What is in NAME?

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Abstract

An interesting and rare case of name syndrome is reported here. A young patient presented with stroke (left side hemiparesis due to cardiac embolic stroke) and cutaneous lesions. Further investigations revealed that left atrial myxoma was the cause of cardioembolic stroke. Skin lesions were also present which included, nevi, ephelides and neurofibroma hence diagnosis of “NAME SYNDROME” was made.

Introduction

The term “NAME” was proposed as an acronym for “nevi, atrial myxoma, myxoid neurofibroma and ephelides”. Rees et al (1973) described a patient of left atrial myxoma and cutaneous lentiginosis. A similar case was reported by Atherton et al (1980). Atherton and colleagues described a patient with a vast number of macular pigmented lesions on the skin, which involved the lips; myxoid neurofibroma; and domed blue nevi. Echocardiograms revealed dense echoes arising from the space between the mitral leaflets; these findings were consistent with the histological features of a myxoma. Koopman and Happle (1991) suggested that the acronym NAME could stand for nevi, atrial myxoma, mucinosis of skin and endocrine activity.

Case History

A 39 year old female patient presented with rapidly progressive left sided hemiparesis, history of intermittent syncope, dizziness, dyspnoea, fatigue, malaise, weight loss for past 8 months. There was history of skin tags all over body present since childhood. There was no history of hypertension, diabetes, autoimmune disease, Tuberculosis, Seizure disorder. No history of cardiac disease or sudden cardiac death in family. No history of similar skin lesions in other family members including mother, father and only elder sister. Clinical examination revealed regular, normal volume pulse, normal BP, multiple skin tags (Figure 1) all over body, nevi on limb and trunk (Figure 2), multiple brownish black macules (Ephelides) on the skin all over body including upper, back, trunk and limbs (Figure 2). Breast and thyroid examination was normal. Respiratory and abdominal examinations were normal. Cardiac examination revealed loud 1st heart sound, normal S2 and a mid diastolic murmur localized to mitral area, on neurological examination there was left hemiparesis (power grade 2/5 in Left UL, 3/5 in left LL). Investigation showed-Haemoglobin-12.1, total leukocyte count-8300, ESR-81 mm in 1st hour, platelet -4.59 lakhs/uL, 3/5 in left LL). Investigation showed Haemoglobin-12.1, total leukocyte count-8300, ESR-81 mm in 1st hour, platelet -4.59 lakhs/cumm., serum sodium and potassium 140mEq/lt, and 3.8mEq/lt respectively. Random blood sugar-95mg/dl, liver function and renal function tests were normal. X-ray chest and USG Abdomen was normal. Noncontrast CT scan head showed hemorrhagic infarct in right basal ganglia region.

In view of clinical examination and investigations provisional diagnosis of cerebrovascular accident with left side hemiparesis secondary to cardioembolic stroke was made, supportive treatment was started and further investigations done. 2D-Echocardiography revealed enlarged LA (5.1cm), Large LA mass attached to intratrial septum with Normal morphology of all cardiac valves (Figure 3). Cardiac CT scan revealed a lobulated frond like pedunculated soft tissue lesion measuring 62×32×46 mm in the left atrium attached to inter-atrial septum. The lesion showed variegated appearance and mild enhancement on delayed images (28HU plane scans and 44 HU on delayed scans), lesion occupied large part of left atrium and was seen partially prolapsing into left ventricle during systole, the findings most probably was suggestive of cardiac tumor (Figure 4). MRI brain showed mild hypo to hyper intense lesions on T1 suggestive of hemorrhagic infarct in Rt.basal ganglia region. Biopsy of cutaneous tags revealed neurofibroma (Figure 5). A resection surgery of cardiac lesion was done after 6 weeks by cardiothoracic vascular surgeon; a mass was excised with stalk attached via pedicle to wall of left atrium near interatrial septum. The mass was about 6×5 c.m.in size gelatinous, friable, pedunculated attached via pedicle to wall of left atrium near interatrial septum. Biopsy of lesion showed minimal cellularity; only scattered polygonal to spindle cells with scant pink cytoplasm were present in a loose myxoid stroma, cells were concentrated below the endocardium, and findings were suggestive of myxoma (Figure 5). In view of detailed clinical examination and biopsy report of lesion, patient was diagnosed as “NAME” syndrome (nevi, atrial myxoma, myxoid neurofibroma, and ephelides) with embolic stroke with left hemiparesis. Review 2D-ECHO was done 4 weeks after surgery which revealed mild mitral regurgitation with mild tricuspid regurgitation with mildly dilated left atrium, there was no evidence of intracardiac mass or thrombus. Patient is in regular follow-up and healthy. Other family members were also screened for myxoma, 2D –ECHO was normal in all of them.

Discussion

A left atrial myxoma was first described by King TW et al in 1845 on autopsy. The echocardiography has greatly facilitated the antemortem diagnosis of cardiac tumors. Supplemental diagnostic imaging methods include computed tomography (CT) and nuclear magnetic resonance imaging (MRI). Atherton first time suggested the term NAME syndrome nevi, atrial myxoma, myxoid neurofibroma, and ephelides in 1980. Koopman and...
Happle (1991) described autosomal dominant transmission of name syndrome; they recommended the following alternative interpretation of “NAME”: nevi, atrial myxoma, mucinosis of the skin, endocrine over activity. Myxoma occurs in all age groups but is particularly frequent between the third and sixth decades of life. In developing countries like India age of onset is slightly lower (mean age-37.1 years) Women predominate in most series. Myxoma usually occur sporadically, but familial myxoma have been reported. The clinical features of myxomas are determined by their location, size, and mobility. Most patients present with one or more of the triad of embolism, intracardiac obstruction, and constitutional symptoms. Embolism occurs in 30 to 40 percent of patients with myxomas. Depending on their size and mobility, myxoma commonly give rise to signs of obstructed filling of the left or right ventricle with subsequent dyspnea, recurrent pulmonary edema, and right-heart failure.

The signs of myxomas mimic the clinical picture of mitral- or tricuspid-valve stenosis. Zabala et al presented 2 cases of similar myxoma and skin lesions in a family. Wilsher et al presented a family syndrome of cardiac myxoma, myxoid neurofibroma, cutaneous pigmented lesions, and endocrine abnormalities, three
cases from a family of four were presented, and all had cutaneous pigmented lesions. Most of the cases of name syndrome reported so far had familial transmission but our case is sporadic because all other family members did not have any cutaneous lesions and screening 2D-ECHO was also normal in all first degree relatives.

In the present case, auscultatory findings were suggestive of mitral stenosis but cutaneous lesions were telling a different diagnosis. Echocardiography was helpful in the diagnosis of the tumor but final diagnosis was done only by biopsy of resected tumor as secondary tumors of heart are more common than primary tumor. The NAME syndrome is although rare but an important syndrome because cutaneous lesions of the syndrome may help in the early diagnosis of the cardiac myxoma which causes severe morbidity and mortality to patients.

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References