Guidelines for Investigation of Elevated Ferritin and Transferrin Saturation and testing for Hereditary Haemochromatosis (HH)

Consider testing for Hereditary Haemochromatosis in patients with:

- Raised serum ferritin (see box 1)
- History of HH in first degree relative
- Unexplained abnormal liver function
- Clinical features which raise clinician's suspicion of HH
  - Arthralgia / arthritis
  - Skin pigmentation
  - Diabetes, amenorrhoea, secondary hypogonadism, loos of libido, impotence
  - Congestive cardiac failure

If serum ferritin elevated (>300mcg/L men, >200mcg/L women) consider

Inflammation
Liver disease
Malignancy
Genetic iron overload: Hereditary Haemochromatosis (HH)

If ferritin is elevated then check fasting transferrin saturation

Fasting transferrin saturation >45% or history of HH in first degree relative
(even if ferritin and fasting TS are normal)

Fasting transferrin saturation <45%

No genetic testing for HH required

HFE genetic testing
http://hospital.blood.co.uk/diagnostic-services/hi/hi-test-request-forms/

Positive → **Refer to MTW HH service**

Negative → **Consider non-HFE mutations**

Non HFE-mutations

These are rare.
May be considered if ferritin and fasting TS raised in absence of HFE mutations e.g. hemojuvelin (HJV), transferrin receptor-2 (TfR2), ferroportin (SLC40A1), hepcidin (HAMP), African iron overload.

These should be discussed with Consultant Haematologist.

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