Case Reports and Reviews

Case Report

Genetic Cause of Chest Pain in Children

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Introduction

Chest pain in children is a challenge. The most important thing in a patient suffering from chest pain is exclude any other symptoms. There is in some cases an association of headache and chest pain. About to name this syndrome as chest pain - hypertension syndrome. There are parameters as history and investigations to diagnose this syndrome. There is must be a familial incidence to diagnose this syndrome. The familial incidence from a paternal side is documented. The genetic test is used to complete the diagnosis in association with high hormonal level of catecholamin. In these cases, the cardiac enzymes mostly are normal.

As a role in a case of chest pain the echocardiography and electrocardiogram are important to be done. Some cases need abdominal ultrasound to exclude tumor releasing hormone. The neuroendocrine tumors pheochromocytoma and paraganglioma are the cause of hypertension in 0.5–2% of pediatric cases [1, 2].

Case presentation

A 8 years old boy was suffering from chest pain at the age of 4 years. The pain at the lateral side of the chest and increase with playing and effort. Examination done and normal all vital signs, no audible murmur. Electrocardiography and ECG was normal. Investigation like creatin phosphokinase (CPK) and troponin 1 was done and normal.

The co-arction of the aorta was excluded and the cardiac function was normal. Ejection fraction and fraction shortening were 70% and 29% respectively. After 2 years the patient suffering from headache. There is no vomiting and not related to any day time. Blood pressure was measured and the recorded as 220/140 mmHg. There is a history of hypertension in the family. The uncles are on antihypertensive medication but not known if primary or secondary type of hypertension. Admission to pediatric intensive care unit was done. The case was started to be treated as hypertensive encephalopathy .

Urgent cannula insertion and hydralazin infusion was taken. No change in blood pressure still 220/140 mmHg even after 4 hours of infusion. At that time all investigations done.

Abdominal ultrasound and electrocardiography was ordered and the result was a mass in the abdomen and depressed cardiac function, respectively. Ejection fraction and fraction shortening were 50 % and 15 % respectively. Left ventricular hypertrophy. The diameter of the interventricular septum was 1.5 cm (Figure 1).

The most important finding in abdominal ultrasound (Figure 2) is supra renal mass. The report was written and show: a well-defined hyperechoic left supra-renal mass , measured (5.5 X 4 cm). It is seen completely separable from the left renal capsule and it shows peripheral vascularity. The recommendation was magnetic resonance image (MRI) abdomen with contrast and confirm the diagnosis (Figure 3)

Urgent order for lab investigation for catecholamine. The level was critical level (Table 1) . Alpha and beta blocker was started and the blood pressure start to response to treatment.

Referral done for high center for resection of the tumor and para aortic lymph node.

After the operation there was smooth course and no hypotension was recorded. Genetic study was done and the result was mutation mutations in genes encoding subunits of succinate dehydrogenase (SDHx) . The interesting thing is this large mass not cause morbidity or mortality for the patient, Although one uncle die due to sever uncontrolled hypertension. Resection done of the tumor and smooth post-operative course.

Discussion

Hypertension in children may be primary or secondary. The association of chest pain with hypertension is one of the red flag symptom that is important for early diagnosis.

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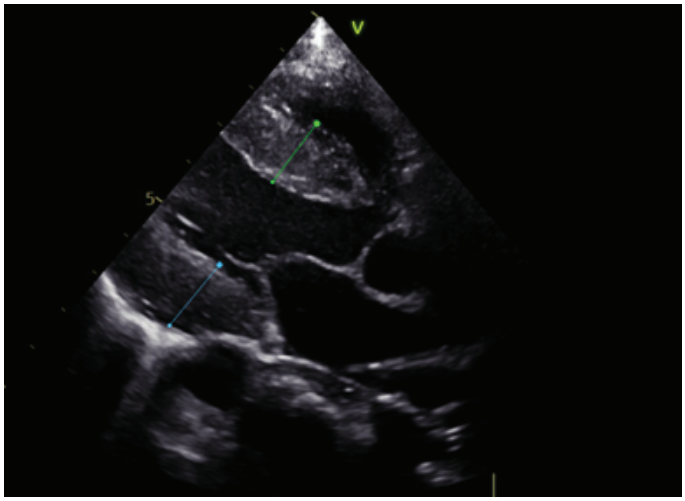

Figure 1: *Transthoracic echocardiography in left parasternal view. The interventicular septum was 1.8 cm (green colour), posterior wall of the left ventricle (blue colour) was 1 cm. This is type of hypertrophic cardiomyopathy.*

