CASE REPORT

Carney's Triad—The Full Syndrome with Four Components: A Case Report

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Abstract

Carney's triad is a rare nonhereditary condition affecting young females. Its prevalence is less than one in a million population.[1] It is characterized by gastrointestinal stromal tumors (GISTs), pulmonary chondroma, and extra-adrenal paraganglioma. The genetic alterations in this triad are yet to be understood. It is more appropriately classified as a novel multiple endocrine neoplasia. We report an interesting case of a young lady incidentally detected with tumors composing the triad.

Keywords: Carney's triad—GIST, Paraganglioma, Pulmonary chondroma.

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Introduction

In 1977, J Aiden Carney first described the multitumoral syndrome, Carney's triad.[2] It is an association of three very rare tumors—pulmonary chondromas, gastrointestinal stromal tumors (GIST), and extra-adrenal paragangliomas. The occurrence of these tumors can be either synchronous or metachronous. There are more than 150 case reports published of this condition with most common presentation being pulmonary chondroma and GIST. The disorder commonly affects young women typically and is nonhereditary.

Case Description

A 38-year-old lady presented with symptoms of upper respiratory tract infection. Chest X-ray (Fig. 1) performed as part of routine evaluation incidentally detected a well-circumscribed lesion in the right lower zone of the lung. She was also detected to have high BP recording of 180 to 100 mm Hg and was started on medical therapy. Further evaluation with CT thorax for the lung nodule detected lesions in right adrenal gland, greater and lesser curvature of stomach. These investigations were done elsewhere.

FDG PET done for metastatic workup (Fig. 2), in addition, detected paraganglioma between posterior wall of duodenum and inferior vena cava. To further characterize the right adrenal lesion better, additional MRI of abdomen was performed (Fig. 3).

Biochemical investigations revealed an elevated plasma metanephrine done elsewhere and elevated 24-hour urinary normetanephrine and serum chromogranin.

A diagnosis of Carney's triad was made (Figs. 4 and 5).

She was started on alpha blockade with T. Prazosin XL 5 weeks followed by beta-blocker 1.5 weeks prior to surgery.

Her blood pressure and heart rate were well controlled with the medications.

She underwent laparotomy (Fig. 5)—excision of paraganglioma with right adrenalectomy and excision of GIST that were three in number.

Postoperatively, her recovery was uneventful, and she did not require antihypertensives.

Histopathology was suggestive of paraganglioma with GAPP score 5, right adrenocortical adenoma, and GIST with low risk.

The pulmonary chondroma is planned for surgical excision at a later date.

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Discussion

Pulmonary chondroma, gastrointestinal stromal tumors, and paraganglioma are individually rare tumors. The occurrence of these tumors either synchronously or metachronously was first described in 1977 by J Aiden Carney.[1] The presence of at least two of these components is necessary to make a diagnosis of Carney's triad.

Carney in 1999 had also reviewed the findings in 79 patients with this syndrome (including 48 patients recognized since 1983) where majority had gastric and pulmonary tumors as the most common combination. Young women are affected most often with a mean age of 20.2 years.[2]

Most often, the triad is incomplete and all three components are seen in only about 25% of cases. Patients presenting with any of these components of this triad should be evaluated to exclude the other components, especially functioning paragangliomas.[3] It's unusual for all the three tumors to manifest at the point of diagnosis.

Carney's triad is a nonfamilial condition with unknown etiology. A possible genetic alteration in the mitochondrial enzyme succinate dehydrogenase (SDH) has been proposed, and some case reports have documented the alteration.[3] Previously, neural crest origin for all three tumors of the triad was suggested.[4,5] There is no evidence of familial occurrence, and hence, screening of asymptomatic

family members is not recommended.[6] Comparative genomic hybridization has shown 1 cenq21 deletion to be the most frequent genetic change; however, further studies are needed.[9]

In 2013, retrospective study by Carney et al. described the existence of the fourth component, the adrenal adenoma.

