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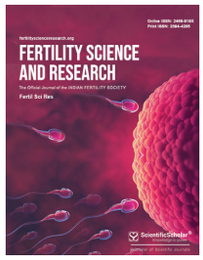


*Case Report*

# Breaking Barriers: A Triumph over Infertility in Swyer's Syndrome

Sudha Prasad[1], Pooja Gupta[1], Saumya Prasad[1]

[1]Artificial Reproductive Technology, Matritava Advanced IVF and Maternity Center, Gurugram, India

***Corresponding author:**
Saumya Prasad,
Artificial Reproductive Technology, Matritava Advanced IVF and Maternity Center, Sector 23, Gurugram - 122017, Haryana, India.

matritava@gmail.com

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## ABSTRACT

This case study examines the comprehensive fertility treatment and eventual successful conception of a patient with Swyer's syndrome. It emphasizes the impact of suitable luteal support on reproductive outcomes. The patient underwent a thorough evaluation including karyotyping, laparoscopy, and gonadectomy. She then pursued donor oocytes in vitro fertilization (IVF) cycles, which were preceded by an estradiol challenge to evaluate endometrial receptivity. Human chorionic gonadotropin (hCG) was used for luteal phase support in the third IVF attempt after two previous unsuccessful cycles. The third IVF cycle, which included injection hCG for luteal support, led to successful implantation and conception. The pregnancy was uneventful, and the patient delivered a healthy baby via caesarean section at 37 weeks gestation. This case underscores the intricate management required for infertility in patients with gonadal dysgenesis. The positive outcome with hCG luteal support suggests its potential role in enhancing endometrial receptivity and implantation success in IVF treatments.

**Keywords:** Donor oocyte IVF, HCG, Luteal support, Swyer's syndrome, Successful conception, XY gonadal dysgenesis

## INTRODUCTION

Swyer syndrome, also known as 46 XY gonadal dysgenesis, is a rare disorder where individuals possess a typically female appearance despite having a male karyotype (46 XY).[1] Patients with Swyer syndrome usually present with primary amenorrhoea and non-functional gonads, often referred to as streak gonads, which are primarily composed of fibrous tissue.[2] Due to the absence of functional ovaries, individuals with Swyer syndrome do not produce endogenous oestrogen, necessitating hormonal replacement therapy to develop secondary sexual characteristics and maintain bone health.[3] Even with hormone therapy, infertility remains a major challenge for these patients because they lack viable oocytes.[4]

Assisted reproductive technologies (ART), especially in vitro fertilisation (IVF) using donor oocytes, have emerged as a viable option for women with Swyer syndrome to achieve pregnancy.[5] However, the success of IVF depends on several factors, including the patient's hormonal response and the receptivity of the endometrium.[6]

Recent research has underscored the importance of luteal phase support with human chorionic gonadotropin (hCG) in improving endometrial receptivity and increasing implantation rates in ART cycles.[7]

This case study explores the fertility treatment of a 22-year-old woman with Swyer syndrome, emphasising the impact of hCG support on achieving a successful pregnancy outcome.

## CASE REPORT

Mrs A, a 22-year-old homemaker from Haryana, married to Mr X, a 27-year-old businessperson, presented with primary infertility. The couple had been married for two years with regular, unprotected vaginal intercourse but failed to conceive. At 15 years of age, she was evaluated for primary amenorrhoea without pubertal changes and prescribed cyclical combined pills, resulting in the development of secondary sexual characteristics.

Following marriage at 20 yrs of age, she was re-evaluated for amenorrhoea at a government-run hospital. Karyotyping revealed a 46 XY karyotype, leading to a laparoscopic evaluation and gonadectomy. Operative findings indicated a normal uterus and tubes with bilateral streak gonads. Histopathology showed dysgenetic gonads with predominantly fibro-connective tissue and a thin rim of ovarian stroma at the periphery.

Her examination revealed that she was 157 cms tall, weighed 55 kgs, and had a BMI of 22.49 kg/m$^2$, indicating a normal weight. Her vital signs were stable, with a pulse of 72/min, a regular rhythm and a blood pressure of 120/80 mmHg. There were no signs of pallor, clubbing, cyanosis, jaundice, oedema, or lymphadenopathy, and no hirsutism was observed. Clinically, her thyroid appeared normal, and she exhibited Tanner stage 5 for breast and pubic/axillary hair development. On abdominal examination, a laparoscopy scar was noted, with no masses or organomegaly palpable. Local examination revealed a normal vulva, nulliparous cervix, and normal vagina. A bimanual examination indicated a smaller-than-usual, mobile uterus with free fornices and a uterocervical length of 5 cm.

