Achieving optimal self-management in hereditary haemochromatosis

Results from a community questionnaire

Jessica Pearce, Robin A Ray, Suzanne McKenzie

Background and objectives

Hereditary haemochromatosis is a chronic inherited iron overload disorder that is primarily treated with venesection. The aim of this study was to investigate factors associated with optimal haemochromatosis self-management.

Methods

A national online questionnaire was developed. Questions covered demographics, and individual haemochromatosis history and management. The theory of planned behaviour also guided question development. Two logistic regression models were developed for the binary outcomes of serum ferritin within target range and intention to continue venesection.

Results

There were 378 complete responses collected. Questions measuring factors of importance ($P = 0.022$; odds ratio [OR]: 2.284; 95% confidence interval [CI]: 1.125, 4.637), preventing complications ($P = 0.017$; OR: 2.967; 95% CI: 1.211, 7.265), feeling involved with doctors' decisions ($P = 0.006$; OR: 1.482; 95% CI: 1.121, 1.958) and control of iron levels ($P <0.001$; OR: 1.868; 95% CI: 1.399, 2.493) were positively associated with dependent variables. Significant factors related to having a positive attitude and perceived control over haemochromatosis management.

Discussion

A positive attitude and sense of behavioural control contribute to successful self-management. Doctors are in an important position to provide self-management support and education.

HEREDITARY HAEMOCHROMATOSIS is a chronic inherited iron overload disorder that affects approximately one in 200 individuals of northern European ancestry.[1,2] The majority (approximately 90%) of individuals affected by haemochromatosis are homozygous for the C282Y mutation; H63D mutations are also associated with haemochromatosis, but to a lesser degree.[3,4]

Haemochromatosis presents variably between individuals. The most frequently reported symptoms include fatigue, joint pain, skin discolouration and loss of libido. Complications such as liver cirrhosis, cardiomyopathy and diabetes can also develop.[5,6] If detected and appropriately managed early through venesection therapy, those with haemochromatosis can avoid adverse consequences of iron overload. Once iron levels have been depleted, patients may variably need to continue regular venesections depending on their individual cases.[3,7] Although venesection is the mainstay treatment of haemochromatosis, it is also considered reasonable for individuals to make dietary choices to decrease iron intake. However, there are no formal dietary guidelines for haemochromatosis.[8,9]

Individuals with chronic diseases such as haemochromatosis should take responsibility for the ongoing management of their health status.[10,11] Self-management involves patients as active participants in their management plans in collaboration with healthcare providers. Patients should be appropriately educated about their condition and how to best use resources and act on any concerns that may arise.[10,12] The need for venesection can be guided by monitoring with iron studies, which is why ongoing collaboration with healthcare professionals is an important component of self-management for those with haemochromatosis.[12]

The intent to manage a chronic condition, such as having repeated venesections in the case of haemochromatosis, is voluntary, and theoretical frameworks can be useful in predicting this intent. The theory of planned behaviour (TPB) links individual intention to perform a particular behaviour back to three underling domains: attitudes, subjective norms and perceived behavioural control. Attitudes relate to whether one has a positive or negative view of the behaviour in question, while subjective norms relate to the social pressures and views of others surrounding the behaviour. Perceived behavioural control relates to self-efficacy and perceived ease of completing the behaviour in question.[13] Past applications of the TPB include predicting the self-management of chronic diseases such as renal disease, diabetes and airway disease.[14,15] This study used the TPB as a framework to guide analysis.

This research follows on from earlier focus group studies, the results of which have been published in *AJGP*.[16]

The aim of this study was to investigate factors associated with optimal self-management in individuals with haemochromatosis.

Methods

Questionnaire development

A structured questionnaire was designed using data from focus groups,[16] a systematic literature review and a guide to writing TPB questionnaires.[17]

Individuals with and without haemochromatosis piloted the questionnaire. Feedback was incorporated into the final questionnaire, which was then approved by James Cook University's Human Research and Ethics Council (H5714). The final questionnaire was released online using SurveyMonkey software (Appendix 1, available online only).

Participants and recruitment

The questionnaire was promoted to adults with a known diagnosis of haemochromatosis. The online questionnaire was advertised through general practitioners (GPs), haemochromatosis support groups and Haemochromatosis Australia, a patient advocacy organisation. Haemochromatosis Australia featured the questionnaire link on its website, social media pages (Facebook and Twitter) and newsletter. Participation was not restricted by genotype or past or current venesection schedules, and the questionnaire was open for 54 days during March and April 2015.

