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Carney Triad: A Rare Multisystem Syndrome of Gastric Gastrointestinal Stromal Tumour, Pulmonary Chondroma and Extra-Adrenal Paraganglioma-Imaging Diagnosis of an Incomplete Form: Case Report with Characteristic Radiological Findings

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Abstract: ***Background**: Carney triad is an extremely rare non-hereditary syndrome characterised by the coexistence of gastric gastrointestinal stromal tumours (GISTs), pulmonary chondromas and extra-adrenal paragangliomas. It predominantly affects young women and is caused by succinate dehydrogenase (SDH) deficiency secondary to SDHC promoter hypermethylation. Complete expression of all three tumour types is uncommon; most patients present with an incomplete form. **Case Presentation**: We report a case of incomplete Carney triad in a young adult female who presented with recurrent epigastric pain and iron-deficiency anaemia. Contrast-enhanced CT demonstrated multiple enhancing submucosal gastric masses consistent with GISTs, a hypervascular para-aortic mass compatible with paraganglioma, and multiple pulmonary nodules with coarse ring-and-arc calcifications typical of pulmonary chondromas. Histopathology of the resected gastric tumours confirmed SDH-deficient wild-type GISTs. **Management and Outcome**: The patient underwent partial gastrectomy for the symptomatic GISTs. The pulmonary chondromas were managed conservatively with surveillance imaging. The para-aortic paraganglioma was resected after biochemical evaluation. Long-term multimodal imaging surveillance was instituted because of the risk of metachronous tumours. **Conclusion**: Recognition of the characteristic imaging constellation of multifocal gastric GISTs, calcified pulmonary nodules and hypervascular extra-adrenal masses in a young woman should prompt consideration of Carney triad. Accurate radiological diagnosis enables appropriate surgical planning, genetic counselling and lifelong surveillance.*

Keywords: Carney Triad, Gastrointestinal Stromal Tumour, Pulmonary Chondroma, Paraganglioma, SDH Deficiency, Multidetector Computed Tomography

1. Introduction

Carney triad is a rare non-familial neoplastic syndrome first described by J. Aidan Carney in 1977. It is defined by the association of three tumours: gastric gastrointestinal stromal tumours (GISTs), pulmonary chondromas and extra-adrenal paragangliomas. Fewer than 100 cases have been reported worldwide, with a prevalence estimated at less than 1 in 1 000 000. The condition shows a striking predilection for young women (female-to-male ratio approximately 9 : 1) and usually becomes clinically apparent in the second or third decade of life.

Although designated a triad, complete simultaneous expression of all three components is rare (less than 2–5 % of patients at initial diagnosis). The most frequent combination is gastric GIST plus pulmonary chondroma. The GISTs of Carney triad are characteristically multifocal, gastric in location, SDH-deficient (loss of SDHB immunohistochemical staining) and wild-type for KIT and PDGFRA mutations. Pulmonary chondromas are benign cartilaginous tumours that frequently exhibit characteristic chondroid (ring-and-arc or "popcorn") calcification. Extra-adrenal paragangliomas are hypervascular and may be functional or non-functional.

The underlying molecular mechanism is epigenetic silencing of the SDHC gene through promoter hypermethylation, leading to SDH complex deficiency. This distinguishes Carney triad from the related but hereditary Carney–Stratakis syndrome (GIST + paraganglioma due to germline SDHx mutations) and from the completely unrelated Carney complex (myxomas, pigmented skin lesions and endocrine overactivity).

Because the tumours may develop asynchronously over many years, lifelong multimodal imaging surveillance is essential once the diagnosis is established.

2. Case Report

A young adult female in her mid-twenties presented with a six-month history of intermittent epigastric pain, early satiety and progressive iron-deficiency anaemia. There was no significant past medical or family history. Physical examination revealed mild epigastric tenderness but no palpable mass. Laboratory investigations confirmed microcytic hypochromic anaemia; urinary and plasma metanephrines were within normal limits.
Upper gastrointestinal endoscopy demonstrated multiple submucosal polypoid masses in the gastric body and antrum, several of which showed central ulceration. Contrast-enhanced multidetector CT of the abdomen revealed multiple enhancing, predominantly exophytic soft-tissue masses arising from the gastric wall, ranging from 1.5 to 4.5 cm in diameter. A discrete, intensely enhancing mass was identified in the left para-aortic region adjacent to the adrenal gland but clearly separate from it. CT of the thorax demonstrated

several well-defined pulmonary nodules, the largest in the left upper lobe, containing coarse ring-and-arc calcifications characteristic of chondroid matrix.

