Hereditary Haemochromatosis (HH) Care Pathway – For GPs

Genetic testing of clinically unaffected individuals.

University Hospitals Bristol and Weston NHS Foundation Trust

- **GP to arrange molecular testing of an <u>Adult</u> individual who has a first degree relative with a clinical diagnosis of hereditary hemochromatosis (HH) or who is known to be a C282Y /C282Y homozygote.**
- **Put '<u>predictive</u> genetic testing of common mutation (C282Y) in HFE gene' on lab form & include family history.**
- **Testing requires 4ml of blood in EDTA.**

Results.

C282Y/C282Y homozygote.

- Has an increased risk of developing HH.
- Baseline bloods; ferritin, fasting transferrin saturation (TS), Full blood count (FBC) & Liver function tests (LFTs).
- Then monitor fasting TS & ferritin, annualy.
- If fasting TS & ferritin abnormal seek advice from secondary care
- Screening of first degree relatives reccommended.

C282Y heterozygote.
'Carrier of HH.'

- Not usually associated with significant health problems.
- No further action needed.
- First degree family members of a carrier not routinely screened.

Further advice regarding:

- Genetic aspects – contact on-duty Genetic Counsellor, Local Clinical Genetics Service.
- Symptoms and iron overload - contact local specialist in secondary care e.g. Gastroenterology, Haematology or Hepatology.

Notes:

- The Bristol laboratory no longer tests for a 2nd common genetic change found in the HFE gene, H63D. Neither homozygotes (H63D/H63D) nor heterozygotes (carriers) are believed to be at risk of Iron overload. C282Y/H63D compound heterozygotes are only at low risk of significant iron overload. It is possible that other labs may check for this. Mild to moderate iron overload may be a risk in association with other factors e.g. alcohol abuse, fatty liver disease and/or metabolic syndrome.
  Seek specialist advice about monitoring if concerned.
- There are rarer forms of HH due to mutations in other genes. Where possible it is important to confirm the diagnosis in the family & the result of any genetic tests.
- 75-85% of individuals who are homozygous C282Y do not develop symptoms.
- There are rarer mutations in HFE, but these are not routinely screened for.