



**Australian Government**  
**Department of Health and Ageing**



Australia and New Zealand Horizon Scanning Network

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AND THE GOVERNMENT OF NEW ZEALAND

# **National Horizon Scanning Unit**

## **Horizon scanning prioritising summary**

**Volume 1, Number 4:**

**Population genetic screening for  
haemochromatosis: Identifying  
asymptomatic “at risk” homozygous  
individuals.**

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The production of these *Horizon scanning prioritising summaries* was overseen by the Health Policy Advisory Committee on Technology (HealthPACT), a sub-committee of the Medical Services Advisory Committee (MSAC). HealthPACT comprises representatives from health departments in all states and territories, the Australia and New Zealand governments; MSAC and ASERNIP-S. The Australian Health Ministers' Advisory Council (AHMAC) supports HealthPACT through funding.

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# PRIORITISING SUMMARY

**REGISTER ID:** 0000006

**NAME OF TECHNOLOGY:** POPULATION GENETIC SCREENING FOR HAEMOCHROMATOSIS

**PURPOSE AND TARGET GROUP:** IDENTIFYING ASYMPTOMATIC "AT RISK" HOMOZYGOUS INDIVIDUALS

## STAGE OF DEVELOPMENT (IN AUSTRALIA):

Genetic Testing for haemochromatosis is established technology, however population screening is not current policy.

- |   |  |
|---|--|
| <input type="checkbox"/> Yet to emerge      | <input type="checkbox"/> Established   |
| <input type="checkbox"/> Experimental       | <input checked="" type="checkbox"/> Established <i>but</i> changed indication or modification of technique |
| <input type="checkbox"/> Investigational    | <input type="checkbox"/> Should be taken out of use  |
| <input type="checkbox"/> Nearly established |  |

## AUSTRALIAN THERAPEUTIC GOODS ADMINISTRATION APPROVAL

- |                              |  |
|------------------------------|--|
| <input type="checkbox"/> Yes | ARTG number  |
| <input type="checkbox"/> No  | <input checked="" type="checkbox"/> Not applicable |

## INTERNATIONAL UTILISATION OF SCREENING FOR HAEMOCHROMATOSIS:

COUNTRY	LEVEL OF USE		
	Trials Underway or Completed	Limited Use	Widely Diffused
Australia	✓		
United States	✓		

## IMPACT SUMMARY:

The Medicare Benefits Schedule (MBS) provides item number 66794 with the aim of detecting the genetic mutation responsible for haemochromatosis, if patients have an elevated transferrin saturation or elevated serum ferritin, or a first degree relative with haemochromatosis; or a first degree relative with homozygosity for the C282Y genetic mutation. The technology is available through private and public pathology providers for individuals considered at risk of hereditary haemochromatosis (HH).

Individuals who are homozygous for a single mutation (C282Y) in the HFE are at risk of developing iron overload and subsequent haemochromatosis. Approximately 60% of C228Y homozygotes develop iron overload but it is unknown what proportion of these patients will go on to develop serious disease. Iron accumulation can damage the heart, liver, pancreatic islets and other tissues. Undetected cases can lead to cirrhosis of the liver, hepatic malignancy, heart failure or diabetes mellitus. Phlebotomy is a safe, effective and acceptable treatment, with possible benefits for the population at large due to the increase in the blood donor population. Early detection and therefore early treatment avoids the sequelae of the disease. A population based screening study in Australia (Olynyk et al 1999) found 0.5% of screened individuals were homozygote for C228Y and half of these individuals presented with clinical features of haemochromatosis. The number of claims processed by HIC for the

MBS item number 66794 has increased from 17,995 in the period July 1999 – June 2000, to 31,583 for the period July 2002 – June 2003.

A number of studies have found screening for HH to be cost-effective but this would require further research in the Australian setting.

### **CONCLUSION:**

Population screening for haemochromatosis has not been assessed, however use of the MBS item number (66794) is increasing and it is showing signs of rapidly diffusing throughout the Australian health system. Limited prevalence studies indicate that population screening for HH has the potential to deliver both long and short-term benefits.

### **HEALTHPACT ACTION:**

Therefore it is recommended that this technology be referred to the Pathology Services Tables Committee.

### **SOURCES OF FURTHER INFORMATION:**

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### **SEARCH CRITERIA TO BE USED:**

Hemochromatosis/epidemiology/\*genetics/\*prevention & control  
Mass screening  
Sensitivity-and-specificity  
Predictive-value-of –tests  
Homozygote  
Prevalence