CASE REPORT

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A Case of Carney Complex with Pontine Glioma

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ABSTRACT

Carney complex (CNC) is a rare autosomal dominant syndrome characterized by skin pigmentation abnormalities, endocrine tumors, and cardiac myxomas. This report presents an 11-year-old girl with a history of pontine glioma treated with chemotherapy and radiotherapy at 2.5 years of age, who presented with weight gain and short stature, along with syndromic features (multiple nevi around the mouth and nose, four café-au-lait spots, and bilateral clinodactyly of the fourth toes) identified during physical examination. Genetic testing revealed a novel pathogenic *PRKAR1A* variant, confirming the diagnosis of CNC. The patient was diagnosed with Cushing's syndrome due to unsuppressed cortisol levels observed in a high-dose dexamethasone suppression test. Pathological evaluation following unilateral adrenalectomy confirmed the presence of primary pigmented nodular adrenocortical disease. This case highlights the importance of recognizing the atypical course of CNC to prevent delays in diagnosis and treatment.

Keywords: Carney complex, pontine glioma, Cushing's syndrome, short stature

What is already known on this topic?

Carney complex is a rare autosomal dominant multiple neoplasia syndrome characterized by skin pigmentation abnormalities, endocrine tumors, cardiac myxomas, and primary pigmented nodular adrenocortical disease, which may cause adrenocorticotropic hormone-independent Cushing's syndrome.

What this study adds?

This case describes the coexistence of Carney complex and pontine glioma, an association not previously reported in the literature, and identifies a novel pathogenic *PRKAR1A* variant. It also highlights the phenotypic variability of Carney complex and the potential for subtle Cushing's syndrome to delay diagnosis.

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Introduction

Carney complex (CNC) is a rare autosomal dominant genetic disorder, first described by Carney (1,2) in 1985. CNC is characterized by cutaneous pigmentation abnormalities, cardiac and cutaneous myxomas, endocrine gland neoplasms or hyperfunction, and schwannomas (1,2). Affected individuals may present with various endocrine manifestations, including primary pigmented nodular adrenocortical disease (PPNAD), growth hormone-secreting pituitary adenomas, prolactinomas, thyroid adenomas or carcinomas, and gonadal tumors, with two or more endocrine tumors often observed (3). The clinical presentation of CNC varies widely, ranging from multiple concurrent features to a single isolated manifestation. The components of CNC may manifest at any age, which may complicate the diagnosis in certain cases. PPNAD is a rare cause of adrenocorticotropic hormone (ACTH)-independent Cushing's syndrome (CS). ACTH-independent CS due to PPNAD has been found to be associated with CNC in 90% of cases (4). It represents the most common endocrine hyperactivity in CNC and occurs in approximately 25% of affected individuals. However, the incidence may be underestimated due to atypical and subclinical disease presentations. In autopsy studies conducted on patients with CNC, PPNAD was observed in nearly all cases (3,4,5).

In this article, we present a case of a patient with a history of pontine glioma who presented with short stature and obesity. Comprehensive evaluations led to a genetic diagnosis of CNC, followed by the identification of CS.

Case Report

An 11-year-old female patient presented with significant weight gain over the past six months and short stature noted over the preceding 1-2 years. Her medical history included a diagnosis of pontine glioma at the age of 2.5 years, identified during the evaluation of neurological symptoms, for which she underwent chemotherapy with temozolomide and radiotherapy (RT) at a total dose of 54 Gy. There was no consanguinity between the mother and the father. No history of pathological short stature or genetic disorders was reported in the family history. On physical examination she had a body weight of 48 kg [+0.98 standard deviation score (SDS)], a height of 136 cm (-1.6 SDS), and a body mass index of 25.95 (+2.06 SDS). Her pubertal development was compatible with Tanner stage 5. The patient, who had a reported age of menarche at 9.5 years, also reported regular menstruation. Clinical findings included proportional short stature, central obesity, multiple nevi around the mouth and nose, four café-au-lait spots with the largest (2.5×1.5 cm) located in the interscapular region. In addition, two smaller café-au-lait spots (<1×1 cm) were present on the posterior aspect of the right upper leg, and one spot (<1×1 cm) at the sacral level, and bilateral clinodactyly of the fourth toes (Figure 1). Given her early menarche potentially indicative of precocious puberty or rapidly progressing puberty, short stature, history of an intracranial mass, obesity, skin lesions, and syndromic features, further investigations were initiated with the preliminary consideration of a disorder from the rasopathy group.

