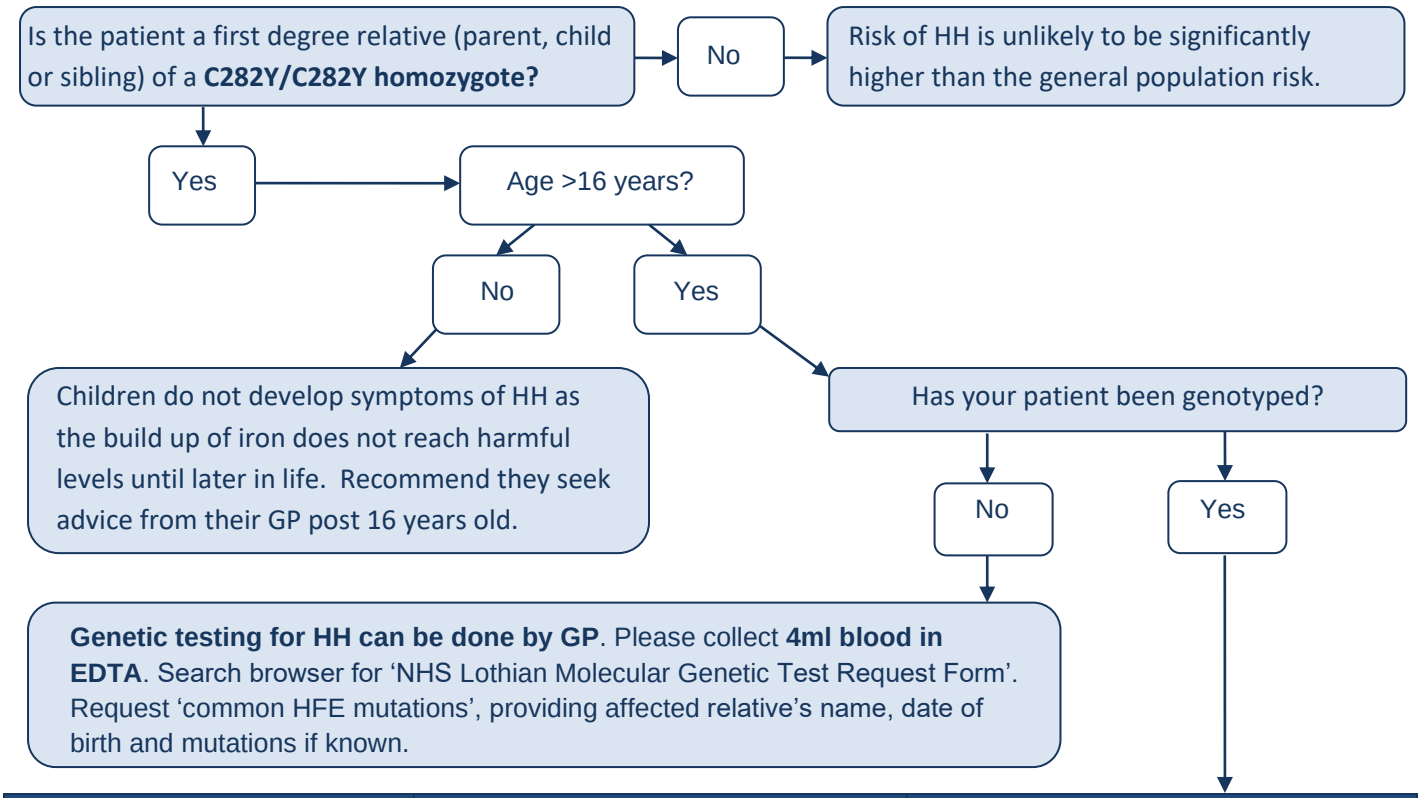
Hereditary Haemochromatosis (HH)

Advice for asymptomatic relatives when a genetic diagnosis has been made in a family member



Genetic test result	Patient management	Family advice
C282Y/C282Y homozygote	Measure transferrin saturation and fasting serum ferritin annually. If both raised, refer to haematology. If either elevated, contact haematology for advice. If both raised and abnormal liver function tests, refer to hepatology.	First degree relatives to seek advice from local GP following this pathway. If C282Y homozygote has multiple children, genetic testing of their partner could clarify if genetic testing is indicated in their children when over 16 years.
C282Y/H63D compound heterozygote	Measure transferrin saturation and fasting serum ferritin every 3 years. If both raised, consider referral to haematology. If either elevated, contact haematology for advice. If both raised and abnormal liver function tests, refer to hepatology.	Testing their children is not indicated as their risk of developing iron overload similar to general population, in absence of liver comorbidity factors
H63D/H63D homozygote or **C282Y** heterozygote (carrier) or **H63D** heterozygote (carrier)	Not likely to be at risk of developing iron overload. No regular follow-up recommended.	Reassure - being a carrier of HH is common in population. Testing their children is not indicated.

Additional guidance

Patients with unexplained increased iron levels can be investigated by a haematologist. Our patient information leaflets can be found through RefHelp > Hereditary Haemochromatosis > Resources & Links.

The gene changes that cause HH are very common. For people of Northern European origin, approximately 1 in 10 people carry one copy of C282Y and 1 in 5 people carry one copy of the milder H63D version of the gene.

Contact address: Clinical Genetics, Western General Hospital, Crewe Road South, Edinburgh, EH4 2XU
Telephone number: 0131 537 1116, Email: WGH.ClinicalGenetics@nhslothian.scot.nhs.uk