Management of children and adolescents with familial hypercholesterolaemia in a specialist out-patients clinic

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Introduction: Heterozygous familial hypercholesterolaemia (FH) is a common autosomal-dominant genetic disorder (1:200) with an increased risk of premature coronary artery disease even in the young. Published criteria exist for diagnosis and management of FH1. We report our findings in paediatric and adolescent patients referred to and treated in the FH Outpatients Clinic.

Methods:
We retrospectively reviewed hospital records of patients diagnosed with genetically confirmed FH under 18 years of age in a single tertiary cardiac centre between January 2006 and June 2014.

Results:
Forty eight patients (25 male) with FH were identified. Median age at diagnosis was 10.1 (range 2.3 - 17.3) years. Reasons for patients’ referral was mainly diagnosis of FH in a relative (79%) and abnormal serum lipid profile in referred patient (8%). Medical records and follow-up data were available in 39 patients. Dietary intervention was commenced in all patients and 29 patients (67%) were started on treatment with a statin at a median age of 12.45 (range 7.6 - 18.6) years. Decrease in total cholesterol (TC) levels was achieved from initial TC (mean (SD)) 7.05(1.47)mmol/L to 5.45(1.19)mmol/L at latest review (p= 0.000001). Similar decrease in low-density lipoprotein (LDL) values was encountered (initial LDL 5.44(1.55)mmol/L, at latest review 3.63(1.08)mmol/L; p<0.0001). No significant changes were seen in high-density lipoprotein (HDL) levels (initial HDL 1.32(0.35)mmol/L, at latest review 1.34(0.29)mmol/L; p=0.75).

Conclusions:
Dietary and medical treatment of children and adolescents with FH has beneficial effects on lipid profile. As early diagnosis and treatment are essential for long-term prognosis2, a detailed family history review focused on hypercholesterolaemia and timely referral to a specialist Outpatients Clinic are mandatory in general paediatric and paediatric cardiology clinics.

1www.nice.org.uk/guidance/cg71 (last review on 10th December 2014).

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