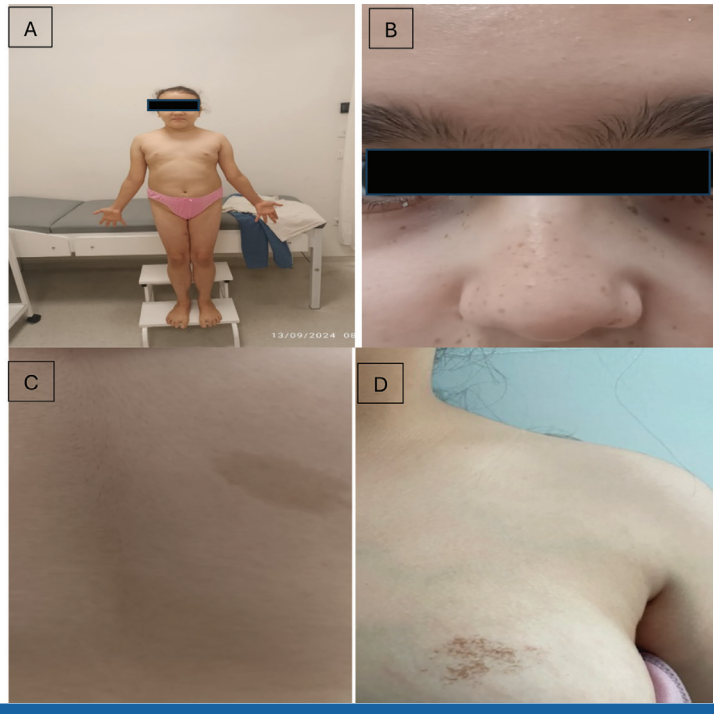

Figure 1. A) Central obesity, **B)** spotty black pigmentation on the cheeks and nose, **C)** café au-lait spot in the interscapular region, **D)** lentiginous skin lesion on the upper left breast

Biochemical tests conducted for the evaluation of short stature were within normal limits, and celiac tests were negative. Her insulin-like growth factor-1 level was low (72.1 ng/mL, -2.35 SDS), and bone age was consistent with 15 years (Table 1). Anterior pituitary hormone levels, assessed due to prior RT to the head and neck region were evaluated as appropriate for her age and pubertal stage.

Genetic analysis, including a broad-spectrum panel to encompass rasopathies, was performed. While CNC is not classified as a rasopathy, this panel was used because of overlapping clinical features. The genetic panel analysis identified a novel, heterozygous pathogenic variant, c.51dup (p.Cys18MetfsTer2), in the *PRKAR1A* gene (ENST00000589228). *PRKAR1A* variants are associated with autosomal dominant CNC type 1. This variant has a reported maximum population frequency of 0.0% in the gnomAD genome database.

The c.51dup (p. Cys18MetfsTer2) *PRKAR1A* variant, caused by a single-base insertion, is predicted to lead to a frameshift mutation and loss of function of the *PRKAR1A* protein. According to the American College of Medical Genetics and Genomics criteria and segregation analysis, this novel variant is classified as "pathogenic" based on PVS1, PM2, and PP1 scores. Segregation analysis confirmed the presence of this variant in the affected father, while it was absent in the mother. A detailed physical examination of the father revealed hyperpigmented macules on the lips as the sole clinical manifestation, with no additional abnormalities detected. The father was subsequently referred to adult endocrinology for comprehensive evaluation and further diagnostic screening. Subsequent to the genetic analysis, further communication with the family disclosed the presence of a previously unacknowledged paternal uncle who had undergone bilateral adrenalectomy and remained under ongoing medical surveillance for a confirmed diagnosis of PPNAD.

