Genetic Haemochromatosis: A Qualitative Exploration of Patients' Experience of Diagnosis in Primary Care

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Background

- Genetic haemochromatosis (GH) is the most common inherited genetic disorder in Caucasians (Bacon et al. 2011), and commonly affects Northern Europeans, especially those with Celtic or Nordic descent, with a ratio of approximately 1:220 - 250 people (Fitzsimmons et al. 2018; Phatak et al., 2008;). Despite the prevalence of GH only 1:5000 people are diagnosed with it (Haemochromatosis UK [HUK] 2019; British Liver Trust [BLT] 2017).
- In GH the body absorbs excess iron which can lead to systematic iron overload within the liver and other internal organs such as the pancreas, heart and joints; eventually causing inflammation and tissue damage. Early symptoms are non specific such as fatigue, abdominal and joint pain and as such, may be considered inconsequential by GP's, resulting in a delay in diagnosis and treatment.
- To date there has been little research examining patient's thoughts and experiences of being diagnosed with GH, a disorder which requires life long treatment with venesection, and which may lead to cirrhosis of the liver and increased risk of hepatocellular carcinoma (Ulvik 2015).

AIM

Explore patient experiences of living with Genetic Haemochromatosis and the diagnosis process and treatment

Design

- Data was collected using semi-structured interviews with a sample of 22 patients with haemochromatosis who responded to a poster advertising the study
- The interview covered their experience of diagnosis and treatment and the effect it was having on their lives
- Patients had been diagnosed between a year and more than 30 years The interviews were recorded and transcribed verbatim. Analysis of the data was conducted using thematic analyis.

Picture 1: Patient undergoing venesection

Results

Emerging Narratives around Diagnosis

There were number of common themes that came out of the interviews around the diagnosis process. Firstly, participants talked about the time taken before a diagnosis was made and a perceived lack of knowledge and information from GP about the disease

my joints were aching mainly and my right hand side of my chest…..I thought there was something not right here so I kept going back to the doctors to get a load of blood tests …and this particular doctor I was seeing said, I can't seem to find anything wrong with you…. And I thought there was something wrong with me. I can feel it inside of me

Time taken for diagnosis

Although a handful of patients were diagnosed relatively quickly, most patients interviewed talked about experiencing a delay, in some cases of many years, from first presentation of symptoms to their GP to final diagnosis.

No. Well he wouldn't know anyway because his knowledge about the subject, because I know I went back about something else and I said to him oh by the way I've got this haemochromatosis. "Oh what's that then?"

I didn't ask the doctors no, it was kind of, I suppose you lose a bit of confidence when you are watching them read Google in front of you as to what it is. And I know they have probably got the background and maybe they have got more that they can access but I didn't go back, it was pretty much she just handed me over

Knowledge and Information from GP

Many of the patients felt that GPs lacked knowledge of genetic haemochromatosis and talked about how GPs were unable to give them any detailed information about the disease .

Discussion

- Early detection and treatment for GH depends on increased knowledge of GPs. This qualitative study identified that patients perceive there to be gaps in understanding GH diagnosis and treatment. Ensuring GPs are aware of GH and the strategies for diagnosis could result in improved patient care.

Relevance

- These findings indicate that improved education for GPs regarding GH may be beneficial in order to improve patient care for this condition and potentially reduce delays in diagnosis

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