The voice of haemochromatosis journeys in regional Australia

A qualitative study exploring self-management

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Background and objectives
Hereditary haemochromatosis is a common inherited disorder of iron metabolism with avoidable long-term sequalae if it is detected early and managed well. The objective of this article was to explore factors influencing self-management of haemochromatosis in regional Australia.

Methods
Semi-structured focus group interviews were held in local community libraries. Data were transcribed verbatim, uploaded to NVivo data management software, descriptively coded and thematically analysed.

Results
Participants talked about living with haemochromatosis as an individual journey that requires balancing their changing symptoms, treatment and the demands of their daily life. Data analysis revealed four major themes: 'Discovering my haemochromatosis', 'Talking about venesection', 'Managing with diet', and 'Living with haemochromatosis'.

Discussion
People living with haemochromatosis had a positive attitude to the condition, but there were individual variations in the ways they engaged in self-management. A doctor–patient partnership is crucial in the management of haemochromatosis, especially in rural areas. Patients' symptom experiences and self-management decisions need to be seriously considered.

HEREDITARY HAEMOCHROMATOSIS is an inherited disorder of unregulated iron metabolism that affects approximately one in 200 Caucasian people.[1] The presenting symptoms, if any, are highly variable, but include fatigue, joint pain and skin discoloration. Progressive iron overload can result in damage to organs, including the heart, liver and pancreas, leading to morbidity and mortality.[1,2] In patients with elevated serum ferritin, haemochromatosis can be effectively managed through venesection. Individuals may also choose to make dietary changes to reduce iron intake; however, no formal guidelines exist and diet does not replace the role of venesection.[3,4]

Self-management requires patients to have sufficient knowledge of their condition and its management, as well as the confidence and self-efficacy to take control.[5,6] Self-management is optimised through collaboration between healthcare professionals and their patients.[5–7] Monitoring symptoms, presenting for venesection and possibly making dietary changes are ways in which individuals can self-manage their haemochromatosis. Regular medical monitoring of iron studies is necessary to guide the need for venesection.[3,4] However, access to healthcare resources in rural and regional Australia can vary, influencing effective disease self-management.[8]

Previous research into the management of haemochromatosis has investigated adherence to venesection therapy, response to treatment[2,9,10] and the general management of haemochromatosis.[11–13] Much of this work has been situated in a European or American context, and has relied on surveys to capture data. No Australian data are available, and no studies have given people living with haemochromatosis a voice to express the self-management of their condition. This study aimed to explore patient-identified factors influencing the self-management of haemochromatosis in regional Queensland, Australia.

Method

As the first phase of a sequential mixed-methods study, we explored participants' experiences of living with haemochromatosis and their self-management strategies, as a basis for a quantitative survey. We used two community-based focus groups to explore the realities of living with haemochromatosis.[14–16]

Ethics

Ethical approval was granted by the James Cook University Human Research Ethics Committee (reference number: H5714).

Participants

Adults with haemochromatosis were purposively sampled for this study. There were no restrictions on genotype, time since diagnosis or venesection frequency, which enabled a broad range of data to be collected.

A three-pronged approach to recruitment occurred through the distribution of information packs by five medical practices, the local haemochromatosis support group, and advertising on Haemochromatosis Australia's website and social media pages (the least useful for local recruitment). Once prospective participants returned

their consent forms, they were contacted and invited to participate in one of the scheduled focus groups.

Focus-group questions

Semi-structured questions, based on a systematic review of the literature and designed to gain insight into each participant's journey of haemochromatosis, were developed. Questions were reviewed and piloted prior to use with the focus groups (Table 1).

Data collection and analysis

Focus groups were held in September 2014 in meeting rooms in local public libraries. Two voice recorders were placed at a distance apart to capture all responses. Participants were reminded of the purpose of the focus group and asked to keep the information shared confidential. Consent was reiterated before commencing the discussion. Questions from the interview guide were introduced in response to the group discussion, enabling the natural flow of conversation to be guided towards the aim of the research. The groups were moderated by JP, and RR was the note taker.

