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Case Report

ACTH-Dependent Cushing's Syndrome in an Adolescent with a Cystic Pituitary Lesion: A Diagnostic Challenge

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Abstract

Cushing's syndrome (CS) is an uncommon endocrine condition in children, most frequently resulting from ACTH-dependent Cushing's disease. We report a 12-year-old female presenting with progressive facial swelling, facial puffiness, and dorsocervical fat pad suggestive of hypercortisolism. Biochemical evaluation showed elevated serum cortisol (28.7 µg/dL) and plasma ACTH (153 pg/mL), confirming ACTH-dependent CS. MRI revealed a cystic sellar lesion with differential diagnosis of Rathke cleft cyst. The patient had persistent hypertension, left ventricular hypertrophy, neutrophilia, elevated liver enzymes, and urinary tract infection with Enterobacter aerogenes. She was managed symptomatically while undergoing further evaluation. This case highlights a diagnostically challenging presentation of pediatric CS, emphasizing the need for early recognition and multidisciplinary management to prevent complications.

Keywords: Cushing syndrome; adolescent; hypertension; blood pressure; magnetic resonance imaging

Introduction

Cushing's syndrome was first described by Harvey Cushing in 1932 [1]. It is characterized by chronic elevation of circulating cortisol levels, which may result from exogenous glucocorticoid administration or endogenous overproduction of cortisol. Excess cortisol is associated with metabolic and systemic effects, including hyperglycemia, increased protein catabolism, immunosuppression, neurocognitive alterations, skeletal complications such as osteoporosis, and mood disturbances like depression. Common clinical features include weight gain, hypertension, and hypokalemia [2]. The reported incidence of Cushing's syndrome ranges from approximately 0.2 to 5.0 cases per million individuals annually, with an estimated prevalence of 39-79 per million across different populations [3]. Endogenous Cushing's syndrome resulting from pituitary or adrenal adenomas is reported to occur approximately three to four times more frequently in females than in males, whereas ectopic Cushing's syndrome shows a similar distribution between both sexes.

The pathogenesis of Cushing's syndrome involves either exogenous exposure to corticosteroids or endogenous overproduction of cortisol, which may be ACTH-dependent or ACTH-independent [4]. Endogenous Cushing's syndrome is broadly classified into ACTH-dependent and ACTH-independent forms, accounting for approximately 80% and 20% of cases, respectively. ACTH-independent Cushing's syndrome primarily results from excess cortisol production due to adrenal causes such as adenoma, carcinoma, or hyperplasia, and may also occur secondary to exogenous glucocorticoid use [5]. The primary treatment for endogenous Cushing's syndrome is surgical removal of the causative tumor. However, additional therapies such as pharmacological treatment, radiotherapy, or bilateral adrenalectomy may be required in certain cases.

Case Presentation

A 12-year-old female patient presented with progressive facial swelling of approximately one-year duration. The swelling was insidious in onset, gradually progressive, and not associated with pain, redness, or local temperature rise. The patient also reported fever of short duration at presentation. Clinical examination revealed facial puffiness and a dorsocervical fat pad (buffalo hump), suggestive of hypercortisolism as shown in Figure 2. Family history was notable for contact with tuberculosis. Personal history was unremarkable, and the patient had not yet attained menarche. Biochemical evaluation demonstrated an elevated morning serum cortisol level of 28.7 µg/dL (reference range: 4.46-22.7 µg/dL) and a markedly elevated plasma ACTH level of 153 pg/mL (reference range: 0-46 pg/mL), consistent with ACTH-dependent hypercortisolism. Given the association between cortisol excess and secondary hypertension, the patient underwent close monitoring with hourly blood pressure charting, which revealed persistently elevated readings.

