THE PREVALENCE OF FAMILIAL HYPERCHOLESTEROLAEMIA IN PATIENTS WITH CLINICAL CORONARY ARTERY DISEASE

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BACKGROUND AND OBJECTIVES

- Familial hypercholesterolaemia (FH) is a dominantly inherited genetic disorder with an estimated population prevalence 1/200 – 1/300 (~0.5%)
- Disease results in congenitally elevated LDL-C levels and, if untreated, results in severe and premature cardiovascular disease
- FH is likely to be over-represented in patients presenting to hospitals with coronary artery disease
- There has been little research into the prevalence of this severe but treatable condition within an Australian population
- The aim of this study was to assess the prevalence of FH within a population of coronary artery disease patients

METHODS

- Cross sectional study, assessing patients ≤ 70 yo at Austin Health, Melbourne, Australia from Feb-June 2018 presenting with:
 - Acute coronary syndrome
 - Elective percutaneous intervention
 - Coronary artery bypass graft surgery
- Diagnosis of FH was made utilising Dutch Lipid Clinic Network criteria
 - "Definite" or "probable" FH if DLCN score > 5
- Cardiovascular disease risk factors: hypertension, diabetes mellitus, smoking status, elevated lipoprotein(a) were also recorded
- Wilcoxon rank-sum test and Fisher's exact test were used to analyse continuous and categorical variables

RESULTS AND TABLES

Table 1. Comparison of demographics and characteristics of patients diagnosed with FH (Definite/Probable FH, n=19) and patients without FH (Non-FH, n=158)

	FH n=19	Non-FH n=158	p value
Age in years, median (IQR)	53 (47 - 60.5)	61 (57 - 67)	0.005
Age less than 60, n (%)	14 (73.7)	65 (41.1)	0.013
BMI, median (IQR)	28.7 (26.6 - 35.6)	29.1 (26.5 - 32.8)	0.821
Pathology Results			
Total Cholesterol in mmol/L, median (IQR)	5.1 (4.9 - 5.9)	4.0 (3.2 - 5.1)	p<0.001
LDL in mmol/L, median (IQR)	3.3 (3.0 - 3.8)	2.1 (1.4 - 3.1)	p<0.001
Lp(a) in g/L, median (IQR)	0.62 (0.19 - 0.96)	0.23 (0.10 - 0.54)	0.040
Lp(a) greater than 0.5 g/L, n (%)	9 (56.3)	35 (29.9)	0.048
Lipid Lowering Medication use			
Statin use at admission, n (%)	18 (94.7)	100 (63.3)	0.004
HbA1c as percentage, median (IQR)	5.8 (5.6 - 7.4)	5.8 (5.4 - 6.8)	0.411
Hypertension, n (%)	8 (44.4)	102 (64.6)	0.123
Smoking status			
Current Smoker, n (%)	8 (44.4)	51 (32.9)	0.431

BMI, Body mass index; LDL, Low-density lipoprotein; Lp(a), Lipoprotein(a); HbA1c, Haemoglobin A1c; IQR, Interquartile range

Table 2. Comparison of FH prevalence, general demographics in patients aged less than 60 (n=79) and those 60 and above (n=98)

	Age <60yo n=79	Age 60+ n=98	p value
Definite/Probable FH, %, (95% CI)	17.7 (10.0 - 27.9)	5.1 (1.7 - 11.5)	0.013
BMI, median (IQR)	28.7 (26.6 - 34.8)	29.3 (26.5 - 32.1)	0.575
Pathology Results			
Total Cholesterol in mmol/L, median (IQR)	4.8 (3.6 - 5.4)	4.0 (3.2 - 5.1)	0.028
LDL in mmol/L, median (IQR)	2.9 (1.8 - 3.6)	2.1 (1.4 - 3.0)	0.004
Lp(a) in g/L, median (IQR)	0.30 (0.13 - 0.78)	0.20 (0.10 - 0.54)	0.390
Lp(a) greater than 0.5 g/L, n (%)	20 (35.7)	24 (31.2)	0.709
HbA1c as percentage, median (IQR)	5.6 (5.4 - 7.5)	5.8 (5.4 - 6.8)	0.803
Hypertension, n (%)	39 (50.0)	71 (72.4)	0.003
Smoking status			
Current Smoker, n (%)	29 (37.7)	30 (31.3)	0.334

FH, Familial Hypercholesterolaemia; BMI, Body mass index; LDL, Low-density lipoprotein; Lp(a), Lipoprotein(a); HbA1c, Haemoglobin A1c; IQR, Interquartile range; 95% CI, 95% confidence interval

- 214 patients assessed during time period, 177 eligible for inclusion in analysis
 - four patients had triglycerides > 4.5 mmol/L, 11 patients had missing LDL-C levels, 21 patients were discharged before interview and one patient was non-English speaking
- Estimated prevalence of FH 10.7% (95% CI 6.6% - 16.3%)
 - FH patients were younger, had higher LDL-C levels and higher rates of statin use
 - 2/19 patients had a prior diagnosis of FH
 - 9/19 FH patients also had Lp(a) > 0.5 g/L
- Estimated prevalence in patients < 60 yo rose to 17.7% (95%CI 10.0% - 27.9%)
 - FH prevalence in patients 60+ was 5.1%, (95% CI: 1.7% – 11.5%) $p=0.013$

CONCLUSION

- FH is over-represented but under recognised in this population
- Despite 18/19 FH patients being already treated with statins, a median LDL of 3.3 suggests sub-optimal treatment of these patients
- Its high prevalence and associated morbidity and mortality makes a strong case for implementation of disease screening
- Diagnosis also allows specialist lipidology referral and cascade screening of undiagnosed family members

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