The patient underwent comprehensive screening for other components of CNC. Echocardiography did not reveal any evidence of cardiac myxomas, and on thyroid and pelvic ultrasonography there were no pathological findings reported. Due to her history of short stature and obesity, along with the well-established association between CNC, PPNAD, and ACTH-independent CS, further evaluation was conducted. Circadian cortisol rhythm analysis showed unsuppressed night-time serum cortisol levels, consistent with an impaired diurnal rhythm (5.29 µg/dL). Suppression tests, including overnight dexamethasone suppression and low-dose dexamethasone suppression, showed unsuppressed cortisol levels (4.02 µg/dL and 4.66 µg/dL, respectively Urinary free cortisol levels were within the reported reference range (16.6 µg/24 h; normal <37 µg/24 h). A high-dose Liddle test revealed a paradoxical increase in urinary free cortisol levels (Table 2).

Table 1. The patient's initial diagnostic tests performed because of presentation with obesity and short stature. Normal ranges are shown in brackets

Glucose	82 mg/dL (70-110)	**TSH**	2.56 mU/L (0.51-4.30)
Bun	8 mg/dL (5-18)	**fT4**	1.3 ng/dL (0.93-1.7)
Creatinine	0.41 mg/dL (0.39-0.73)	**FSH**	4.41 U/L (0.4-8.6)
Sodium	140 mmol/L (136-145)	**LH**	3.51 U/L (0.9-13.3)
Potassium	4.8 mmol/L (3.5-5.1)	**Estradiol**	53.6 ng/L
LDL	42 mg/dL (0-130)	**ACTH**	19.9 pg/mL
HDL	42 mg/dL (35-55)	**Cortisol**	13.2 µg/dL
Total cholesterol	173 mg/dL (92-200)	**IGF-1**	72.1 ng/mL (-2.35 SDS)
Triglyceride	54 mg/dL (0-150)	**Prolactin**	8 µg/L (4.79-23.3)
Urine	Density: 1019 pH: 5	**Insulin**	6.83 mU/L

BUN: blood urea nitrogen; LDL: low-density lipoprotein cholesterol; HDL: high-density lipoprotein cholesterol; TSH: thyroid stimulating hormone; fT4: free thyroxine; FSH: follicle stimulating hormone; LH: luteinizing hormone; ACTH: adrenocorticotropic hormone; IGF-1: insulin-like growth factor-1

Table 2. Results of the low and high dose dexamethasone suppression tests

	Low dose dexamethasone supression test		High dose (Liddle) dexamethasone test		
	Basal	**Final**	**Basal**	**Day 1**	**Final**
Plasma cortisol (µg/dL)	13.2	4.66	15.5	13.4	**6.47**
Free cortisol in urine (µg/24 h)	8.56	6.66	17.8	14.5	**26.6**

For the low-dose dexamethasone suppression test, plasma cortisol <5 µg/dL and urinary free cortisol <10 µg/day are considered normal; alternatively, urinary free cortisol should be suppressed by >50%. For the high-dose (Liddle) dexamethasone test, urinary free cortisol may decrease by approximately 90% from baseline, and serum cortisol measured the morning after the last dose is suppressed by >90% from baseline.

Dynamic contrast-enhanced computed tomography (CT) of the adrenal glands demonstrated a 6×5 mm lobulated lesion in the body of the left adrenal gland and a millimetric calcified focus in the medial limb of the right adrenal gland. The patient underwent unilateral adrenalectomy of the left adrenal gland Histopathological examination revealed multiple pigmented adrenocortical micronodules (most <1 mm), consistent with PPNAD (Figures 2 and 3).

