ORIGINAL RESEARCH PAPER

Endocrinology

MOSAIC SWYER SYNDROME: A CASE REPORT OF A 2-YEAR-OLD WITH KYPHOSCOLIOSIS AND NORMAL HORMONE LEVELS

KEY WORDS:

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ABSTRACT

Mosaic Swyer syndrome (MSS) is a rare form of sex chromosome disorder, characterized by a 46,XY karyotype with varying degrees of gonadal dysgenesis. Patient with swyer syndrome are genotypically male and phenotypically female. It typically presents with a lack of puberty development in individuals with female external genitalia. some cases of apparent Swyer syndrome present with a 46,XY karyotype in lymphocytes, in fact, result from low-level, tissue-specific45,X/46,XY mosaicism leading to abnormal gonadal development.The presence of the Y chromosome increases the risk of germ cell neoplasms This report discusses the clinical presentation of a 2-year-old child reared as male, presenting with kyphoscoliosis and normal hormone levels.The case highlights the importance of early recognition and the challenges in managing patients with atypical manifestations of the syndrome, underscoring the need for a multidisciplinary approach in care.

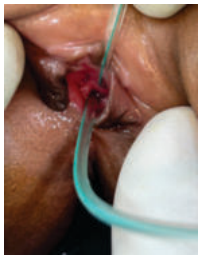
INTRODUCTION

Mosaic Swyer syndrome, also known as XY gonadal dysgenesis, is a genetic disorder affecting individuals with a 46,XY karyotype who exhibit female phenotype characteristics. The syndrome is typically diagnosed when individuals present with primary amenorrhea, absent or underdeveloped secondary sexual characteristics, and gonadal dysfunction. However, the clinical manifestations can vary, and some individuals may show atypical features such as skeletal abnormalities, as seen in this case.

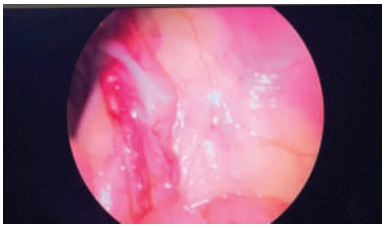
Case Report

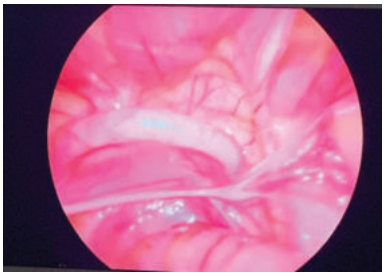
A 2-year-old female child was referred to our pediatric surgery OPD from a periphery rural hospital with a history of ambiguous genitalia and abnormal skeletal development, including kyphoscoliosis, which was noted by her pediatrician at birth. The child was not assianged any gender at the time of birth owing to the ambiguity of external genitalia due to clitoromegaly.The child's physical examination revealed a height and weight within the normal range for her age, but with noticeable spinal deformities.

Clinical History and Symptoms: The child had no history of any major illnesses, and no family history of genetic disorders. Her birth weight and length were appropriate for gestational age. Over the first 2 years of life, her developmental milestones were reported to be appropriate, but her pediatrician noted the kyphoscoliosis and ambigous genitalia at the time of birth. The child was reared as a male child by parents.



Hormonal Assessment: A comprehensive hormonal evaluation was conducted to assess for signs of pubertal development. Her hormone levels, including estradiol, testosterone, luteinizing hormone (LH), and follicle-stimulating hormone (FSH), were found to be within normal limits for her age, with no evidence of premature or delayed pubertal onset.





Karyotype Analysis: A karyotype analysis was performed, revealing a 46X/46,XY pattern, consistent with a diagnosis of mosaic Swyer syndrome. The mosaicism identified in the karyotype was of a relatively low percentage, but the presence of this genetic abnormality explained the patient's phenotypic presentation.

Radiological Findings: X-rays confirmed the diagnosis of kyphoscoliosis, with the spinal deformities being mild to moderate in nature. No other skeletal anomalies were detected, and the patient's bone mineral density was within the expected range for her age. Ultrasonography was suggestive of normal uterus and ovaries

Diagnostic Laparoscopy

Diagnostic laproscopy was suggestive of normal uterus and ovaries and rest of the abdomen was unremarkable. No testis were seen. Decision to preserve the ovaries were taken as there have been cases reported for pregnancy in swyers.

DISCUSSION

Mosaic Swyer syndrome is a rare and heterogeneous disorder. The most common presentation is primary amenorrhea and absent sexual development in a 46,XY individual, who would otherwise appear female. However, atypical presentations, such as skeletal deformities like kyphoscoliosis, are less commonly reported but should be considered in the differential diagnosis.

The pathophysiology of Swyer syndrome involves mutations or deletions in the SRY gene, which is responsible for initiating male sexual differentiation. In mosaic cases, only a proportion of cells exhibit the mutation, which can result in variable phenotypic expression. This variability can include the presence of additional features like skeletal abnormalities, which may complicate the clinical picture.

Kyphoscoliosis, while not typically associated with Swyer syndrome, has been observed in other genetic disorders such as Turner syndrome and is thought to result from abnormal bone development due to underlying chromosomal or hormonal disruptions. In our case, the kyphoscoliosis was an isolated finding without additional musculoskeletal abnormalities.

The hormonal profile in Swyer syndrome patients is usually abnormal, with low or absent sex steroids and elevated gonadotropins, which was not the case in this patient. The presence of normal hormone levels may indicate that the patient's gonadal dysgenesis had not yet manifested as a failure of pubertal development at this young age, which remains a point of ongoing monitoring.

CONCLUSION

This case of a 2-year-old female with mosaic Swyer syndrome and associated kyphoscoliosis emphasizes the clinical variability of this condition. While hormonal levels were normal for her age, the presence of skeletal abnormalities highlights the need for thorough evaluation and monitoring in patients diagnosed with MSS. Cases with mosaic swyer syndrome have reported to have pregnancies through IVF. Early multidisciplinary intervention is crucial for optimal management and ensuring that the child's developmental needs are met. Further research into the long-term outcomes of mosaic Swyer syndrome is needed to better understand its impact on skeletal and endocrine health.

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