Case Report

Case report of familial hypercholesterolemia with internal carotid neck swelling

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Abstract

Familial hypercholesterolemia is a multi-gene disorder affecting 1 in 200-500 individual which is characterized by increased level of total cholesterol and low density lipoprotein (LDL) which deposit cholesterol reach protein leading to xanthomas, corneal arcus and prone to develop risk for atherosclerosis and coronary heart disease. 11 year female, born of consanguineous marriage attended with yellowish patches (Xanthelasma) bilaterally present over upper eyelid, buttock since 5-6 years. In family history younger girl along with both parents has Xanthelasma. In investigations, routine blood, urine and ECG& ECHO was normal except lipid profile of patient and sibling was abnormal. Child was treated with atorvastatin and dietary advice. In follow up after 3 month, child had complained of neck pain and swelling to left side. Then USG neck and Color Doppler showed reduced diastolic flow of left ICA and type two plaques and type 111 plaque in Rt common carotid artery. Anti-platelets was added and referred to cardiologist for coronary angiography which showed LAD – proximal mid distal diffuse plaque, LCX- proximal eccentric plaque. This case highlights the significance of conscious and proactive genetic screening for familial hypercholesterolemia especially in consanguineous marriage, to diagnose, treat and prevent mortality with premature coronary artery disease.

Keywords: Atherosclerosis, behavior modification, coronary disease, familial hypercholesterolemia, xanthomas

Introduction

Familial hypercholesterolemia is a multi-gene disorder.[1] This is characterized by increased level of total cholesterol and low-density lipoprotein (LDL)[2] which lead to deposit in the form of xanthomas, corneal arcus and risk for atherosclerosis and coronary heart disease. The most widely accepted criteria for diagnosis of definite FH for children less than 16 year is Simon Broome's criteria which contain the presence of total cholesterol level > 260 mg/dl or LDL-C > 155 mg/dl plus at least one physical finding = tendon xanthomas, or tendon xanthomas in first- or second-degree relative or DNA-based evidence of an LDL-receptor mutation, familial defective apo B-100 or a PCSK9 mutation.[3] An increase in cholesterol level is the condition which initially no sign and symptom but important risk factor for atherosclerosis leading to premature ischemic heart disease (PIHD, coronary vascular disease (CVD) and cerebrovascular atherosclerosis, subsequently premature death.[1,2] We are describing a case of familial hypercholesterolemia with strong family history in which early detection and aggressive treatment along with first-degree family screening may reduce morbidity and mortality.

Case Description

11-year-old female Muslim, birth of consanguineous marriage visited OPD with complain of yellowish patches bilaterally present over upper eyelid, buttock and elbow since 5-6 years. child had no history of fever, cough, breathlessness and chest pain. Child had no previous history of hypothyroidism, nephrotic

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Figure 1: Showing xanthelasma over eye lid

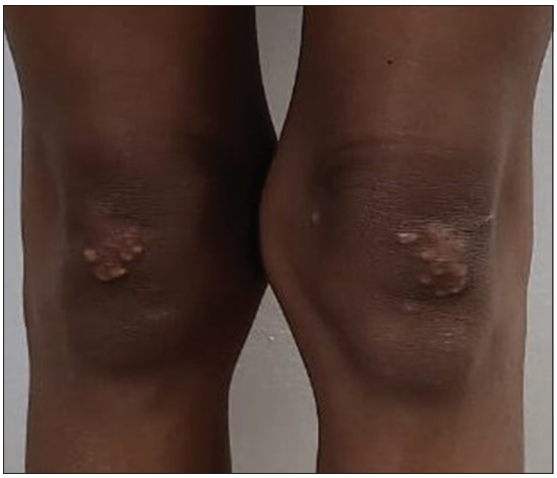

Figure 2: Showing xanthelasma over B/L knee

syndrome or any long duration of drug intake. In family history younger girl sibling has similar complain of xanthelasma. Mother and father have also history of xanthelasma. On examination child had xanthelasma present as previous described [Figures 1 and 2]. In anthropometry child height, weight and BMI were within normal limit. Vitals including BP were within normal limit. On the basis of above description familial hypercholesterolemia diagnosis was suspected.