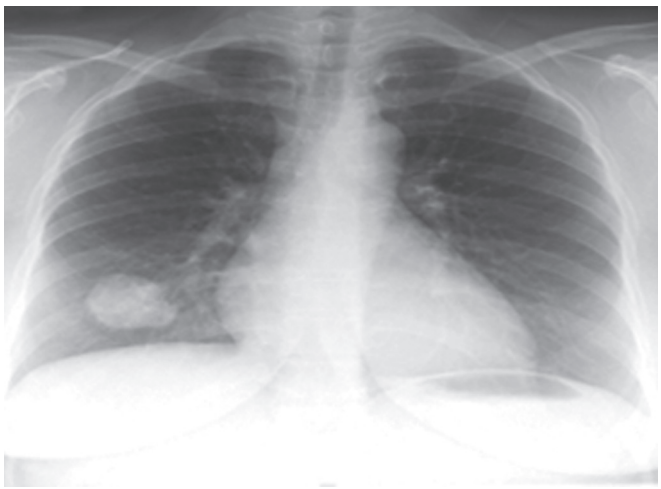


Fig. 1: Well-defined low-density calcified lesion in right lower zone

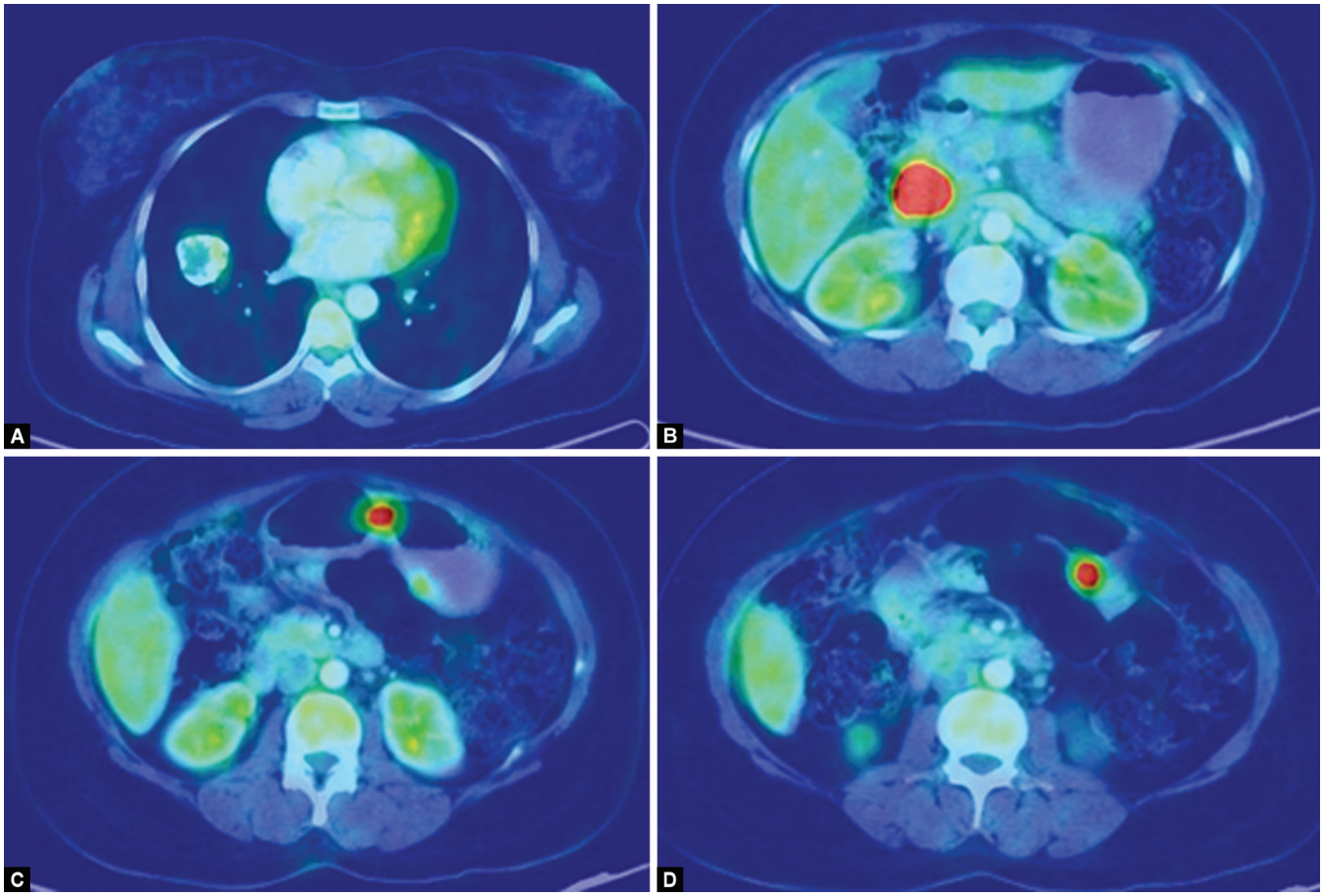
Most often, GIST presents as palpable abdominal mass. These tumors may also present with hemorrhage and anemia and is caused by ulceration of the submucosal lesion into the gastric mucosa.

Although the treatment of choice is surgical, GIST in Carney's triad is indolent. These tumors occur early, can have extensive intra-abdominal metastasis which progresses slowly. The mean age is 12.4 years between initial presentation and recurrence. If recurrence is noted, then surgical treatment is a must as they are known to respond poorly to adjuvant systemic therapy such as imatinib.[7]

