MAPHM Symposium 2017

20th October 2017

Abstract Title:

Initial data from a newly set-up Familial Hypercholesterolaemia Register

First author:

Dr Myra Tilney

Other Authors:

Conrad Azzopardi

Abstract

Background

Familial hypercholesterolaemia (FH) is a common monogenic disorder, resulting in premature cardiovascular morbidity and mortality. It has potentially preventable outcomes and yet is underdiagnosed and undertreated worldwide, in spite of the availability of cost-effective therapy. Cardiovascular diseases are an important cause of premature mortality nationally, accounting for 17.1% of Premature Years of Life Lost under 65 years (PYLL-65) in 2013; FH is likely to be a major contributor to this. Based on international prevalence, we estimate that up to 2000 individuals are likely to be affected in our community, but do not have the impression that these have been fully identified.

Methods

Over the last year an FH register has been established, with all the necessary authorisations (primary and secondary care administration, data protection and ethics), and patient consent. As an initial step, we started recruiting patients attending Lipid Clinics, and using cascade testing to identify likely affected relatives. The Dutch Lipid Clinic Network criteria are being used for diagnostic purposes. These are being entered into an Excel spreadsheet, enabling ongoing review and analysis.

Results

Data collection commenced in January 2017;to date 27 patients have been identified, including 8 men and 19 women, with 7 classifying as 'Definite Familial Hypercholesterolaemia using the DLCN classification. 5 of these 7 patients have achieved target control based on the European Society of Cardiology 2016 Guidelines. For an autosomal dominant condition, it is noteworthy that more women than men have been identified, although the numbers are small. This sex difference is in keeping with overseas data, which have identified that the discrepancy is due to premature male mortality.

Conclusions

Within this short time frame, the FH register has included a number of patients who satisfy the DLCN criteria for Definite FH, and others who classify as 'Probable' and 'Possible' FH. The FH register provides a tool that will facilitate better recognition and management of such patients- and

hopefully impact on cardiovascular outcomes. However, it will require better recognition and referral to provide a complete picture of   FH in Malta.