Genetic Haemochromatosis

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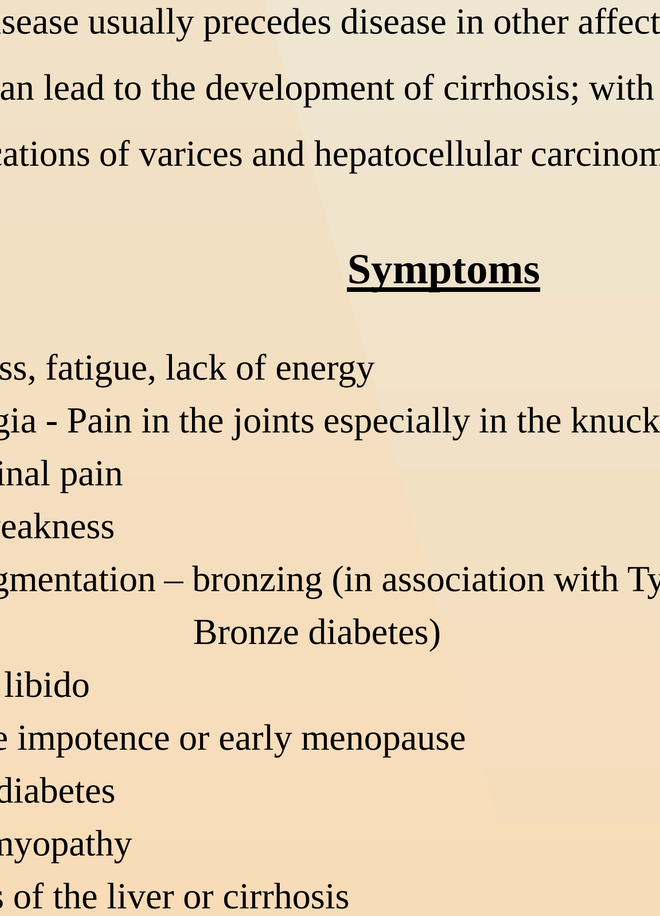
Genetic Haemochromatosis: what is it?

- Genetic haemochromatosis (GH) is the most common inherited genetic disorder in Caucasians (Bacon et al. 2011).
- Worldwide distribution but commonly affects Northern Europeans, especially those with Celtic or Nordic descent; ratio of approximately 1:220-250 people (Phatak, Bonkovsky and Kowdley 2008; Adams et al. 2005).
- The proportion of people diagnosed with this condition in Britain and Ireland is higher; approximately 1 in 200 British Liver Trust {BLT} 2017).
- Despite GH being the most common genetic disorder, approximately only 1:5000 people are diagnosed with GH, making it an under diagnosed condition (BLT 2017).
- GH causes the body to absorb more iron from the diet than is required and as the liver is the main storage for iron, any excess is transported there (Powell Deckington and Deugnier 2016).
- Over time, GH causes systemic iron overload not only within the liver but within other internal organs such as pancreas, heart and joints; eventually causing inflammation and tissue damage, which could lead to heart disease and diabetes.
- Liver disease usually precedes disease in other affected organs (Ulvik 2015) which can lead to the development of cirrhosis; with the associated complications of varices and hepatocellular carcinoma (HCC).

Symptoms

- Tiredness, fatigue, lack of energy
- Arthralgia - Pain in the joints especially in the knuckles of the first 2 fingers
- Abdominal pain
- Limb weakness
- Skin pigmentation – bronzing (in association with Type 2 diabetes called Bronze diabetes)
- Loss of libido
- Possible impotence or early menopause
- Type 2 diabetes
- Cardiomyopathy
- Fibrosis of the liver or cirrhosis
- Hepatomegaly
- Varices

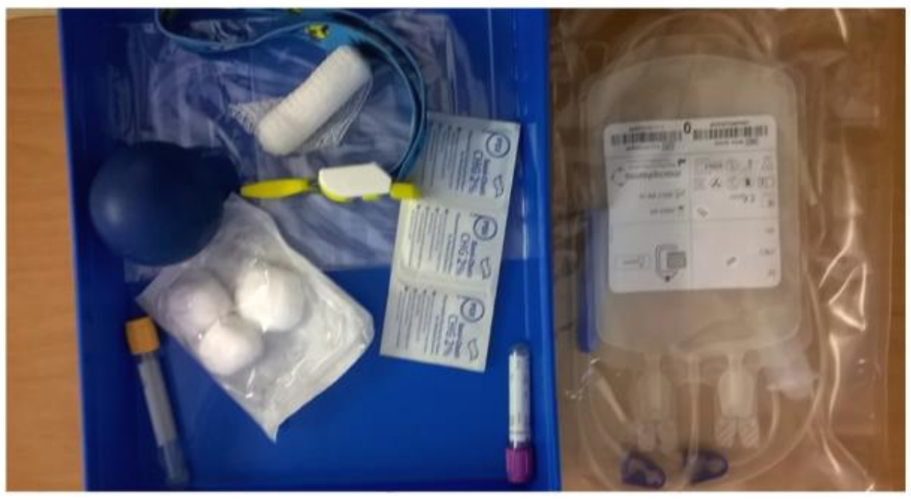


Picture 1: Venesection

(BLT 2017)

Treatment

- Treatment of GH is to simply to remove iron from the body.
- This is done by regular phlebotomy known as venesection. Approximately 400 to 500ml of blood is removed at one time once commenced can continue life-long.
- Informed, signed consent is usually undertaken for the first venesection to explain this, as well as the risk associated with phlebotomy.
- The main risk although extremely rare is ulna nerve damage. The more frequent minor risks are that of potential bruising and discomfort.



Picture 2: Venesection equipment

Research idea

There is little research which examines patient's thoughts and feelings of being diagnosed with a life-long disorder which requires life-long treatment in the form of venesection and which may lead to cirrhosis of the liver.

Exploring patient symptoms prior to, and after venesections has not been studied fully, nor the implication if they are diagnosed with cirrhosis.

In the initial phase of the disease venesections are undertaken weekly for many weeks/months. This may have a huge impact re time off work to attend treatment, cost of parking at the hospital etc. Understanding the effect of this on patients will enable the NHS to improve patient care.

Aims and Objectives of the Study

Aim

To examine the perceptions/perspective and experiences of those living with genetic haemochromatosis.

Objectives

- To ascertain how and when patients were first diagnosed with the disease and the effect of this on ontological security (patients sense of who they are)
- To examine patients experience of living with the disease and how they have had to make adjustments to daily life and work.
- To examine patients experiences of receiving treatment for the disease.
- To provide guidance to the NHS on how to improve patient care for this condition

References

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