ORIGINAL RESEARCH PAPER
Volume-8 | Issue-3 | March-2019 | PRINT ISSN No 2277 - 8179
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

FAMILIAL HYPERCHOLESTEROLEMIA-CASE REPORT OF RARE CAUSE OF DYSLIPIDEMIA

Paediatrics

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ABSTRACT

Familial hypercholestremia is an autosomal dominant genetic disorder due to mutations in LDL receptor gene resulting in high serum cholesterol & accumulation in tissues. Familial hypercholestremia is an important risk factor for atherosclerosis & premature coronary artery disease. we report a case of 13 year old boy with history of multiple xanthomas and severe hypercholestremia. Both parents had hypercholestremia with family history of similar complaints & death of sibling. Lipid profile & Doppler study of carotid arteries showed abnormality. The child was treated accordingly (lipid converting agents & life style modification and was on followup.

KEYWORDS

Familial hypercholestremia, coronary artery disease, xanthomas.

INTRODUCTION:

Familial hypercholesterolemia (FH) is a monogenic autosomal codominant disorder characterized by strikingly elevated LDL cholesterol, premature cardiovascular disease and tendon xanthomas(1).

CASE REPORT:

13 year old boy admitted in our hospital with chief complaints of multiple nodular lesions over shoulders, upper and lower limbs,buttocks and interdigital web spaces since the age of 4 years. Child underwent multiple checkups in various hospitals and was underdiagnosed. Child has strong family history with similar lesions in his sibling.

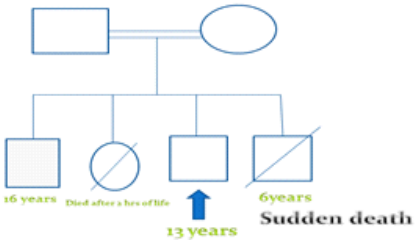


FIGURE 1: family history of child

Anthropometry: Weight: 30kg Expected: 39kg
Height: 139cm Expected:149cm
BMI: 15.54

Vitals: Pulse rate: 80/min, regular, normal rhythm, no radio femoral delay, all peripheral pulses are felt equally.
Blood pressure:110/80 mm Hg.

Head to Toe Examination:

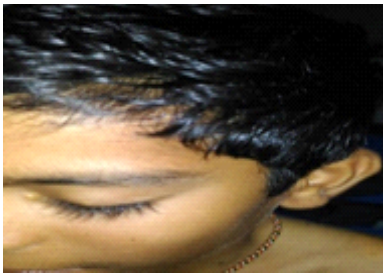
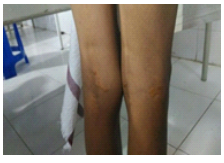


Figure 2: Xanthelesma present over both upper eye lids



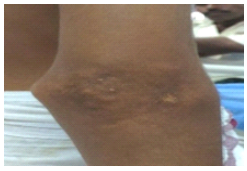


Figure 3: Xanthomas present over extensor aspect of elbows, B/L popliteal fossa.

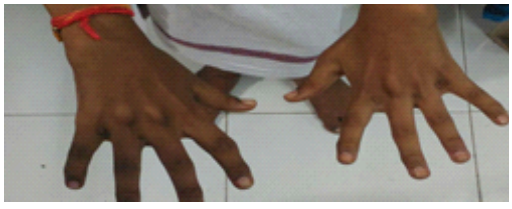


figure 4: Tuberous xanthomas over interdigital web spaces.

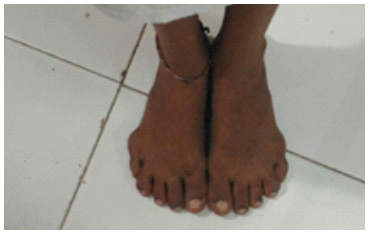


Figure 5: Tendinous xanthomas on lateral aspect of foot.

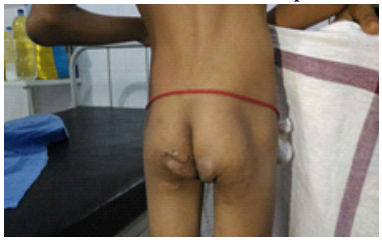


Figure 6: Tuberoeruptive xanthomas present over both buttocks.

Systemic Examination:

- Ejection systolic murmur is heard in aortic area radiating to carotids.
- Bruit is heard over both carotids.
- Rest of examination is unremarkable.
- Ophthalmic examination is normal.

Investigations:
Basic haematological parameters, liver enzymes, fasting blood sugar levels, thyroid function tests were within normal range.

Lipid profile	Patient	father	Mother
TOTAL CHOLESTEROL	547 mg/dl	338 mg/dl	350 mg/dl
HDL	40 mg/dl	41 mg/dl	44 mg/dl
LDL	492 mg/dl	265 mg/dl	280 mg/dl
VLDL	15 mg/dl	31 mg/dl	30 mg/dl
TRIGLYCERIDES	77 mg/dl	157 mg/dl	130 mg/dl

- 2D Echo was normal
- ECG was normal
- Carotid Doppler had shown diffuse atherosclerotic changes.
- The child was diagnosed by clinical and laboratory investigation as a case of homozygous familial hypercholesterolemia.
- He was started on Tab Atorvastatin 20mg,Tab Ezetimibe 10mg, Tab Ecospirin 75 mg and dietary advise given.
- Specific mutational analysis and LDL receptor studies were not done due to limited available facilities.

Follow up:
We recommended regular follow up, including Doppler echocardiographic evaluation of heart and aorta annually.

DISCUSSION:
Familial hypercholesterolemia is one of the most common and most severe form of monogenic hypercholesterolemia. It was the first genetic disease of lipid metabolism to be clinically and molecular characterized2. The incidence of heterozygous FH is 1:500 whereas homozygous FH is rare occurring in 1:1,000,0003.

The patients with this disorder usually present with appearance of cutaneous xanthomas, formation of corneal arcus, development of atherosclerosis at very early age and enlarged tendon achilies. The current management of this disorder are lifestyle modification, statin treatment and lipoprotein apheresis. These patients should have psychological support. Surgery is required for the removal of large cutaneous or tuberous xanthomas. Homozygous FH epitomizes the potentially fatal cardiovascular consequences of exposure to extreme hypercholesterolemia from birth onwards. So, these patients should have regular follow up.

Conclusion:
Since, it is the striking physical sign which aids in diagnosis, it is very important to identify xanthomas early in life in order to commence treatment early which would improve the outcome.

Conflict of Interest:
There is no conflict of interest among the authors.

Funding Statement:
There is no funding for the case report.

Ethical Approval:
Necessary approval was taken from the Institution and the patients for carrying out this work

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