Case Report

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A Rare Case of Carney's Complex in an Adult

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ABSTRACT

A 35 year old male came to medical attention when he was seen to have prominent jaw with frontal bossing and large doughy hands. Clinical examination revealed features of acromegaly and multiple lentiginous hyper-melanotic patches on the upper and lower extremities. Imaging revealed multiple left atrial myxomas and a diffusely enlarged adenohypophysis. A diagnosis of Carney's Complex was made, which is a rare autosomal dominant condition, characterized by the triad of endocrinopathies, atrial myxomas and lentiginous skin lesions. He was surgically managed for the same and is currently asymptomatic. He continues to remain under regular follow up at our centre. With about 150 described cases on contemporary literature1, this rare condition, often requiring a multidisciplinary approach for accurate management, remains both a therapeutic challenge and an enigma.

Keywords: *Carney, Endocrine, Acromegaly, Growth, Myxoma, Transnasal, Trans-Sphenoid, Adenoma*

Introduction

First described in literature by J. Aiden Carney in 1985 (who also described Carney's triad, which is entirely different) Carney's complex is a rare autosomal dominant condition with variable penetrance, caused due to the mutation of the PRKAR1A gene[2], encoding the R1-alpha subunit of protein kinase A. A plethora of both cardiac and extra-cardiac involvement co-exist, with the propensity to remain undiagnosed, mainly owing to the erroneous attribution of signs and symptoms as incidental. Our patient, who was diagnosed 05 years ago, presented with classical features, and through our diagnostic and therapeutic endeavours, has given us valuable insight into this rare condition.

Case Report

The patient was apparently asymptomatic 05 years ago, when he visited the hospital for consultation for a next of kin. He was told by our senior colleague to undergo evaluation for his abnormally prominent jaw and frontal bossing, with large doughy hands. This was in addition to his tall stature. However, since the patient had no physical symptoms at the time, he did not seek medical attention. The patient came to medical attention almost a year later. He presented to us with complaints of excessive sweating of six months duration. Examination revealed other acromegalic features, including prognathism, mandibular enlargement, coarse facial features, increased heel pad thickness and a large fleshy nose. Large spade like hands were conspicuously present. A deep and hollow sounding voice was noted. However, arthropathy, kyphosis, carpel tunnel syndrome or acanthosis nigricans were notably absent. A loud S1 was heard. Visual field charting was normal. Additionally, multiple hyper-melanotic skin lesions, each measuring 2-4 mm on the flexor aspect of his right wrist and anterior aspect of right shin.

Magnetic resonance imaging of the brain and heart revealed a pituitary macro-adenoma and a left atrial myxoma, respectively. Histo-pathological examination of above mentioned anomalies, confirmed the prima-facie assumptions made at imaging. The patient underwent resection of the multiple atrial myxomas and a TNTS of the pituitary adenoma, with an uneventful postoperative period. Subsequent evaluation showed no recurrence on yearly follow up visits. However, a non suppressible growth hormone test, suggested persistence of acromegaly. Serial MR imaging of adrenals showed 06 mm nodular lesion in right adrenal gland. USG scrotum was normal. DEXA scan did not suggest increased fracture risk. A right upper lid swelling, detected a year after surgery, was biopsied, which turned a report of benign squamous papilloma.

At present the patient continues to have the skin lesions, but is otherwise asymptomatic. He is under regular follow up and as on date, shows no sign of recurrence, as evidenced by serial MR imaging and hormone assay.

Discussion

Carney's complex, may also present with myxomas of the breast and skin. There may an associated primary pigmented nodular adreno-cortical disease. Psammomatous melanotic schwannomas, sertoli cell tumours and other tumours involving thyroid and other glands and ducts have also been described. These and other described variants, such as cutaneous myxomas (involving face and external auditory meatus) were absent in our patient.. While our patient never exhibited symptoms of hyperprolactinemia, it must be mentioned that the same is possible in Carney's, classically

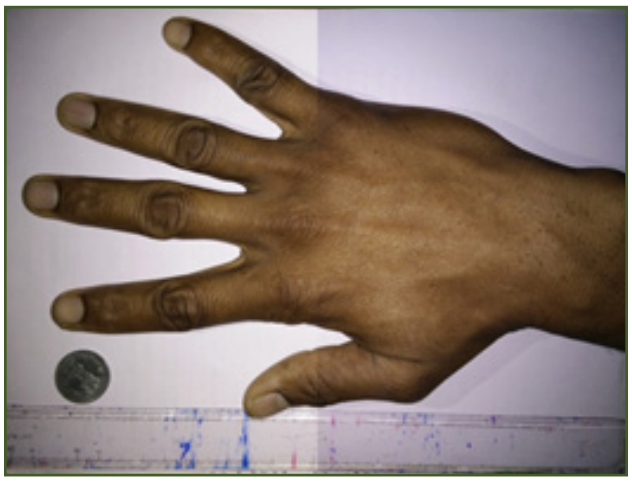

Fig. 1: Large spade like hands.

