Hereditary Haemochromatosis Care Pathway

Aim: Cascade test so that C282Y homozygotes and C282Y/H63D heterozygotes are identified

Wessex Clinical Genetics Service

Hereditary haemochromatosis is a genetic condition. There are two common genetic variants (mutations) in the *HFE* gene that cause hereditary haemochromatosis; these are called C282Y and H63D. Affected individuals usually have two C282Y variants (homozygotes) or a C282Y and a H63D variant (compound heterozygotes).

Further information for health professionals and patients is available on the Haemochromatosis UK website: https://www.haemochromatosis.org.uk/

How to Request Nationally Funded Genotyping as a GP:
Genetic testing forms can be accessed via the Wessex Regional Genetics Laboratory website: www.wrgl.org.uk by clicking on the 'Referral Forms' tab, selecting 'Rare and Inherited Disease Genetics' then printing the form titled "Rare and Inherited Disease referral form". The National Genomics Test Directory test code for "Iron overload – hereditary haemochromatosis testing" is **R95**. GP to complete patient details and own details under "Referring Consultant".

Patient is 1st degree relative or partner of:
- C282Y/C282Y (homozygote)
- C282Y/H63D (compound heterozygote)
- C282Y (carrier)

Age > 18 years?

Yes

No

Please can GP request genotyping from Wessex Regional Genetics Laboratory (see above box in red outline)

Children do not develop symptoms of HH as the build up of iron does not reach harmful levels until later in life. **GP to consider testing at 18 years.**

Patient with diagnosis of iron overload

Genotyping done?

Yes

No

- C282Y carrier
- H63D carrier
- H63D/H63D (homozygote)

- C282Y/C282Y (homozygote)
- C282Y/H63D (compound heterozygote)

Please refer to genetics:
- A genetic counsellor appointment will be offered

GP actions:
- GP to request genotyping on adult siblings
- GP to refer patient to hepatologist

Patient with incidental finding of:
- C282Y carrier
- H63D carrier
- H63D/H63D (homozygote)

Common variants not identified

Was patient referred with iron overload?

Yes

No

GP to refer patient to hepatology

No further action indicated

Do not need to be seen by genetics – please do not refer
- Carriers of the C282Y gene mutation should have their **fasting serum ferritin and transferrin saturation levels checked by their GP**. If these are normal they should be measured again in 5 years time to see if iron accumulation has occurred. If these levels are high, a referral to a hepatologist should be considered.
- H63D homozygotes and heterozygotes do not require regular testing of iron levels in the absence of symptoms.
- **GP could consider cascade testing for adult family members and partners**

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