

# Early recurrent left atrial myxoma in a teenager with *de novo* mutation of Carney complex

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We report a case of an extremely early recurrence of left atrial myxoma in a 13-year-old girl. On hospital admission, the clinical presentation was of cerebral embolism with noticeable spotty skin pigmentation and hypertelorism. The left atrial myxoma originated from the roof of the left atrium. The histology specimen showed typical finding of a myxoma. Six months later a new intracardial mass was evacuated, the postoperative result showing the same type of myxomatous tissue. Genetic investigations demonstrated Carney complex. The genetic analysis of the child's family was negative, demonstrating *de novo* mutation of this rare disorder.

**Key words:** Carney, left atrium, myxoma, recurrence

per year, accounting for 45% primary cardiac tumors in adults and 15% in children.<sup>[3]</sup> Recurrence is reported in sporadic (4%-7%) and familial cases (10%-21%) with the interval between the formation of the new tumor of more than 4 years.<sup>[4]</sup> Although well documented in the adult population, information about cerebral embolism in the pediatric population is still limited.<sup>[5]</sup> The following case illustrates a combination of the two rare diseases clinically presenting with a cerebral stroke and an extremely rapid recurrence of the left atrial myxoma in a teenager.

## Introduction

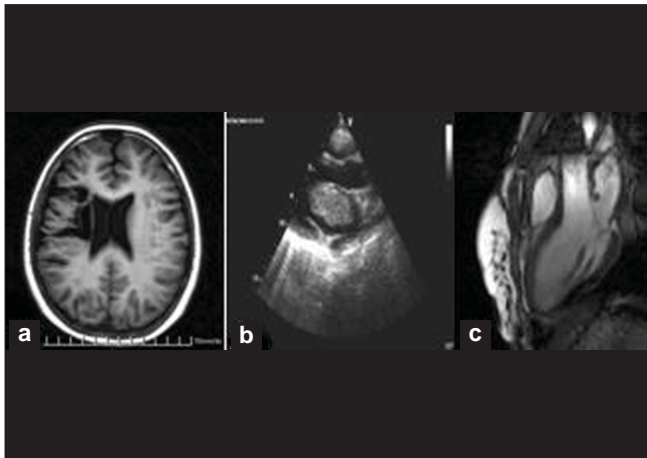
The Carney complex (CNC)<sup>[1]</sup> is a rare dominantly inherited syndrome characterized by skin pigment abnormalities, endocrine over activity and cardiac myxomas. About 600 patients worldwide have been reported by the National Institute of Health (NIH) - Mayo Clinic (USA) and the Cochin centre (France) by January 2008.<sup>[2]</sup> Cardiac myxomas are rare benign tumors with an estimated incidence of 0.5-1 per million population

## Materials and Methods

An athletic, dark-haired 13-year-old girl was admitted to our hospital with an acute migraine attack with vomiting, dispnoea, abdominal pain, and left-sided body weakness. Anamnesis revealed frequent migraines within the previous 2 years associated with vomiting and bulbar motion abnormalities. On admission, she was somnolent, pale, with left facial nerve paresis, left-sided body weakness, hyperactive deep tendon reflexes, and positive plantar reflex. Multiple hyperpigmented skin spots and hairy forearms and thighs were also noted without visible evidence of neurofibromas. Cardiac auscultatory findings, the chest X-ray and the computed tomography (CT) of the brain performed on admission were normal. Electroencephalogram (EEG) showed low voltage delta activity. The repeated CT scan after 12 h demonstrated a massive right-sided fronto-parietal ischemic zone [Figure 1a]. Heart ultrasound

