Familial Hyperlipidemia -Case Series

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Introduction

Familial hyperlipidemias can present with simple manifestations like gall stones to life threatening events like myocardial infarction,CVA or severe acute pancreatitis.It is hence important to identify these at the earliest and start appropriate medications to avoid future catastrophe.Here are some of the cases encountered in our practice.

Case 1

26 year old morbidly obese female came with complaints of abdominal pain ,vomiting and breathlessness for 3 days.On examination patient was conscious ,tachypneic and dyspneic .CVS -s1 s2 + ,RS-b/l ocassional crepts +P/A -epigastric tenderness + CNS -NFND.B/P -126/80 PR-118/min spO2-92 % in room air 97% in 8 L O2 ,CBG -340 mg /dl .investigation revealed amylase ,lipase -1262/2359 triglyceride-1174,total cholesterol -546 ,urine acetone was negative ,ABG revealed type 1 respiratory failure .CT chest was suggestive of ARDS AND CECT Abdomen showed acute pancreatitis .total cholesterol /Apo B >0.2 .Patient was diagnosed to have type 2 Dm ,type 5 hyperlipoprotenemia.she was kept on NPO and treated with cpap care, insulin infusion ,aspirin ,atorvastatin,fenofibrate and fish liver oil.Genetic studies did not reveal any one gene defect.

Case 2

18 year old female ,born of non consaguinous marriage presented with right hypochondrial pain and ocassional vomiting .she had tendon xanthomas .otherwise system examination was normal.her father had history of taking statins for dylipidemia found during routine check up.Usg abdomen showed choledocholithiasis and borderline distension of cbd ,her total cholesterol was 304,HDL -39 ,LDL -259.lipoprotien a level was 46.total cholesterol /HDL -73,LDL/HDL-6.6.Genetic analysis showed defect in LDL receptor gene .hence she was diagnosed to have familial hypercholestrolemeia (type 2 liperlipoprotenemia).she was treated with statins ,ezetimibe and laporoscopic removal of gb stone was done

Case 3

38 year old male with not a smoker /alcoholic presented with typical cardiac chest pain .ECG was suggestive of anterior wall MI and troponin was elevated .Immediate coronary angiogram showed 80 percent stenosis in LAD and primary PCI was done .family history of MI in father and uncle at 40 years of age.Further work up showed triglyceride -634 total cholesterol-345.cholestrol :apo b >0.2 .genetic analysis did not reveal particular gene defect, he was diagnose to have familial combined hyperlipidemia (type 3 hyperlipoprotenimia).he was treated with antiplatelets and statins .

Conclusion :

Early identification of familial hyperlipidemias and prompt treatment with statins and other lipid lowering drugs can drastically reduce future atherosclerotic cardiovascular disease.

Studies have proved that only lower is better but the more younger the better too.hence lowering lipid profile to mimimum at age as young as possible has proven to prevent future cardiovascular events.Hence screening siblings,children and other family members of these patients can also help initiate treatment in people before any adverse events sets in.