Scheme 5B – Interpretative HFE Genotyping and Hereditary Haemochromatosis

General Comments Scenarios 1 and 2 /2016 **Distributed February 2016**

Scenario 1

It was surprising that a number of labs did not comment on the current ferritin in the reference range. Determination of a fasting transferrin saturation would be useful as an earlier indicator of whether there is iron accumulation. Although an immediate TfSat may not affect the short-term management of the patient guidelines suggest that TfSat should be part of normal diagnostic workup and it would be an earlier phenotypic indicator of biochemical penetrance of the genotype.

An appropriate monitoring interval is essential. Advising “regular" monitoring is not sufficiently informative.

A GP could manage this patient until iron overload is detected. Suggesting a specialist referral is ok, but should not be overly directive, although local practice may vary in this respect.

Scenario 2

The key is that some action on the raised ferritin must be suggested. There is a danger that the HFE test will do harm if the statement that hereditary haemochromatosis is unlikely is seen as a reason to do nothing. Although suggesting immediate referral is not very bad, an interpretive report should earn its money by suggesting an economical approach which is first to do more appropriate tests.

A repeat fasting TfSat is advisable to clarify the iron status, as the serum iron concentration has a high biological coefficient of variation.