Magnetic resonance imaging (MRI) of the brain with pituitary protocol demonstrated a well-defined cystic sellar lesion measuring approximately 13.8 × 18.2 × 18.8 mm with peripheral enhancement. Radiological differentials included Rathke cleft cyst and sellar epidermoid cyst. The lesion is seen abutting the optic chiasma, suggesting a probable Rathke cleft cyst with a potential risk of future visual impairment secondary to optic pathway compression (Figure 1). Echocardiography revealed concentric left ventricular hypertrophy with grade 1 diastolic dysfunction and preserved ejection fraction (55%), likely secondary to chronic cortisol excess. Abdominal ultrasonography was unremarkable. Laboratory investigations showed elevated hemoglobin levels and neutrophilia, consistent with cortisol-mediated physiological effects. Liver function tests revealed elevated SGOT. Urine examination demonstrated proteinuria (1+) and pus cells. Urine culture showed significant bacteriuria with growth of Enterobacter aerogenes (>10^5 CFU/mL), likely facilitated by cortisol-induced immunosuppression.

During hospitalization, the patient was managed symptomatically and for associated complications. Antihypertensive therapy with amlodipine and enalapril was initiated for blood pressure control. Empirical intravenous ceftriaxone was administered for urinary tract infection. Pantoprazole was prescribed for gastric protection, while paracetamol and ondansetron were used for symptomatic relief. Intravenous fluids (DNS) were provided for supportive care. Dexamethasone was administered as part of the endocrine evaluation, and tramadol was used for analgesic support when required. The patient remained under close vital and blood pressure monitoring.

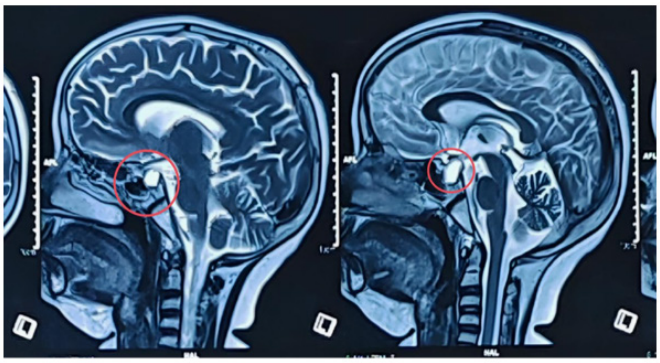

Figure 1: Sagittal MRI showing a sellar cystic lesion (circled), suggestive of a Rathke cleft cyst, abutting the optic chiasma with potential risk of visual impairment.

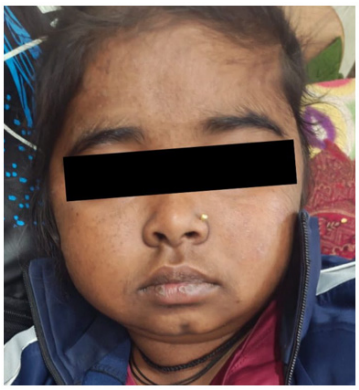

Figure 2: Clinical photograph showing facial puffiness and moon facies suggestive of hypercortisolism.

Discussion

Cushing's syndrome (CS) is an uncommon endocrine condition caused by sustained exposure to elevated cortisol levels, arising either from endogenous overproduction or exogenous glucocorticoid administration [6]. While Cushing's syndrome is well documented in adults, its occurrence in children is rare and often characterized by subtle, variable features that can lead to delayed diagnosis [7,8]. The incidence of Cushing's syndrome in children is low, with ACTH-dependent disease due to a pituitary corticotroph adenoma being the most common cause in those older than 5-7 years [7,9]. Our 12-year-old patient falls within this age group, supporting ACTH-dependent hypercortisolism as the most probable etiology. In this case, typical clinical features of hypercortisolism were observed, including progressive facial puffiness and a dorsocervical fat pad. Prolonged cortisol excess results in centripetal fat redistribution, facial rounding, hypertension, and various metabolic abnormalities [6,10].

