

## Haemochromatosis Study -Protocol Summary for Requesting Doctors

To evaluate efficacies of different testing strategies for identification of individuals with haemochromatosis and clinical iron overload amongst the following high risk groups:

### Strategy 1: Patients with clinical symptoms associated with haemochromatosis

Patients with clinical symptoms associated with haemochromatosis (see table below), unexplained by other causes, may benefit from iron studies investigation. Iron studies (including Fe, TIBC, % iron saturation, ferritin) can be requested on routine PaLMS form. Recommended follow up for abnormal results summarised in flowchart at back of this page.

### Possible clinical indications for haemochromatosis investigation

<b>1° relative</b>	Diagnosed as homozygous/ compound heterozygous for HFE mutations or with clinical haemochromatosis
<b>Liver disease</b>	<ul style="list-style-type: none"><li>enlarged liver, fibrosis, carcinoma</li><li>cirrhosis, liver failure</li></ul>
<b>Diabetes Mellitus</b>	
<b>Cardiac disease</b>	<ul style="list-style-type: none"><li>cardiomyopathy</li><li>arrhythmias</li></ul>
<b>Excessive skin pigmentation</b>	<ul style="list-style-type: none"><li>bronzed or slate grey</li></ul>
<b>Arthropathies</b>	<ul style="list-style-type: none"><li>polyarthritis</li></ul>
<b>Hypopituitarism</b>	<ul style="list-style-type: none"><li>Hypogonadism (amenorrhoea, loss of libido, testicular atrophy, impotence)</li><li>Adrenal insufficiency</li><li>Hypothyroidism /Hypoparathyroidism</li></ul>
<b>Iron overload related conditions</b>	<ul style="list-style-type: none"><li>Thalassaemia, Sideroblastic anaemia</li><li>Porphyria cutanea tarda</li><li>Hereditary acaeruloplasminaemia</li><li>Chronic excessive alcohol consumption</li></ul>

### Strategy 2: Asymptomatic Caucasian population

Iron screen (Fe, TIBC, % iron saturation) performed on asymptomatic Caucasian individuals, who have not had previous testing and have no known affected relatives.

Specific sticker to be included on PaLMS request form by medical practitioner when referring pt for enrolment. Patient should read 'Haemochromatosis Patient Information Sheet' and complete consent form, including brief questions verifying asymptomatic status, to be eligible for enrolment.

Results are reported to referring doctor with comments on suggested follow up of abnormal results.

### Strategy 3: 1° relatives of patients diagnosed with haemochromatosis, or homozygosity/ compound heterozygosity for HFE mutations

Patients diagnosed with haemochromatosis may be referred to PaLMS' associated clinical network (see contact details below) for long term monitoring, clinical management and advice on appropriate contact and referral of relatives for investigation.

Under current Health Insurance Commission regulations, DNA testing of 1° relatives for HFE mutations is covered by a Medicare rebate.

#### Contact details:

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## Recommended Testing Strategy

