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

**General Comments Scenarios 1 and 2/2022** **Distributed April 2022**

**Case Number 1:**
John Naismith, male, D.O.B. 22/02/1952, Unique identifier 908 788 1356
Patient's address: 3 Park Lane, Upper Fifield, RC12 4JH
Referring GP: Dr Moira McFadyen, Fifield Surgery, Upper Fifield, RC12 6GH

Mr Naismith has a younger brother George, who has been shown to be homozygous for the p.Cys282Tyr mutation in the HFE gene. Consequently, John Naismith has been tested for the mutation. The test result shows that he is heterozygous for the p.Cys282Tyr mutation and heterozygous for the p.His63Asp variant. Mr Naismith is married and has two grown up children and 4 grandchildren. Mr Naismith's fasting Tsat is 50% and serum ferritin is 350µg/L. He has a little bit of arthritis but his health is otherwise good. The GP wishes to know what further actions need to be taken.

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**Case Number 2:**
Freya Jones, female, D.O.B. 12/12/1956 Unique identifier 879 645 4312
Patients address: The Hollies, Glebelands, RH6 5VQ
Referring GP: Dr Nicholas Farndon, The New Surgery, Glebelands, RH6 7TY

Mrs Jones has a brother-in-law in New Zealand who has been found to have haemochromatosis. Consequently, her sister has been tested and found to be a carrier of the p.Cys282Tyr mutation. Mrs Jones' GP has requested serum iron measurements prior to arranging a genetic test. The fasting Tsat is 60% but the serum ferritin is 250µg/L. The GP subsequently arranged for a genetic test and the result has come back that she too is a carrier of the p.Cys282Tyr mutation. The family want to know who now needs to be tested and what blood samples and tests are required. The family in the UK consists of a brother, husband and one son who is married.

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**General feedback/advice from the assessors:**

1. Please read the scenario carefully. It is rather like a school exam question; you need to address all the points raised. Don't address points that aren't in the referring letter.
2. A number of labs are using a mix of nomenclature. HGVS (Human Genome Variation Society) expect the use of the three letter amino acid abbreviation as opposed the one letter amino acid code. Please avoid the use of H/D and C/Y, this is not HGVS nomenclature.
3. Please include your laboratory number in the report to aid assessment.
4. Please number your pages, especially if the report runs to more than one page.
5. Although examples of "good report layouts" have been previously shared, we appreciate that some labs may have specific pre-set report layouts which are unable to be changed.