DIAGNOSIS AND MANAGEMENT OF HEMOCHROMATOSIS

Summary of the Clinical Practice Guideline | February 2010

OBJECTIVE

Alberta clinicians are assisted to

- Diagnose patients at risk for HFE gene-associated hemochromatosis
- Prevent end-organ disease by early diagnosis
- Optimize clinical investigations followed by genetic testing
- Counsel and manage patients with a definitive diagnosis

TARGET POPULATION

Adults 18 years and older

EXCLUSIONS

Population screening for hereditary hemochromatosis

Children under 18 years of age

RECOMMENDATIONS

SCREENING

✓ Consider patients* for screening if they have:

- Unexplained bronze or gray skin discoloration
- End-organ disease
- Family history of confirmed hemochromatosis (see Algorithm)

* Hemochromatosis is more common in people of Northern European heritage

✓ Screen using percent iron saturation and ferritin, in the fasting state, on one occasion if suspicion is low. If suspicion is high, screen twice to confirm.

✓ Suspect genetic hemochromatosis if iron saturation > 45%, particularly if ferritin is > 300 ng/mL in males or 200 ng/mL in females

X DO NOT measure ferritin alone. Ferritin is NOT an adequate measure since it is an acute phase reactant which can be elevated in acute and chronic infection, inflammation and neoplasm

GENETIC TESTING

✓ Perform the genetic test if screening is suggestive of genetic hemochromatosis. The genetic test determines mutations in the HFE gene – C282Y and H63D (or S65C).

✓ Use the Molecular Diagnostic laboratory requisition for Edmonton or Calgary to order the test

✓ Provide basic genetic information to the lab prior to ordering genetic testing

✓ Consider the potential for insurance discrimination for asymptomatic patients

✓ Refer to Medical Genetics Clinics in Edmonton or Calgary if further counselling is necessary

MANAGEMENT

- ✓ Ferritin should be lowered to:
  - 50-100ng/mL in patients with evidence of end-organ damage
  - < 200ng/mL in women without evidence of end-organ damage
  - < 300 ng/mL in men without evidence of end-organ damage
- ✓ Maintain hemoglobin above 110 g/L
- ✓ Encourage regular blood donation if the patient is a suitable blood donor. Hemochromatosis is NOT transmissible through a blood donation.

ALGORITHM