Variables

Optimal self-management of haemochromatosis within the questionnaire was defined using two dependent variables: a self-reported serum ferritin within normal range (binary outcome yes versus no/unsure) as well as the intention to continue to have venesections as necessary (yes versus no/unsure). Multiple choice questions included demographics and details of haemochromatosis diagnosis and management. Five-point Likert scales were used to explore the domains of TPB, with 1 being the most negative response through to 5 being the most positive (Appendix 1, available online only).[17,18]

Statistical analysis

Data analysis was conducted using Statistical Package for the Social Sciences (SPSS) version 23. Descriptive statistics were used to express frequencies of responses to demographic questions and details of haemochromatosis diagnosis and management. The Likert scale results of TPB were analysed to find the mean and standard error of the mean (SEM) for each variable. Independent TPB variables were then correlated with each of the two dependent variables using independent t-tests. A P value of <0.05 indicated statistical significance in this study; however, for the multiple comparisons for the t-tests, a Bonferroni-corrected P value of <0.003 was calculated. Two logistic regression models regressed TPB variables and selected sociodemographic variables (age, sex and education) onto each of the dependent variables. Odds ratios (ORs) were calculated with 95% confidence intervals (CI).

Results

A total of 406 questionnaires were attempted; 378 remained after data cleaning (removal of incomplete or duplicate responses). Of the 378 respondents, 354 individuals indicated that they required repeated venesections either currently or in the future. Twenty-eight respondents did not intend, or were unsure about, continuing venesections. The remaining 24 (6.3%) respondents reported not requiring repeated venesections.

Sociodemographics

The majority of participants (56.3%) were aged between 45 and 64 years. There were representatives from all age groups from 18 years to over 75 years, and 249 (65.9%) respondents were women (Table 1).

A small proportion (13.8%) of respondents had been diagnosed within the previous 12 months. Approximately one-third (33.6%) had been diagnosed between the past one and five years, with the remainder having known about their haemochromatosis for over five years. The reported genotypes of the respondents were: 161 (42.6%) C282Y homozygotes, 48 (12.7%) C282Y/H63D heterozygotes, and 103 (27.2%) unaware of their genotypes. The remaining 17.5% were heterozygotes, with either C282Y or H63D and one normal gene (Table 1).

Haemochromatosis management

GPs were the most common specialty of doctors that individuals consulted regarding their haemochromatosis management. Specialist haematologist involvement came second, while 5% of participants were not seeing a doctor about their haemochromatosis. Almost half (47.9%) were continuing to see their primary doctor at least every five months (Table 1).

Following a diagnosis of haemochromatosis, initial venesections to reduce serum ferritin levels were completed more frequently than once a month in 143 (37.8%) participants. Repeated venesections to maintain serum ferritin levels most commonly occurred between every three and five months (38.1%).

The theory of planned behaviour

The TPB-based questions were treated separately during analysis, with the underlying theoretical framework being used to interpret outcomes (Table 2).

Attitudes

The results show that participants indicated a positive attitude towards venesection. The mean Likert scale responses for all questions representing attitude were higher for those with a self-reported serum ferritin within the normal range and those intending to continue venesection. The differences between TPB attitude variables were statistically significant to the intention to continue venesection.

Subjective norms

The advice on managing haemochromatosis given to respondents by their doctors was shown to be of

Table 1. Sociodemographics and history of haemochromatosis results

		All responses (n = 378)
Demographics		
Sex	Male	129 (34.1%)
	Female	249 (65.9%)
Age (years)	18–24	10 (2.6%)
	25–34	25 (6.6%)
	35–44	59 (15.6%)
	45–54	101 (26.7%)
	55–64	112 (29.6%)
	≥65	71 (18.8%)
Highest level of education	Did not complete Year 10	18 (4.8%)
	Completed Year 10	51 (13.5%)
	High school graduate	41 (10.8%)
	Trade, technical or vocational	113 (29.9%)
	Bachelor's degree	91 (24.1%)
	Postgraduate degree	64 (16.9%)
Employment status	Employed	231 (61.1%)
	Retired	90 (23.8%)
	Other*	57 (15.1%)
Postcode	New South Wales	104 (27.5%)
	Australia Capital Territory	6 (1.6%)
	Victoria	57 (15.1%)
	Queensland	123 (32.5%)
	South Australia	35 (9.3%)
	Western Australia	21 (5.6%)
	Tasmania	21 (5.6%)
	Northern Territory	4 (1%)
	Not specified	7 (1.8%)
Respondent's haemochromatosis history		
Years since diagnosis	<1	52 (13.8%)
	1–5	127 (33.6%)
	5–10	93 (24.6%)
	>10	106 (28%)
Genotype	C282Y/C282Y	161 (42.6%)
	C282Y/H63D	48 (12.7%)
	Unsure of genotype	103 (27.2%)
	Other	66 (17.5%)
Primarily managed by general practitioner		236 (62.4%)
Initial venesection frequency at least once a month		143 (37.8%)
Ongoing venesection frequency every 3–5 months		144 (38.1%)