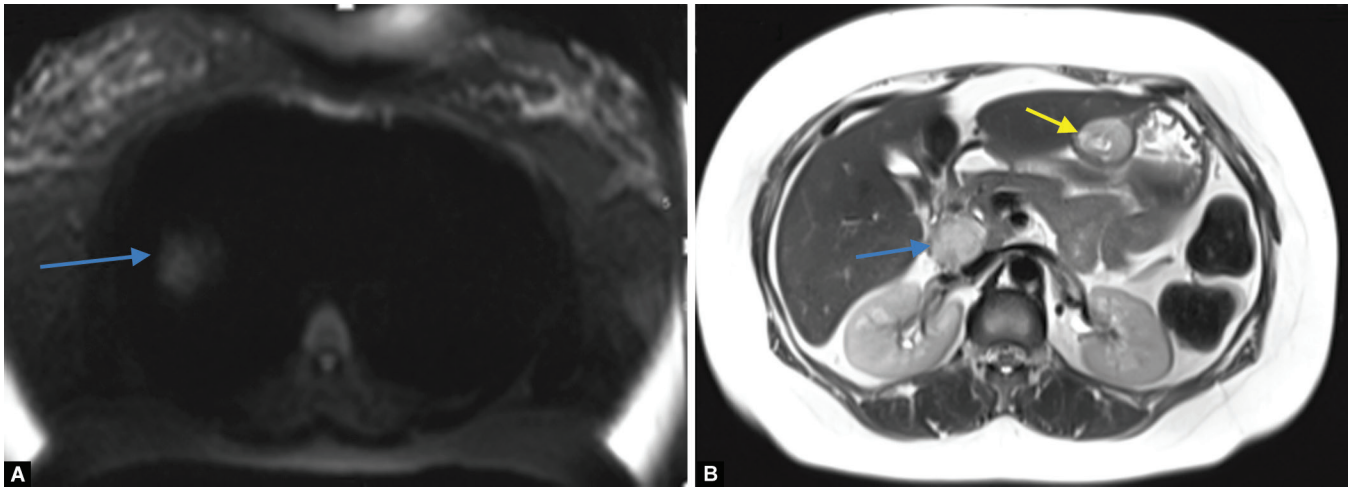
Pulmonary chondromas are typically asymptomatic and present as incidental radiographic findings. They are more often unilateral (83%) and often multiple. These tumors can be followed by annually because of their benign nature.[10] This is only indicated if lung function is impaired and features are suggestive of malignant transformation.[8]

The paraganglioma is the least frequent of the three tumors and are usually catecholamine secreting. These tumors occur in younger patients, are multifocal in nature, and behave benign.[2] Surgical enucleation is the treatment of choice. Metastasis occurs in 10% of patients.

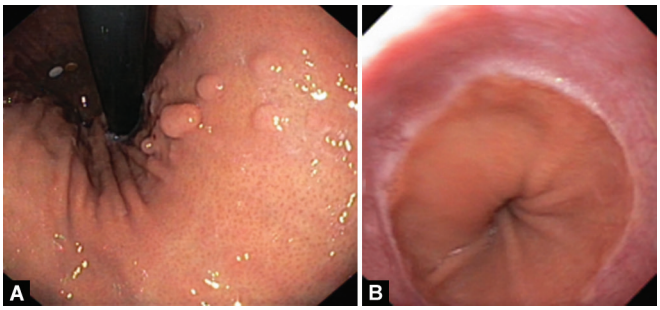
To the best of our knowledge, this is the first case report from India reporting all the components of the Carney's triad, including the adrenal adenoma.



Figs 2A to D: Lung: Exophytic and calcified lesion 32 × 31 mm (SUV 4.60). Retroperitoneal: (3.4 × 3.3 cm) between posterior wall of duodenum and inferior vena cave suggestive of a paraganglioma (SUV 46.24) with a well-marginated, soft tissue enhancing. Right adrenal lesion: 28 × 36 mm (SUV 5.02). GIST: 30 × 24 mm soft tissue lesion in the anterior perigastric region (SUV 45.63). 12 × 13 mm another enhancing soft tissue lesion in the inferior part of stomach—GIST (SUV 16.76). Stomach with two well-marginated exophytic lesions in greater and lesser curvature of stomach



Figs 3A and B: Calcified right lung nodule. A hyperintense lesion in the anterior wall of stomach (arrow in yellow). Another hyperintense lesion in right adrenal gland (arrow in blue)



Figs 4A and B: Although serum gastrin levels were within the normal limits, upper GI scopy showed gastric polyps with erythematous incisura

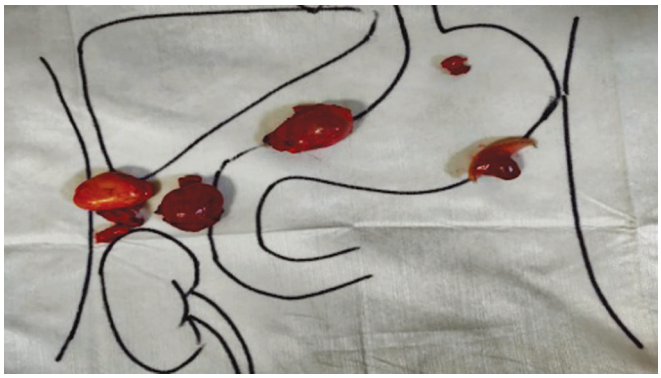


Fig. 5: Intraoperative findings

Conclusion

Low incidence, nonhereditary, young generation, and female predominance make this syndrome unique. Presence of any one of these tumors, especially in patients younger than 40 years, should warrant baseline evaluation to look for the other components. Long-term follow-up is necessary as there could be long interval between presentation, intervention, and relapse.

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