REPORT OF WORKING GROUP SET UP BY THE TÁNAISTE IN MARCH, 2006 TO EXAMINE THE NATURE AND EXTENT OF HAEMOCHROMATOSIS IN IRELAND AND TO ADVISE HER ON THE ACTION NECESSARY TO ADDRESS THE PROBLEMS CAUSED BY HAEMOCHROMATOSIS.

DEPARTMENT
OF HEALTH & CHILDREN
AN ROINN SLÁINTE
AGUS LEANAÍ

June 2006

Report of Working Group set up by the Tánaiste in March, 2006 to examine the nature and extent of Haemochromatosis in Ireland and to advise her on the action necessary to address the problems caused by Haemochromatosis.

Department of Health & Children
June 2006

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CHAIRMAN'S INTRODUCTORY LETTER TO THE TÁNAISTE

JUNE, 2006

Ms Mary Harney TD
Tanaiste and Minister for Health and Children
Hawkins House
Dublin 2

Dear Tanaiste,

I have pleasure in enclosing the report of the working party established by you in March 2006 to examine issues relating to Hereditary Haemochromatosis (HH).

HH is a potentially life threatening illness, which affects a sizeable number of Irish people. It is also an illness which can be easily and inexpensively detected and if caught on time, responds to relatively easy and inexpensive treatment. It is an illness which exists largely below the radar, its existence is unknown to most people, including people in the medical profession, policy-makers and opinion formers.

This short report examines the nature of HH, a disease characterised by a genetic predisposition to absorb excessive dietary iron, and its knock-on effects in liver and heart disease and in diabetes. It attempts to establish the extent of the problem, the steps needed to get a more accurate picture, especially since current figures almost certainly underestimate the extent of the problem in Ireland. The report examines such issues as the need for screening and awareness programmes, support for existing sufferers of HH, the role of the Irish Haemochromatosis Association, the use of blood donated by HH patients, discrimination by insurance companies and other related issues.

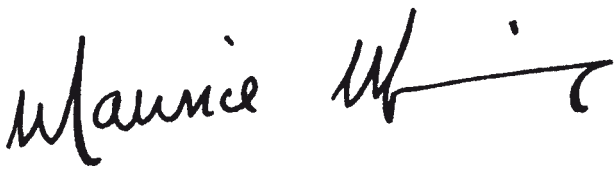
This report represents a serious opportunity to focus public attention on HH and to put in place, at relatively low cost, a series of measures which will have positive consequences out of all proportion to the cost involved. It is clear that, in most cases, once detected the disease can be effectively treated and that identification of the predisposition early in the course of the disease permits effective prevention. We are aware that this is not an exhaustive report. It is however a wake-up call proposing a series of easily implementable recommendations which need to be implemented with urgency.

The working party in grateful to you Tánaiste, for your speedy and generous response to our request for the establishment of this working group and we ask you to treat our report as a matter of urgency. We believe that if these measures, outlined in the report, are implemented, lives will be saved and a better quality of life will be possible for very many people who currently suffer from HH.

This working party would not have been possible without the crusading work of Ms Margaret Mullett, who with her colleagues in the Irish Haemochromatosis Association, have focused attention on the illness and campaigned for change. I would also like to thank the other members of the working party, Dr Suzanne Norris, Dr William Murphy, and Mr Brendan Gallagher. In particular, I would like to express my very sincere thanks to Ms Mary Jackson of the Department of Health and Children for her professionalism and dedication.

With best wishes,

Yours sincerely,



Maurice Manning

CHAPTER 1

Background to and Terms of Reference of the Working Group

BACKGROUND

It was agreed at a meeting on 15th February, 2006, between Ms Mary Harney, TD, Tánaiste and Minister for Health and Children and Dr Maurice Manning that a Working Group would be set up to examine all of the issues relating to haemochromatosis in Ireland and to advise her on the actions necessary to address these issues. Accordingly a Working Group was set up with membership as follows:

Dr Maurice Manning (Chairperson)
Dr William Murphy, National Medical Director of Irish Blood Transfusion Service
Dr Suzanne Norris, Consultant Hepatologist, St James's Hospital
Ms Margaret Mullett, Irish Haemochromatosis Association
Mr Brendan Gallagher, Irish Haemochromatosis Association
Ms Mary Jackson, Principal Officer, Blood Policy Division, Department of Health and Children

TERMS OF REFERENCE

The Terms of Reference of the Working Group were to:

1. Advise on steps needed to establish the extent of HH in Ireland;
2. Examine the screening procedures currently in place and advise on steps needed to put in place adequate screening procedures;
3. Examine current education and awareness programmes, both among the public and among GPs and advise accordingly;
4. Examine issues relating to insurance and advise accordingly;
5. Examine issues relating to the donation of blood by people with HH and advise accordingly;
6. Examine the future role of the Irish Haemochromatosis Association and advise accordingly;
7. Consider the advisability of restricting access to iron supplementation without a prescription;
8. Examine any other issues as may properly arise and advise accordingly.