Transcripts of the audio recordings were reviewed to ensure these were true and accurate to the original recordings. Data were de-identified before uploading to NVivo version 10 qualitative data management software. Descriptive codes were grouped into categories and emerging themes identified.[17–19] Researchers independently coded the data, and a consensus process was used to verify the final codes and themes.

Results

Thirteen participants from the region, aged 30 years to post-retirement, comfortably shared their experiences of living with and managing haemochromatosis (Table 2). No participants were related to one another. Four major themes emerged from the data:

- Discovering my haemochromatosis
- Talking about venesection
- Managing with diet
- Living with haemochromatosis.

Subthemes were also identified for each major theme, enabling further distinction of key issues.

Discovering my haemochromatosis

Each participant shared their own story about how they came to be diagnosed with haemochromatosis. The majority of participants were diagnosed either through their general practitioner (GP) investigating their symptoms, or following the diagnosis of a family member. The symptoms preceding the diagnosis ranged from debilitating ('I just wanted to go to sleep all of the time') to non-existent ('I had no particular symptoms that are normally associated with haemochromatosis'), which was consistent with the expected wide variation in presentation.[1,2]

A delayed diagnosis

The difficulty in achieving a diagnosis and resulting frustration were evident when four participants explained how their GPs initially missed the diagnosis of haemochromatosis. Non-specific symptoms of haemochromatosis, which are often present, were misdiagnosed as vitamin B12 deficiency, asthma, a part of the ageing process or dismissed as no concern:

> *I got to the stage where I was staggering and slurring, my whole body ached … but it took nine years for my doctor to diagnose me.*

Familiarity with patients over time may mask the diagnosis. In some cases, a change of doctor or a fresh approach led to the diagnosis of haemochromatosis.

> *Sometimes, it's really sort of a blessing really when they [regular GP] go on holidays and you get a locum because they're looking at you through totally fresh eyes … But, I mean, I could imagine that if someone fronted up to see me every month and I knew their story inside out back to front, that it's perfectly human that you continue on … there needs to be a bit of lateral thinking.*

Talking about venesection

The topic of venesection was discussed at length throughout the focus groups. All participants, except two who were newly diagnosed, had experienced venesection. Although some participants described the process itself as being unpleasant, the improvement in symptoms and appreciation of the importance of venesection were clearly evident in the data.

Continuing with venesections

All participants agreed that for those who needed it, organising their venesection is an important part of managing haemochromatosis.

> *I've accepted long term that I've always got to have venesection, and I think that's about it. I think as long as I keep doing them and getting checked I'll keep my ferritin low.*

Feeling better

Some participants identified increasing fatigue leading up to their next venesection and reduced fatigue post-venesection.

> *Two days after I have [venesection], I wake up and I feel like a whole big weight's lifted off of me. I feel lighter and not so tired.*

Challenges with venesection

Several participants discussed how the initial stages of having their iron stores reduced was challenging. During this initial stage, venesections occurred as often as every week for months. Venous access following repeated venesections was also highlighted as an issue for those with haemochromatosis. The role of nurses who are experienced in venesection technique is important for sustaining regular venesection.

> *When they [venesections] were weekly, I felt like 'Which arm do I use and which vein?', and I found that some of the nurses were particularly good at finding veins and some of them weren't.*

> *When I was giving it [blood] every two weeks, you feel like a bit of a pin cushion … it pays to have someone that's good to do it [venesection].*

Participants also expressed self-management knowledge about preparation for venesection.

Table 1. Semi-structured questions guiding focus group discussions

Can you share something about your haemochromatosis journey? How did you find out that you had haemochromatosis? What has been happening since your diagnosis?
How do you view the importance of having the right levels of iron in your blood? What do you do to manage your haemochromatosis?
Would anyone like to talk about their experience of venesection? How often do you have blood taken?
What are the positive aspects of having your blood taken?
Are there any problems anyone encounters with having their blood taken?
Other than venesection, is there any other way anyone manages their haemochromatosis? Does anyone use diet as a way of controlling iron levels?
Do you feel like you have control over the choices you make in managing your haemochromatosis?
How do other people in your life influence how you manage your haemochromatosis?
What does your doctor do?
What do you expect to happen with your haemochromatosis long term?