*Other employment statuses include: unemployed, student, volunteer, disabled/unable to work, home maker, carer

importance, with mean Likert scale ratings higher than the other subjective norm variables. However, this difference was not of statistical significance. There was a significant difference ($P = 0.001$) in the importance of family or friends' advice for those who reported serum ferritin being within the normal range (2.82; SE: 0.08) versus those who did not (3.26; SE: 0.11).

Perceived behavioural control

Feelings of being involved with doctor decisions were significantly associated with serum ferritin being within the normal range. Differences between ease of access to venesection and planning venesection around schedules reached statistical significance for those intending to continue venesection (both $P < 0.001$).

Logistic regression model 1: Serum ferritin self-reported as being within normal range

Believing that it is possible for someone with haemochromatosis to have normal iron levels ($P < 0.001$; OR: 1.868; 95% CI: 1.399, 2.493) and feeling involved with doctors' decisions ($P = 0.006$; OR: 1.482; 95% CI: 1.121, 1.958) were positively associated with self-reported serum ferritin within the target range (Table 3). Importance of doctors' advice showed statistical significance ($P = 0.021$), but with an OR of 0.632 (95% CI: 0.632, 0.429). This model is based on the Nagelkerke R^2 value and estimates 24.9% of variance in whether a person reports their serum ferritin being within the normal range or not.

Logistic regression model 2: Intention to continue with venesection

Belief that venesection is important ($P = 0.022$; OR: 2.284; 95% CI: 1.125, 4.637) and able to help prevent complications of haemochromatosis ($P = 0.017$; OR: 2.967; 95% CI: 1.211, 7.265) were positively associated with an intention to continue with regular venesection as necessary (Table 3). This model estimates 69.1% of variance in the dependent variable (Nagelkerke R2).

Discussion

Management of chronic conditions such as haemochromatosis can extend beyond the care offered by healthcare professionals and focus on how individuals choose to (or not to) engage in health-related behaviours.[10]

The TPB framework assisted with interpretation of the questionnaire results in this study. Attitudes and perceived behavioural control have been found to be significant in other studies investigating predictors of intention to adhere to required management regimens for chronic medical problems.[14,15] Subjective norms are the third domain from the TPB. However, we found that this was not a significant predictor of optimal disease control in haemochromatosis when variables were analysed individually (Table 2) and in regression models (Table 3) compared with the other two domains. Similar findings were evident in a study of renal transplant patients.[14] Yet, in studies of patients with chronic airways disease and diabetes, subjective norms are significant predictors of optimal disease management.[15] In chronic diseases with a management focus on medical interventions rather than lifestyle behaviour, support from family and friends may be less important than factors such as a positive attitude and perceived behavioural control. The results of this study show the importance of understanding how positive attitudes towards venesection and a sense of perceived behavioural control are relevant to optimising haemochromatosis self-management education. None of the sociodemographic factors included in the regression models (age, sex and education) reached statistical significance.

Not all individuals with haemochromatosis require repeated venesections, as indicated by 6.3% of questionnaire respondents. The results of this study indicate that education surrounding the importance of venesection and its role in the self-management of haemochromatosis play a part in increasing personal intention to continue venesection. Individuals with haemochromatosis could benefit from being educated about the effectiveness of venesection and how it is possible to remain in control of serum ferritin levels. The value of venesection can be brought into further context through explaining the potential complications that are being prevented through repeated venesection. This could also be an important educational target area for those who are unsure about or do not intend to continue venesection.

