Scottish Government Health Directorates Chief Scientist Office

CHIEF SCIENTIST OFFICE

FOCUS ON RESEARCH

DEVELOPMENT OF A LABORATORY PATHWAY TO ASSIST THE DIAGNOSIS OF HEREDITARY HAEMOCHROMATOSIS IN PRIMARY CARE

Researchers
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Aim
To develop a hospital laboratory pathway to improve the diagnosis of Hereditary Haemochromatosis (HH) in Primary Care.

Project Outline/Methodology
HH, also known as the 'Celtic Curse', is a single gene disorder most common in North European and Celtic populations. One in 8 Scots are unaffected carriers of HH but 1 in 250 are at risk of HH. It is characterised by high iron levels which can damage the liver, the pancreas, the joints and other tissues. HH seldom presents before 30yrs but the diagnosis is often missed or made late in the disease. There is then a need to improve diagnosis as all complications can be prevented or lessened by simple blood letting (similar to blood donation).

Serum ferritin (SF) is a blood test of body iron stores and is frequently measured by GPs in Primary Care. This study examined patient samples sent by GPs in Greater Glasgow and Clyde with raised SF values.

A further blood iron test, Transferrin saturation (Tsat) was performed on samples with high SF . Samples with high SF and high Tsat were further tested for the HH gene (HFE gene).

Key Results
3734 samples from patients more than 30yrs were studied. HFE gene testing was performed on 1745.

878 males were tested for HFE gene.
58 patients with HH were detected. 57 of these had SF above 300umol/l and 52 had Tsat above 50%. Combining these SF and Tsat results allowed us to identify a group of male patients with a 1 in 5 chance of having HH.

867 females were tested for HFE gene.
74 patients with HH were detected. All 74 had SF above 200umol/l and 68 of these had Tsat above 40%. Combining these SF and Tsat results allowed us to identify a group of female patients with a 1 in 6 chance of having HH.

Conclusions
We would now propose:

1. The large number of SF requests received from Primary Care should be utilised to improve the diagnosis of HH.
2. The following algorithm (investigation pathway) should be applied to samples sent from Primary Care.

Patients above 30yrs;
Females SF above200µmol/l, males SF above 300µmol/l, should have iron studies performed. If Tsat above 50% (males) or above 40% (females); samples should be referred for HFE gene testing.

What does this study add to the field?
This study has developed and tested a laboratory algorithm which hugely enriches the detection of clinically relevant HH in primary care.

Implications for Practice or Policy
Improvement in the detection of HH will enable earlier treatment and help to prevent organ damage caused by iron overload. It will also help to identify other patients with HH through family screening after diagnosis in the index case.
The Scottish National Blood Transfusion Service should also benefit. Patients with treated HH can become Blood Donors. This will control their HH and also provide Blood for hospital patients.

Where to next?
We hope to trial the algorithm in a larger patient population in several Scottish healthboards with the final aim of developing a national algorithm for use in all Scottish Health Boards.

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