No complications were observed after the operation and the patient remained under regular follow-up. Four months after unilateral adrenalectomy, the serum cortisol levels were measured and were 10.3 µg/dL at 08:00 AM and 2.35 µg/dL at 11:00 PM. The 24-hour urinary free cortisol level was 10.1 µg/24 h (normal <37 µg/24 h). At present, she has no additional complaints. However, ongoing follow-up is being conducted to monitor existing findings, evaluate the response to treatment, and identify the emergence of new manifestations. For endocrine complications of CNC, annual measurement of urinary free cortisol levels is planned for PPNAD monitoring. In cases of clinical suspicion, diurnal cortisol levels (with samples taken at 11:30 PM, 12:00 AM, 7:30 AM, and 8:00 AM), a dexamethasone suppression test (modified Liddle's test), and an adrenal CT examination will be conducted. Annual thyroid ultrasonography is recommended as part of routine surveillance in patients with CNC (3). If there is clinical suspicion for ovarian tumors, a suprapubic ultrasound will be performed. In cases of suspected gigantism and/or acromegaly, pituitary magnetic resonance imaging will be performed to evaluate for pituitary adenomas. In addition, a 3-hour oral glucose tolerance test and a 90-minute thyrotropin-releasing hormone stimulation test will be conducted, if necessary. Parental consent was obtained for publication of this case report.

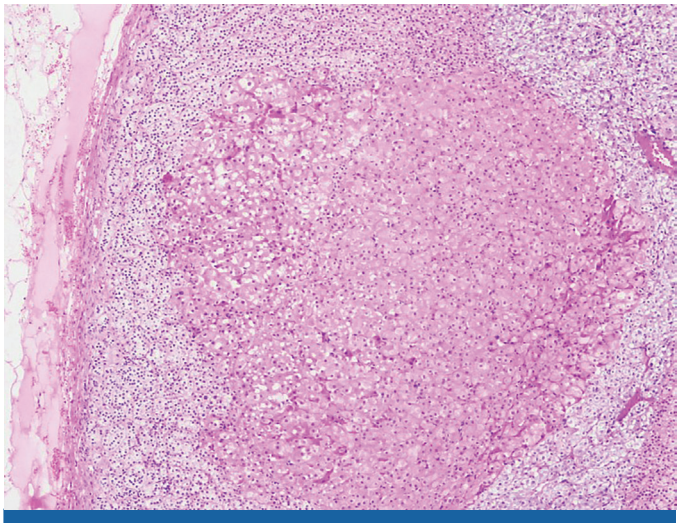

Figure 2. A micronodule (<1 cm) in the adrenal cortex, well-defined and composed of cells with eosinophilic cytoplasm (H&E stain, x100 magnification)
H&E, hematoxylin and eosin

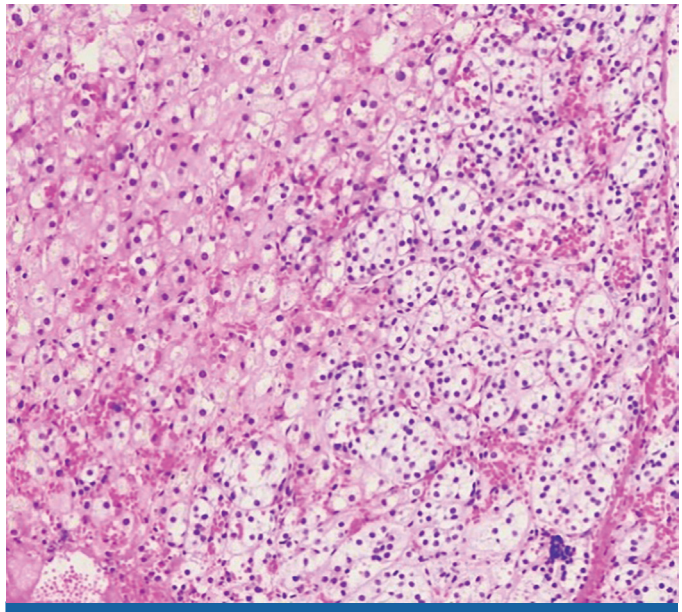

Figure 3. Cytoplasmic lipofuscin pigment in adrenal cortical micronodules (H&E stain, x200 magnification)
H&E, hematoxylin and eosin