As respondents of the questionnaire valued involvement with doctors' decisions, doctors are in the prime position to play a vital part in delivering self-management education in collaboration with their patients. However, there is some contradiction in this finding, as the results also show that those who find doctors' advice more important may be less likely to have their serum ferritin within the normal range (OR: 0.632). The majority of respondents were visiting their GP at least annually. This provides GPs with a chance to check levels of understanding and opportunistically educate patients about self-management. Doctors also need to ensure their knowledge concerning venesection routines remains current. As the results of this study suggest a lack of influence of subjective norms including patients' families, significant others may be of less value when supporting patients and educating them about self-management. Ensuring that individuals optimally self-manage their conditions and keep serum ferritin levels within normal ranges will improve quality of life by reducing the effects of morbidity associated with iron overload for patients, their families and the healthcare system.[12]

Self-reported data for this study were collected as a cross-sectional questionnaire, and therefore it is important to recognise the potential for recall bias. There are possible inaccuracies in respondents' answers when recalling the details surrounding initial diagnosis and whether their ferritin was to target or not. The use of patient advocacy organisations and GPs to promote the study has the potential to bias the questionnaire towards individuals who are more proactive in managing their haemochromatosis. This was identified as an area of weakness in the study and addressed in part by the use of social media for advertising the questionnaire, which allowed the questionnaire to reach a wider number of patients with haemochromatosis who may not have had previous involvement with haemochromatosis organisations. It is difficult to calculate an expected response rate, as the actual number of people with haemochromatosis in Australia is not known, but the questionnaire was completed by people from a broad range of ages, locations and employment categories, indicating a useful spread across possible data sources. Another potential area of bias in this study could be the over-representation of women (65.9%); however, the results show that sex was not a significant predictor of disease management. Only 42.6% of respondents identified as being C282Y homozygotes, as many were unsure of their genotypes. Consequently, the results of this study could not be related back to genotype. Ongoing research into the predictive factors for optimal self-management of chronic disease will be able to provide further insight into factors that can be addressed to improve education resources and their delivery. Investigating the factors that underpin self-management from the healthcare professionals' point of view could also improve consistency between patient and practitioner understanding.

Conclusion

The results of this survey have provided information on predicting optimal self-management of haemochromatosis. This study has also shown new insight into the views of those with the condition. The results indicate that positive attitudes and perceived behavioural control contribute to an increased likelihood to self-manage the condition. While doctors are in an important role to help guide management, patient experience of haemochromatosis appears to vary. The role of family and friends in a patient's disease management appears to be of less influence.

Table 2. Differences in scores between theory of planned behaviour items, between reporting and not reporting serum ferritin within normal range, and intending and not intending to continue venesection (corrected *P* <0.003)

| | Serum ferritin self-reported as being within normal range (n = 378) | | | | | | Intention to continue venesection* (n = 354) | | | | | |
| | Yes n = 255 | | No/unsure n = 123 | | | | Yes n = 326 | | No/unsure n = 28 | | | |
	Mean	SE	Mean	SE	t-test	Sig†	Mean	SE	Mean	SE	t-test	Sig†
TPB – Attitudes												
Venesection is important	4.37	0.07	3.94	0.13	–3.02	0.003	4.54	0.04	2.32	0.24	–9.02	<0.001
Venesection is beneficial	4.35	0.06	3.97	0.12	–2.79	0.006	4.51	0.05	2.43	0.23	–8.58	<0.001
Venesection is tolerable	3.90	0.06	3.61	0.10	–2.52	0.012	3.93	0.06	2.82	0.18	–5.55	<0.001
Venesection improves symptoms	3.91	0.05	3.72	0.07	–1.98	0.048	3.94	0.05	3.18	0.10	–6.74	<0.001
Venesection prevents complications	4.39	0.05	4.19	0.07	–2.29	0.022	4.43	0.04	3.39	0.13	–7.06	<0.001
TPB – Subjective norms												
Others influence decisions	2.00	0.06	2.31	0.10	2.61	0.10	2.08	0.06	2.21	0.22	0.68	0.495
Importance of family/friends' advice	2.82	0.08	3.26	0.11	3.23	0.001	2.93	0.07	3.18	0.25	0.99	0.320
Importance of doctor's advice	4.33	0.05	4.38	0.07	0.56	0.572	4.40	0.04	3.75	0.25	–2.52	0.017
Importance of support of others	3.47	0.07	3.67	0.09	1.75	0.081	3.53	0.06	3.71	0.23	0.87	0.382
TPB – Control beliefs												
Belief it is possible to have normal iron levels	3.98	0.06	3.48	0.08	–5.04	<0.001	3.83	0.05	3.57	0.17	–1.42	0.156
Feeling involved with decisions being made by doctor	4.21	0.06	3.74	0.10	–4.22	<0.001	4.15	0.05	3.29	0.25	–3.36	0.002
Times of feeling less motivated to have venesection	2.13	0.07	2.52	0.11	3.00	0.003	2.24	0.06	2.75	0.28	1.78	0.085
Ease of access to venesection services	3.80	0.07	3.59	0.11	–1.65	0.099	3.83	0.06	3.04	0.28	–3.56	<0.001
Ease of planning venesection around schedule	3.49	0.07	3.14	0.10	–2.83	0.005	3.47	0.06	2.64	0.19	–3.76	<0.001

