

Author's response to reviews

Title: Best Practice Guidelines for the Molecular Genetic Diagnosis of Type 1 (HFE-Related) Hereditary Haemochromatosis

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To the Editor, BMC Medical Genetics

RE: Manuscript ID 3651609531027284

Best Practice Guidelines for the Molecular Genetic Diagnosis of Type 1 (HFE-Related) Hereditary Haemochromatosis

We are pleased that the reviewers felt that this was an important and useful manuscript. As no compulsory changes were requested by the reviewers, we are now re-submitting the manuscript unchanged.

Dr Sly suggested no changes, and found the manuscript to be a "clear and useful guideline."

Dr Rochette made two minor points regarding the manuscript, both suggesting that the guidelines should recommend further mutation screening in individuals with clinical/biochemical symptoms of haemochromatosis but who are not homozygous for C282Y. Dr Rochette states that "this is now the strategy in this situation".

We do not agree that this is now the standard of care; scanning the HFE gene (or other genes) for mutations causing haemochromatosis remains in the area of research, because the clinical relevance of mutations other than C282Y and H63D has not been fully established. The decision on whether to screen patients for further mutations as part of a research protocol is one for individual genetics centres and is not covered by these guidelines.

Dr Rochette also makes a minor comment "Unless it is obvious in the UK, it should be stated in the paper that a quality control has to be set up in and between laboratories when mutation detection is involved."

Quality control is of course a very important and necessary part of diagnostic testing, but it is covered by specific guidelines published by the Clinical Molecular Genetics Society and others, and does not need to be repeated here.

Your e-mail mentions the requirement for informed consent for genetic testing. These guidelines do not report the results of any genetic testing, so the issue of informed consent does not arise. Informed consent for diagnostic genetic testing in general is outside the scope of these guidelines.

David Barton
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