General Comments

Scenario 3

The clinical details and HFE test result could be consistent with a form of haemochromatosis other than the common HFE C282Y homozygous or HFE C282Y/H63D compound heterozygous forms.

Detailed further investigation is required, perhaps after referral to a specialist unit. This is an unusual case and so non-type 1 hereditary haemochromatosis is not unlikely. Quite a few reports did not address the possibility of secondary iron overload, which may be more likely.

Only a few reports suggested determining a fasting tfsat. It may be unlikely to be normal after two very high results, but maybe he takes daily iron supplements.

Scenario 4

This genotype is strongly associated with type 1 haemochromatosis.
The ferritin is currently normal and this suggests that the patient is not iron overloaded at present and reports should say this. Many reports did not do so. If no other symptoms of HH are present only monitoring is indicated. The patient should have fasting transferrin saturation and ferritin tests annually (or up to 3 years). Patients with this genotype may require regular phlebotomy to prevent iron overload but in female patients this is often unnecessary before menopause.

Referral to a specialist unit may not be necessary at this stage or it may be recommended local practice. The value of an interpretive report is to advise the GP what to do, particularly if he or she chooses not to refer at this stage.

Describing the patient's iron status as normal is technically wrong as there is wide agreement on a tfsat of 45% as suggestive of possible iron accumulation. However as the clinical details do no specify whether it was a fasting tfsat, and there is lab to lab variation, these results could be considered borderline.