

early diagnosis provides opportunities for fertility preservation counseling, as patients possess a functional uterus, making oocyte donation and assisted reproductive technologies viable options. Advances in genetic testing, imaging, and hormonal assessments continue to enhance clinical awareness, leading to improved patient outcomes [4].

A small number of successful pregnancies achieved with oocyte donation have been reported in patients with Swyer syndrome [3].

Case Report

A 17-year-old female patient (Figure 1) sought medical attention at S Nijalingappa Medical College and HSK Hospital in Bagalkote, Karnataka, India, due to the absence of menstruation. The patient had no notable surgical background. She reported no recurrent abdominal discomfort, exposure to hormones or radiation, or neurological symptoms like headaches or vision problems. She was the eldest child of parents who were second-degree relatives. Her younger sibling was a healthy male. Physical assessment revealed her height as 167 cm and a body mass index of 15.8 kg/m². Evaluation of her sexual development showed bilateral streaky ovary and Tanner stage 2 breast growths, underdeveloped secondary sexual characteristics like lack of axillary hair, but well-formed pubic hair.

Inspection of the external genitalia indicated female characteristics, with a visible vaginal opening. External genitalia shows unambiguously female, the vagina and uterus are present but underdeveloped. Importantly, there are no congenital anomalies or dysmorphic features beyond the lack of secondary sexual characteristics. Subsequent tests revealed elevated serum hormone levels: follicle-stimulating hormone at 47.3 mIU/l and luteinizing hormone at 34.96 mIU/l. Estradiol was measured at 66.969 mIU/l. Imaging studies showed abnormal reproductive organs: ultrasound detected a rudimentary uterus (Figure 2), while magnetic resonance imaging revealed an underdeveloped uterus lacking fallopian tubes and with streak-like ovaries, without distinct endometrial or myometrial layers. Both kidneys appeared normal.

Genetic analysis through karyotyping (Figure 3) identified a 46XY genotype. To validate this finding and rule out potential false results, fluorescent *in situ* hybridization (FISH) (Figure 4) was performed, which confirmed the presence of one X and one Y chromosome in all examined cells. These findings were consistent with patients' presentations.

Mosaicism was specifically ruled out through karyotype and FISH analysis, as it can present with overlapping but distinct clinical features, such as asymmetry in gonadal development, mixed gonadal dysgenesis, and a higher risk of ambiguous genitalia or Turner-like features. Ruling out mosaicism is critical because it affects gonadal

malignancy risk assessment and also aids in individual management decision which includes the timing of gonadectomy and long-term hormonal therapy.

Discussion

Swyer syndrome affects individuals with XY chromosomal make-up; nevertheless, they have a female appearance. The exact incidence is unknown. The diagnosis of Swyer syndrome is made based on thorough clinical evaluation, detailed patient history, identification of characteristic findings (e.g., amenorrhea and streaky gonads), and a variety of tests including karyotyping and FISH. Accurately differentiating Swyer syndrome (46, XY complete gonadal dysgenesis) from other disorders of sexual development is critical due to distinct implications for management, prognosis, and patient counseling. Several conditions, such as Turner syndrome (45X or mosaic variants), mixed gonadal dysgenesis (45X/46XY), and androgen insensitivity syndrome, can present with overlapping features but require additional investigations. The height



Figure 1. 17-year-old girl with primary amenorrhea suggestive of Swyer syndrome.

of the patient is particularly helpful in distinguishing patients with pure 46XY gonadal dysgenesis from those with 45XO/46XY mosaicism but without the classic features of Turner syndrome [5]. The chromosomal analysis in Swyer syndrome shows a male karyotype (46XY) and FISH signals for males.

This female approached us at the appropriate age and time. This helped us with the proper workup and management. A study by Michala et al. [3] also shows that many females experienced delays in reaching accurate diagnosis, often several years after the presentation to their general practitioners. It is suggested that health professionals should update their scientific knowledge and be aware of sexual development disorders. Early diagnosis

is important for a number of reasons: 1. Because of the risk of gonadal malignancy. 2. Early institution of hormonal therapy is vital for the induction of puberty. 3. HRT is required to prevent osteoporosis [1].

