

A Paediatric Medical Mystery: Early Puberty and Sacrococcygeal Anomaly

Versha Rani Rai, Sanagar Ali, Moshina Noor Ibrahim and Zubair Khoso Ahmed

Department of Paediatric Medicine, National Institute of Child Health, Karachi, Pakistan

ABSTRACT

This case report presents the diagnostic journey and management of a 1-year male child who presented with precocious puberty (PP) and a swelling in the left hip region. PP is described as the development of pubertal changes at a younger age specifically before the age of 8 years in girls and 9 years in boys with early progression of secondary sexual features, rapid bone maturation, lower final height, unsuitable bodily appearance, and psychological behavioural disorders. A thorough clinical examination, radiological investigations, and laboratory tests revealed a complex condition involving PP and the presence of a sacrococcygeal germ cell tumour. The tumour was histologically confirmed through an ultrasound-guided biopsy and immunohistochemistry, and further staging workup revealed distant metastases. The patient was subsequently initiated on chemotherapy with regular follow-up evaluations. This case underscores the importance of a holistic approach to paediatric care in addressing such perplexing clinical scenarios.

Key Words: *Precocious puberty, Germ cell neoplasms, Chemotherapy.*

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INTRODUCTION

Precocious puberty (PP) involves early activation of the hypothalamic-pituitary-gonadal axis, causing premature development of secondary sexual characteristics in children.¹ Genetic, environmental, and hormonal factors contribute to its complex aetiology. The threshold for early puberty is typically before 8 years in girls and 9 years in boys, necessitating comprehensive evaluations for deviations.^{2,3} Sacrococcygeal anomalies, diverse congenital malformations at the spine's base, pose diagnostic challenges in paediatrics. Coexisting with PP, they create a rare yet clinically challenging scenario.^{4,5} This case report details a paediatric patient experiencing both early puberty and a sacrococcygeal anomaly, emphasising the diagnostic journey, management challenges, and the need for a multidisciplinary approach.

CASE REPORT

A 1-year male child was brought to the Endocrinology outpatient department with a chief complaint of PP. The parents reported that over the past six months, they had noticed several alarming changes in their child's physical development.

Notably, there had been a significant increase in penile size, accompanied by a deepening of his voice and the development of hirsutism. These symptoms raised concerns as they were not in line with the typical developmental milestones expected at this age.

Furthermore, the child's parents also drew attention to a swelling in the left hip region that had been gradually enlarging over the course of two months (Figure 1). The swelling was described as non-tender, with no signs of inflammation, scar, or excoriation. Notably, there was no evident change in skin colour in the affected area, and hair distribution appeared normal when compared to other parts of the child's body. However, tufts of hair were observed along the lower spine, adding complexity to the clinical presentation.



Figure 1: Swelling on gluteal region.

Correspondence to: Dr. Versha Rani Rai, Department of Paediatric Medicine, National Institute of Child Health, Karachi, Pakistan

E-mail: versharai.sg@gmail.com

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Table I: Serum hormones levels.

Test	Result	Reference
Serum FSH	1.62 mIU/mL	1.4 - 15.4 mIU/mL
Serum LH	<0.00 IU/L	1.2 - 7.8 IU/L
Serum Testosterone	>52 nmol/L	Adult male (9-34 nmol/L) Adult female (0.52-2.43 nmol/L) 1-5 years. male (0.07-0.87 nmol/L) 1-5 years. female (0.07- 0.35 nmol/L)
Alpha Fetoprotein	152.0 ng/mL	Male (0.56 - 11 ng/mL) Female (0.78 - 9.3 ng/mL)
Beta HCG	12823 mIU/mL	Male (Up to 2.6 mIU/MI) Children: Newborn-03 months <5.0 mIU/mL 3-18 months <1.0 mIU/mL

On the initial anthropometric assessment, the child’s weight was recorded at 8 kg, which fell within the normal range but was slightly below the mean ($z = -1.69$ SDs). His height was 70 cm, indicating a more significant deviation from the mean ($z = -2.49$ SDs). These measurements collectively suggested that the child was experiencing growth-related issues.

General physical examination revealed a child whose facial features exhibited a medial convergent squint. The oral examination revealed a high-arched palate with no pigmentation abnormalities.

The examination of the chest, back, spine, musculoskeletal system, and skin revealed no other deformities or remarkable findings. The child's skin was notably free from rashes or any visible alterations.