Discussion

The presented case, who was referred to our clinic because of short stature and obesity but was subsequently diagnosed with CNC, is of interest because of the atypical features of CNC. To the best of our knowledge, this is report in the literature to describe the coexistence of pontine glioma and CNC. In addition, we suggest that in this case with precocious puberty and a history of cranial RT, the CS findings may have been overlooked due to their subtlety, which may explain her short stature. The very mild paternal CNC findings, despite harboring the same variant, also highlight the difference in expression. We suggest that because of the potential for atypical presentation, such as with short stature as in the presented case, it is beneficial to keep CNC in mind and at least investigate the additional findings of CNC. CNC is classified as a multiple neoplasia syndrome with an autosomal dominant inheritance pattern (1). Patients may present with the characteristic clinical features of CNC, but, as demonstrated in this case, the condition can also be identified in pediatric patients presenting to endocrinology clinics with nonspecific complaints, such as short stature although she had a history of cranial neoplasia. The clinical manifestations of CNC are highly variable, even among members of the same family (3).

Approximately 50% of CNC families reported in the literature have mutations in the *PRKAR1A* gene located on chromosome 17q22-24. The *PRKAR1A* gene, novel variants of which were identified in the patient and her father, encodes the regulatory subunit type 1α (R1α) of protein kinase A (PKA), a classical tumor suppressor protein. PKA is integral to numerous endocrine

signaling pathways. The R1α subunit inhibits PKA activity, and mutations in *PRKAR1A* may result in the production of a truncated, nonfunctional protein. This leads to increased PKA activity, resulting in intracellular signaling dysregulation and subsequently, endocrine hyperfunction or tumorigenesis (3,6,7).

Currently, approximately 62% of CNC cases have identifiable pathogenic *PRKAR1A* variants, with about 70% of these cases attributed to mutations in *PRKAR1A* (60% detected through sequencing and 10% via deletion-duplication analysis). In the remaining 30% of cases, the genetic basis remains unknown. Bertherat et al. (8), in the largest study to date, identified *PRKAR1A* mutations in 114 of 185 CNC families (62%), reinforcing the critical role of this gene in CNC pathogenesis.

Although universal diagnostic criteria for CNC have not yet been established, a revision was reported in 2001 that introduced major and minor diagnostic criteria, enhancing diagnostic sensitivity to approximately 98% (3). In the presented case, a comprehensive clinical evaluation and thorough patient history satisfied the diagnostic criteria, even prior to the confirmation of the diagnosis through genetic testing.

The association between CNC and CS has been well-documented. A study by Cazabat et al. (9) demonstrated that among CNC patients presenting with CS secondary to PPNAD, the frequency of identifying pathogenic *PRKAR1A* variants increased to 80%. CS is a significant and potentially life-threatening component of CNC. While CS can present with overt clinical symptoms, it may also manifest in an atypical manner, as observed in the presented case, where only mild Cushingoid features were present alongside disrupted diurnal cortisol rhythms. Diagnostic confirmation can be achieved using high-dose dexamethasone suppression testing, which would show cortisol non-suppression, or by identifying paradoxical increases in 24-hour urinary free cortisol levels.