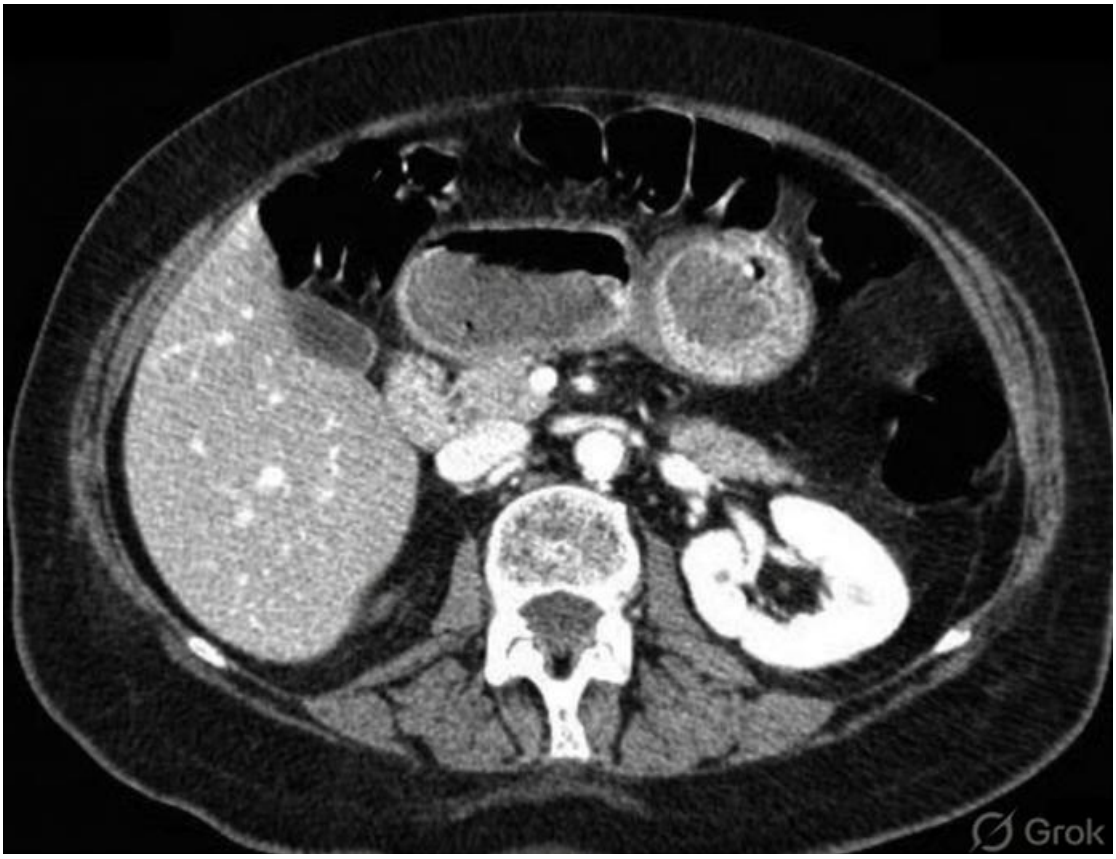

Figure 1: Axial contrast-enhanced CT image of the upper abdomen demonstrating an enhancing submucosal/exophytic gastric mass (consistent with gastrointestinal stromal tumour) together with additional smaller gastric lesions. The constellation of multifocal gastric GISTs in a young woman raises the possibility of Carney triad.

The combination of multifocal gastric masses, a hypervascular extra-adrenal mass and calcified pulmonary nodules in a young woman was considered highly suggestive of incomplete Carney triad. Endoscopic ultrasound-guided biopsy of a gastric lesion confirmed a spindle-cell neoplasm with immunohistochemical loss of SDHB staining and absence of KIT/PDGFRA mutations, establishing the diagnosis of SDH-deficient GIST.

The patient underwent partial gastrectomy with complete resection of the symptomatic GISTs. Histopathology confirmed multiple SDH-deficient GISTs without high-risk features. The para-aortic mass was subsequently resected and proved to be a paraganglioma. The pulmonary chondromas were left in situ and monitored with serial chest CT. The patient remains under long-term surveillance with annual cross-sectional imaging.