> *As soon as I hit the air-conditioning, I could feel my veins disappear, so I'd make sure I drank lots of water before and wore a coat while I was waiting for them to do the venesection.*

Access to venesection

Participants described having their venesections done across a variety of locations, including at general practices, local hospitals, specialist clinics and blood bank. However, those from more rural locations (up to 200 km away from the regional city centre) had more of a challenge accessing venesection; these patients expressed difficulties, such as needing to travel to a regional centre.

> *Most of them [practices] are only open 8am to 4pm, business hours and no weekends, so it's a bit harder to try and get access, especially if you're working away.*

Managing with diet

Participants eagerly shared about taking control of their diet and discussed this in terms of changes made and challenges encountered.

Dietary changes

Many participants made their own choice to reduce red meat consumption.

> *Well, I probably had meat every day previously … I now eat mostly chicken, fish, tuna, that sort of stuff … I rarely eat steak much now.*

Avoiding vitamin C because of its link with increasing iron absorption was another common dietary change.[4]

> *I have definitely cut out vitamin C, because I used to take vitamin C all the time, and that's one thing you just stop that altogether.*

> *I'd been taking multivitamins to kind of pick me up. When I was diagnosed, the GP said to me, 'Oh you'd better cut out the multivitamins because it's too much vitamin C'.*

Dietary challenges

Three participants described how making their usual dietary choices became difficult when travelling away from home.

> *If I know that I'm going to be travelling, then I might for the week or so before just really concentrate on having no red meat … I kind of balance it out that way.*

The need to travel and fit venesection appointments into a rural lifestyle also posed some challenges.

> *I know when in certain periods where I travel a lot, especially when you go out west … there isn't a lot to choose from, mostly steak or something like that. So, I find if I travel a lot in six months and then I go and get tested [for iron] again to do another bleed, it tends to be a little bit higher.*

Living with haemochromatosis

The impact of haemochromatosis on everyday life was discussed at length. Participants recognised that haemochromatosis is a manageable condition that has an ongoing effect on their lives.

It is manageable

While participants largely expressed confidence in managing their condition, there were also periods when the required changes almost became too much to manage.

> *In terms of my self-management, I'm sort of floundering a bit. I'm smart enough and I can be committed enough to do a good job. But it's like this combined with everything else just makes me get exhausted out of the blue, my brain doesn't seem to be able to focus, and I haven't quite got to the point of throwing my hands up and saying, 'Oh, stuff it' … but sometimes I think I come a bit close to it.*

Frustration was also expressed about finding the best management plan that works for their individual health needs.

> *I'm just hoping I find the right thing, whether it's swimming three times a week, pilates once a day, no cheese, I just wish I knew what the right mix was for me. I'm not thinking it's going to kill me or anything, I just want to survive each day with a bit less pain.*

Table 2. Focus group participant genders and recruitment methods

	Focus group 1 (n)	Focus group 2 (n)
Total participants	10	3
Male	4	2
Female	6	1
Recruitment method		
General practice	4	1
Support group	5	1
Haemochromatosis Australia advertising	1	1

In rural areas, consistent medical support for self-management was also a consideration. Having rapport with a regular doctor can be difficult in towns served by locums.

> *[specialist physician] said, 'Go and find a doctor to make friends with, and not one of these locums passing through'.*

Responding to my body

Most participants were aware of how their symptoms indicated the need for venesection and the consequences of delaying venesection, and were proactive in planning ahead.

> *I do feel better after ... I start to come good and feel really good, and I think I can tell when I need to have it.*

> *At one stage, the doctor wanted to try four-monthly ... well, the four-monthly venesections didn't work out because my ferritin level elevated, that's when I started having all these problems ... so he put me back to every three months now, and that's where I'm staying. I'm not budging.*

Taking control

Taking control and accepting responsibility for their haemochromatosis was a persistent theme. Participants emphasised that managing haemochromatosis is very personal, and they felt empowered to take charge and do what they could to improve their condition. Participants sought information from their doctors, the internet and others with haemochromatosis. Those who had been diagnosed for some years and/or were members of the support group spoke confidently about self-management.

