

## A Basic Screening Test for Hereditary Hemochromatosis

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### Short Communication

Hereditary Hemochromatosis (HH) is a multiorgan disease defined as systemic iron overload, as a result of a reduction in the serum concentration of the hormone hepcidin which causes increased activity of ferroportin, the only identified cellular iron exporter [1]. This results in increased iron absorption from the diet and raised plasma iron levels leading to cellular iron accumulation in hepatocytes, cardiomyocytes and pancreatic cells. Clinical symptoms of HH include fatigue, right upper quadrant abdominal pain, arthralgia, symptoms of heart failure and diabetes. Physical manifestations of the condition include hepatomegaly, congestive cardiac failure and skin pigmentation. These signs and symptoms cause considerable debilitating morbidity in patients. The sequelae of this disease process and treatment modalities (such as venesection and iron chelation therapy) add to the significant financial burden already on the National Health Service (NHS) dealing with patients with chronic illness. It is the most common single gene disorder in North European populations with a prevalence of approximately 1 per 220-250 individuals [2].

The prevalence is greater in people from a Nordic or Celtic background. 85% to 90% of cases are homozygous for C282Y mutation in the *HFE* gene on chromosome 6 with up to 35% of the homozygotes asymptomatic—hence its importance in early testing [3]. Diagnosis is non-invasive and includes clinical examination, assessment of plasma iron parameters, and genetic testing. Once a HH diagnosis has been made, imaging techniques can be utilised to identify organ damage. Magnetic Resonance Imaging (MRI) has emerged as the standard imaging modality for the detection and quantification of hepatic iron deposition, as ultrasound (US) and computed tomography (CT) findings are nonspecific and influenced by multiple confounding variables [4]. Initial testing in a primary care setting widely recommends including a combination of serum ferritin (upper reference values, Men 300 µg/L; Women 200 µg/L) and serum transferrin saturation (>45%) assessment. Serum ferritin used in isolation is a marker of iron storage but raised levels can be associated with a number of false positives for iron overload as it is regulated by inflammation and may be increased in many disease conditions [5]. These biochemical investigations are far cheaper and more feasible than genetic testing initially, which is complicated by multiple molecular and clinical studies in the field.

*HFE* genotyping allows decisive and non-invasive diagnosis. However, multiple clinical genetic studies have led to the identification of genes other than *HFE* in patients with inherited diseases associated with increased hepatic iron storage [6]. This adds complexity to a diagnostic approach to patients with suspected hemochromatosis [6]. A General Practitioner (GP) in the NHS can play a pivotal role in the process of informing patients as well as relatives about genetic disorders such as HH. The GP is in a position to play a prompt referral role towards a specialised haematological and/or genetic consultation. Furthermore, primary care staff can also ensure more effective follow-up of the information procedures undertaken by its patients thanks to the medical follow-up that it carries out [7]. Given the prevalence of HH in the UK population, we suggest that an ad hoc testing of serum ferritin and transferrin in a primary care setting, especially in males >35 years would assist in mitigating against the long-term consequences of this disease in that it would flag up early those individuals that require further investigation. This in turn would reduce the significant financial burden to the NHS related to this disease's therapy and management.

### References

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