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A rare case of carney's complex

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Abstract
Thirty-five-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. Investigations 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 atrial myxomas and pituitary macroadenoma and is currently asymptomatic. He continues to remain under regular follow up. With about 750 described cases on contemporary literature[1], this rare condition remains a diagnostic and therapeutic challenge and often requires a multidisciplinary approach for accurate management.

Keywords: carney's complex, endocrinopathy, acromegaly, Myxoma, adenoma

Introduction
Carney's complex (CNC) 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. The victims of this rare ailment often remain undiagnosed because of a significant variability in the both cardiac and extra cardiac involvement leading to non-specific and vague signs and symptoms that are erroneously ignored as incidental. The syndrome was first described by J. Aiden Carney in 1985. CNC was earlier also called as NAME (nevi, atrial myxoma, ephelides) and LAMB (lentigines, atrial myxoma, blue nevi) syndrome.

Case Presentation
The patient was apparently asymptomatic five years ago, when he visited the hospital for consultation for a relative. He was noticed to have tall stature, abnormally prominent jaw and frontal bossing, with large doughy hands by the attending consultant and was advised to undergo evaluation. 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 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 (Fig. 1). Large spade like hands were conspicuously present (Fig. 2). A deep and hollow sounding voice was noted. There was no history of visual disturbances, galactorrhoea or loss of libido. Arthropathy, kyphosis, carpel tunnel syndrome or acanthosis nigricans were absent. Cardiac auscultation revealed loud first sound in mitral area. Visual field charting was normal. Additionally, multiple hyper-melanotic skin lesions, each measuring 2-4 mm on the flexor aspect of his right wrist (Fig. 3) and anterior aspect of right shin.
Magnetic resonance imaging (MRI) of the brain and heart revealed a pituitary macro-adenoma (Fig. 4) and a left atrial myxoma, respectively, both confirmed by histopathology (Fig. 5). The patient underwent resection of the multiple atrial myxomas and a trans-nasal-trans-sphenoidal (TNTS) excision of the pituitary adenoma. The postoperative recovery was uneventful. However, a non- suppressible growth hormone test, suggested persistence of acromegaly. Serial MR imaging of adrenals showed six 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. 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.

Illustrations

Fig 1: Acromegalic facies, with frontal bossing, mandibular enlargement and large fleshy nose

Fig 2: Large spade like hands

Fig 3: Hypermelanotic lesions on wrist

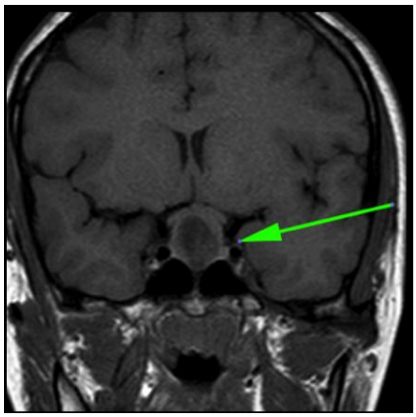

Fig 4: MRI image showing T2 hyperintense lesion in sella and suprasellar region replacing pituitary gland

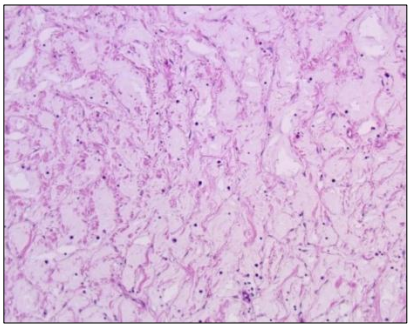

Fig 5: H and E section showing atrial myxoma

Discussion

In addition to atrial myxomas, 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. Our patient did not exhibit the symptoms of hyperprolactinemia. Yet, the same is possible in Carney's complex, classically accompanied by visual disturbances, galactorrhea and loss of libido. Also, interestingly, 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 and therefore necessitates yearly review of the patient, with MRI of the brain and transthoracic echocardiography to detect recurrence, if any. 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 MRI of the adrenals. A careful wait and watch approach, in addition to urinary VMA assessment is the current course of action. Our management team consists of an endocrinologist, cardiologist, neurosurgeon and dermatologist.

Conclusion

Carney's complex is a true, though rare, multisystem disease. The diverse and multisystem involvement seen in this syndrome emphasizes the importance of multi-disciplinary team approach as also highlights the limitations and futility of imaginary 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 the treatment. He is concerned regarding the longevity of his life, owing to the well-known cardiovascular complications of acromegaly, the managing team is doing all that is necessary to allay these concerns. As mentioned above, he continues to remain under regular follow-up.

Consent

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

Competing interests: None

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