## Scheme 5B – Interpretative HFE Genotyping and Hereditary Haemochromatosis

**General Comments Scenarios 3 and 4 /2020** **Distributed January 2021**

**Report format**
The clearest reports were concise and well -formatted, with sub-headed sections and a tabulated or highlighted results summary.

It is important to include all relevant information in the reason for referral.
Relevant phenotypic information provided – such as the ferritin / TSAT should be re-stated in the report.

**Scenario 3/2020**
Jonathan Briggs, male, D.O.B. 27/01/1984; Unique identifier 543 345 2134
Address: 118 Lairg Road, Newbigging, PH12 1RG
Referral from GP, Dr G Wright, Broad Street Surgery, Newbigging, PH11 8PP

Jonathan's wife 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.
Serum ferritin 450µg/L
HFE genotype result: p.Cys282Tyr not detected, p.His63Asp not detected

This patient requested testing as his partner is compound heterozygous for C282Y and H63D and laboratories were expected to comment on risk to offspring as requested.
He also had raised ferritin which should have been addressed in the report

The most valuable reports recommended further action to investigate his raised ferritin and stated that that there was a 50% risk that offspring would be heterozygous for either C282Y or H63D and also that these genotypes do not represent a clinically significant risk.

**Scenario 4/2020**
Daljit Singh, male, D.O.B 01/01/1994; Unique identifier 478 213 5489
Address: 87 Witney Way, Knill, LD8 1DD
Referral from GP, Dr T Humphries, Sutton Wick Practice, Knill, LD9 7AB

Clinical details:
Daljit says he has been tested for HFE previously and thinks his results were normal. He has a persistently raised serum ferritin level of 850µg/L and a Transferrin Saturation of 45%.His GP has requested a repeat HFE genotype.
How would you process this request?

This scenario involved a patient with presumed non-Caucasian origin who has raised ferritin and borderline TSAT levels. He understands that a previous HFE genetic test was normal. The GP has requested a repeat HFE genotyping test.

The expectation was for laboratories to inform the GP (either through a letter or laboratory report after repeat testing) that C282Y is unlikely to underlie abnormal iron indices in this patient (due to presumed ethnicity), and to suggest further investigation for other genetic or non-genetic causes.

It is appreciated that the wording of the clinical scenario could have been more prescriptive, so laboratories that described their laboratory procedure but did not provide an informative letter or report were not scored in this instance.