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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 **30th September 2025**.
- Reports can be added to the Participant's Portal by following the instructions in the user guide found at: <https://uknegashandi.org.uk/scheme-5b-results-submission-help/>

Clinical Scenario 3/2025

GP Surgery: Dr Greet, Park Avenue Practice, Stirling, FK8 2AU

Patient Name: Rebecca Weeks (female)

DOB: 01/09/1980

NHS Number/Patient ID: 578 357 1342

Address: 271 Bellfield Road, Stirling FK8 3KU

Clinical Details/Reason for testing: Rebecca's husband has been identified as compound heterozygous for C282Y p.(Cys282Tyr) and H63D p.(His63Asp), she has requested testing and wishes to know the risk to their offspring.

Serum ferritin 450µg/L

HFE genotyping results: p.Cys282Tyr not detected, p.His63Asp not detected

Please prepare an interpretive report with appropriate advice for the requesting GP.

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

Clinical Scenario 4/2025

GP Surgery: Dr Mustard, Trevithick Surgery, Camborne, Cornwall, TR14 8TT

Patient Name: Charu Shah (female)

DOB: 04/08/1968

NHS Number/Patient ID: 984 757 8560

Address: 59 Barlowena, Camborne, Cornwall, TR14 7RP

Clinical Details/Reason for testing: Charu has been diagnosed with diabetes and has had both hips replaced. Her serum ferritin is 5000mg/l. The GP wanted to eliminate haemochromatosis as a possible explanation. The GP wants to know whether there is any further action required to rule out other forms of inherited iron overload.

HFE genotyping results: The HFE genotype came back as normal. The C282Y p.(Cys282Tyr) mutation was not detected.

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