The risk of tumor development in Swyer syndrome is significant. A 20%-30% incidence of tumor in Swyer syndrome was reported [5]. The most common tumors involved in this condition are gonadoblastoma, dysgerminoma, and embryonal carcinoma were also reported [2]. Due to the risk of tumors, an extensive search for the rudimentary gonads is needed and bilateral gonadectomy is advisable. As for this patient, since we could not detect any rudimentary gonads, she requires a close follow-up in the long run. An interesting finding in this patient is that despite the late induction of puberty and non-compliance to hormonal treatment, there is marked improvement in her uterine development. However, her uterine size was smaller than in normal controls. This finding was similar to the study carried out by Michala et al. [3]. The small uterine size however does not appear to have an adverse effect on the fertility outcome in Swyer syndrome [2]. A small number of successful pregnancies have been reported in this group of patients [1,3]. Pregnancies were possible through oocyte donation and hormonal treatment. The presence of the XY genotype and the H-Y antigen does not affect the normal uterine and endometrial response. Thus, the possibility of maintaining normal pregnancy and delivery confirms the physiological ability of the uterus to accommodate and maintain successful pregnancy in patients with XY dysgenesis [6]. Swyer syndrome can be sporadic or linked to mutations in SRY, NR5A1, or MAP3K1, making genetic counseling valuable for family planning [4]. This case was considered significant due to the high risk of gonadal malignancy associated with Swyer syndrome,



Figure 2. Ultrasound image showing rudimentary uterus.

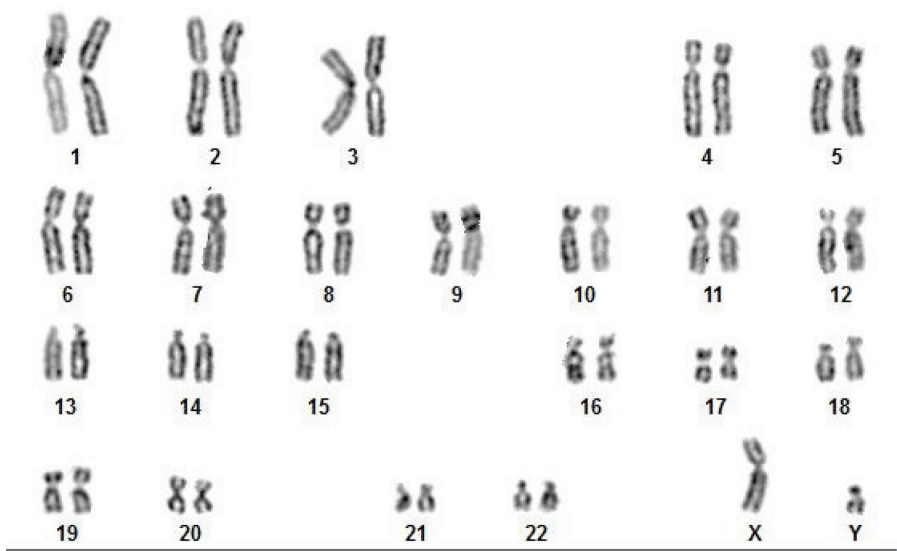


Figure 3. Karyotype showing 46XY.

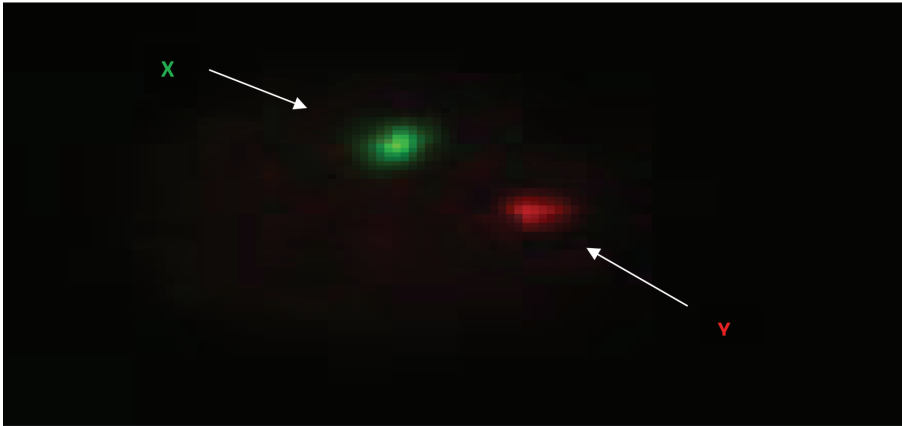


Figure 4. Fluorescent in situ hybridization showing signals for XY.