The Working Group met formally on 3 occasions, with a number of other sub-group meetings.

CHAPTER 2

Nature and Extent of Haemochromatosis in Ireland.

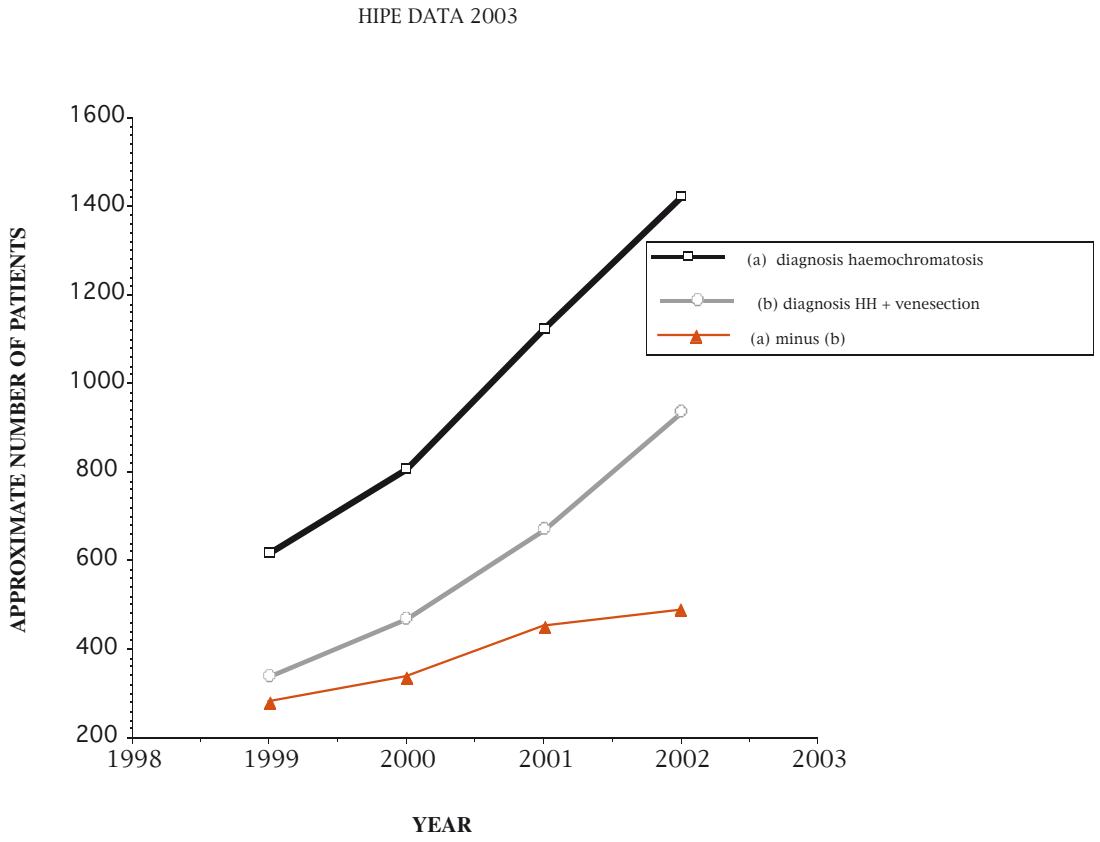
Hereditary haemochromatosis (HH) is the most common inherited autosomal recessive disorder in the white population with a prevalence of 0.2 - 0.5% in those of northern European origin, particularly those of Nordic or Celtic ancestry in whom it occurs with a prevalence of 1 in 200 of the population[1]. It is characterised by excessive absorption of dietary iron and a progressive increase in total body iron stores. The phenotype (or clinical expression) is due to iron accumulation in the parenchymal cells of the liver, pancreas, heart and anterior pituitary. In its most extreme form, the disease may manifest with potentially life-threatening complications such as cirrhosis, diabetes, cardiac failure and arrhythmias, and hepatocellular carcinoma (HCC). Other manifestations include skin pigmentation, a destructive arthritis, and hypogonadism[1]. These conditions are commonly reported in newly diagnosed patients with HH: diabetes 10-30%, arthritis 30-40%, arrhythmias 20%, and cardiac failure 15-35%.

