

Director: Mrs D Pritchard
Manager: Miss A De'Ath

Tel: +44 (0) 1443 622185
Email: ukneqashandi@wales.nhs.uk
Web: www.ukneqashandi.org.uk

UK NEQAS for H&I
Welsh Blood Service
Ely Valley Road
Talbot Green
Pontyclun
CF72 9WB

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 **02nd June 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 1/2026

GP Surgery: Dr. Julian Thorne, 22 Kingfisher Way, Mallowford, TH48 9PL

Patient Name: Rosalind Finch (female)

DOB: 14/05/1976

NHS Number/Patient ID: 270 705 4761

Address: 7 Windmill Row, Elderberry Lane, Mallowford, TH48 8BW

Clinical Details/Reason for testing: Rosalind says she has been tested for HFE previously and thinks the results were normal. She has a persistently raised serum ferritin level of 900µg/L and a Transferrin Saturation of 40%. The GP has requested a repeat HFE genotype.

How would you process this request?

HFE genotyping results: Unknown

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 2/2026

GP Surgery: Dr. Helena Mycroft, Marshland Surgery, 11 Low Street, Cinderbridge-on-Marsh, Drakeshire, DR15 1XX

Patient Name: Sebastian Vane

DOB: 22/02/1994

NHS Number/Patient ID: 497 510 9969

Address: 5 Thistlethwaite Mews, Cinderbridge-on-Marsh, Drakeshire DR15 3QT

Clinical Details/Reason for testing: Sebastian's wife was found to be compound heterozygous for p.(Cys282Tyr) and p.(His63Asp). Therefore, he has requested testing and wishes to know any risk to their potential offspring.

He has a serum ferritin of 350µg/L.

HFE genotyping results: p.(Cys282Tyr) not detected, p.(His63Asp) heterozygous

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.