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UK NEQAS for H&I
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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 **29th September 2026.**
- 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 3/2026

GP Surgery: Dr Helen Morris, Heathfield Medical Centre, Norwich, NR6 8AD

Patient Name: David Lewis (male)
DOB: 14/08/1978
NHS Number/Patient ID: 762 491 3084
Address: 44 Ash Grove, Norwich, NR7 8YT

Clinical Details/Reason for testing: David was referred to hepatology after abnormal liver function tests were identified during investigation of obesity and type 2 diabetes. Serum ferritin was reported as **1450 µg/L** with a transferrin saturation of **52%**.

To investigate possible hereditary haemochromatosis, HFE genotyping was requested.

HFE genotyping results: p.Cys282Tyr not detected

p.His63Asp homozygous

At review six months later, lifestyle modifications had resulted in some improvement in liver enzymes, however ferritin remained elevated at 1200 µg/L.

David's GP has contacted the laboratory asking:

- Whether the HFE result adequately explains his biochemical findings.
- Whether any additional investigations should be considered.
- Whether testing of his 42-year-old brother is indicated.

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.

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CLINICAL SCENARIO 4/2026

GP Surgery: Dr Sarah Whitfield, Riverside Practice, Durham, DH2 4PL

Patient Name: Margaret Hughes (female)
DOB: 12/03/1962
NHS Number/Patient ID: 417 832 9056
Address: 7 Manor Court, Durham, DH1 4RW

Clinical Details/Reason for testing: Margaret was referred to rheumatology because of progressive arthropathy affecting the second and third metacarpophalangeal joints and longstanding fatigue.

Biochemical investigations demonstrated:

- Ferritin: 1850 µg/L
- Transferrin saturation: 82%

The rheumatologist requested HFE testing because hereditary haemochromatosis was suspected.

HFE genotyping results: Homozygous for p.Cys282Tyr

Margaret has one brother, one sister and two adult children.

The GP requests guidance regarding:

- The clinical significance of the HFE result.
- Further investigations or specialist referral.
- Which family members may require assessment.

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.