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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 **05th November 2024**.
- 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 3/2024

GP Surgery: Dr Linda Noone, Green Town Medical Centre, 120 Woodford Road, Harrogate, HG1 4JP
Name: David Power
DOB: 25/08/1991
NHS number/Patient ID: 475 890 1458
Address: 11 Greenlea Apartments, Woodford Road, Harrogate, HG1 4JP

Clinical details: Fatigue. High ferritin (500µg/L on 01.07.2024 & 450µg/L on 20/09/2024). TS% not available.

HFE genotyping result: No p.Cys282Tyr or p.His63Asp detected

Participants are required to write a genetic report for the above patient using HGVS nomenclature, indicating specifically to the GP if any further action is advisable.

CLINICAL SCENARIO 4/2024

GP Surgery: Dr Stephen Jones, Bridgley Surgery, Bridgley, BY12 6XL
Name: Ranveer Singh
DOB: 27/01/1984
NHS number/Patient ID: 553 345 2134
Address: 10 Station Road, Bridgley, BY11 9DN

Clinical details: Ranvir's wife (Rebecca) has been identified as compound heterozygous for C282Y p.(Cys282Tyr) and H63D p.(His63Asp), he has requested testing and wishes to know the risk to their offspring. He has a serum ferritin level of 450µg/L

HFE genotype result: p.Cys282Tyr not detected, p.His63Asp not detected

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