The urogenital examination was particularly significant, as it revealed signs of pubertal development in a child of this age. The distribution of pubic hair corresponded to Tanner stage 2, indicating early pubic hair growth. Additionally, the size of the child's penis measured at 4.5 cm, consistent with Tanner stage 3 development. Both testes were palpable in the scrotum, were firm to the touch, and measured approximately 3 ml in volume. There was a single, normal urethral opening. Findings were consistent with peripheral PP.

The most concerning physical finding was the irregular swelling located in the left buttock, measuring 10 x 11 cm (Figure 1). This swelling exhibited no signs of inflammation, tenderness, or increased temperature. It appeared hard upon palpation. The presence of this swelling caused asymmetry in the natal cleft, and tufts of hair were noted in the lower spine area, further complicating the clinical presentation.

The combination of PP symptoms and the presence of a large sacrococcygeal mass raised significant concerns, prompting further investigation into the underlying aetiology of these multifaceted clinical findings.

An initial X-ray of the wrist for bone age assessment revealed a bone age of approximately 2 years according to Greulich and Pyle's method, which was inconsistent with the child's chronological age of 1 year.

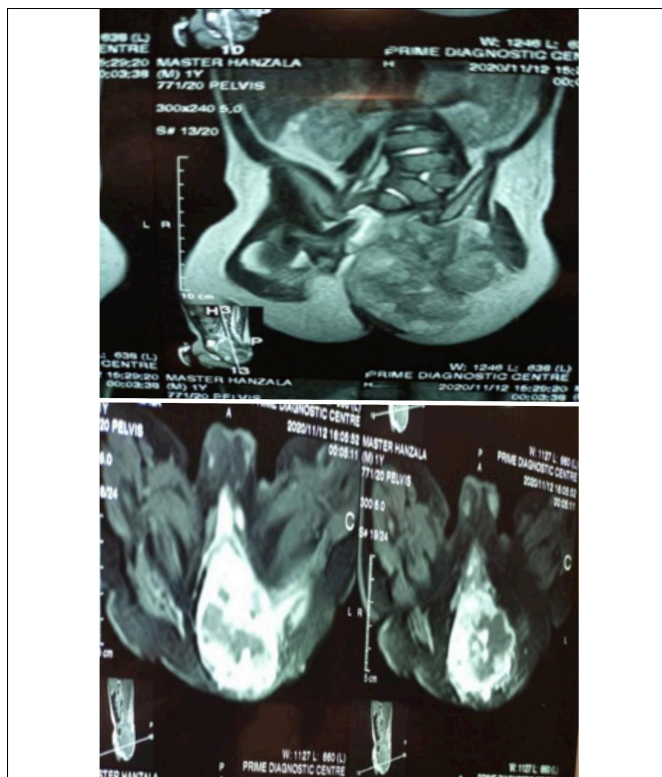


Figure 2: MRI Sacrococcygeal region and pelvis with contrast.

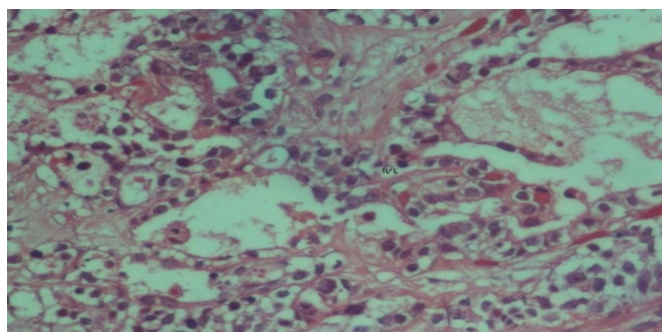


Figure 3: Histopathology of the tumour. The tumour is composed of cuboidal cells arranged in tubules and cystic pattern with round regular nuclei (H&E, x200).

Thyroid function tests indicated that free thyroxine (FT4) and thyroid-stimulating hormone (TSH) levels were normal, eliminating thyroid dysfunction as a potential cause of PP. An analysis of serum hormone levels revealed that follicle-stimulating hormone (FSH) was 1.62 mIU/ml, and luteinising hormone (LH) was undetectable. Notably, serum testos-

terone levels were markedly elevated at greater than 52 nmol/L, far exceeding the normal range for a child of this age (Table I).

An ultrasound of the left buttock region unveiled a substantial solid heterogenous mass measuring 8.1 x 7.3 cm. Although the colour Doppler study indicated subtle flow within the mass, the findings raised the possibility of a neoplastic lesion.

Ultrasound of both testes revealed no significant abnormalities, except for minimal hydrocele on the right side.

