Are we adhering to Simon Broome's Criteria for referrals for genetic mutation for familial hypercholesterolaemia at Queen Alexandra Hospital and are there any clear differentiators between the two outcome groups?

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Objectives:

We aimed to analyse our cohort of patients referred for FH genetic testing at Queen Alexandra Hospital from 2014 to 2015. We looked at patients tested positive as well as those tested negative. We aimed to identify the criteria used to refer patients for FH genetic testing, mainly looking at whether Simon Broome's Criteria were met.

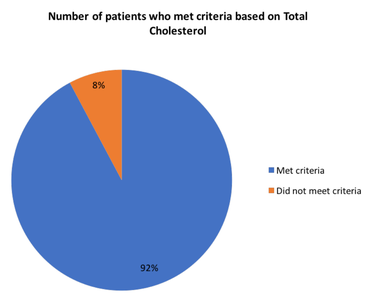
Methods:

A list of patients referred from Queen Alexandra Hospital and tested positive and negative respectively for FH genetic mutation was generated by the FH Specialist Nurse. The software Diabeta 3 was used to access clinic letters to obtain clinical information. ICE software was used for blood results. Unfortunately, clinical information was lacking for some of the patients referred from GP.

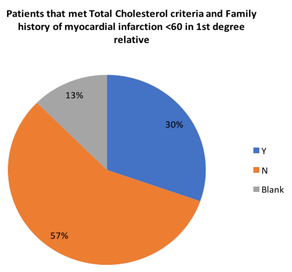
Results:

- In total, 80 patients were identified for the audit. Out of those, 61 tested negative for FH and 19 patients tested positive for FH. Out of 61 patients for those tested negative, clinical information was only available for 59 patients (N=59). Out of those tested positive for FH, data for analysis for our audit purposes were only available for 18 patients(N=18).
- Based on Total Cholesterol value, the majority (94%) of those tested negative met the criteria. Out of those tested positive, 83% met the criteria. In total, from the data available for all the patients referred for FH gene testing (N=77), 92% met the criteria for the Total Cholesterol value.
- 60% of those tested positive for FH did met Simon Broome's Criteria for Total Cholesterol and presence of Xanthomas compared to only 82% of those tested negative. There was hardly any difference in yield if both of these criteria were met (5% in the Negative arm and 7% in the positive arm).
- With regards to Total Cholesterol value and family history of MI in second degree relative aged less than 50, there was only a marginal difference in outcome between the 2 arms. 16% of those tested negative met both of these criteria and 13% of those tested positive met both of these criteria.
- However, overall, out of all referrals, 67% did not have a Family History of MI in second degree relative below the age of 50.
- We found that almost similar proportion of each outcome had a family history of raised cholesterol
- Nearly half of the total population (51%) did not have a family history of MI in first degree relative below the age of 60. However, analysing both outcomes separately, 46% of those tested positive had a family history of MI in first degree relative below age of 60 compared to only 30% of those tested negative.

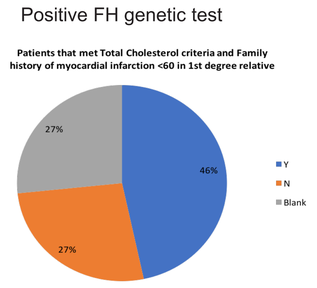
Graphs and tables



Negative FH genetic test



Positive FH genetic test



Conclusions:

- The Simon Broome Criteria are used in the majority of cases.
- 92% of patients met criteria based on Total Cholesterol. Out of the 6 patients who did not meet the criteria for Total Cholesterol, 5 did not meet criteria for LDL cholesterol either.
- Within the limitation of this audit, we can conclude that the most striking difference between positive and negative outcomes is a family history of MI in first degree relative aged below 60 (46% in positive outcome versus 30% in negative outcome).
- Using only presence of Xanthelasma or other family history of CVD not in-keeping with Simon Broome criteria alongside high cholesterol level definitely yielded a negative result.

References:

https://www.nice.org.uk/guidance/cg71/evidence/full-guideline-appendix-f-pdf-241917811

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