Experience of the Establishment of a Dedicated FH Service in the Yorkshire & Humber region

P Sutton[1] R Dunn[1] R Desborough[1] J Webster[2] K Mitchell[2] C Burton[3] M Mansfield[3] J Barth[3] D Chandrajay[4] C Tuson[4] A Bandyopadhyay[4] K Haralambos[5] D Narayanan[1]

[1] Hull University Hospitals NHS Trust [2]Calderdale and Huddersfield NHS Trust [3]Leeds Teaching Hospitals NHS Trust [4]York Teaching Hospitals NHS Foundation Trust [5]Cardiff and Vale University Hospitals Board, Wales.

Introduction

In 2017 a dedicated genetic service was established to screen for Familial Hypercholesterolaemia (FH) in the Yorkshire & Humber population to find adults with FH and therefore at increased risk of premature cardiovascular disease. The Commissioning Groups entered into an agreement with the Lead CCG to commission a dedicated service across four hospital hubs (Hull, York, Leeds & Huddersfield) for 3 years; four WTE nurses funded by the British Foundation for 2 years provided genetic counselling with the full service costs covered by the CCG's in year 3. Aims were to identify patients with a proven FH genetic variant (Index), rapidly identify/test family members (Cascades) of positive index cases at a reduced cost and identify barriers to identification and testing of high risk patients.

Methods

Patients were identified predominantly from Lipid/Endocrinology Clinic heritage lists as having a clinical diagnosis of 'Possible' or 'Definite' FH as per the Simon Broome Criteria. Those identified were scored using the Welsh Lipid Clinic Score (a modified Dutch Lipid Clinic Network Score) after secondary causes were excluded. Patients who scored ≥ 6 and resided in a funded area were invited for counselling and the opportunity for FH genetic testing as an index patient (£330). 1st degree relatives of patients found to have a genetic variant were offered screening at a reduced cost (£100). Four cascade genetic tests were predicted for each positive index patient.

Results

The use of the heritage lists and a systematic approach to identification/testing of high risk patients resulted in 638 Index patient tests with a 48% pick-up rate for positive/VUS mutations for FH (305 in 2 years). A further 504 Cascade patient tests resulted in 260 patients identified as having a positive/VUS mutation for FH (260 in 2 years) at a significantly reduced cost.

