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A Rare Case of Familial Hypercholesterolemia with Triple Vessel Disease in a Young Medico who Underwent CABG

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Abstract

A 22-year-old male- normotensive, euglycemic, diagnosed with familial hypercholesterolemia with tuberous xanthomas at six years. He had recurrent angina with ECG showing abnormalities. Echo and TMT were done. X-ray chest was normal. Echo, LV normal, no PHT, EF65%, No RWMA, TMT positive. He was taken up for coronary angiogram, which revealed significant triple vessel disease. He was given anti-lipid measures and underwent coronary bypass surgery.

This case is being presented for rare manifestations of active coronary artery disease in a very young boy with lipid abnormality requiring CABG.

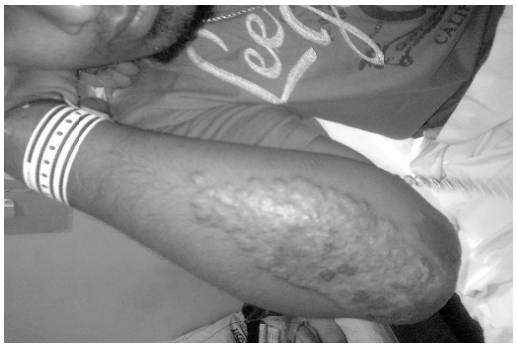


Fig 1

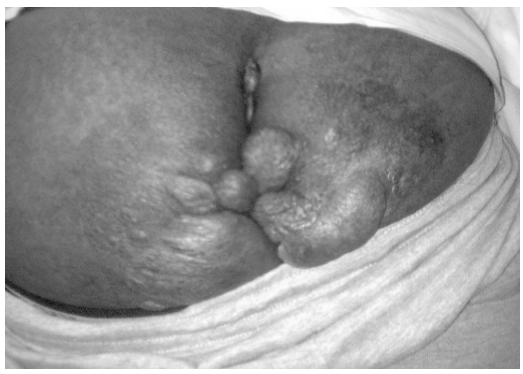


Fig 2

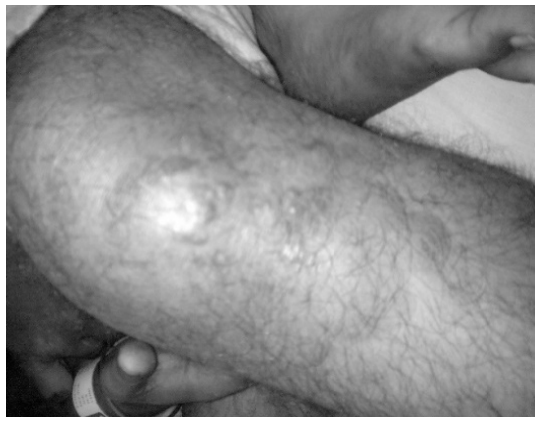


Fig 3

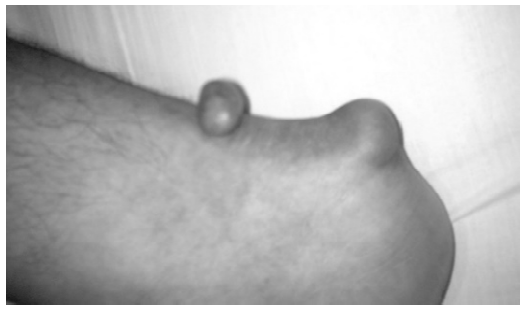


Fig 4

Lipid Profile

- TC: 489 mg%
- HDL: 31 mg%
- LDL: 397 mg%
- TG: 98 mg%
- TC /HDL: 15.8
- BP: 130/80 mmHg
- PR: 72/min; T: 98.4
- CXR: normal
- ECG: minimal ischemic changes
- CBC, Urea, Creatinine: normal
- ECHO: No RWMA, LV normal in size and function, no PAH,
- LVEF: 68%