Investigation and treatment

In initial CBC, KFT, LFT were normal. Random blood sugar was normal. TSH and T4, T3 and routine examination of urine were normal. X-ray chest, ECG and ECHO was normal. Lipid profile of patient and sibling were abnormal as mention in Table 1.

Child was treated initially with atorvastatin and dietary advice and genetic counseling were done but due to poor affordability genetic study was not done.

Follow up

After 3 month, child had come with complain of neck pain confined to left side along with swelling 1.5 cm into 1 cm tender. In oral cavity examination was normal and one week of amoxicillin treatment symptom was not improved. Then USG neck shows vascular swelling followed by Color Doppler showed reduced diastolic flow of left ICA and type II plaque and type III plaque in right common carotid artery [Figure 3]. Anti-platelets were added and referred to cardiologist for coronary angiography and further management. In coronary angiography at higher center showed LAD – proximal mid-distal diffuse plaque, LCX-proximal eccentric plaque.

Discussion

Hyperlipidemia is occurred due to increased concentration of plasma lipoprotein and changes resulting from genetic defect are classified as primary disorder of lipoprotein metabolism.

Table 1: Lipid profile of patient and parent

	Patient	Sibling	Mother	Father
Cholesterol level	587	578.4	263.9	307.8
Triglyceride	244	235.7	404.4	372.6
HDL	30	41.9	35.5	31.9
LDL	492	489.4	147.6	201.3
VLDL	48.8	47.1	80.9	74.5

FH is a monogenic, autosomal dominant that manifested in heterozygous form in approx. 1 in 500 individuals. Homozygous are occurred approximately 1 in a million.[4] The total cholesterol level is much higher in homozygous variant (500-1000 mg/dl) with respect to heterozygous variant (325-450 mg/dl). Incidence of premature coronary artery disease is an early manifestation in homozygous variant (second decade) and xanthoma occurred in first decade.[5] In recent meta-analysis (18 studies), it has found that risk of PIHD, CVD, cerebrovascular event and premature death has significantly increased in patients suffering from familial hypercholesterolemia.[6] My index case is clinically homozygous in which total cholesterol 587 mg/dl, LDL-C 492 mg/dl and sibling 578.4 mg/dl and xanthomas manifested at 5 yrs. of age and internal carotid atheroma and left coronary artery atherosclerosis at 11 year.

The index case as a homozygous variant of FH would improve little to drug therapy, so high dose along with combination of HMG CoA inhibitor, Nicotinic acid and fibrates should be used. If multidrug treatment doesn't show adequate therapeutic result, then LDL apheresis is used.[7]

Conclusion

This case report highlights the significance of conscious and proactive genetic screening for familial hypercholesterolemia especially in consanguineous marriage, to find the condition, guide and management to reduce morbidity and mortality related with it.

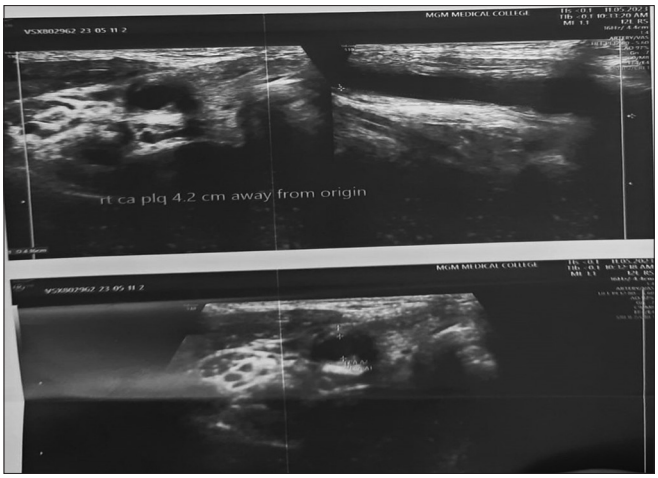

Figure 3: Color Doppler showing plaque in common carotid artery

What's adding this study?

1. The genetic screening especially in consanguineous marriage for familial hypercholesterolemia for early detection and better management.
2. Color Doppler of internal carotid artery to find intimal thickening or atherosclerotic plaque which can predict the coronary artery syndrome.
3. Early aggressive treatment of FH and control of risk factor will help to reduce the morbidity and mortality associated with the disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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