Case report

Pheochromocytoma presenting as a rare and reversible cause of Dilated Cardiomyopathy in a young Adolescent

ABSTRACT

Dilated cardiomyopathy is the most common type of cardiomyopathy in children and adolescents. Most of the cases are idiopathic, with very poor outcome. Identifying a correctable etiology is very crucial as appropriate management could reverse the disease process with complete recovery. Pheochromocytoma and Paragangliomas are catecholamine secreting tumours derived from sympathetic and parasympathetic nervous system. Clinical presentation is highly hetrerogenous, from asymptomatic with incidental finding on imaging or with significant cardiovascular and/or neurological complications.Acute or chronic catecholamine exposure can lead to certain myocardial alterations causing various forms of cardiomyopathy. Most of the such patients are hypertensive. An ultrasound abdomen is a simple, non invasive and easily available investigation which can give a clue to the diagnosis of pheochromocytoma. Early diagnosis and surgical resection of pheochromocytoma is important and can reverse the cardiomyopathy as in our case.

Keywords- dilated cardiomyopathy, pheochromocytoma, paraganglioma, pediatric

INTRODUCTION

Dilated cardiomyopathy (DCM) is a disorder of myocardium with dilatation of left ventricle and poor contractility. This is an important cause of heart failure in pediatric age group. In almost half of the cases the cause is idiopathic and heart transplantation remains the mainstay of therapy[1].High incidence of idiopathic DCM may lead to less attention to look for underlying etiology and thus lead to poor prognosis. Pheochromocytoma and Paragangliomas as such are rare neuroendocrine

catecholamine secreting tumours[2]. Cardiovascular manifestations of pheochromocytoma generally are palpitation, hypertension and left ventricular hypertrophy. Congestive cardiac failure (CCF) secondary to raised cathecholamine level is rare presentation of pheochromocytoma[3]. We are presenting here a case of adolescent male who presented with features of CCF with hypertension and was diagnosed to have pheochromocytoma. Early diagnosis and surgical resection of pheochromocytoma is important and can reverse the cardiomyopathy as in our case.

CASE REPORT –A 14 year old boy presented with intermittent episodes of vomiting, pain in abdomen, headache, palpitations and excessive sweating for last three years. For last 10 days he had heaviness in chest, palpitation, respiratory distress and leg swelling. Family history was not significant. On examination, the pulse rate was 130 beats per minute, blood pressure of 220/130 mm Hg (no significant difference in upper and lower limb) with features of CCF.

Electrocardiogram showed sinus tachycardia. Transthoracic echocardiography revealed dilated Left ventricle (LV) with preserved left ventricular thickness, moderate mitral regurgitation and moderate LV systolic dysfunction with ejection fraction - 35% (Fig -1). Good pulsatile flow in descending aorta ruled out coarctation of aorta. Renal arterial Doppler study was normal. USG Abdomen showed solid to cystic mass over upper pole of left kidney, irregular surface and borders. Plasma free normetanephrines value was very high (4670 pg/ml). Contrast enhanced computed tomography of abdomen showed poorly circumscribed, heterogenous enhancing, soft tissue density focus infiltrating the left adrenal gland and anterior aspect of left kidney measuring about 6.1x3.7x5 cm (Fig -2). Similar conglomerated, necrotic foci seen in the retroperitoneal and the left pre-sacral spaces. Genetic testing and Positron emission tomography (PET) scan was planned but could not be done due to financial constraints.

Treatment:-The patient was initially managed with diuretic, nitroglycerine infusion, oral alpha blocker and beta blocker for the control of blood pressure and CCF. After adequate preoperative preparation the patient underwent surgical excision of paragangliomas and left pheochromocytoma (Adrenalectomy).After surgical resection of tumor, the patient was followed up on an alpha blocker and a beta blocker initially. Plasma normetanephrine level done after 6 weeks of surgery was normal.

Later on all antihypertensive medications were phased out over a period of 5 months. The LV function and size also become normal in follow up period of 8 months.