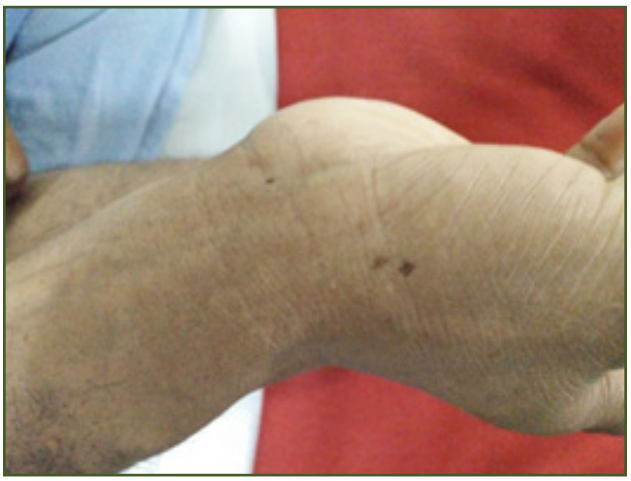

Fig. 2: Hypermelanotic lesions on wrist.

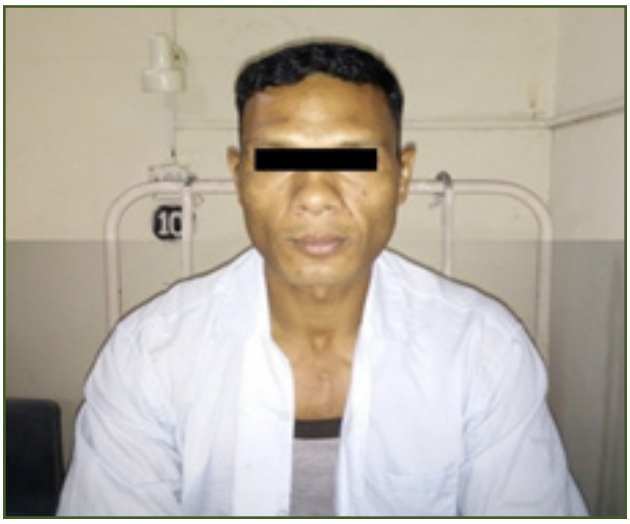

Fig. 2: Hypermelanotic lesions on wrist.

accompanied by visual disturbances, galactorrhea and loss of libido. Also interesting is the fact that the classic spotty pigmentation of the vermillion border of lips occurred, in our patient, more than a year after surgical management. This probably suggests ongoing disease process under the aegis of the mutated PRKAR1A gene. This prompts yearly review of the patient, with MR imaging of the brain and transthoracic echocardiography to preclude recurrence. As on date, our primary concern, in addition to recurrence of primary disease, is the possibility of primary pigmented nodular adreno-cortical disease (PPNAD) suggested by MR imaging of the adrenals. A careful wait and watch approach, in addition to urinary VMA assessment, is probably the best course of action.

Conclusion

Carney's complex is a true, albeit rare, multisystem disease. Our management team consists of an endocrinologist, cardiologist, neurosurgeon and dermatologist. Such diverse and multi-disciplinary involvement is a fitting testament to this syndrome, which, like many others, refuses to identify the compartmentalisation of specialities, that has come to be the hallmark of modern medicine.

Patient's Perspective

The patient has experienced symptomatic relief during the course of our treatment. While his concerns regarding the longevity of his life, owing to the well known cardiovascular complications of acromegaly are well founded, our medical and surgical management aims to allay these concerns. As mentioned above, he continues to remain under frequent review.

Consent

A comprehensive written consent was obtained from the patient for the purposes of this publication.

Competing Interests

None

References

1. Vezzosi D, Vignaux O, Dupin N, Bertherat J. Carney Complex. Clinical and genetic update. Ann Endocrinol (Paris). Dec 2010; 71(6):486-93
2. Groussin L, Horvath A, Jullian E, Boikos S, Rene Corail F, Lefebvre H et al. A PRKAR1A mutation associated with primary pigmented nodular adrenocortical disease in 12 kindreds. J Clin Endocrinol metab. May 2006;91(5):1943-9
3. Urban C, Weinhausel A, Fritsch P, Sovinz P, Weinhandl G, Lackner H et al. Primary pigmented nodular adrenocortical disease and pituitary adenoma in a boy with sporadic Carney complex due to a novel, de novo paternal PRKAR1A mutation. J pediatr endocrinol metab. Feb 2007;20(2):247-52.

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