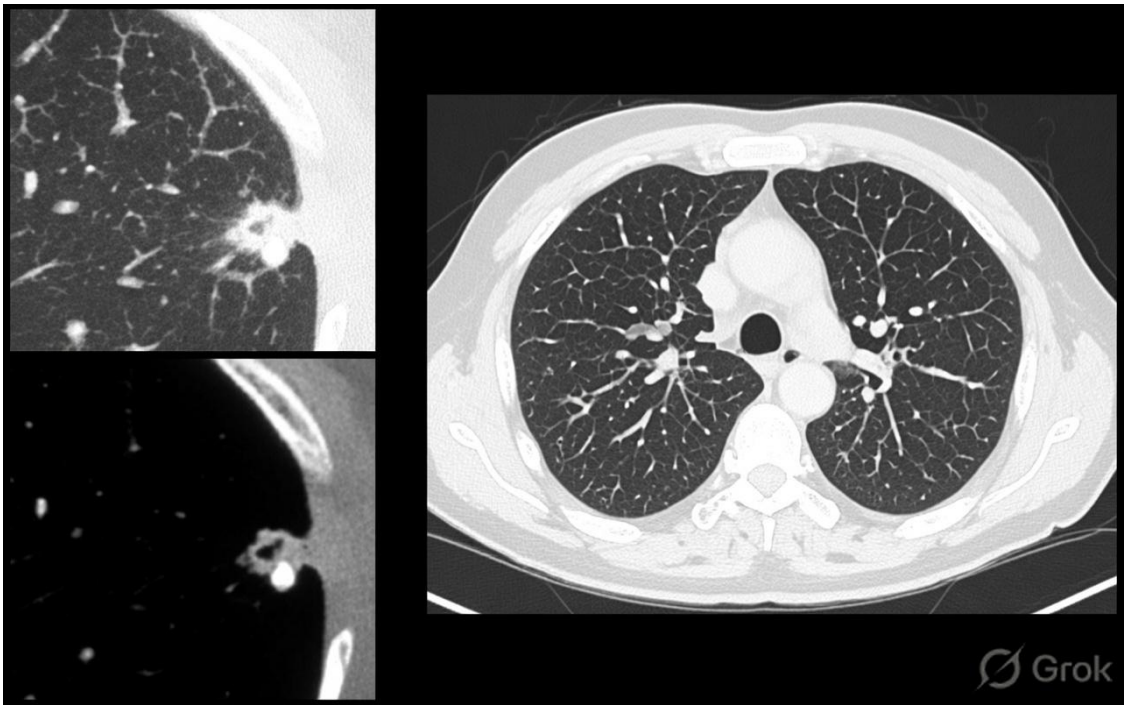

Figure 2: Axial CT images of the chest demonstrating well-defined pulmonary nodules, some containing coarse calcifications. In the appropriate clinical context these findings are consistent with pulmonary chondromas, a key component of Carney triad.

3. Discussion

Carney triad remains one of the rarest tumour syndromes encountered in clinical practice. The radiologist plays a pivotal role because the diagnosis is frequently first suggested by the characteristic combination of imaging findings rather than by clinical suspicion alone.

Gastric GISTs in Carney triad are typically multifocal and may appear as enhancing submucosal or exophytic masses, often with ulceration. Metastases, when present, usually involve the liver or peritoneum rather than lymph nodes. Pulmonary chondromas are the most distinctive thoracic finding; the presence of ring-and-arc or popcorn calcification within otherwise well-circumscribed nodules in a young patient should immediately raise the possibility of the syndrome and prompt a search for gastric and paraganglionic lesions. Extra-adrenal paragangliomas appear as intensely enhancing masses on CT or MRI and may demonstrate the classic "salt-and-pepper" appearance on T2-weighted imaging.

Differential considerations include sporadic multifocal GISTs, Carney–Stratakis syndrome, and metastatic disease. The absence of a family history, the presence of pulmonary chondromas, and the SDH-deficient wild-type molecular profile of the GISTs help distinguish Carney triad from these entities.

Management is primarily surgical for symptomatic or functional tumours. Pulmonary chondromas are benign and rarely require resection unless they cause local complications. Because additional tumours may develop decades after the initial diagnosis, indefinite imaging surveillance is mandatory.

4. Conclusion

Carney triad is an exceptionally rare multisystem neoplastic syndrome whose diagnosis rests heavily on the recognition of a characteristic imaging pattern. The combination of multifocal gastric GISTs, calcified pulmonary nodules and hypervascular extra-adrenal masses in a young woman should prompt the radiologist to suggest the diagnosis, thereby facilitating appropriate surgical management, molecular confirmation and lifelong surveillance.

References

[1] Carney JA, et al. The triad of gastric leiomyosarcoma, functioning extra-adrenal paraganglioma and pulmonary chondroma. N Engl J Med. 1977.
[2] Stratakis CA, Carney JA. The triad of paragangliomas, gastric stromal tumours and pulmonary chondromas (Carney triad). Familial Cancer. 2009.
[3] Sayyouh M, Agarwal P, Lee E. Carney Triad. Radiol Cardiothorac Imaging. 2020;2(4):e200029.
[4] Gaillard F, et al. Carney triad. Radiopaedia.org.
[5] Zhang L, et al. Gastric stromal tumors in Carney triad are different clinically, pathologically, and molecularly from sporadic gastric gastrointestinal stromal tumors. Am J Surg Pathol. 2010.
[6] Boikos SA, et al. Molecular subtypes of KIT/PDGFRA wild-type gastrointestinal stromal tumors. JAMA Oncol. 2016.
[7] Matyakhina L, et al. Genetics of Carney triad. J Clin Endocrinol Metab.
[8] Orphanet. Carney triad. ORPHA:139411.