In the absence of treatment, liver disease is the commonest cause of death in patients with HH. Hepatocellular carcinoma accounts for 30% of all deaths in HH, while other complications of cirrhosis account for an additional 20%. In a cohort of 251 patients studied between 1945 and 1991, cumulated survival at 5 and 10 years was 93% and 77%, significantly below the expected survival rates for age- and sex-matched normal population[2]. In untreated patients, the risk of mortality increases dramatically after the age of 45 years in men and 55 years in women[3]. The morbidity and mortality of HH can be reduced by early diagnosis and treatment to remove excess iron (venesection). **Early diagnosis is therefore important, as life expectancy in treated non-cirrhotic patients is normal.** The observation that HCC is rare in non-cirrhotic patients provides an additional argument for preventive therapy prior to the development of cirrhosis. However, there is still an average delay of 10 years between onset of symptoms (lethargy, arthralgia) and diagnosis of HH, because of the non-specific nature of the symptoms and the unfounded belief among health professionals that HH is rare[4].

In Ireland, HH is underdiagnosed and underreported. A review of 150 homozygous patients attending the haemochromatosis clinic at St James's Hospital over an 18 month period identified diabetes in 17%, cirrhosis in 20% of those biopsied, and HCC in five males as the presenting symptoms of HH. While acknowledging the small numbers, ascertainment bias, and impact of local environmental factors, nonetheless these data highlight that the diagnosis of HH is frequently made at a late stage in the clinical course of the disease. Published data from hospital registries or family studies report signs and symptoms of HH (arthralgia, fatigue, elevated liver biochemistry) for 2.3 to 7.3 years before the diagnosis is made[5]. Lack of early detection is a concern, as treatment does not reverse end-organ damage such as cirrhosis or diabetes. **Based on prevalence of 1 in 100[6], and with a population of 4,000,000, some 40,000 individuals in Ireland are homozygous for the C282Y mutation**. In the absence of a national register, estimating the numbers of persons with known disease is difficult and must rely on secondary sources. Figure 1 represents the number of

individuals with a diagnosis of haemochromatosis (based on genotyping) admitted to hospital in Ireland for the period 1999 to 2002 (source: HIPE). Despite a greater than 100% increase from 1999 to 2002, the numbers of individuals diagnosed (1,500) is low compared to the estimated target number of approximately 40,000. Haemochromatosis would therefore appear to be an ideal condition to consider for population screening in Ireland.

Figure 1.



Recommendations

1. **The Department of Health and Children should set out a policy endorsing a framework for management of haemochromatosis.**
2. **The Health Service Executive should aim to ensure early diagnosis of HH in order to prevent the onset of a range of serious medical conditions.**
3. **An action plan to address all aspects of HH should be set out and managed by the Health Service Executive.**

CHAPTER 3

Screening for Haemochromatosis

A gene called the HFE gene is involved in the regulation of iron absorption. Mutations of this gene cause HH. The main mutation is known as C282Y and a second mutation is H63D. About 1 person in 10 has a single C282Y mutated gene (heterozygote). About 1 in every few hundred people carry two C282Y mutated genes (homozygote).

HH fulfils criteria established by the WHO for population screening for a disease[7]:

- the homozygous genotype is common and potentially fatal if untreated;
- the disease has a lengthy phase of asymptomatic iron accumulation followed by a period of iron overload with reversible organ injury;
- treatment (which is safe, effective and low-cost) during these periods restores life expectancy to normal;
- it can be detected by measurement of the fasting transferrin saturation (TS).

The identification and treatment of asymptomatic persons in whom iron indices are elevated but HH is not clinically apparent has been recommended as a potentially cost-effective strategy for preventing HH-associated morbidity and mortality. Furthermore, consensus regarding the value of screening by simple biochemical parameters such as transferrin saturation (TS) in selected populations is high. Haemochromatosis should be suspected when the fasting TS is greater than 45%, a cut-off value which correctly identified 97.9% of Australian homozygotes with no false positives among the normal population[8].

An Irish study, which screened 330 people for HH, reported high sensitivity using a TS result above 45% as indicative of a positive test, i.e. highly suggestive of haemochromatosis[9]. Phenotypic screening of the general population using these standard iron indices with genotypic confirmation of HH in those with elevated biochemical markers has been reported to be a cost-effective strategy for identification of C282Y homozygotes in other countries. This strategy has a high predictive value even when it was assumed that fewer than 20% of the cases would develop life-threatening complications of the disease. In Ireland, there is no published large-scale study of the phenotypic expression (assessment of iron levels) of the C282Y mutation. Such a study would be useful in determining the necessity for nation-wide population screening. However, in countries where the prevalence of HH is high and where the majority of affected individuals are homozygotes, genotype screening has been recommended by professional bodies and associations (European Association for the Study of Liver Disease (EASL) Guidelines).

The primary objective of a screening proposal would be to determine the prevalence of HH homozygosity in Ireland by screening 10,000 subjects, and to establish a phenotype/

genotype correlation in affected persons. The establishment of a large cohort of HH patients with different levels of clinical expression provides a unique opportunity for translational research into the role of genetic modifiers of iron loading in disease expression.