TPB, theory of planned behaviour
Means and standard error of the mean (SE) of Likert scale data values 1–5 representing least positive to most positive responses
*Of participants who are currently receiving venesection or are likely to need venesection in the future
†Significance following two-tailed t-test comparing TPB variables, adjusted P value <0.003 based on Bonferroni correction

Implications for general practice

Education resources targeting increasing understanding of haemochromatosis that specifically address attitudes and perceived behavioural control on an individual basis could increase self-efficacy and successful ongoing self-management. This can ultimately lead to improved patient outcomes throughout life with haemochromatosis.

Authors

Jessica Pearce MBBS (Hons), College of Medicine and Dentistry, James Cook University, Townsville, Qld. jessica.pearce1@my.jcu.edu.au

Robin A Ray BEd, MHSc, PhD, College of Medicine and Dentistry, James Cook University, Townsville, Qld. robin.ray@jcu.edu.au

Suzanne McKenzie MBBS, MMedSci, GCULT, PhD, FRACGP, College of Medicine and Dentistry, James Cook University, Townsville, Qld. suzanne. mckenzie1@jcu.edu.au

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Table 3. Logistic regression models

	Model 1 Serum ferritin self-reported as being within normal range				Model 2 Intention to continue venesection when needed			
	P value	OR	95% CI Lower	95% CI Upper	*P* value	OR	95% CI Lower	95% CI Upper
TPB – Attitudes								
Venesection is important	0.174	1.319	0.885	1.964	0.022	2.284	1.125	4.637
Venesection is beneficial	0.905	1.025	0.682	1.541	0.059	2.099	0.972	4.535
Venesection is tolerable	0.677	0.940	0.704	1.256	0.107	2.053	0.856	4.922
Venesection improves symptoms	0.175	1.263	0.901	1.769	0.661	1.260	0.448	3.545
Venesection prevents complications	0.439	0.863	0.595	1.252	0.017	2.967	1.211	7.265
TPB – Subjective norms								
Others influence decisions	0.058	0.767	0.583	1.009	0.590	0.794	0.343	1.838
Importance of family/friends' advice	0.275	0.874	0.686	1.113	0.447	1.274	0.683	2.378
Importance of doctor's advice	0.021	0.632	0.632	0.429	0.436	1.356	0.630	2.919
Importance of support of others	0.705	1.055	0.799	1.394	0.447	0.727	0.320	1.653
TPB – Control beliefs								
Belief it is possible to have normal iron levels	<0.001	1.868	1.399	2.493	0.758	1.130	0.519	2.458
Feeling involved with decisions being made by doctor	0.006	1.482	1.121	1.958	0.493	1.249	0.661	2.361
Times of feeling less motivated to have venesection	0.150	0.828	0.640	1.071	0.879	1.056	0.521	2.142
Ease of access to venesection services	0.374	0.882	0.670	1.163	0.650	0.856	0.437	1.676
Ease of planning venesection around schedule	0.224	1.204	0.892	1.624	0.061	2.186	0.965	4.952
Socio-demographics								
Age	0.485	1.075	0.877	1.317	0.673	1.123	0.655	1.924
Sex	0.206	0.691	0.390	1.225	0.400	0.491	0.093	2.576
Completed Year 10	0.962	1.031	0.295	3.603	0.318	0.156	0.004	5.974
High school graduate	0.341	1.892	0.509	7.033	0.355	0.164	0.004	7.580
Trade, technical or vocational training	0.413	1.631	0.505	5.263	0.081	0.041	0.001	1.477
Bachelor's degree	0.336	1.821	0.537	6.178	0.531	0.328	0.010	10.699
Postgraduate degree	0.262	2.067	0.582	7.345	0.852	0.656	0.008	55.564

CI, confidence intervals; OR, odds ratio; TPB, theory of planned behaviour

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correspondence **ajgp@racgp.org.au**