Figure 1 – ECHO at the time of admission (EF-35%)

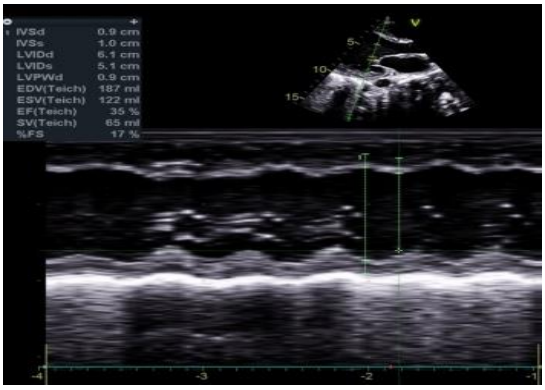


Figure 2 – CECT abdomen showing left adrenal pheochromocytoma

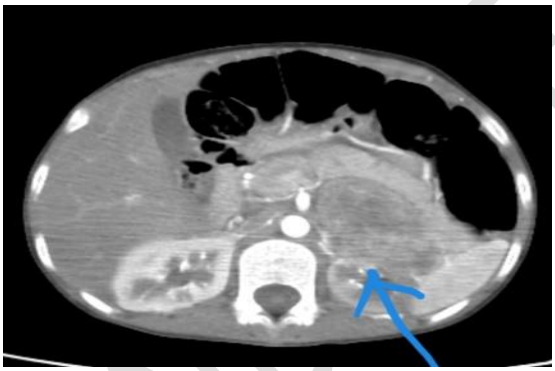
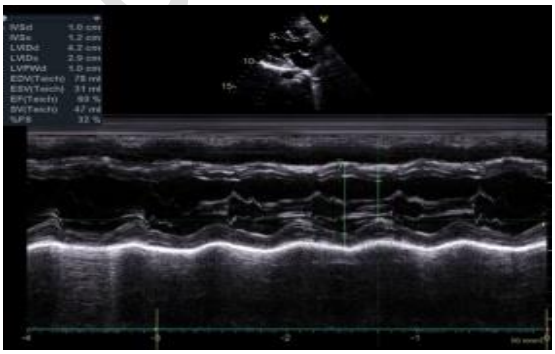


Figure 3 – ECHO image after 8 month of surgey (EF-63%)



DISCUSSION

Nearly half of the cases of DCM are idiopathic and pheochromocytoma as a cause of DCM is very rare. Pheochromocytoma is a rare tumor and seen only in 0.2-0.5 % cases of hypertension, which if missed or not treated properly can be fatal to the patient[4]. DCM is seen in 8-11% of patients with pheochromocytoma[5]. *A* study reported 163 cases of pheochromocytoma and cardiomyopathy and resection of the pheochromocytoma led to improvement of the cardiomyopathy in 96% of cases[6].The typical features of pheochromocytoma include headache, palpitation, excessive sweating and intermittent or persistent hypertension[7]. Cardiomyopathy in pheochromocytoma can be due to elevated level of catecholamines causing downregulation of beta adrenergic receptors, direct toxic effect on myocardial fibers or secondary excessive stimulation of adrenergic level causing hypertensive cardiomyopathy[8]. The sensitivity of plasma normetanephrines and metanephrines was 100% and the negative predictive value of normal plasma metanephrines was 100%. Pheochromocytoma and paraganglioma are localized by a CT/MRI of the adrenal glands and abdomen; I-123-MIBGscintigraphy and 18F-DHPA-PET are complementary[9]. Screening for genetic alterations is also important. Preoperative preparation with α-blockade followed by laparoscopic and adrenal sparing surgical resection is the treatment of choice and usually curative. In malignant pheochromocytomas, radiotherapy and chemotherapy are palliative treatment options[2]. This is followed by resolution of the cardiomyopathy as the catecholamine excess is resolved.

CONCLUSION

Although rare, diagnosis of pheochromocytoma should be considered in cases of DCM associated with hypertension . Early management of pheochromocytoma can lead to reversal of cardiomyopathy.

Disclaimer (Artificial intelligence)

Author(s) hereby declare that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc.) and text-to-image generators have been used during the writing or editing of this manuscript.

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