Regarding investigations, routine blood and urine investigations were within normal limits. She had blood group B+, while her husband had blood group A+. Chest X-ray, electrocardiogram (ECG), and cervical culture were all within normal limits. Endometrial biopsy and tuberculosis polymerase chain reaction (TB PCR) were negative, with the endometrium showing a secretory phase. Her serum levels of follicle-stimulating hormone (FSH) were 75.8 IU/L, luteinising hormone (LH) was 15.47 IU/L, oestradiol was 18.48 pg/mL, and thyroid-stimulating hormone (TSH) was 2.06 mIU/L. On imaging, transvaginal ultrasound revealed a uterus measuring 6 × 2.5 cm with a thin endometrium, and bilateral ovaries were not visualised. Ultrasound of upper abdomen was within normal limits. She was diagnosed with XY complete gonadal dysgenesis (Swyer's syndrome) with post-gonadectomy status and a smaller-than-normal uterus.

The husband's evaluation showed no coital difficulties and a normal semen analysis (3 mL volume, 80 million/mL total sperm concentration, 60% motility with A: 20%, B: 25%, C: 20%, and 60% normal morphology). His karyotype was normal (46 XY).

The couple was counselled for a donor oocyte IVF cycle following three months of cyclical oestrogen and progesterone therapy. Repeat ultrasound showed improved uterine size (7.5 × 3 cm, uterocervical length of 6 cm). She underwent an oestradiol challenge test, which resulted in a satisfactory endometrial buildup of 8 mm with a triple-line pattern by the 10th day of oestradiol valerate 2 mg, taken three times daily. A mock embryo transfer was performed without difficulty. The couple received counselling for a donor oocyte IVF cycle following three months of cyclical oestrogen and progesterone therapy. A follow-up ultrasound demonstrated an improvement in uterine size to 7.5 × 3 cm and uterocervical length (UCL) of 6 cm.

After her last withdrawal bleed, her endometrium was prepared using oestradiol valerate (Tablet Estrabet 2 mg three times a day [max. 12 mg/day], Abbott India Ltd.) followed by injectable progesterone (Injection Susten 100 mg intramuscular daily once a day, Sun Pharma Laboratories Limited, India) from the day of donor oocyte pickup. The endometrial thickness was 8 mm with a triple-line flow zone 2. Four donated oocytes were inseminated with her husband's washed sperm four hours post-oocyte pickup. Two good-quality day-3 embryos were transferred on the fourth day of progesterone injections. Luteal support was maintained with oestradiol valerate tablets (dose-4 mg three times a day) and both injectable (in. Susten 100 mg intramuscularly once a day) and vaginal progesterone (capsule Suten, Sun Pharma, India, 400 mg twice a day). Unfortunately, this cycle did not result in a pregnancy. A repeat frozen thaw embryo transfer cycle was performed. Three grade-A day-3 embryos were transferred once again, following similar endometrial preparation and support. However, this cycle was also unsuccessful.

In the third cycle, the patient was transferred with two good-quality embryos again, but this time luteal support included injectable HCG (Injection Fertigyn 2000 IU intramuscular, Sun Pharmaceuticals Industries Limited, India) along with oral oestradiol valerate (dose-4 mg three times a day) and vaginal progesterone (capsule susten 400 mg twice a day). Fourteen days post-transfer, the pregnancy test was positive, and subsequent beta-hCG levels showed normal doubling. A transvaginal ultrasound at 4 weeks post-embryo transfer confirmed a single live intrauterine gestation corresponding to 6 weeks. Hormonal support continued throughout pregnancy. The pregnancy progressed uneventfully until 37 weeks when the patient developed

**Table 1:** Studies reported successful pregnancy and its outcome.

| Sr No. | Study | No. of Patients | Singleton/ Multiple Pregnancy | Complication | Mode Of Delivery | Indication of C-Section |
|---|---|---|---|---|---|---|
| 1 | Gupta et al. [12] | 1 | Single | None | Not available | Not available |
| 2 | J. Taneja et al. [13] | 1 | Singleton | None | C-section | Demand |
| 3 | A Chrysostomou et al. [14] | 1 | Single | None | C-section | Demand |
| 4 | Tulic et al. [15] | 1 | Singleton | Breech | C-section | Breech |
| 5 | Kalra A et al. [16] | 1 | Single | Preeclampsia | C-section | Severe preeclampsia |
| 6 | Creatsas et al. [17] | 1 | Single | Preeclampsia | C-section | Fetal distress |
| 7 | Kalra A et al. [18] | 3 | 1st –twins<br>2nd –Single<br>3rd –Single | 1st-None<br>2nd – PPROM<br>3rd- Superimposed Preeclampsia | 1st- PTVD<br>2nd- C-section<br>3rd- C-section | -<br>2nd- PPROM and foetal distress<br>3rd – Superimposed Preeclampsia |
| 8 | Urban A et al. [19] | 2 | Single | None | C-section | 1st- Non-progress of labour<br>2nd – Periurethral abscess |
| 9 | Winkler et al. [20] | 1 | Twins | 1st- None<br>2nd – IUGR | C-section | |
| 10 | Jindal et al. [21] | 1 | Single | Oligohydramnios, Gestational Hypertension, Fetal Growth Restriction | C-section | Oligohydramnios, Gestational Hypertension, Fetal growth Restriction |

PTVD: Preterm vaginal delivery, PPROM: Preterm premature rupture of membranes, IUGR: Intrauterine growth restriction.

high blood pressure. A caesarean section was performed, delivering a healthy 2.6 kg female baby. Postoperatively, both mother and baby did well, with successful lactation.