These data will lead directly to the development of an Irish model of clinical care for patients with HH. The structure of the proposal coupled to stakeholder involvement will lead to the development and implementation of guidelines for the diagnosis and treatment of this common disease at primary, secondary and tertiary levels through national networks thereby providing equity of access to care for all persons.

RECOMMENDATION

4. **Funding must be prioritised to develop a HH screening programme. Information obtained from this programme will provide essential guidance to policy makers in structuring a countrywide programme for HH service development.**

CHAPTER 4

Education and Awareness and the role of the Irish Haemochromatosis Association

PUBLIC KNOWLEDGE OF HH

The knowledge that haemochromatosis is a hereditary disorder, which is one of the most common genetic disorders in the Irish population and that this condition, once it is detected at an early stage, can be easily treated to prevent organ damage and allow a normal life expectancy, should be the driving force for action.

A survey of 300 people in Kilkenny, carried out by the pupils of Scoil Aireagail, Ballyhale, Kilkenny, as a winning entry in the Young Scientist Exhibition in the RDS earlier this year set out to answer a number of questions relating to the condition. These questions and the answers to them are set out below.

How many people were aware of HH?
34% of those interviewed had heard of HH but only 19% could mention any one fact about the condition.

Did they know the seriousness and prevalence of it?
Were people aware of the connection between this disease and excess dietary iron?
Only 3.6% knew of the serious consequences for HH and only 6% made the connection with iron.

Were people aware of which foods contain high levels of iron?
Only 3 people (1%) were aware that breakfast cereals were a source of dietary iron.

How many people took vitamin/mineral supplements?
43% of women and 24% of men take iron tables when tired, without medical advice. The corresponding figures for vitamin/mineral supplements are 64% and 36%.

How did those who know about HH get their information?
Only 3 people had heard about HH in their General Practitioner's surgery.

The recommendations from the study, which were endorsed by the Irish Haemochromatosis Association are that **there should be a major Government-funded awareness and education campaign, that supplements containing significant amounts of iron should be available on prescription only, the iron content of all supplements should be highlighted, the**

fortification of foods should be investigated and the issue of screening for HH should be examined.

IRON ON PRESCRIPTION

Campaigning for 'iron on prescription' was one of the priority areas identified by the European Federation of Associations of Patients with Haemochromatosis. In a number of countries such as Austria, Holland, Belgium, France and Portugal, prescriptions are required to buy iron formulations with an iron content of 60 – 100 mg per tablet. In Ireland most pharmacies contain a range of products, some with high iron contents, which may be bought without a doctor's prescription. Some are promoted as being beneficial for reducing tiredness, which is one of the symptoms of haemochromatosis. **It is evident that regular consumption of these products can exacerbate the symptoms of HH** and could result in serious damage to persons unaware of their HH status.

ROLE OF THE IRISH COLLEGE OF GENERAL PRACTITIONERS

With education and awareness about the condition many more people than the number already diagnosed with the condition may discover that they have HH. **Collaboration with the Irish College of General Practitioners to ensure regular updating of its members on the need to screen for haemochromatosis would help to achieve this aim.**
As pointed out in the Chapter 2, failure to detect the condition and initiate treatment can lead to major long-term life threatening complications such as cirrhosis, diabetes, cardiac failure, arrhythmias and hepatocellular (liver) cancer.

ROLE OF THE IRISH HAEMOCHROMATOSIS ASSOCIATION

The Irish Haemochromatosis Association is a support group for HH patients and their families. Its main aim is to educate its members, the general public and members of the medical profession about the condition. Funding to date for the Association has been mainly from voluntary contributions, with €10,000 once-off funding provided in both 2003 and 2004 from the Department of Health and Children for the production of educational materials and a once-off grant of €25,000 in 2006 from the National Lottery towards administrative costs.

During 2005 Ireland became a founder member of the European Federation of Associations of Patients with Haemochromatosis (EFAPH), which comprises eleven countries including the UK, France, Belgium, Holland, Germany, Spain, Portugal, Italy, Norway and Sweden. The Federation will examine common issues and work together to tackle these. An initial priority area identified by the Federation is societal discrimination of patients with HH by the insurance industry.

In order to foster further education and awareness and to provide ongoing support to persons with HH the Working Group recommends that the Health Services Executive provide ongoing funding to the Irish Haemochromatosis Association so that it may appoint an executive assistant and clerical support to assist it with its work, which to date has been done on a voluntary basis from people's homes. An annual grant of €150,000 is required to appoint these staff and to fund the ongoing production and circulation of awareness materials.

