Management of Hereditary Haemochromatosis (C282Y/C282Y homozygotes)

- Baseline
  - All individuals should have LFTs and HbA1C checked
  - If symptoms – consider ECHO, serum testosterone, joint xrays

- Assess appropriateness for venesection
  - It is reasonable to defer venesection until hyperferritinaemia develops in individuals identified during family screening.
  - Consider co-morbidities that may affect tolerability of venesection
  - Asymptomatic individuals of advanced age are unlikely to benefit from a venesection programme whose aim is longterm prevention of clinical problems.

- Refer patient to New Zealand Blood Service for venesection where appropriate (click here for referral form)
  - Initially venesections are usually performed every 1-2 weeks but frequency can be adapted to both the initial levels of hyperferritinaemia and to the patient's tolerance.
  - The volume for venesection should be adapted to the patient's weight: 7ml/kg body weight, not exceeding 500ml per venesection.
  - Venesect until ferritin normalises (target ferritin 50-100). Monitor Hb and Haematocrit before each venesection. Delay venesection if these fall below normal. Check ferritin after every 4 venesections.
  - Maintenance – maintain ferritin 50-100. This is usually achieved with 3-6 monthly venesections.
  - Management is based on ferritin levels. Transferrin saturation levels can fluctuate and are acceptable when levels <75%. Transferrin saturation levels should not guide management.

- If baseline ferritin<1000 and normal LFTs individuals can remain under the care of their GP with initiation of a venesection programme as outlined above.

- If baseline ferritin >1000 or raised LFT at baseline arrange liver ultrasound and refer to gastroenterology for further assessment as such individuals are at increased risk of cirrhosis.

- Individuals with confirmed cirrhosis require monitoring for hepatocellular carcinoma. Referral should be made to gastroenterology and they have should have 6 monthly ultrasound of liver and 6 monthly AFP monitoring.

- Individuals should be educated regarding risk to family members and should encourage siblings to see their GP for screening. Screening of offspring should be deferred until they are adults with iron studies performed in the first instance.

- If unable to tolerate venesection, chelation therapy may be appropriate. Referral should be made to gastroenterology for consideration on an individual basis.

- The continuing appropriateness of venesection should be kept under intermittent review with particular reference to new diagnoses and medication changes that may affect tolerability of the procedure.