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Primary hereditary haemochromatosis (HH) is an autosomal recessive condition predisposing to pathological iron overload.

CLINICAL PROBLEM	ACTIONS	IMPLEMENTATION
<p>1 First degree relative with HH (note 1)</p>	<p>Genetic testing (note 2)</p>	<p>If diagnostic of HH manage as below</p>
<p>2 Iron overload (note 3) i.e. <u>fasting</u> iron saturation above normal range for age and sex or serum ferritin above normal range for age and sex (in absence of concurrent iron therapy) (note 4)</p>	<p>Genetic testing</p>	<p>If diagnostic of HH manage as below</p> <p>Discuss with Haematologist or Gastroenterologist whether liver biopsy indicated if <u>fasting</u> iron saturation and/or ferritin suggest HH, but genetic test normal</p> <p>Refer to Gastroenterology to assess extent of liver disease if presenting serum ferritin >1000 mcg/L or LFT abnormal regardless of genetic test results</p>
<p>3 Hereditary Haemochromatosis diagnosed</p>	<p>All patients:</p> <ul style="list-style-type: none"> • CBC • <u>Fasting</u> iron saturation • Serum ferritin • Liver function tests • Fasting blood glucose • Alpha-fetoprotein <p>Patient <70 years, no evidence of iron overload:</p> <ul style="list-style-type: none"> • Venesection unnecessary • Monitor annually for iron overload and end organ damage <p>Patient <70 years with iron overload (note 3), commence venesection (note 5)</p> <p>Patient >70 years with no evidence of end organ damage discuss need for venesection with patient (note 6)</p> <p>Patient >70 years with evidence of end organ damage, commence venesection</p> <p>Patients having venesection (note 5)</p> <ul style="list-style-type: none"> • Review annually • Check venesection adequate • Bloods – liver function, fasting blood glucose, alpha-fetoprotein • Check cardiac function, enquire about sexual dysfunction and arthropathy 	<p>Refer Haematology if</p> <ul style="list-style-type: none"> • Evidence end organ damage (other than hepatic) (note 1) or • GP uncertain about diagnosis or management <p>Refer Gastroenterology if</p> <ul style="list-style-type: none"> • Abnormal LFT, in absence of other hepatotoxins <p>Discuss with Haematologist / Gastroenterologist if uncertain</p> <p>Commence venesection and refer to gastroenterology if there is evidence of liver damage, or to haematology if evidence of other end organ involvement</p>

NOTES

- 1 All first degree relatives of patients with HH should be investigated because diagnosis and appropriate management, prior to developing significant end organ damage such as diabetes, cirrhosis, hepatocellular carcinoma, cardiac dysfunction, arthropathy and impotence, is associated with normal life expectancy.
- 2 The gene for haemochromatosis has 3 common mutations: C282Y, H63D and S65C, all of which can be tested for. Homozygosity for C282Y, or less commonly double heterozygosity, (heterozygous for 2 mutations) accounts for >95% of patients with HH. Heterozygosity of a single gene defect alone does NOT usually result in excessive accumulation of tissue iron, unless associated with alcoholic liver disease, chronic active hepatitis or porphyria cutanea tarda.
- 3 Definition of iron overload
 - Fasting iron saturation – if above the normal limit for age and gender, suspicious of HH

or

 - Serum ferritin – if above the normal range for age and gender suspicious of HH (may be falsely elevated as an acute phase protein or with underlying liver disease).
- 4 Serum ferritin may be falsely elevated as an acute phase protein or reflect underlying liver disease, so does not always reflect iron excess (especially when the iron saturation is normal).
- 5 Venesection
 - a) Initial
 - Every 1-2 weeks until ferritin 20-50 mcg/L (and fasting iron saturation <50% where ferritin may be falsely elevated e.g. liver disease). Venesection can be accessed, at no cost to the patient, via community laboratories, Waikato Hospital Lab or Blood Transfusion Service (some haemochromatosis patients may be eligible blood donors). Older patients may not tolerate such frequent venesection. For them, less frequent venesection e.g. every 4 weeks may be more appropriate.
 - Monitor with CBC, ferritin and iron studies.
 - Check the haemoglobin result prior to each venesection and aim to maintain the haemoglobin at >115 g/L for women and >130 g/L for men.
 - b) Maintenance
 - Frequency of venesection determined by individual circumstances, but generally 3-4 times/year once controlled.
 - Monitor with CBC, ferritin and iron studies.
 - Check the haemoglobin result prior to each venesection and aim to maintain the haemoglobin at >115 g/L for women and >130 g/L for men.
 - Aim to keep ferritin 20-50 mcg/L (and fasting iron saturation <50% where ferritin may be falsely elevated e.g. liver disease).
 - If the patient becomes anaemic, or the Hb falls by >20 g/L consider other diagnoses.
- 6 If the patient is >70 years and has no end organ damage, particularly if ferritin <1000 mcg/L, venesection may not be necessary. Morbidity associated with venesection e.g. vasovagal episodes, anaemia is likely to cause more harm than good.
- 7 General advice. No iron or vitamin C supplements, minimal alcohol intake.
- 8 Patient Support Group

IRONZ
New Zealand Haemochromatosis Support and Awareness Group
PO Box 23072
Papatoetoe
Auckland
URL: www.ironz.org.nz
Membership \$20/annum. Can provide patients with venesection record books.