleton gestations. Twin pregnancies are extremely rare and pose significant obstetric challenges. We report a rare case of a successful twin pregnancy in a woman with Swyer syndrome.

2. CASE REPORT

A 32-year-old phenotypic female presented with primary amenorrhea. She had normal female external genitalia and a well-developed vagina. There was no history of virilization or cyclic abdominal pain. Secondary sexual characteristics had been induced earlier with hormone replacement therapy.

Cytogenetic analysis revealed a 46,XY karyotype. Pelvic ultrasonography demonstrated a normally formed uterus with streak gonads. Diagnostic hysteroscopy showed a normal uterine cavity.

Following appropriate counseling, the patient underwent assisted reproductive treatment using donor gametes and conceived successfully. A twin intrauterine pregnancy was confirmed at 6 weeks scan. Cervical length was found to be reduced at NT scan and prophylactic cervical cerclage was performed in view of the high-risk of pre term labor

At 24 weeks of gestation, the patient developed pregnancy-induced hypertension and was started on labetalol 100 mg twice daily, with good blood pressure control. Serial antenatal surveillance revealed satisfactory fetal growth and well-being. There were no episodes of preterm labor or premature rupture of membranes.

At 37 weeks of gestation, an elective lower segment cesarean section was performed due to twin pregnancy with breech presentation of the first twin. Two healthy female babies were delivered with good Apgar scores. The postoperative period was uneventful.

3. DISCUSSION

Swyer syndrome is a rare condition, and successful pregnancies in these women remain uncommon. The presence of a functional uterus allows implantation and gestation following donor gamete conception. However, such pregnancies are considered high risk due to abnormal uterine development and lack of endogenous hormonal support.

Twin pregnancy further increases the risk of complications such as cervical insufficiency, hypertensive disorders, and preterm delivery. In the present case, early identification of short cervical length and

timely cerclage, along with strict blood pressure monitoring, played a key role in achieving a favorable outcome.

This case demonstrates that with careful planning, multidisciplinary management, and close antenatal surveillance, even twin pregnancies in women with 46,XY gonadal dysgenesis can result in successful maternal and neonatal outcomes.

4. CONCLUSIONS

Women with 46,XY complete gonadal dysgenesis can achieve successful pregnancy through assisted reproductive techniques using donor gametes. Twin pregnancies in such patients are extremely rare and associated with increased obstetric risk. Early risk identification, vigilant antenatal care, and planned delivery are essential for optimal outcomes.

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