particularly the potential for gonadoblastoma and dysgerminoma in the dysgenetic gonads. Studies indicate that up to 30%-40% of individuals with Swyer syndrome develop gonadal tumors, with the risk increasing over time if the streak gonads are not removed. The malignancy risk is primarily due to the presence of Y chromosome material, which has been linked to oncogenic factors promoting abnormal cell proliferation in dysgenetic gonads.

A structured, multidisciplinary long-term follow-up plan ensures early detection of complications, optimization of hormone therapy, and overall well-being of patients with Swyer syndrome. Regular monitoring of hormone levels, bone health, and cardiovascular risks is critical for improving quality of life and long-term outcomes.

Conclusion

Swyer syndrome is extremely rare and invariably causes primary amenorrhea. Genetic testing plays a very important role in the diagnosis of Swyer syndrome. Early diagnosis is of crucial importance for various reasons, including the risk of gonadal malignancy and the need for removal of the gonads. Early institution of HRT for induction of puberty, improving bone mineral density and also counseling regarding fertility options.

What is new

Recent updates in Swyer syndrome include advances in early diagnosis and risk management, optimized hormone replacement therapy, emerging fertility prospects through assisted reproduction, and the importance of multidisciplinary care.

Conflicts of interest

The authors declare that they have no conflict of interest regarding the publication of this case report.

Funding

This study is not funded.

Consent for publication

Written consent was obtained from the parents of the patient.

Ethical approval

Ethical approval is not required at our institution to publish an anonymous case report.

Author details

- H. Ashwini¹, Sujayendra Kulkarni¹, Naveen Charantimath², Anil Satyaraddi², Suvarna Tapashetti³
1. Lecturer, Cytogenetic Laboratory, S Nijalingappa Medical College and HSK Hospital, Bagalkot, India
 2. Department of Endocrinology, S Nijalingappa Medical College and HSK Hospital and RC, Bagalkot, India
 3. Kuntoji Maternity Hospital, Bagalkot, India

References

1. Negar K, Saber G, Mahdi E. Swyer syndrome in a woman with pure 46, XY gonadal dysgenesis and a hypoplastic uterus: a rare presentation. *Fertil Steril*. 2010;93:1. <https://doi.org/10.1016/j.fertnstert.2009.09.062>
2. Granados H, Phulwani P. Absent visualization of a hypoplastic uterus in a 16 year old with complete 46 XY gonadal dysgenesis (swyer syndrome). *Endocrinol Metab Syndr*. 2013;2:1143.
3. Michala L, Goswami D, Creighton SM, Conway GS. Swyer syndrome: presentation and outcomes. *BJOG*. 2008;115:737–41.
4. Giri KP, Thapa S, Rawat R, Rouniyar S. Unanticipated diagnosis of Swyer syndrome: a case report. *Ann Med Surg*. 2024 Sep 5;86(10):6293–9. <https://doi.org/10.1097/MS9.0000000000002535>
5. Nadereh B, Mojgan KZ. Dysgerminoma in three patients with Swyer syndrome. *World J Surg Oncol*. 2007;5:71. <https://doi.org/10.1186/1477-7819-5-71>
6. Babre VM, Bendre K, Niyogi G. A rare case of swyer syndrome. *Int J Reprod Contracept Obstet Gynecol*. 2013;2:485–7

Summary of case

1	Patient (gender, age)	Female, 17
2	Final diagnosis	Swyer syndrome
3	Symptoms	Primary amenorrhea
4	Medications	HRT
5	Clinical procedure	NIL
6	Specialty	Endocrinology