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Service information: Hereditary Haemochromatosis

Gene/Locus: *HFE* (6p21.3) **OMIM:** #235200

Referrals: Clinical Geneticists, Haematologists, Hepatologists, Gastroenterologists, Specialist Haemochromatosis Nurse, General Practitioners

Testing: Diagnostic testing offered in clinically affected patients, carrier testing offered to partners of affected/carrier patients, presymptomatic testing in patients at risk of developing haemochromatosis. Under 16s are not routinely tested for haemochromatosis.

Target Reporting Times:

- Diagnostic 28 calendar days
- Carrier 28 calendar days
- Presymptomatic 14 calendar days

Test Details:

- **HFE mutation analysis:** Allele-specific PCR for the mutations p.Cys282Tyr (C282Y) and p.His63Asp (H63D).

Service Details:

Over 90% of UK haemochromatosis patients have two copies of the p.Cys282Tyr mutation and less than 5% have one copy of each of the mutations p.Cys282Tyr and p.His63Asp (compound heterozygotes). Approximately 6% of the normal Caucasian population carry one copy of p.Cys282Tyr.
(UK Haemochromatosis Consortium. 1997 Gut: 41, 841-44) (King and Barton. 2006 BMC Medical Genetics: 7:81).

In the general population, it has been estimated that the risk of developing clinical haemochromatosis with two copies of p.Cys282Tyr is approximately 1%. However, a study of relatives of haemochromatosis patients found that 85% of male and 69% of female relatives with this genotype had iron overload, which may potentially lead to the disease

Information and advice can be obtained from the Nottinghamshire Haemochromatosis & Gastroenterology Service, c/o Endoscopy Centre, Nottingham University Hospitals NHS Trust, City Hospital Campus, Hucknall Road, Nottingham, NG5 1PB. Tel: 0115 9691169 Ext: 54139.

Sample Requirements:

- **EDTA** blood sample (1-4 ml), labelled with patient's **full name**, **date of birth** and **NHS number** or genomic DNA (20µl at ≥30ng/µl).
- Samples should be accompanied by a **fully** completed referral card which should include the patient's full name, date of birth and NHS number.
- Please also include details of the test, relevant clinical details and full details of the referring clinician and centre

Consent:

Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient's relatives and their health professionals. Following testing, a sample of the patient's DNA may also be used anonymously to validate new tests and for internal quality control purposes.