> *Well, I believe that you've got to try and do what you've been told to do. Get plenty of physical exercise, eat right, don't touch soft drink or grog or anything like that.*

> *It's a holistic approach really ... it's the same thing as walking across the road when the light's the right way at the pedestrian crossing.*

Throughout the study, participants were active in monitoring their symptoms, preparing themselves for venesection and staying in contact with their doctor as ways to having some control over their management.

Discussion

For participants in our study, living with haemochromatosis in a regional area required lifestyle adjustments to achieve self-management, supported by timely medical intervention. Experiences of haemochromatosis varied for different presentations, and factors relevant to each person's context. Our data highlight important factors influencing self-management and the experience of living with haemochromatosis that have implications for patients and medical practitioners.

Patients living with haemochromatosis consider self-management to include dietary modifications, regular venesection and ongoing monitoring of symptoms. The attitudes to living with and managing haemochromatosis were generally positive, with indications of accepting the condition particularly evident in the subthemes 'It is manageable' and 'Continuing with venesection'. There were strong indications that patients wanted to take control of their symptoms, as evidenced in the subthemes 'Responding to my body' and 'Taking control'. In contrast to other studies of health behaviours,[20] our data suggest that the journey of having haemochromatosis is an individual process, and the role of family and friends does not seem to have a significant influence. However, having a regular doctor (in most cases a GP or haematologist) who is familiar with the individual's case is important for successful collaborative disease management.

Participants consistently reported dietary changes that could be implemented into their management routine, although they understood that venesection was the main intervention for managing haemochromatosis. It was apparent that in a condition such as haemochromatosis, which relies predominantly on medical management through venesection, patients may also implement their own disease control measures through monitoring symptoms, making dietary changes and maintaining, where possible, healthy lifestyles.

Many participants were well aware of their fluctuating symptoms and the role of venesection as an important component of self-management.[21] This was particularly evident when they reported an increase in symptoms leading up to venesection and relief after the procedure. This is consistent with earlier work by O'Connell and Sheahan,[22] whose participants conveyed an accepting attitude towards venesection and recognised the importance of this intervention. The findings of our study have relevance for the timing of venesection, highlighting the need for coordination of personal symptom monitoring and medical intervention,

and emphasising the need for a close doctor–patient relationship. Lack of a regular doctor had a negative impact on the journey of some participants, and was a further challenge to those in remote areas serviced by rotating locum doctors. Physical access to venesection facilities may also be an issue in some cases for those who work away or need to travel from more rural locations. While participants described the steps they could take to physically prepare for venesection, they clearly valued the role of health professionals who are skilled in performing the venesection procedure. Further work is needed to explore the efficacy of telehealth to enable patients in rural and remote locations to access consistent medical care.

While the focus groups included a range of participants recruited from the community, all participants were from one regional centre in Queensland, Australia, and were actively engaged in the management of their condition. Their level of understanding may be greater than that of those who do not participate in support groups or attend regular clinical care. Although only a small proportion of local general practices were accessed for recruitment, participants were evenly distributed for gender. Most participants recalled their experiences from diagnosis to the present, enabling a range of insights to be gained across the disease trajectory. The in-depth data resulting from the focus group discussion provided a wide range of data for the development of a detailed survey. Free text data from the subsequent nation-wide survey (unpublished data, JP, RR, SMcK) did not reveal any new themes, indicating that the views captured by the focus groups were representative of this cohort.

Conclusion

The results of this study suggest that living with haemochromatosis is an individual journey that requires consistent, medically supported self-management strategies guided by a positive attitude and awareness of their condition to achieve optimal disease management.

Implications for general practice

Doctors should work closely with individuals with haemochromatosis to educate them about the importance of self-management, and help guide the need for venesection therapy. However, further research is still required to measure the impact of self-management on the clinical outcomes of haemochromatosis, especially in rural areas where consistent doctor–patient relationships and access to venesection may be more difficult to maintain.

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Competing interests: None.

Provenance and peer review: Not commissioned, externally peer reviewed.

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