UK NEQAS
International Quality Expertise

Histocompatibility & Immunogenetics

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Scheme 5B - Interpretive: HFE Genotype and Hereditary Haemochromatosis

Clinical Scenario 5B 03/2024

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.

Scenario/scoring rationale from the assessors:

Diagnosis of haemochromatosis excluded, risk low, exclude other causes of raised ferritin.

Clinical Scenario 5B 04/2024

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.

Scenario/scoring rationale from the assessors:

Must contain advice re: risk to offspring and state whether testing is appropriate for adult offspring.