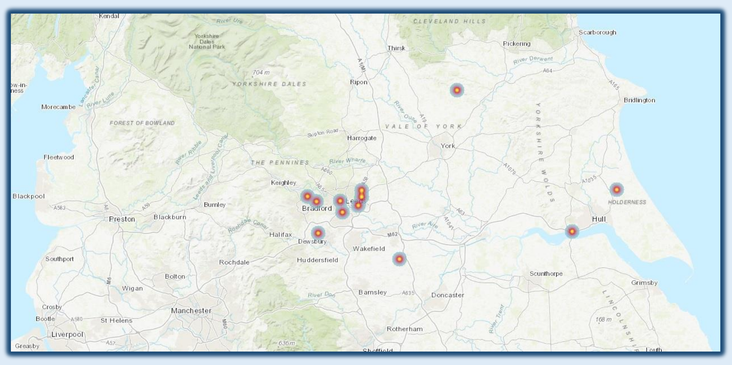

Fig. 2 Distribution intensity of positive index patients April 2017

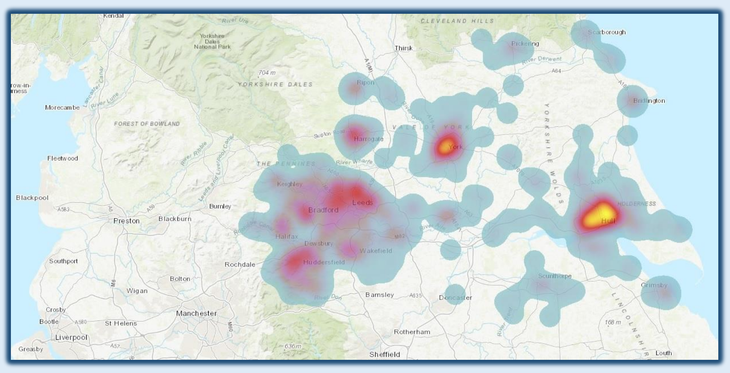

Fig. 3 Distribution intensity of positive index patients April 2019

Fig. 1 Heat Map Key

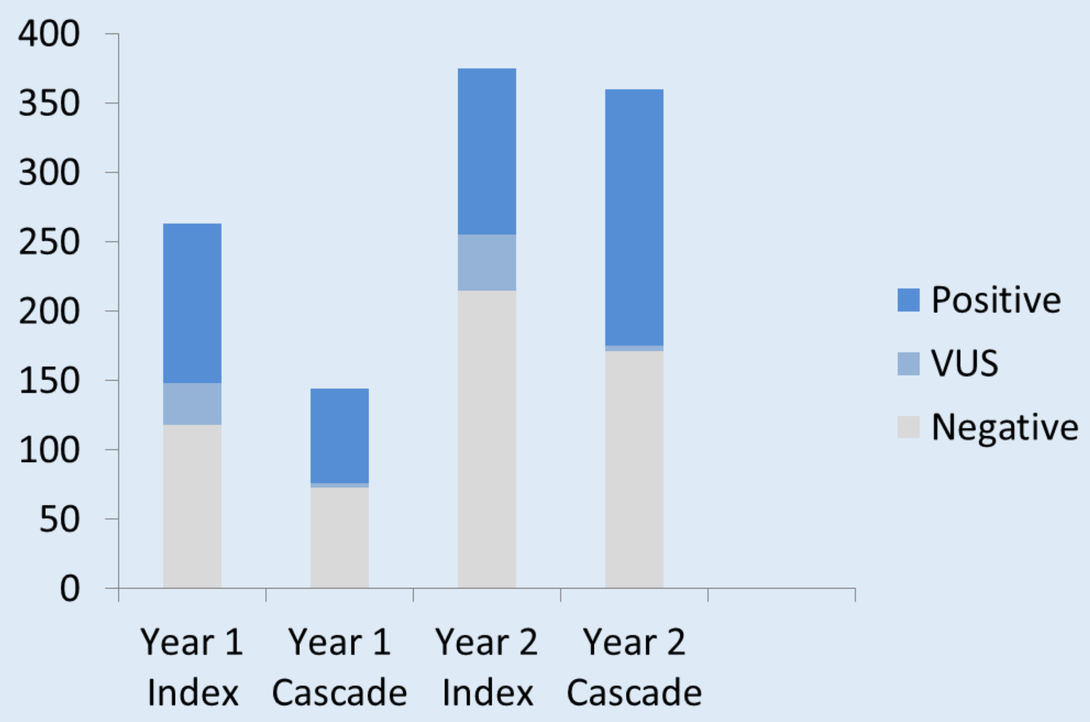

Fig. 4 Distribution of genetic testing results of index and cascade patients

Discussion

Taking into account the risk of premature cardiovascular disease in untreated FH and the funding for a set number of genetic tests (quota), these results demonstrate that cascade testing is effective in identifying FH patients within a resource limited service. Not all CCG's were commissioned in year 1 (14 of 18) resulting in potential geographical health inequalities for identification and treatment of FH. Data from year 1 demonstrated the effectiveness of cascade testing and recruited a further 3 CCG's into the programme. Areas without Lipid Clinics had lower identification rates, highlighting the need to increase awareness in primary care and explore the use of primary care data to identifying people at increased risk of FH. Some local populations had large stationary families necessitating > 4 cascade tests per index, creating additional patient workload not previously considered. A selection bias and modified DLCN score was in part contributory to the success of the FH programme, raising awareness in primary care and utilising primary care case finding software will be needed to continue this success. Children under the age of 16 years at risk of FH were identified but could not be screened by the Y&H service. Following this the service moved to secure funding to establish a Paediatric FH Service.

References

1. Familial hypercholesterolaemia: identification and management. NICE Guidance CG 71. www.nice.org.uk/guidance/cg71