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Scheme 5B - Interpretive: HFE Genotype and Hereditary Haemochromatosis

Please note: The patient information given is fictitious and for UK NEQAS for H&I external quality assessment purposes only

- Please make a report using your usual reporting format.
- The report should contain sufficient information to be of use to clinical staff involved in the patient's care and treatment.
- Reports should be uploaded to the Participant's Portal by **27th May 2025**.
- Reports can be added to the Participant's Portal by following the instructions in the user guide found at: <https://ukneqashandi.org.uk/scheme-5b-results-submission-help/>

Clinical Scenario 1/2025

GP Surgery: Dr Alice Hope, The Hope Surgery, 17 Victoria Ave, Georgetown GE79

Patient Name: James Bolton

DOB: 17/1/2004

NHS Number/Patient ID: 475 890 1872

Address: 179 Elisabeth Square, Georgetown GE79

Clinical Details/Reason for testing: James was forwarded for HFE genetic testing as his father has previously tested as homozygous for the p.Cys282Tyr variant in the HFE gene. James has an elevated Ferritin level of 600ug/L.

HFE genotyping results: James has tested as a carrier of the p.Cys282Tyr variant only.

The GP has contacted the laboratory in relation to the significance of James's genetic result. James also has two siblings, Laura age 9 and Luke age 18. Please provide an interpretative report for James Bolton with clear information on any genetic risk of developing clinical signs of haemochromatosis, and any other advice that may be needed for his siblings.

The report should contain sufficient information for the GP to determine what further action is required.

Clinical Scenario 2/2025

GP Surgery: Dr Marie Ellory, The Nuneham Health Centre, Nuneham NZ12 7RR

Patient Name: Lara Benach

DOB: 18/03/69

NHS Number/Patient ID: 475 890 6000

Address: 11 Maple Yard, Nuneham NZ12 7RR

Clinical Details/Reason for testing: Lara was forwarded for HFE genetic testing on the basis of an elevated TS% of 57% and Ferritin level of 400 ug/L.

HFE genotyping results: Lara has negative for both p.Cys282Tyr and p.His63Asp variants.

Please provide an interpretive report with appropriate advice for the GP. As Lara is Spanish, please indicate if it is advisable to perform other genetic tests aside from HFE testing .

The report should contain sufficient information for the GP to determine what further action is required.