Angiogram revealing triple Vessel disease

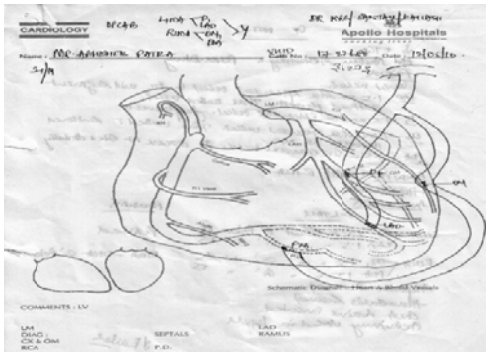


Fig 5

Coronary bypass graft given

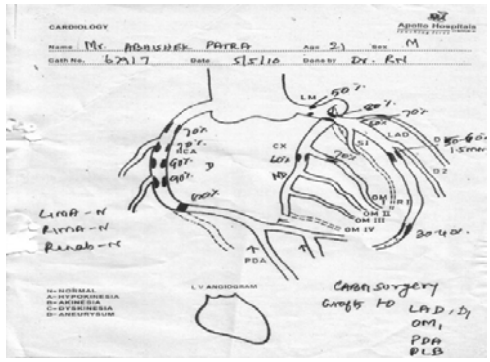


Fig 6

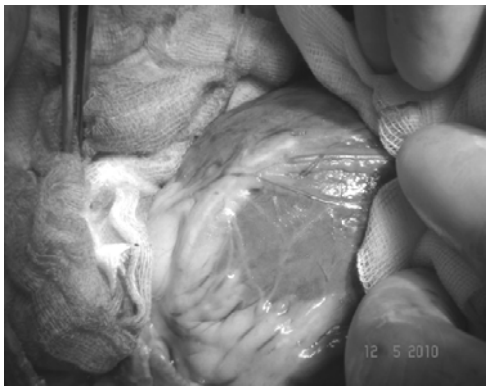


Fig 7

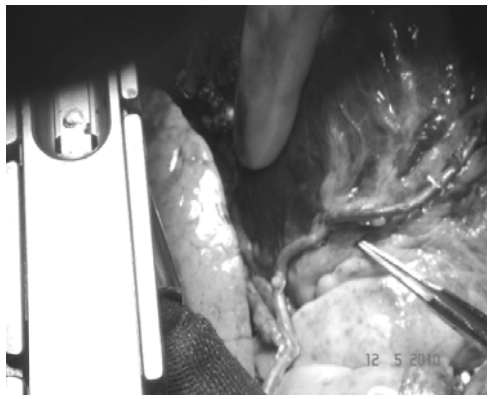


Fig 8

CAG was done because of hyperlipidemia, symptoms of CAD, family history of hyperlipidemia and CAD, high lipids and many Xanthomas, coronary angio was done

Case report:

We saw a 22-year-old male as the OP with xanthomas all over his body (Fig 1 and 2). He also experienced frequent anginal pain. Investigations revealed hypercholesterolemia with stress test positive and ECHO abnormality. He was given anti-lipid drugs and found to have the familial disease. A coronary angiogram (Fig 5- 11) was done, which revealed significant triple vessel disease, and he underwent coronary bypass surgery. The postoperative period was uneventful. He was commenced on anti-lipid measures, and he is doing well.