Recommendations:

5. **There should be a major Government-funded awareness and education campaign. A key part of this would be a Haemochromatosis Awareness Week. The Health Service Executive should link with the Irish Haemochromatosis Association in devising this campaign.**

6. **Supplements containing high doses of iron (greater than 60mg) should be available on prescription only.**

7. **The iron content of all supplements should be highlighted by manufacturers.**

8. **Manufacturers of foods which are fortified with iron should alert persons with HH of this fact.**

9. **Participation by the Irish College of General Practitioners in a country-wide education campaign of its members is desirable.**

10. **General practitioners should refer HH patients and those testing positive for the C 282Y gene to specialist services, as required, for support and advice.**

11. **Ongoing mainstream funding should be provided through the HSE to the Irish Haemochromatosis Association to allow it appoint an executive assistant and clerical support and to provide education and support to persons with HH and their families.**

CHAPTER 5

Haemochromatosis and Insurance

INFORMATION FROM THE IRISH INSURANCE FEDERATION ABOUT LIFE ASSURANCE COVER

The Irish Insurance Federation has advised that where underwriting of insurance is concerned each application is looked at individually, getting as much detail as possible about a person's health and the decision to:

- offer cover,
- offer cover at an increased premium or subject to special conditions,
- or to decline to offer cover,

depends on the facts of each particular case.

With the passing of the Disability Act in 2005, the results of genetic tests (positive or negative) need not be disclosed to insurers and cannot be taken into account by the underwriter. However, the applicant is still obliged to disclose to the insurer full details of symptoms, treatment and family history. Where a blood test is normal and sufficient time has elapsed since diagnosis to establish that the condition is well controlled, an individual should generally be able to obtain life cover (which includes mortgage protection cover) at the standard premium. However, if there are complicating factors, e.g. there is a high iron level or organ damage, an extra premium may be applied and there may also be a small number of cases where the individual is uninsurable.

For patients with HH, the individual's current state of health at the time of the insurance application is the key issue. Under the Irish Insurance Federations's Code of Practice, genetic test results need not be disclosed where an individual is seeking life cover of up to €381,000. For life cover above this amount (or for other types of cover) haemochromatosis test results will not be taken into account in the underwriting process. The Code applies only to the results of the genetic test – the legal obligation to disclose all relevant information means that full details of symptoms, treatment, and where asked, family history, must be provided.

Where somebody is diagnosed with a condition or where his/her health deteriorates after taking out a policy, this will have no impact on any existing policies that he/she may have (as long as he/she has answered all questions fully at he time of applying for the policy and continues to pay the premiums due).

DISCRIMINATION AGAINST PEOPLE WITH HAEMOCHROMATOSIS

Information available to the Irish Haemochromatios Association, however, would indicate that a number of people with Haemochromatosis (HH) and others who carry the gene for the condition may be unfairly discriminated against by the insurance industry. Information has been obtained in

relation to the main insurance products such as life assurance, mortgage protection, critical illness, income protection and health cover.

The Working Group acknowledges, however, that the Equal Status Act allows people to be treated differently where insurance is concerned when this treatment is based on reliable actuarial or statistical data or other relevant underwriting or commercial factors.

Insurers differ, but the minimum deferral period for someone following diagnosis of the HH condition, is from six months to two years. Even when considered eligible for cover, applicants could expect to pay a loading of up to 100% on standard rates.

Voluntary Health insurance Healthcare (VHi)

Premia for members with HH are not loaded irrespective of duration of membership because VHI offers insurance coverage on a community rated basis. VHi has confirmed that any person with a diagnosis of HH is eligible to become a member of VHI and there is no loading of premia for such members, as it operates a policy of open enrolment and lifetime cover, as provided in legislation. There are restrictions on the provision of benefits for conditions that pre-existed the membership. These restrictions apply for a minimum period of time depending on the age at which the member joined. If a member joins under 55 years the minimum period is 5 years. This increases to 7 years for those aged 55 -59 and rises to 10 years for those aged 65 and over. Once these minimum periods have expired benefits are then provided for conditions that pre-existed the membership. This applies to all members with a pre-existing diagnosis of HH in the same way as it would apply to any other member with a pre-existing condition.

VHi Healthcare's approach to cover for genetic diseases is that if a member is enrolled at birth, s/he would be covered for any genetic diseases identified subsequently. All other members however are subject to the pre-existing rule. When considering whether a condition is pre-existing VHi has particular regard to the date of onset of the condition, as opposed to the date of diagnosis or onset of symptoms/signs of the condition. This is a key factor when considering whether a condition has pre-dated the membership. A genetic predisposition to a particular condition does not necessarily imply that the pre-existing condition rule will be relevant. This is because some genetic conditions have been identified as having patho-physiological mechanisms that are manifest from birth and these are subject to the pre-existing rule. VHi considers HH, when genetic in origin, to be consistent with this given that it is generally considered that the increased absorption of iron from a normal diet exists from birth. The VHi contends that with other conditions where the onset of patho-physiological mechanism is not as clearly defined, these are not subject to the pre-existing condition rule, unless the patient had knowledge of the condition before joining.