A retrospective analysis conducted at the Mayo Clinic identified 37 patients with CNC. These patients were classified into four categories: classical CS (consistent clinical and laboratory findings); subclinical CS (absence of clinical symptoms but laboratory findings consistent with CS); possible CS (presence of some clinical findings with borderline results in one or more tests defining classical CS or adrenal pathological findings); and no CS (absence of both clinical and laboratory findings indicative of CS). Among the 17 patients diagnosed with classical CS, 15 underwent surgical intervention, including bilateral adrenalectomy (9 patients), subtotal adrenalectomy (2 patients), and partial unilateral adrenalectomy (4 patients), while two cases were diagnosed through autopsy reports. All patients who underwent bilateral adrenalectomy achieved complete remission. In the subtotal adrenalectomy group, remission was achieved in both patients, although one developed adrenal insufficiency. In the partial unilateral adrenalectomy group, two patients experienced recurrence of CS during follow-up and subsequently underwent total adrenalectomy. For the 12 patients with subclinical or possible CS, long-term follow-up without intervention, ranging from 1 to 36 years (mean: 10 years), demonstrated no progression to overt CS. Similarly, among the eight patients classified as having no CS, follow-up over 2 to 49 years (mean: 19.5 years) revealed no evidence of CS development. Notably, one patient remained untreated for nearly 30 years, while another exhibited spontaneous remission of CS. Furthermore, PPNAD was identified in two patients who exhibited no clinical features of CS, underscoring the potential for subclinical adrenal involvement in CNC (10). CS due to PPNAD must be treated to manage the consequences of cortisol overproduction. In rare cases, anti-cortisol medical treatments, such as ketoconazole or mitotane, have been used, but surgical interventions remain the primary approach currently (11,12) Although bilateral adrenalectomy remains the standard treatment for PPNAD, literature reports indicate heterogeneous clinical responses following unilateral adrenalectomy, with rare cases demonstrating sustained remission of CS for up to nine years postoperatively (10,13,14,15). These findings suggest the need to reconsider performing partial rather than total bilateral adrenalectomy in selected patients with CNC and CS.

In the presented case, considering the patient's young age, the presence of a 6×5 mm left adrenal lesion on imaging, and existing evidence suggesting relatively prolonged remission periods following unilateral adrenalectomy in selected cases, a comprehensive evaluation of potential clinical outcomes was conducted in consultation with the patient's family. Following a multidisciplinary consensus with the surgical team, unilateral adrenalectomy was deemed the most appropriate therapeutic approach. In our patient who underwent unilateral adrenalectomy, the most recent evaluation showed regression of CS findings, with no evidence of adrenal insufficiency.

Upon reviewing the existing literature, no documented association between CNC and pontine glioma was found. In the presented case, the pontine glioma has been considered a different presentation of CNC, which may be biologically reasonable given the tumor suppressor function of PRK. However, further research is needed to elucidate the potential relationship between CNC and intracranial masses, including pontine gliomas.

Previous studies have highlighted that CNC can exhibit clinical variability even among members of the same family and that the diagnostic process may be delayed for years. To expedite diagnosis, increasing awareness of CNC's multifaceted manifestations, obtaining a detailed medical history of patients and their families, and being vigilant about diagnostic pitfalls are essential. Notably,

the presence of unusual symptom combinations should prompt clinicians to consider rare endocrinopathy syndromes such as CNC (16). This case reinforces the importance of recognizing CNC as a rare but clinically challenging condition with systemic and endocrine manifestations. Early diagnosis, comprehensive screening for associated features, and appropriate management strategies are essential to mitigate potential complications and improve patient outcomes.

Conclusion

CNC represents a rare etiology of endocrine tumors. Although the majority of CNC cases develop CS at some stage, its atypical clinical course frequently results in delayed diagnosis. In patients presenting with ACTH-independent CS, PPNAD should be considered as a potential underlying cause. Given that CNC and its associated complications can manifest at any point during a patient's lifetime, comprehensive screening for PPNAD is mandatory in all cases. Furthermore, genetic testing and surveillance should be extended to family members of affected individuals to facilitate early or missed detection and mitigate the risk of life-threatening complications.

Ethics

Informed Consent: Parental consent was obtained for publication of this case report.

Footnotes

Authorship Contributions

Surgical and Medical Practices: Gülümay Vural Topaktaş, Emrullah Arslan, Tayfun Çinleti, Özlem Anlaş, Ebru Pala, Benay Turan, Eren Er, Bumin Nuri Dündar, Concept: Gülümay Vural Topaktaş, Emrullah Arslan, Data Collection or Processing: Gülümay Vural Topaktaş, Emrullah Arslan, Bumin Nuri Dündar, Analysis or Interpretation: Gülümay Vural Topaktaş, Emrullah Arslan, Literature Search: Gülümay Vural Topaktaş, Emrullah Arslan, Tayfun Çinleti, Özlem Anlaş, Ebru Pala, Writing: Gülümay Vural Topaktaş, Emrullah Arslan, Özlem Anlaş, Bumin Nuri Dündar.

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