Unlike adults, pediatric Cushing's syndrome typically presents with growth retardation along with weight gain, which is considered a key distinguishing feature. Delayed pubertal development and menstrual abnormalities can result from suppression of the hypothalamic-pituitary-gonadal axis. In this 12-year-old patient, the absence of menarche may be attributed to cortisol-induced hypogonadism. Biochemical assessment in this case revealed elevated morning serum cortisol along with significantly increased plasma ACTH levels, confirming ACTH-dependent Cushing's syndrome. Measurement of plasma ACTH is crucial for distinguishing between ACTH-dependent and ACTH-independent etiologies. Current diagnostic approaches recommend initial biochemical confirmation followed by targeted pituitary imaging for lesion localization. Pituitary microadenomas are often small and may not be easily detected even with high-resolution MRI. The presence of a cystic sellar lesion in this patient raises differential diagnoses such as Rathke cleft cyst.

Differentiating cystic adenomas from non-functioning cystic lesions remains a diagnostic challenge, and radiological findings should always be interpreted in conjunction with biochemical data. The presence of hypertension in this patient represents a recognized systemic effect of prolonged cortisol excess. Glucocorticoids enhance vascular responsiveness to catecholamines and promote cardiovascular remodeling. The observed persistent hypertension and concentric left ventricular hypertrophy are in line with the documented cardiovascular complications of Cushing's syndrome, which can also occur in pediatric patients. Early identification is crucial to minimize long-term cardiovascular risk. The urinary tract infection observed in this case may be attributed to cortisol-induced immunosuppression. Chronic hypercortisolism impairs cellular immune function while promoting neutrophil demarginating, thereby explaining the coexistence of increased infection susceptibility and neutrophilia. An elevated risk of infectious and metabolic complications has been reported across various clinical contexts, including cases associated with pregnancy.

In addition to its acute clinical features, pediatric Cushing's syndrome is associated with long-term effects on growth, metabolic function, and psychosocial health. Residual comorbidities and reduced quality of life may persist even after treatment [11]. These observations highlight the need for early diagnosis and sustained long-term follow-up. Definitive treatment of ACTH-dependent Cushing's syndrome typically involves transsphenoidal removal of the pituitary adenoma, while medical therapy or additional interventions may be required in cases of persistent disease. Early management in pediatric patients is essential to reduce the risk of long-term metabolic, cardiovascular, and reproductive complications. In summary, this case highlights a typical presentation of ACTH-dependent Cushing's syndrome in an adolescent, supported by clinical findings, biochemical evidence, and imaging features. It emphasizes the rarity of pediatric Cushing's syndrome, the predominance of pituitary-related causes, and the widespread systemic effects of sustained cortisol excess.

Conclusion

This case represents a typical presentation of ACTH-dependent Cushing's syndrome in an adolescent female, with progressive Cushingoid features, elevated serum cortisol accompanied by markedly increased ACTH levels, and a sellar lesion identified on imaging. The occurrence of secondary hypertension, left ventricular hypertrophy, and infection reflects the extensive systemic effects of prolonged hypercortisolism. Pediatric Cushing's syndrome is uncommon and often presents with subtle, gradually progressive features, which may delay diagnosis. Therefore, early clinical suspicion, prompt biochemical assessment, and appropriate imaging are essential to minimize long-term metabolic, cardiovascular, and reproductive complications. Timely multidisciplinary management plays a key role in improving outcomes in affected children.

Ethical approval

Institutional review board approval is not required.

Patient Consent Declaration

The authors certify that they have obtained all appropriate patient's consent.

Funding Declaration

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Conflict of interest

There are no conflict of interest

Clinical Trial Number

Not applicable.

Clinical Trial Registration

Not applicable.

Acknowledgment

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Author Contribution

DC conceptualized the study, performed extensive literature review, curated and analyzed the data, and drafted the original manuscript. HS contributed to data analysis, literature review, and assisted in manuscript writing. PK was involved in study design, data collection, and critical revision of the manuscript. AR contributed to data curation, literature search, and manuscript editing. RS assisted in data collection, interpretation, and final proofreading of the manuscript. All authors have read and approved the final version of the manuscript. All authors meet the criteria for authorship and confirm that the work presented is original and conducted with integrity.

Use of Artificial Intelligence

The authors did not use artificial technology to write and prepare the manuscript.

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