## DISCUSSION

This case report presents the complex management and successful outcome of infertility in a patient with Swyer syndrome, a rare condition characterised by XY gonadal dysgenesis. Swyer syndrome patients possess a 46 XY karyotype but develop as phenotypic females due to dysfunctional gonads that fail to produce sex steroids, necessitating hormone replacement therapy for the development of secondary sexual characteristics and maintenance of female reproductive organs.[1]

The initial diagnosis of Swyer syndrome was confirmed through karyotyping, revealing a 46 XY chromosomal pattern.[7] The patient's gonadectomy was essential to prevent the malignant transformation of dysgenetic gonads, a common risk in Swyer syndrome.[3] This aligns with existing literature, which reports a 20–30% risk of gonadoblastoma in individuals with Swyer syndrome.[2]

Infertility management in Swyer syndrome often involves assisted reproductive technologies due to the absence of functional ovaries. In this case, the use of donor oocytes was necessary, aligning with standard practices for patients with gonadal dysgenesis.[8] The oestradiol challenge test demonstrated good endometrial receptivity, which is crucial for successful embryo implantation.[9] However, initial IVF attempts failed, highlighting the challenges and variability in endometrial receptivity and implantation success.

The eventual successful IVF cycle incorporated daily hCG injections for luteal support, which has been shown to enhance endometrial receptivity and improve implantation rates.[10] The literature indicates that hCG plays multiple roles in pregnancy, including promoting angiogenesis, immunosuppression, and endometrial differentiation, which are critical for successful implantation and maintenance of early pregnancy.[11] Studies have shown that hCG administration in the luteal phase can significantly improve pregnancy rates in IVF cycles.[7]

According to a review of ten studies, a total of 13 patients with Swyer syndrome (excluding our case) achieved successful pregnancies [Table 1]. However, one patient was lost to follow-up, limiting further outcome analysis.[12] Several studies reported distinct pregnancy outcomes with varied complications. Taneja et al. and A Chrysostomou et al. described an uncomplicated singleton pregnancy that was electively delivered via caesarean section at the patient's request.[13,14] In contrast, Tulic et al. performed a caesarean

due to breech presentation, demonstrating the importance of careful foetal monitoring for non-cephalic deliveries.[15]

Cases involving pregnancy-related complications were also documented. Creatsas et al. and Kalra et al. both reported instances of preeclampsia, which required prompt Caesarean sections to address maternal or foetal distress. These cases highlight the prevalence of hypertensive disorders in pregnancies achieved through ART in Swyer syndrome patients.[16-18] Urban et al. documented two successful and uncomplicated pregnancies in a woman, both delivered via cesarean section—the first due to non-progression of labor and the second due to a periurethral abscess.[19] Additionally, Winkler et al. documented a twin pregnancy complicated by intrauterine growth restriction (IUGR) in one foetus, managed surgically to minimise risks.[20] This case underscores the higher risks and complexities of multiple gestations in patients with Swyer syndrome, further emphasising the need for individualised care and close monitoring throughout pregnancy. Another study by Jindal et al.[21] in 2023 reported oligohydramnios, gestational hypertension, and foetal growth restriction for which a caesarean section was done. Hypertension is more frequently observed in pregnancies achieved through assisted reproductive technologies, possibly due to altered placental development.[5] The timely caesarean section ensured the safe delivery of a healthy baby, and both mother and child had an uneventful postoperative period with successful lactation initiation.

Our case contributes to the evidence regarding successful pregnancy in women with Swyer syndrome. It emphasises the role of hCG in enhancing implantation and endometrial receptivity; hence, it is important that more research and clinical trials are conducted towards improving the treatment protocols and the outcomes in similar cases. Additionally, the need for ongoing monitoring and tailored supportive care throughout pregnancy is emphasised to mitigate risks associated with assisted reproductive technologies.

## CONCLUSION

This case report highlights the successful management of infertility in a patient with Swyer syndrome using donor oocyte IVF and personalised hormonal support. The positive outcome achieved through the addition of hCG to the treatment protocol underscores its potential benefits in improving endometrial receptivity and implantation rates. Further research and clinical studies are warranted to establish optimised treatment protocols for patients with similar conditions, aiming to enhance their reproductive outcomes and overall quality of care.

### Author contribution

SU.P: Research idea, design of the study; SU.P, P.G: Acquisition of data for the study, analysis of data; SU.P, P.G, S.P: Drafting of the manuscript, revising it critically for important intellectual content and final approval of the version to be published.

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