Haemochromatosis Australia
Inherited Iron overload disorder

Submission to the Standing Committee on Health

Best practice in chronic disease prevention and management.

Haemochromatosis Australia

Haemochromatosis Australia is the not for profit support, advocacy and health promotion group for people with the genetic disorder haemochromatosis and their families.

Our vision is that no Australian will suffer harm from haemochromatosis.

www.haemochromatosis.org.au

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SUBMISSION TO THE SENATE STANDING COMMITTEE ON HEALTH:
Best practice in chronic disease prevention and management.

Best practice for haemochromatosis detection

Introduction
Haemochromatosis (inherited iron overload disorder) is the most common genetic disorder in Australia, affecting over 100,000 people. It is a significant cause of preventable chronic disease and can be fatal.

Approximately 1 in 200 Australians of European origin (106,000) have the genetic pre-disposition to haemochromatosis (homozygous for the C282Y mutation in the HFE gene, or 'homozygous C282Y') *see attached table*

This condition causes the body to absorb and store too much iron. If it is unmanaged 50% of men (26,000) and 10% of women (5,300) in this group will develop some or all of the following chronic conditions -

- Liver damage: fibrosis, cirrhosis or carcinoma
- Cardiac arrhythmia
- Diabetes
- Hormonal changes / impotence

An additional significant proportion will develop

- Chronic Fatigue
- Arthritis
- Generalised joint pain

Iron overload and these symptoms are easily prevented if the genetic predisposition to haemochromatosis is detected in young adults.

Where the genetic predisposition is detected iron levels can be easily monitored with routine blood tests to measure iron studies. Venesection (similar to blood donation) is the accepted and uncontroversial means of avoiding or reducing iron overload.

Various studies suggest that between 50% and 100% of haemochromatosis is currently undiagnosed[1-9]. In many cases diagnosis occurs as a result of the development of the morbidities listed above, all of which could have been prevented.

Optimal practice
Optimal practice would ensure that all people who are homozygous C282Y would

- Be diagnosed before the onset of the chronic conditions listed above,
- Have annual monitoring of iron studies, and
- Where iron overload is detected, commence venesection as necessary.

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Recommendation

Term of Reference 6 - *Innovative models which incentivise access, quality and efficiency in chronic disease prevention and management.*

We recommend that –

- the Medicare rebate available for genetic testing for haemochromatosis be extended to all 18-40 year olds, and
- general practitioners be financially rewarded for the initial episode of detection of people aged 18-40 who are homozygous C282Y, and
- A relevant 'Health Pathway' type resource be made available to all general practitioners to assist in achieving best practice response. (See http://www.healthpathwayscommunity.org/ for examples).

Rationale

Increasing the rate of early, presymptomatic, testing for haemochromatosis, and treatment as appropriate, will

- prevent a significant amount of chronic disease.
- lead to a significant reduction in the number of liver transplants required to treat end-stage haemochromatosis-related liver disease
- lead to a significant increase in the number of young, healthy, regular, lifetime blood donors to the Australian Red Cross Blood Service.

Incentivising GPs by rewarding only positive tests will

- focus effort on at risk populations, being Caucasians and in particular people of Celtic descent.
- Avoid encouraging testing of populations of low risk eg people of non-European descent.
- It will particularly encourage the cascade testing of family members of people with a positive test.

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Table 1: Cases of Haemochromatosis Identified During Studies in Australia and New Zealand 1990-2012

Study, Year [Reference] "Study Name"	Population	People screened, how	Previously known to be C282Y+/+	Newly identified through screening	% Undiagnosed C282Y+/+ prior to study	Prevalence of C282Y+/+ in the study
Delatycki et al, 2012 [3] **"ironXS"**	Victorian high school children aged 15-16, 65.6% Caucasian	5757 children were tested for the C282Y mutation via cheek-brush DNA testing	0	28	28/28 **100%**	28/5757 **1 in 206**
Allen et al, 2008 [4] **"HealthIron"** substudy of Melbourne Collaborative Cohort Study (MCCS)	Melbourne, Vic, prospective cohort study over a 12 year period, 31192 adults who were born in Australia/ Ireland/UK/NZ, 100% Caucasian	of 31192 enrolled adults, 29676 were tested for the C282Y mutation using blood extracted from neonatal-type blood-spot cards	12	78	78/90 **85%**	203*/31192 **1 in 154** *52 identified during MCCS before HealthIron; 61 were lost to follow-up
McCullen et al, 2008 [5]	Brisbane, QLD, tertiary public hospital patients who had blood samples sent to the pathology laboratory, ethnicity not stated	for all 18779 adult blood samples, TS checked, if >40%, same blood sample was also tested for the C282Y mutation	14	21	7/35 20%	35/18779 **1 in 537**
Delatycki et al, 2005 [6] **"HaemScreen"**	Victorian work places, mix of white & blue collar, mix of rural, metropolitan, CBD, 63.7% Caucasian	of 11307 enrolled adults, 11197 were tested for the C282Y mutation via cheek-brush DNA testing	4	47	47/51 **92%**	51/11307 **1 in 221**
Olynyk et al, 1999 [7]	Busselton, WA, random sample of Caucasians in a longitudinal population study, 100% Caucasian	3011 adults with blood in storage from 1994 were tested for the C282Y mutation using blood extracted from neonatal-type blood-spot cards	4	12	12/16 **75%**	16/3011 **1 in 188**
Burt et al, 1998 [8]	Christchurch, NZ, random sample of adults from the electoral roll, 96.0% Caucasian	1064 adults consented to blood tests and were genotyped for the C282Y mutation using blood samples	0	5	5/5 **100%**	5/1064 **1 in 213**
Cullen et al, 1997 [9]	Brisbane, QLD tertiary hospital neonatal screening unit, "predominantly Caucasian sample"	1660 newborns were tested for the C282Y mutation using blood extracted from neonatal blood-spot cards	0	8	8/8 **100%**	8/1660 **1 in 208**
Pooled total of **representative general population screening studies** *(excluding McCullen et al 2008 as not representative of a general population)*			20	178	158/178 **89%**	**311/53991 1 in 173**

References

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