

# DELAYED DIAGNOSIS OF CARNEY COMPLEX -A 30 YEAR JOURNEY

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## BACKGROUND:

Carney Complex is rare multiple endocrine neoplasia syndrome affecting adrenal, pituitary and thyroid. Was described for the first time by J. Aidan Carney in 1985[1]

## CASE:

A 54 years old lady had an episode of paroxysmal atrial fibrillation and was referred for routine echocardiogram which she had in December 2014. Echocardiogram showed large mobile mass (myxoma) in left atrium with a well maintained systolic function. She underwent urgent excision of atrial myxoma by Cardiothoracic surgical team.

Post surgery, she went into fast atrial fibrillation and was treated with amiodarone which chemically cardioverted her. She was discharged home after 7 days hospital stay. She presented to hospital again 2 months later complaining of cardiac sounding chest pain and shortness of breath. Her chest was clear, when she was assessed, ECG sinus rhythm, troponin was negative.

She was reviewed by Cardiologist and was for outpatient follow up.

She was reviewed by endocrine team and had pigmented spots on her face (freckles) and coarse facial features and big hands. She is currently on cabergoline 250mcg once weekly, hydrocortisone 15mg am 5mg midday and 5mg evenings, levothyroxine 125mcg od, Bisoprolol 5mg od

Her background include Acromegaly treated by transphenoidal surgery in 1979, followed by radiotherapy, goitre for which she had partial thyroidectomy, hypothyroidism, and family history of myxoma

## CASE PROGRESSION:

The patient remains stable with no evidence of growth hormone excess with growth hormone level of 0.39mcg/L, FT4 17.5 and normal cortisol day curve.

Her last MRI pituitary showed resolution of the pituitary adenoma (minimal amount of pituitary gland tissue in the pituitary fossa)



Fig.1-Echocardiogram showing myxoma – Apical 4 chamber view

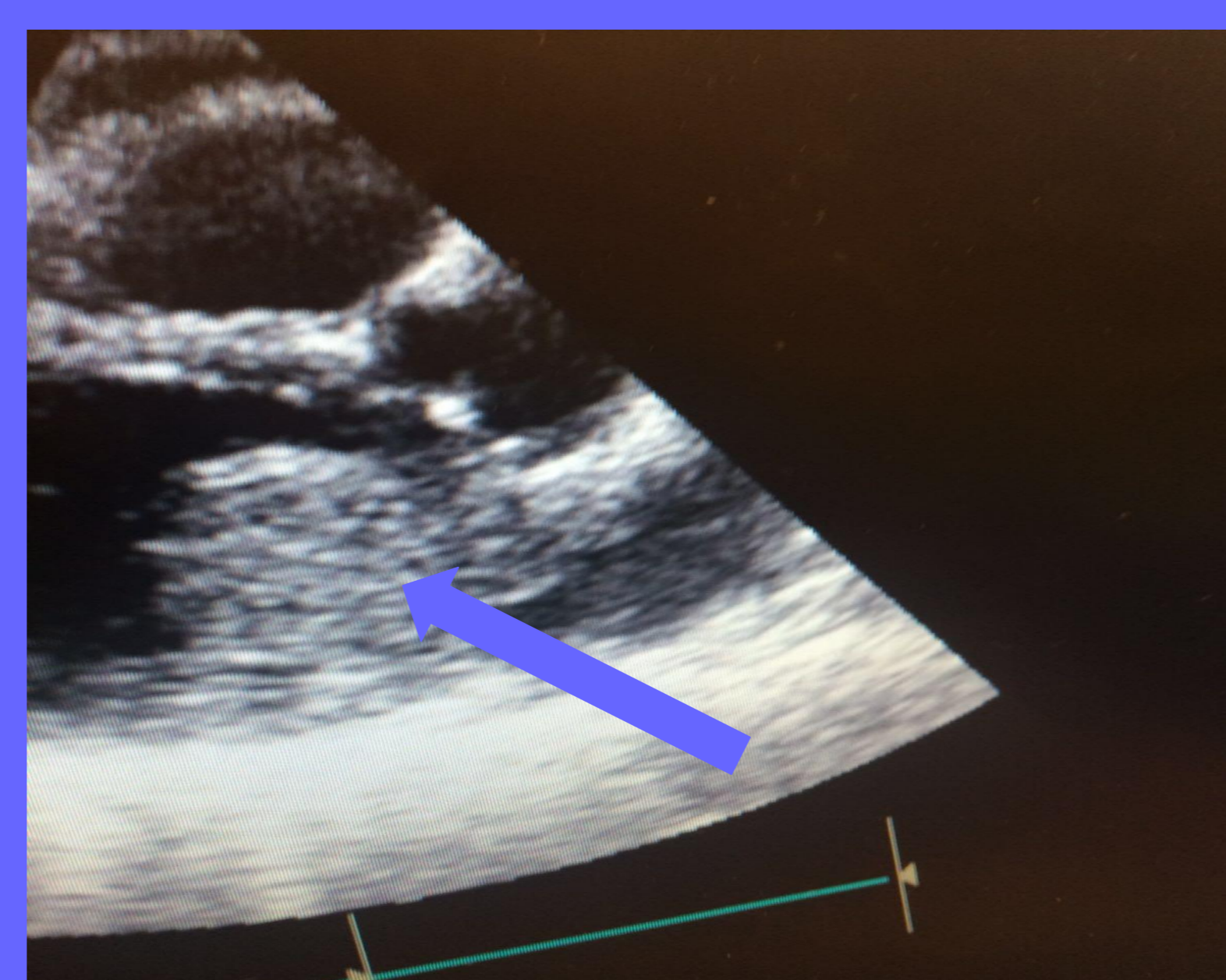


Fig.2-Echocardiogram showing myxoma-Parasternal long axis view

## DISCUSSION

Carney Complex is rare multiple endocrine neoplasia syndrome affecting adrenal, pituitary and thyroid.

It's associated with other non-endocrine tumours such as cardiac, skin, mucosal or breast myxomas, testicular tumours, melanotic shwanomas and abnormal pigmentation (spotty skin pigmentation/freckles) [2,3]

It's an autosomal dominant inherited condition [2,3]

The commonest endocrine manifestation is primary pigmented nodular adrenal cortical disease causing Cushing's syndrome. Others are Large cell calcifying Sertoli cell tumours, as well as growth hormone and prolactin secreting pituitary tumours, thyroid adenoma and ovarian cysts.

## CONCLUSION:

- Patients with Carney Complex should have annual review and annual blood test for IGF1, prolactin and thyroid function test.
- They also need to have annual echocardiogram (ECHO) as well as pituitary MRI, thyroid and testicular/ovarian ultrasound.
- Annual colonoscopy is recommended if acromegaly is part of Carney complex
- Reaching to a unifying diagnosis can be difficult and delayed as demonstrated in this case report
- Our patient's record shows that she had echocardiogram in 2008 and 2010 and there was no mention of atrial abnormality.
- This case report highlights the importance of annual echocardiogram during endocrine clinic review.

## REFERENCES:

1. Carney JA, Gordon H, Carpenter PC, Shenoy BV, Go VL: The complex of myxomas, spotty pigmentation and endocrine overactivity. *Medicine (Baltimore)* 1985;64:270-283.
2. Strakis CA, Kirschner LS, Carney JA: Clinical and molecular features of the Carney complex: diagnostic criteria and recommendation for patient evaluation. *J Clin Endocrinol Metab* 2001;86:4041-4046
3. Oxford handbook of Endocrinology and Diabetes page 610