BUPA Ireland

Any member of BUPA who is diagnosed with a new condition after two years is fully covered. If a persons has symptoms of a conditions (irrespective of whether the condition has been diagnosed) they are covered for treatment following the first 26 weeks of membership in the case of persons

aged under 55 or following the first 52 weeks of membership in the case of the person over 55. If the person had symptoms of a condition before they joined BUPA Ireland then the condition would not be covered for 5 years for persons aged under 55, 7 years for persons aged between 55-59 and 10 years for persons aged over 60. Those waiting periods are not specific to haemochromatosis, they apply to persons taking out insurance for the first time, or have let their health insurance lapse for a period greater than 13 weeks. They do not apply to persons transferring from another health insurance company who have already served waiting periods nor to those joining from their dates of birth. If the person is transferring directly from another health insurer (or has been a member from birth) and has served all applicable waiting periods then that person is fully covered. No additonal charges are levied for any type of condition which the individual may have.

BUPA Ireland has stated that genetic testing cannot be used for the purpose of establishing a pre-existing condition for health insurance. Just as some people have a family history of cancer and so have a higher risk of being diagnosed with cancer, someone with a genetic predisposition of HH has exactly the same rights to health insurance as someone who does not have the genetic predisposition.

VIVAS Health

VIVAS Health responded to the Group that under the Health Insurance Act the rates of premium charged are the same for all adult members, irrespective of medical condition. In addition, persons cannot be denied coverage (except in very limited circumstances relating to fraudulent claims). There are no questions or tests for a member's medical condition or predisposition to an illness prior to or during their contract of insurance with VIVAS health, in compliance with the Health Insurance Act. The sole issue would be the application of pre-existing medical conditions in accordance with its handbook of membership.

Whether or not a person would be covered for treatment for HH would be dependent on the length of time they had health insurance coverage. An initial waiting period of 26 weeks applies to all new members who have never been previously insured (by any insurer) or have had a break of cover in excess of 13 weeks. This period rises to 104 weeks when a person is aged over 65. In addition, pre-existing conditions' waiting periods may apply if a person has not been with some insurer for at least 5 years.

GENETIC ISSUES AND DISCRIMINATION

The issue of genetic discrimination is a significant problem, particularly for first degree family members in the 25-35 age group who should have genetic screening but who are vulnerable to financial penalty by insurance companies if they have a positive gene test, even without evidence of iron load, without definite evidence of penetration of the C282Y gene mutation, or evidence that they will develop iron overload in the future.

Doctors should be obliged to advise that correct medical management is likely to make life insurance significantly more expensive even though pre-emptive diagnosis and treatment by preventing illness must save the insurance industry money in the long term. This is a very unsatisfactory situation, as the likelihood of a significant premium increase deters many people from having the genetic test. In Australia the insurance industry will provide cover as long as there is no long term damage i.e. cirrhosis etc.

A number of persons with a genetic predisposition to HH or in early stages of the condition are discriminated against because of poor information available to insurance companies on HH and morbidity/mortality risks. There appears to be a data network similar to that used by financial institutions whereby a reference check may be made on any potential applicant to establish if he/she has ever been refused cover by another company.

Voluntary Health Insurance's protocol means that members with existing policies of 5 years or longer, when diagnosed with HH, shall not be entitled to Venesection cover for a period of five years. This seems unduly discriminatory and the Group recommends that VHi review its protocol in this regard.

RECOMMENDATIONS

12. **In order to accommodate and resolve the discrimination experienced by HH sufferers/carriers it is essential that the Irish Insurance Federation examine the facts in this report and recommend to members that they should apply a non-discriminatory policy towards persons who have tested positive for the HH gene.**

13. **If a positive HH diagnosis is known, the criteria set out in the Irish Insurance Federation's Code of Practice should be applied, whereby younger applicants in particular, who are in treatment and have therefore no significant health risks, are not unfairly loaded when seeking insurance cover.**

14. **The issue of a consistent policy regarding insurance cover must be dealt with before a screening programme is initiated.**

15. **Voluntary Health insurance Healthcare should review its protocol whereby members who are diagnosed with HH must wait for a period of 5 years for cover for venesection.**

CHAPTER 6

Collecting blood for transfusion from donors with haemochromatosis.

Collecting and transfusing blood from people with HH is done in several countries including EU and EFTA states, the USA and Canada, and Australia and New Zealand. It is allowed under the Directives without any special conditions.

There is no biological reason for excluding such donors in the absence of any of other current deferral criteria, and it is certain that many current active donors are unaware they have HH, and that these include many who are among our more frequent donors.

The crux of the matter lies in the pressure on the donor to withhold important information either for his or her own well being, or more likely for recipient well being, so as to allow what is not a wholly altruistic act to proceed, and in the donor's own interests.