The MRI of the lumbosacral spine showed a large soft tissue density mass lesion within the sacrococcygeal region, which exerted extrinsic compression on the rectum, displacing it toward the right side and extending into the pelvic cavity (Figure 2). Moreover, there was minimal extension of the lesion into the sacrococcygeal spinal canal, highlighting the invasive nature of the mass.

An ultrasound-guided biopsy of the mass was performed, revealing a germ cell tumour (GCT). The tumour exhibited a solid and cystic arrangement of cells, polygonal nuclei, vacuolated cytoplasm, and eosinophilic hyaline globules. Schiller-Duval bodies were also noted (Figure 3). Immunohistochemistry confirmed the diagnosis of yolk sac tumour (YST). Serum markers for AFP and HCG were raised (Table I).

CT scans of the chest and abdomen with contrast revealed multiple intrapulmonary nodules and lymph node involvement, while MRI of the sacrococcygeal region and pelvis demonstrated extensive infiltration of nearby structures. A bone scan suggested focal bone pathology involving the sacrum. These findings indicated advanced disease with distant metastases, necessitating an oncology consultation.

The child was promptly registered in the oncology department, where the parents received counselling on the diagnosis and treatment options. Chemotherapy using the JEB regimen was initiated, with regular follow-up assessments including imaging and tumour marker monitoring to guide ongoing management.

DISCUSSION

The simultaneous occurrence of early puberty and sacrococcygeal anomalies in paediatric patients is rare, making this case a noteworthy medical mystery. This patient's presentation highlights the importance of comprehensive evaluation and interdisciplinary collaboration in managing complex clinical scenarios.

Early puberty is typically attributed to the premature activation of the hypothalamic-pituitary-gonadal axis, leading to accelerated sexual development. In this case, the coexistence of early puberty and a sacrococcygeal anomaly raised questions regarding potential underlying hormonal or

genetic factors that may have contributed to this unique presentation.

In comparison to a study featuring a suprasellar hCG-producing germinoma in a 10-year male, this case diverges significantly in tumour location, age of onset, and disease progression.³ Unlike the favourable outcomes postoperatively in that study, this case showcases an advanced disease state with distant metastases affecting multiple sites, such as the lungs, lymph nodes, and bones. Similarly, a different study involving a pineal mass diagnosed as a β -hCG-secreting GCT in a 5-year boy contrasts with this case in terms of patient age and tumour location.⁴ The pronounced disease advancement with distant metastases, in this case, underscores the aggressive nature of the pathology compared to more localised conditions. In contrast to a case report discussing a malignant steroidogenic tumour arising from a sacrococcygeal teratoma in a 5-year girl, the present case outlines a distinct tumour type and gender difference, highlighting the diverse presentations within sacrococcygeal neoplasms leading to PP.⁵ Moreover, a study detailing a mixed germ cell-sex cord-stromal tumour associated with an ovarian YST in a 4-year girl further exemplifies differences in tumour site (sacrococcygeal region) and gender, showcasing an advanced disease state with distant metastases.⁶

Contrasting with a study involving the management of a female neonate with a giant sacrococcygeal teratoma, this case emphasises gender disparity and distinct tumour types, accentuating the challenges associated with an advanced disease state and distant metastases in a male child.⁷

Similarly, in comparison to a retrospective review of sacrococcygeal teratomas in multiple children, this singular case underscores the complexities and challenges of managing an advanced disease state with distant metastases in a male child.⁸

Finally, in contrast to a study discussing pre-school age girls with PP amid various underlying conditions, this case highlights the rarity of a male child presenting with an advanced disease state of sacrococcygeal GCT leading to PP.⁹

These comparative analyses underscore the multifaceted nature and uniqueness of this case among diverse presentations of PP associated with different tumour types, sites, ages, and genders. The complexity and aggressiveness of this pathology necessitate comprehensive and tailored treatment regimens, interdisciplinary management, and ongoing monitoring to address the challenges posed by an advanced disease state with distant metastases. The distinctiveness of this case calls for further exploration of the underlying mechanisms driving such atypical presentations, emphasising the need for continued research in this rare subset of paediatric oncology and endocrinology.

PATIENT'S CONSENT:

Informed consent was taken from parents of the patients for the publication of pictures and case.

COMPETING INTEREST:

All authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

VRR: Manuscript writing.

SA: Writing and compiling.

MNI: Discussion writing and literature.

ZKA: Formatting and final revision.

All authors approved the final version of the manuscript to be published.

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