Haematology – Clinical Guidelines: Suspected Haemochromatosis – GP Referral Guidelines

Introduction

Hereditary haemochromatosis is an autosomal recessive condition predisposing to pathological iron overload which may affect the liver, pancreas, heart, pituitary gland and joints. Over 90% of cases are caused by homozygous (C282Y) mutation of the HFE gene which can be detected by PCR. A raised ferritin may also be reactive to other conditions, particularly other causes of liver disease, alcohol excess, infection, inflammation or neoplastic disease.

The following should be referred urgently for outpatient assessment:

- Elevated ferritin with evidence of otherwise-unexplained 'end organ damage': congestive cardiac failure, liver dysfunction, diabetes or hypogonadism

Appropriate investigation in primary care for patients not meeting criteria for urgent referral:

- Repeat ferritin measurement in 4-6 weeks
- Check liver biochemistry, fasting glucose, transferrin saturation
- Careful alcohol history
- Consider 'reactive' cause: infection, inflammation, neoplasia

Referral for specialist opinion should be considered for:

- Persistent unexplained raised ferritin >600 mcg/L, and/or transferring saturation >50%.
- Genetic counselling / screening of first degree relatives of hereditary haemochromatosis cases