While several countries have tried to circumvent this, attenuate it, or simply ignore it in their approach to the issue, the principle remains that removal of the protection of a wholly altruistic donation is associated with increased risk of disease transmission, at least in the context of monetary reward; it is reasonable to conclude that failure to separate the donor's interests from those of the recipient is not in the recipient's interests.

A recent proposal by a Belgian group suggests a way forward that does not compromise the principle of a wholly altruistic blood donation – uncoupling the phlebotomy from the donation. Under this scheme, the person first presents for phlebotomy; once this has been completed satisfactorily they are then asked if they would like to donate the blood to the transfusion service, at which point they undergo the blood donor checks, interviews and consent process.

The major drawbacks are practical rather than philosophical – the donor does have to undertake an act of pure altruism, but the initial phlebotomy will have had to have been performed under rigorous blood transfusion service conditions of material control, personnel, premises and documentation control, donor and donation unique identification, and procedural exactness. The costs of this are not trivial – the collection bag will have to be an expensive Irish Blood Transfusion Service (IBTS) one rather than a cheaper dry pack for every collection, and staff will have to be deployed from IBTS resources to screen and treat many non-donors.

Nevertheless there are advantages: it moves the IBTS position on HH to one of engagement rather than standoff, which is appropriate for an organisation so solidly dependent on community good will and cooperation. It provides a source of blood, which may be of considerable benefit in the future; and it provides an opportunity for the IBTS to develop a position in providing therapeutic

phlebotomy as a service to the HSE and the private sector that may result in an overall economic benefit or balance.

The Working Group has been informed that the IBTS plan is to devote a special clinic to collecting blood from people with HH who require therapeutic phlebotomy. This clinic will be held regularly at key points – initially in Dublin and Cork, but they could be held in any major centre if there is demand and utility - for example every Tuesday evening in Stillorgan, every Thursday in D'Olier Street, etc. Donors would present with a prescription for phlebotomy, and would be assessed for fitness to undergo phlebotomy on the day. They would then be registered on a special panel, and undergo a standard IBTS phlebotomy – donor and donation number would be assigned prior to phlebotomy. Collection complete, they will be given their follow-up instructions and appointment. They would then be given an opportunity to donate the blood, which would remain in their possession at this point. If they opt to donate they then enter a separate process to fill out the donor questionnaire, undergo interview, and complete a donor assessment. At the end of this process the donation is accepted, or assigned as technically unusable, as appropriate.

A medical officer will complete the initial assessment for phlebotomy and provide the follow up plan; the actual phlebotomy and the donor assessment thereafter will be done according to current IBTS protocols.

Beyond this, regular HH donors who have been through this process a set number of times will be able to attend regular clinics – they will have an identifiable record as a HH donor, and additional questions can be asked under set and validated procedures by the Registered Nurse or Medical Officer to ensure that all is in order to allow a routine donation to proceed – the default position is that the donor can return to the special clinics. In due course, although it will take several years, the haemochromatosis procedures could be incorporated into regular clinics.

Constant surveillance of disease marker rates will give an indication over time of the validity of this approach, particularly when a new marker is introduced for a disease that should have been avoided by questionnaire and assessment.

When a screening programme for HH commences it will identify a number of donors who have the condition, either directly or though sibling tracing. It will be important for the IBTS to have a developed position in the event this happens to avoid unnecessary loss of committed, safe, wholly altruistic donors.

Pending availability of funding in the IBTS budget for 2007, planning and training will begin in 2006 to begin the first clinics early in 2007.

RECOMMENDATIONS

16. **The IBTS must set out its policy in relation to retention/deferral of donors who have HH, so that healthy donors are not lost.**

17. **Venesection services should be established by the Irish Blood Transfusion Service at the earliest date and not later than during 2007.**

CHAPTER 7

Venesection and costs for patients

Venesection (phlebotomy) means the removal of blood, just like giving a blood donation. It is the treatment of choice and the most effective strategy for management of HH. During venesection excess iron is removed. The earlier iron stores in the blood are depleted, the better the prognosis for the patient.

The only known drug that will safely remove iron from the body is desferrioxamine (Chelation therapy). Chelation therapy is used in some special circumstances, but venesection is the preferred treatment for HH at the present time.

There is a considerable variation in the cost of venesection depending on whether a person attends a hospital or their GP. If a patient is a medical card holder, there is no charge for venesection in a public hospital. The costs of venesection treatment in hospitals for people who do not have a medical card range from €60 up to maximum of €600 per annum**.**

Venesection is not covered by the GMS if carried out in a GP surgery.
A medical card holder will have to travel to the nearest hospital for phlebotomy. This may involve a very long bus/car journey at considerable expense and time to the patient. These patients may be reluctant to pursue their treatment and consequently they may jeopardise their health. This is not an acceptable situation. Ideally HH, which is a life long illness should be classified as such and treatment should be free to all patients, and through GP surgeries, if preferred. This is strongly recommended by the Irish Haemochromatosis Association.

