the degree of atherosclerosis in monogenic FH patients versus polygenic individuals with hypercholesterolaemia.

Method: Carotid Intima Media Thickness (cIMT) was measured by B-mode ultrasound in three different segments of carotid arteries (common carotid artery, bulb and internal carotid artery) in 86 individuals (53 female and 33 male) with a clinical diagnosis of FH (LDL-C mean+SD: 5.8 ± 1.3 mmol/l). 56 patients had monogenic FH with a known mutation in the LDLR or APOB gene and 30 patients had no mutation but they had a score in the top two quartiles of a six LDL-C-raising SNPs gene score.
Results: The monogenic patients were younger than polygenic individuals (50 ± 14 years vs 57 ± 12 years, p = 0.03). There was no significant difference in total cholesterol level, LDL-C and HDL-C between the two groups. Triglyceride level was significantly higher in polygenic compared to monogenic group [1.6 ± 0.7 mmol/l vs 1.2 ± 0.5 mmol/l, p = 0.01]. After adjustment for age and gender, the mean of all the cIMT measurements was significantly higher in monogenic than polygenic patients [0.74 mm (0.7–0.79) vs 0.66 mm (0.61–0.72), p = 0.03].
Conclusion: The severity of atherosclerosis as measured by cIMT is higher in monogenic FH individuals than the polygenic group. While LDL-C levels need to be reduced in both groups, the greater degree of carotid atherosclerosis supports aggressive management of their LDL-C levels with potent statins and other LDL-C lowering modalities in combination.

RELATIONSHIP BETWEEN MEASUREMENTS OF NON-HDL-CHOLESTEROL AND LDL-CHOLESTEROL IN FAMILIAL HYPERCHOLESTEROLAEMIA

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Background: Familial Hypercholesterolaemia (FH) is a monogenic disorder of Low Density Lipoprotein (LDL) metabolism which can be specifically diagnosed by genetic testing. However LDL-cholesterol (LDL-C) estimation has the drawback that it requires a fasting sample, because the Friedewald formula uses a factor relating to fasting triglyceride. Also other cholesterol containing lipoproteins are atherogenic and should be considered as part of cardiovascular risk assessment. Recent guidelines from National Institute for Health and Care Excellence (NICE) recommend non-HDL cholesterol (non HDL-C) for assessment of cardiovascular risk. The Joint British Societies (JBS3) guideline also recommends non-HDL cholesterol and proposes a conversion factor: Non-HDL-C = 1.24 × LDL-C. This project assessed the application of this factor in patients who have presented to lipid clinics as possible FH.

Method: The Wales FH service offers genetic testing to patients presenting to lipid clinics who meet criteria for possible FH. Of 711 index patients who had complete pre-treatment lipid results, 171 had a pathogenic FH mutation and 540 did not. The majority of these “mutation negative” patients are likely to have a polygenic basis for their hypercholesterolaemia.
Results: In the overall group (n = 711) non-HDL-C was 1.13xLDL-C. In mutation negative patients the factor was 1.14 compared to the mutation positive patients 1.10 (p < 0.001). For the group as a whole the factor was lowest in those with high LDL-C and low triglyceride (Table).

Lipid Concentration	Triglycerides < 2 mmol/L	Triglycerides ≥ 2 mmol /L
LDL-C ≥ 6.5 mmol/L	1.08 (n = 244)	1.15 (n = 157)
LDL-C < 6.5 mmol/L	1.11 (n = 188)	1.21 (n = 122)

Conclusion; These data suggest that the JBS3 conversion factor of 1.24 is too high for patients with possible FH especially for those with very high LDL and low triglyceride.

WHERE HAVE ALL THOSE FAMILIAL HYPERCHOLESTEROLAEMIA PATIENTS GONE?

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Best evidence currently supports cascade testing as a means of tracing family members to identify at the very least, first and second degree affected relatives of known FH patients. Previously we presented our experience on the development of a telephone-screening clinic for family members of DNA positive proband to enhance the effectiveness of the cascade testing process.

NICE recommend, that all individuals identified as definite FH are referred to a specialist and that a structured clinical review is carried out annually. The role of the FH Clinical Nurse Specialist (CNS) is to ensure this process is followed. The confirmed genetic diagnosis is communicated to the patient's GP quoting NICE CG71, requesting that their patient is referred to an FH specialist locally. We aimed to evaluate how effective this process is by exploring whether or not these patients were being referred to specialist care.
All 62 patients with a positive DNA diagnosis of FH who had attended the telephone outreach clinic were selected for evaluation. A number of patients were identified as being referred to the lead for FH services within the trust. The remainder of patients were sent a letter requesting a call to the FH service to discuss referral post identification of FH. The response rate was poor at 3%. Therefore, in order to ascertain figures, the remaining patients were approached via telephone and the results are as follows:

Referred to specialist care	48 (77%)
GP managing patient	3 (5%)
No referral	5 (8%)
Patient or parent declined referral	3 (5%)
Lost to follow up	3 (5%)

Reassuringly, the majority of patients are referred to specialist care. Recent funding from the British Heart Foundation has secured the appointment of a full time CNS to this service. This will enable further development of patient follow up and through education and awareness sessions with patients and their GP's we believe these figures can be further improved.

PEDIATRIC CHOLESTEROL SCREENING IN ITALY: THE SPIF PROJECT

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