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 sequelae 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. 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. 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.

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. Self-management is optimised through collaboration between healthcare professionals and their patients. 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. However, access to healthcare resources in rural and regional Australia can vary, influencing effective disease self-management.

Previous research into the management of haemochromatosis has investigated adherence to venesection therapy, response to treatment and the general management of haemochromatosis. 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.

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. 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.12

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 veins?’, 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.
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.

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.

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.

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.
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, 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. 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, 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,
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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References


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.