RECOMMENDATIONS

18. **HH should be classified as a life-long chronic illness and treatment should be covered by the General Medical Service.**

19. **The cost of up to €600 for venesection treatment in public hospitals is a dissuasive element, especially for younger people with HH. The Department of Health and Children should consider introducing a standard fee (or removing this fee altogether) in order to attract more people to attend for regular venesection.**

20. **Arrangements should be made by the HSE to reimburse GPs for providing venesection in their surgeries. This would help to reduce the number of persons attending hospital outpatients unnecessarily.**

CHAPTER 8

Summary of Recommendations

1. The Department of Health and Children should set out a policy endorsing a framework for management of HH.

2. The Health Service Executive should aim to ensure early diagnosis of HH in order to prevent the onset of a range of serious medical conditions.

3. An action plan to address all aspects of HH should be set out and managed by the Health Service Executive.

4. Funding must be prioritised to develop a HH screening programme. Information obtained from this programme will provide essential guidance to policy makers in structuring a countrywide programme for HH service development.

5. There should be a major Government-funded awareness and education campaign. A key part of this would be a Haemochromatosis Awareness Week.

6. Supplements containing high doses of iron (greater than 60mg) should be available on prescription only.

7. The iron content of all supplements should be highlighted by manufacturers.

8. Manufacturers of foods which are fortified with iron should alert persons with HH of this fact.

9. The Irish College of General Practitioners should update its members of how HH can be detected and treated.

10. General practitioners should refer HH patients and those testing positive for the C 282Y gene to specialist services, as required, for support and advice,

11. Ongoing mainstream funding should be provided through the HSE to the Irish Haemochromatosis Association to allow it appoint an executive assistant and clerical support and to provide education and support to persons with HH and their families.

12. In order to accommodate and resolve the discrimination experienced by HH sufferers/ carriers it is essential that the Irish Insurance Federation examine the facts in this report and recommend to members that they should apply a non-discriminatory policy towards persons who have tested positive for the HH gene.

13. If a positive HH diagnosis is known, the criteria set out in the Irish Insurance Federation's Code of Practice should be applied, whereby younger applicants in particular, who are in treatment and have therefore no significant health risks, are not unfairly loaded when seeking insurance cover.

14. Voluntary Health insurance Healthcare should review its protocol whereby members who are diagnosed with HH must wait for a period of 5 years for cover for venesection.

15. The issue of a consistent policy regarding insurance cover must be dealt with before a screening programme is initiated.

16. The IBTS must set out its policy in relation to retention/deferral of donors who have HH, so that healthy donors are not lost.

17. Venescection services should be established by the Irish Blood Transfusion Service at the earliest date and not later than during 2007.

18. HH should be classified as a life-long chronic illness and treatment should be covered by the General Medical Service.

19. The cost of up to €600 for venesection treatment in public hospitals is a dissuasive element, especially for younger people with HH. The Department of Health and Children should consider introducing a standard fee (or removing this fee altogether) in order to attract more people to attend for regular venesection.

20. Arrangements should be made by the HSE to reimburse GPs for providing venesection in their surgeries. This would help to reduce the number of persons attending hospital outpatients unnecessarily.

REFERENCES:

1. EASL international consensus conference on haemochromatosis. J Hepatol 2000;33:485-504.

2. Niederau C, Fischer R, Purschel A, et al. Longterm survival in patients with hereditary hemachromatosis. Gastroenterol 1996;110:1107-19.

3. Yang Q, McDonnell SM, Khoury MJ, et al. Haemochromatosis-associated mortality in the US from 1979 to 1992: an analysis of multiple-cause mortality data. Ann Intern Med 1998;129:946-53.

4. McDonnell SM, Preston BL, Jewell SA, et al. Survey of 2851 patients with haemochromatosis: symptoms and response to treatment. Am J Med 1999;106:619-24.

5. Adams PC, Valberg LS. Evolving expression of hereditary haemochromatosis. Semin Liv Dis 1996;16:47-54.

6. Byrnes V, Ryan E, Barrett S, et al. Genetic haemochromatosis, a Celtic disease: is it now time for population screening? Genet Test 2001;5:127-30.

7. Tavill AS.AASLD Practice Guidelines: diagnosis and management of hemochromatosis. Hepatol 2001;33:1321-28.

8. McLaren CE, McLachlan GJ, Halliday JW, et al. Distribution of transferrin saturation in an Australian population: relevance to the early diagnosis of haemochromatosis. Gastroenterol 1998;114:543-49.

9. O'Hara R, Cavanagh N, Cassidy M, Cullina M. The role of transferrin saturation as a screening test for hereditary haemochromatosis in an Irish population seeking medical care. Ann Clin Biochem 2003;40:169-74.

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