Scheme 5B – Interpretative HFE Genotyping and Hereditary Haemochromatosis

General Comments Scenarios 1 and 2 /2018 **Distributed March 2018**

Scenario 1

Overall relatively few penalty points were given for this scenario. The commonest reason for penalties was suggesting referral to a specialist when the clinical details state that the request is from a specialist: these are expected to be interpretive reports taking account of all information provided.

The patient had raised ferritin and transferrin saturation, although no information was given about whether these indices had been raised persistently. We therefore would have liked the reports to mention the possibility of other types of genetic haemochromatosis but to state that this should only be investigated if iron indices were persistently raised.

Although the patient might be referred back to general practice, we did not expect the further investigation of causes of the signs and symptoms to be stressed as the patient is being investigated by a gastroenterologist.

Scenario 2

The commonest reason for giving penalty points in this scenario was inadequate advice about family screening. Some reports did not recommend any screening: this may have been influenced by the Porto 2016 guidelines that suggest it is not appropriate to recommend testing of relatives of p.C282Y/p.H63D compound heterozygotes. In this case, however, the son's genotype is known and this has implications, particularly for any other siblings of the son and for the son's father; it is particularly misleading to state explicitly that no screening is indicated. We do not feel that it is acceptable to assume that because the mother has been screened that all appropriate screening has been done in this family.

In line with the Porto 2016 guidelines we allowed both the testing of iron indices and not doing so, although we penalised the suggestion that lifetime monitoring should be suggested without iron indices being tested immediately. We feel that immediate testing of iron indices is probably useful in a patient of this age and would be cheap in time and money, while lifetime monitoring may be of low value and is expensive. For those who favour lifetime monitoring we suggest that advice to a GP should include a recommended interval: the fact that it is difficult for specialist laboratory to choose a sensible interval does not mean that this should be left to a GP.