Reviewer's report

Title: Patient accounts of diagnostic testing for familial hypercholesterolemia: comparing responses to genetic and non-genetic testing methods

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Reviewer: gerald watts

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An important study based on qualitative methods suggesting that genetic and phenotypic testing for FH does not have a discernible adverse impact on patients' lives. Endpoints confirm continuity of perspectives, reduction in uncertainty, lack of prominence of FH relative to other conditions and only minor social impact. So all is well with testing for FH by which ever method, but how generalizable are the results, given the small sample size and selected group?

Design appropriate, but method of selection of volunteers unclear. ? randomly selected from parent population?

Clinical characteristics of patients not specified. Table required. Were these all people without CHD or low risk FH. Not all FH mutations are the same in UK; non-pathogenic mutations may not cause concern, whereas major deletion with very high cholesterol can. Details required.

Results would be easier to follow in Summary Table, rather than sentences with annotations that remind one of The Tractatus.

Page 5 para 2 line 3: 100-fold relative risk of fatal CHD in FH is a gross exaggeration emanating from initial analyses from Simon Broome Registry. HRs are now accepted to be lower, ranging from 10 to 15.

The language is in part somewhat Baroque and arcane: are words like 'ineluctable', 'provenance' and 'salience' really necessary. The simpler word would do better and have greater impact with non-specialist readers in this reviewer's opinion

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.