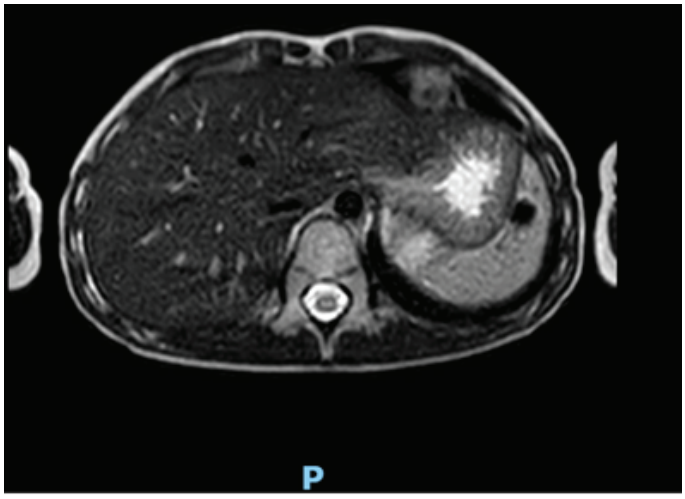

Figure 2: *Abdominal computed tomography with contrast show a left mass at the supra renal gland.*

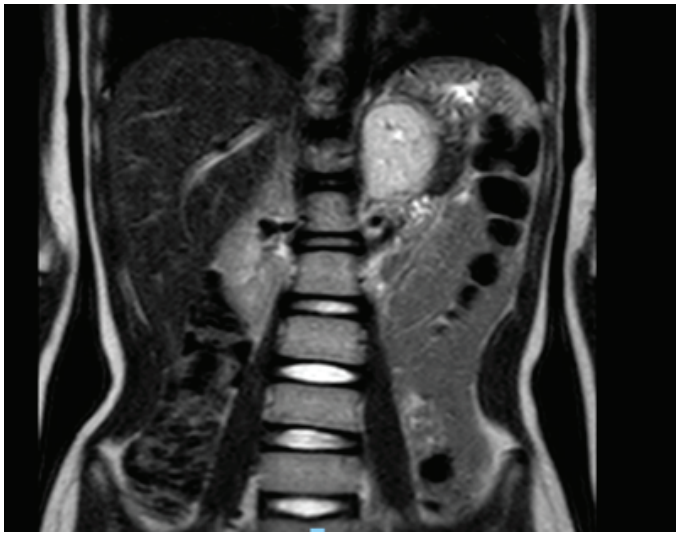

Figure 3: *Magnetic resonance image shows a huge mass in the left adrenal gland.*

Table 1: *Critical level of chatecholamins.*

Test	Result	Unit	Ref. Range
Urine Volume	450	ml	
Adrenaline	7.31 H	ug/24 H	4-20
Noradrenaline	676.78 H	ug/24 H	23-105
Dopamine	490.65 H	ug/24 H	2-144

Paragangliomas (PGLs) and pheochromocytomas (PCs) are rare tumors arising from cells of the autonomic nervous system and adrenal medulla, respectively. PGLs can originate from parasympathetic and sympathetic paraganglia of the head and neck, thorax, abdomen, and pelvis. Most head and neck PGLs (HNPGLs) are paraympathetic in origin and do not secrete any catecholamines, whereas PCs and abdominal PGLs are of sympathetic origin and often secrete catecholamines. Only approximately 2% of PGLs occur in the thoracic cavity or mediastinum; however, these tumors often exhibit an aggressive phenotype [3].

In our case the genetic test was done and the diagnosis was mutations in genes encoding subunits of succinate dehydrogenase (SDHx). In this mutation there is family inheritance of the hypertension. The importance of this test to be done in the family member is pattern of inheritance and association of other type of mutation. Other causes of hypertension are clinically excluded according to the clinical picture or the gene study.

The case was diagnoses as hereditary PGL syndromes. Several distinct familial PGL syndromes (PGL 1-5) are caused by mutations in genes encoding subunits of succinate dehydrogenase (SDHx) itself or factors necessary for the correct assembly of the SDH complex [4].

The massage from this case is if there is chest pain in children and even no hypertension . Important to follow up the patient in the outpatient clinic on regular base as the early hypertension that can be detected in the clinic can discover an early respectable neuroendocrine tumors.

The detailed family history is important and should be take in clear and detailed manner

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