Steps of CABG surgery

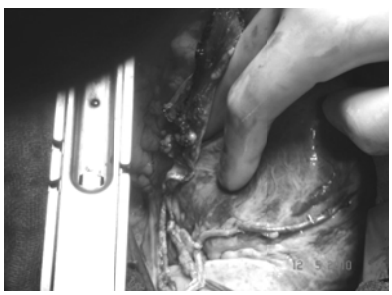


Fig 9

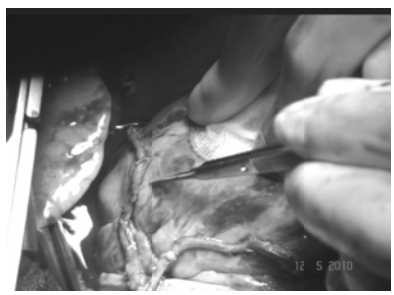


Fig 10

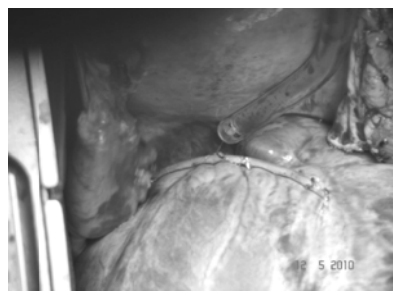


Fig 11

Normal BP, obese, xanthomas on examination on many parts of the body, including hands, elbow, ankle, knee, and perianal region.

Investigations:

He had undergone complete investigations for lipid status, diabetes, thyroid function, X-ray chest, ECG, ECHO, and TMT. His cholesterol level was very high (489mg%) and hence started on medications.

Treatment:

Internal mammary artery grafting in combination with postoperative intensive lipid-lowering treatments, including low-density lipoprotein apheresis, facilitates good long-term results of coronary revascularization in patients with FH. Since he had familial hyperlipidemia with symptomatic coronary artery disease, he decided to do a coronary artery bypass.

Discussion- CABG in young occurs in:

1. **Kawasaki's disease,** coronary artery aneurysms.
2. **Familial hypercholesterolemia** Autosomal dominant

- Mutations in LDL receptor gene
- High LDL-C and normal TG
- Tendon xanthomas common
- Premature coronary atherosclerosis is a complication

3. Homozygous FP

- Occurs 1 in 1 million
- Total cholesterol > 500mg%
- Coronary artery disease occurs before the 2nd decade
- Cerebrovascular and peripheral vascular atherosclerotic disease
- Skin biopsy confirms
- Molecular assays helpful
- Rarely do they survive beyond 2nd decade

4. **Heterozygous FH**

- Occurs in 1 in 500
- Most common single gene disorder
- LDL 200 – 400mg
- Adulthood disease
- 50% of siblings are affected
- Premature Coronary artery disease in family members
- Molecular assays are helpful

5. Anomalous origin of the coronary artery from the pulmonary artery **(Bland white garland syndrome) fig 13 and 14**
6. **Progeria-** Familial hyperlipidemia with premature aging. (fig 15)

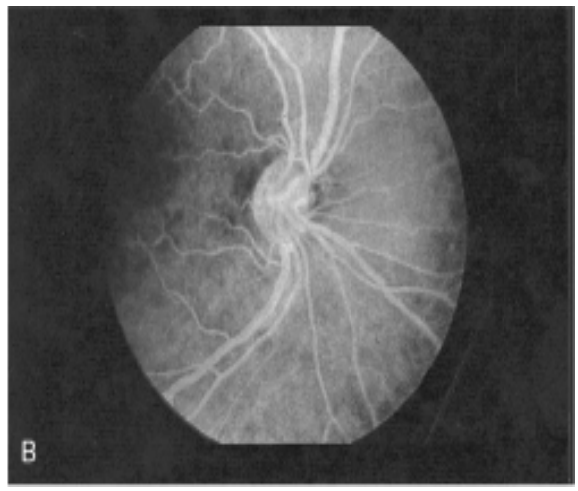


Fig 12 shows a case of hyperlipidemia with lipemiaretinalis

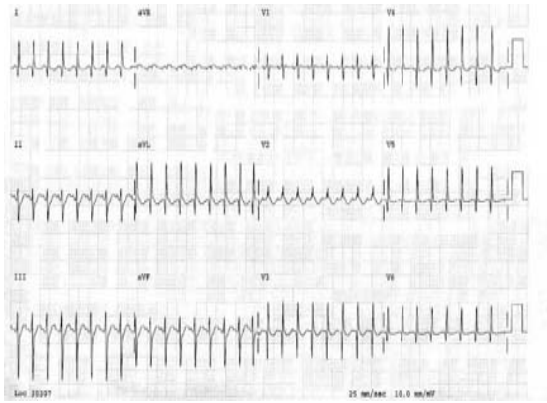
Lipid-lowering strategies:

- Diet therapy
- Drugs - Statins, Cholesterol absorption inhibitors, BAS, Niacin
- Lomitapide
- LDL apheresis
- Orthotopic liver transplant
- Evolution of stenotic lesions following revascularization in patients with FH can be controlled significantly by lipid-lowering therapy to maintain a TC level of <220mg/dl, and if diet alone cannot achieve it, aggressive medication and even LDL apheresis might be justified.

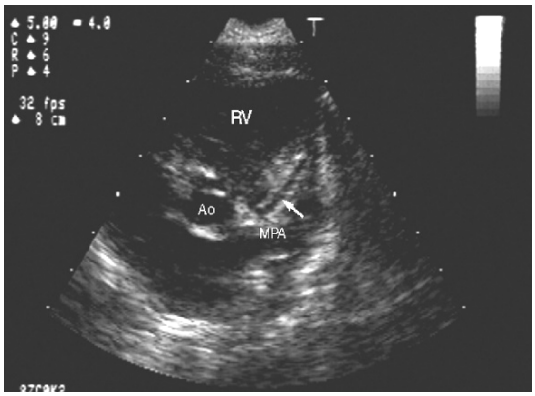
HELP has been found to reduce LDL-C acutely and drastically and decrease high-sensitivity C-reactive protein (HS-CRP) and fibrinogen by 65%.[4] Both of these inflammatory markers have been associated with acute and chronic CVD. A

recent observation is that it is safe to keep LDL below 100 and in co-morbid cases below 70.

Arterial grafting improved the long-term freedom from reoperation in patients with familial hypercholesterolemia.



ECG in a case of bland white garland syndrome Fig 13



Anomalous origin of coronary artery in bland white garland syndrome Fig 14

Progeria Fig 15

Types:

- Type 1 - eruptive xanthomas, no CVS. Lipemiaretinalis, plasmapheresis, diet
- Type 2 - planar, atherosclerosis, abnormal arcus cornea bile acids, statins, niacin, fibrates
- Type 3 - palmar, atherosclerosis, abnormal glucose tolerance statins, and fibrates
- Type 4 - eruptive tuberous, atherosclerosis, lipemiaretinalis obesity, statin fibrates
- Type 5 - eruptive tuberous atherosclerosis obesity lipemiaretinalis, obesity, hyperinsulinemia
- Tangier - macular rash, atherosclerosis, corneal infiltrations, enlarged orange tonsils nodes

Two more additions

- Apolipoprotein a-1 and c deficiency - atherosclerosis, corneal clouding, CNS, CVS normal.
- HDL def with planar xanthomas, atherosclerosis corneal opacity
- Therapy
- Familial lipoprotein def - familial polygenic hyperlipoproteinemia or C 3 def
- No drug therapies
- Familial hypercholesteremia - statins, bile acid sequestrants, ezetimibe
- Alirocumab and efalizumab subcutaneous injected – pcsk 9 protein convertasesubtilisin
- Kexin type inhibitors for heterozygous familial hyperlipoproteinemia

Conclusion:

This rare case of familial hypercholesterolemia resulting in significant coronary artery disease requiring bypass surgery in a very young boy is presented. Similarly, young people develop coronary artery disease due to anomalous origin of the coronary artery from the left pulmonary artery, progeria with premature

aging, hyperlipidemia, Kawasaki's disease with coronary artery aneurysm, and juvenile diabetes mellitus. The familial lipid abnormality is very difficult to control, and most of them end up with coronary artery disease. It is very rare to see tuberous xanthomas in many parts of the body, and his mother also has a similar problem (on medical management). Awareness, early detection, and treatment can avoid early mortality.

Reference:

1. J ThoracCardiovascSurg 2000; 119:1008-14.