VOLUME - 11, ISSUE - 05, MAY - 2022 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

General Medicine

CARNEY'S COMPLEX : A CASE REPORT

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ABSTRACT Carney complex (CNC) is an uncommon genetic syndrome of pigmented skin lesions, endocrine dysfunction and myxoma. CNC is often misdiagnosed , identification of some special clinical features and imaging features may help with the diagnosis. Early diagnosis of CNC would vigilant ongoing surveillance of tumors ; the prognosis of CNC may thereforebe enhanced by early treatment.

KEYWORDS :

CASE DESCRIPTION

A 52 year old male came to our cardiology OPD with the chief complaints of chest pain and shortness of breath since 10 days . Patient was apparently asymptomatic before 10 days later he developed chest pain which was intermittent in onset retrosternal in origin and shortness of breath of grade 3 according to NYHA classification , associated with platypnea . He is a known case of hypertension since 5 years and using regular medication .In the past he had history of intestinal polyps for which had a surgical procedure of ressection and anastomosis .

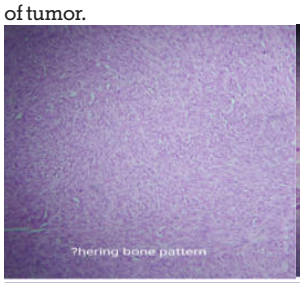
No significant family history

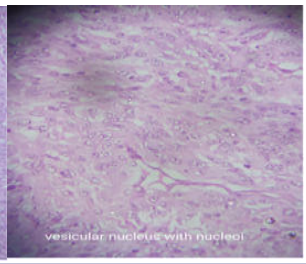
H/o weight loss and fever present. :
O/E:blackish spotty pigmentation present on lips and conjuctiva
CBP report showing anemia with haemoglobin - 8g/dl and other parameters were within the limits
CRP was positive , **ESR** - elevated
ECG was suggestive of left atrial enlargement with sinus rhythm
On 2D-Echo - left atrium dilated, CRHD , severe MR, large pedunculated mass in LA freely mobile

CORONARY ANGIOGRAM: CAD with severe Triple vessel disease

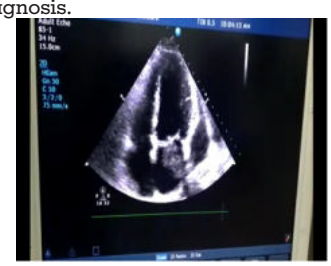
CT chest - hypo dense lesions in right lobe of thyroid and lytic lesions in the vertebral bodies

Radioisotope scan of thyroid gland showing cold nodules suggestive of malignancy & biopsy done showing features of thyroid carcinoma. Patient underwent surgery in our cardiothoracic surgery department for MVR, CABG, excision of tumor.





Histopathology report : d/d undifferntiated pleomorphic sarcoma , fibrosarcoma were given and advised IHC for definitive diagnosis.



DISCUSSION

In the above case we thought a diagnosis of probably CARNEY COMPLEX as the patient had cardiac mass in the left atrium and thyroid neoplasm and spotty pigmentation of lips. CNC is an autosomal dominant disorder, described by multiple endocrine tumors, skin and heart involvement. The diagnostic criteria for CNC are: two MAJOR or one MAJOR with one SUPPLEMENTAL CRITERIA

MAJOR CRITERIA :

- spotty skin pigmentation commonly seen on {lips, conjunctiva and inner or outer canthi, vaginal and penile mucosa}
- myxoma {cutaneous and mucosal}
- cardiac myxoma,
- breast myxomatosis
- PPNAD{ paradoxical positive response of urinary glucocorticoid to dexamethasone administration}
- Acromegaly [secondary to growth hormone [GH] producing adenoma]
- Large-cell calcifying Sertoli cell tumor [LCCSCT] ,
- Thyroid carcinoma
- Psammomatous melanotic schwannoma,
- Blue nevus,
- Breast ductal adenoma,
- Osteochondromyxoma

SUPPLEMENTAL CRITERIA :

- Affected first-degree relative
- Inactivating mutation of the PRKAR1A gene)

Carney complex contributes for 7% of all cardiac myxomas. Eventhough primary tumours in heart are rare still we need

consider them as differentials

Cardiac tumours are classified as primary and secondary tumors. In primary tumors >80% are benign, myxoma is most common among them . Myxomas are commonly sporadic, many autosomal dominant disorders with both lentiginosis and cardiac myxomas are described . In past named , as **LAMB** (lentigines, atrial myxomas, mucocutaneous myxomas, and blue nevi) syndrome **NAME** (nevi, atrial myxoma, myxoid neurofibroma, and ephelides) syndrome, presently grouped under the vast category of CNC

The Carney complex gene identified as the regulatory subunit 1A of protein kinase A **(PRKAR1A)** situated at **17q22-24**. An inactivating heterozygous germ-line mutation PRKAR1A has been documented in about two thirds of individuals with CNC Endocrine hyperactivity is one of the main manifestations of this syndrome. Corticotropin hormone–independent Cushing syndrome because of primary pigmented nodular adrenocortical disease is a vital characteristic

Cardiac myxomas in the CNC usually are many , can occur in any chamber of heart with chance of reoccurance after resection . eventhough they are commonly benign, but related with notable morbidity because of stroke due to tumor embolization and heart failure due to intracardiac valvular obstruction.

CLINICAL SYMPTOMS & SIGNS

Skin features of CNC involve

- Pigmentation { blue nevi of the face, lips, sclera, trunk, or genital mucosa}
- Cutaneous myxomas
- Obesity, cushingoid appearance

Masses

Masses in relation with CNC include

- Cutaneous
- Thyroid
- Breast
- Testis

Neurologic

Stroke due to tumor emboli.

Cardiac

on examination,

- loud first heart sound
- Diastolic apical rumbling murmur (similar to mitral stenosis)
- Holosystolic murmur heard at apex radiation to axilla (mitral regurgitation)
- Tumor "plop"

Constitutional symptoms seen systemically

Diagnosis

Laboratory studies

- Complete blood count (CBC)
- ESR
- Thyroid function tests
- Growth hormone
- 24-hour urinary cortisol excretion test and dexamethasone stimulation test- PPNAD diagnosis as part of CNC

2D-Echo is the investigation of choice for cardiac involvement in the CNC

Mass lesions in CNC require biopsy/resection for histopathological diagnosis.

- **COMPILICATIONS OF CNC**
- Recurrance of myxomas
- Congestive heart failure
- Stroke
- Peripheral embolization,
- Pulmonary hypertension
- Intracardiac myxomas may cause ball-valve obstructions leading to syncopal attacks, cardiac instability and sudden death

Management

In CNC medical treatment is limited for endocrine overactivity, which is usually seen.

- Surgery is performed to resect intracardiac myxomas
- Extracardiac myxomas and nonmyxomatous benign lesions only resected if they produce discomfort through local extension. Large or symptomatic skin myxomas and benign lesions can be excised
- malignant tumors may require resection, along with possibly, adjunctive therapy

CONCLUSION :

This case has been diagnosed as probable carneys complex as patient had spotty pigmentation of lips and conjunctiva ,cold nodules in the thyroid scan suggestive of thyroid carcinoma confirmed with biopsy which fulfills criteria for diagnosis of carneys complex and patient also had history of intestinal polyps and tumor in the left atrium .Patient was operated and had good recovery .HPE report given as a undifferentiated pleomorphic sarcomas

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