Audit of patient selection for Familial Hypercholesterolaemia (FH) genetic testing over a 2-year period with retrospective comparison of the Simon-Broome criteria vs the Dutch Lipid Clinic Network (DLCN) criteria vs the Wales FH service genotype scoring criteria.

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Abstract

Background: Familial hypercholesterolaemia (FH) is a genetic disease resulting in elevated levels of low-density lipoprotein cholesterol that predisposes to early onset cardiovascular disease. Three main scoring systems exist to facilitate patient selection for genetic testing. This audit aims to discern which criteria perform best within a specialised lipid clinic.

Methods: 70 patients that received FH testing were retrospectively scored against each system. Comparisons between the three systems were made and the number of patients that met testing criteria, positive rates, and false negative rates noted.

Results: Wales criteria allowed for the least number of genetic tests to be done and had the highest positive rate (29.4%), however had among the greatest number of false negatives. DLCN criteria had the lowest number of false negatives but required testing a larger patient population. Simon-Broome criteria had the lowest positive rate (12.5%).

Conclusion: Wales criteria allows for the least number of tests to be done with the highest positive rate. False negatives using the Wales criteria can be reduced using clinical judgement and by having a complete family history of lipid levels.