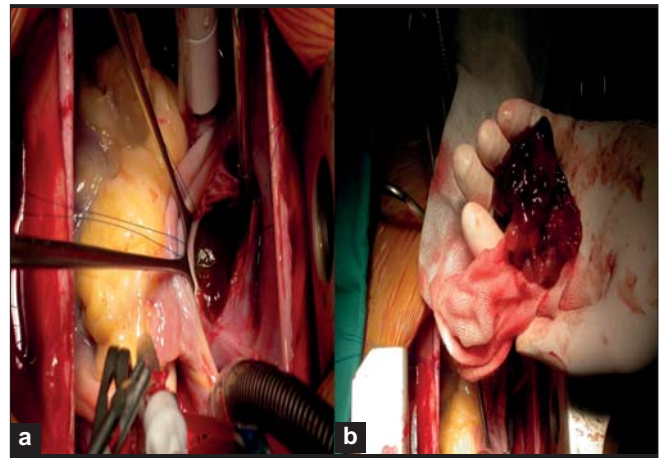
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**Figure 1: (a) computed tomography brain scan. (b) Cardiac Heart ultrasound of primary myxoma. (c) nuclear magnetic resonance imaging of recurrent myxoma**

(ECHO) showed a huge left atrial mass ( $28 \times 37 \times 57\text{mm}$ ) on a short peduncle, arising from the roof of the left atrium protruding through the mitral valve into the left ventricle [Figure 1b]. She was operated after stabilization of her cardiac and cerebrovascular status. The mass was enucleated in toto including a piece of the underlying atrial septum via the left and right atriotomy [Figure 2a, b]. The histopathological findings demonstrated typical cardiac myxoma. The postoperative course was uneventful and the girl was discharged to a rehabilitation centre. Her postoperative cardiac ECHO examination on day 15, 2, and 4 months after surgery showed no residual tumor. She was readmitted to our hospital six months after the initial operation with severe headache, choking and speech difficulties. The cardiac ECHO demonstrated a new tumorous mass ( $8 \times 10\text{ mm}$ ), hanging on a long peduncle from the midportion of the interatrial septum. The finding was confirmed by nuclear magnetic resonance imaging (NMR) [Figure 1c]. On reoperation, the mass was found to originate from the foramen ovale region. The tumor was resected with the underlying septum. The operative and postoperative courses were uneventful. The histology confirmed an identical myxoma as the primary mass. More attention was paid to her hypertelorism and pigmentation but no endocrine abnormalities (thyroid and parathyroid gland, pituitary and adrenal gland) were detected by routine imaging and laboratory investigations. The gynecological findings were within normal limits for a teenage female. The DNA samples were sent to the referent centre



**Figure 2 : (a) Intraoperative view of the tumor (b) The enucleated myxoma**

for Carney complex (Dr Stratakis, National Institute of Health, University of Washington, and Seattle)<sup>[6]</sup> where it was confirmed protein kinase A regulatory subunit 1A mutation (PRKAR1A), c418\_419delCA het in exon 4 in our patient. The genetic investigation of the family was negative, the girl obviously being a new mutation.

Her routine follow up at 1, 3, 6, 12, and 24 months showed no new cardiac masses.

## Results

Intracardiac tumors in the pediatric age group are rare with an incidence of 0.17% in children. Myxomas account for 6% of all pediatric cardiac tumors after rhabdomyomas (63%) and fibromas (6%). Recurrence of the tumor has been reported with the majority of patients being reoperated more than four years after the initial surgery.<sup>[7]</sup> Carney complex is a genetic disease inherited in an autosomal dominant manner, one of the additional criteria being a myxoma (cutaneous, cardiac, breast or bone).<sup>[8]</sup> Our patient with atrial myxoma had a *de novo* Carney complex diagnosed after an extremely short recurrence period of the cardiac tumor.

## Comment

The Carney complex associated with myxoma of various locations is being extensively studied in connection with associated genetic abnormalities.

The most interesting controversy is a rather frequent recurrence of myxoma after complete resection, the fact not observed in most benign neoplasms. In spite of low incidence of the Carney complex, its diagnosis is often established after several years of neurological symptoms and abnormal and clearly visible skin pigmentation. This report supports the possibility of an early recurrence of cardiac myxoma, more likely to be attributed to its underlying genetic nature, rather than incomplete surgical resection.

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