Hereditary Haemochromatosis Care Pathway
Aim: Cascade test so that C282Y homozygotes and C282Y/H63D heterozygotes are identified.

1. **Referral of 1st degree relative of:**
   - C282Y/C282Y homozygote
   - C282Y/H63D compound heterozygote

   Age > 18 years?
   - No → Triage. Advice by letter: Standard letter (1) to referrer CC: parent
   - Yes → Genotyping done?

2. **Referral with diagnosis of iron overload**
   - C282Y/C282Y homozygote or C282Y/H63D compound heterozygote
   - Yes → Genotyping done?

3. **Referral for incidental finding of:**
   - C282Y heterozygote
   - H63D heterozygote
   - H63D homozygote

   Referral with raised serum ferritin and/or raised transferrin saturation and/or other medical problems?
   - Yes → Standard result/information (3) letter to GP, CC: Patient
   - No → Triage. Advice by letter: Standard letter (5)

4. **Referred with raised serum ferritin and/or raised transferrin saturation and/or other medical problems?**
   - Yes → Referral for diagnosis of iron overload
   - No → Triage. Advice by letter: Standard letter (2) to GP to arrange genetic testing with iron studies

5. **Genotyping done?**
   - No → Re-refer if C282Y/C282Y homozygote or C282Y/H63D compound heterozygote
   - Yes → C282Y/C282Y homozygote or C282Y/H63D compound heterozygote

   1. Genetic counsellor appointment
   2. Suggest referral of adult siblings and adult children to GP for genotyping*
   3. Ensure referral to hepatologist

   Standard information letter (4) CC: GP/referrer (ensure referral to hepatologist)

   *NB: Partners of C282Y/C282Y and C282Y/H63D not advised for testing unless presenting with symptoms of iron overload