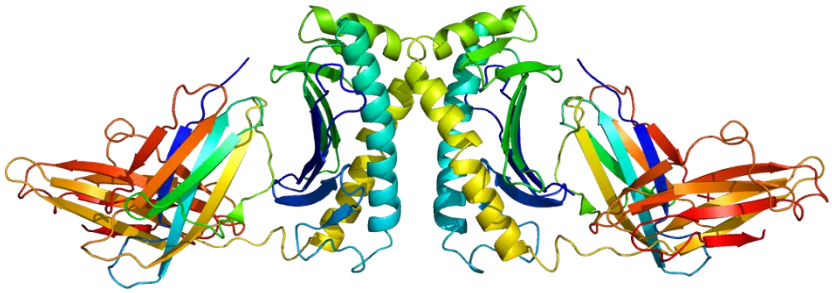
English for Nurses: Answers

Hereditary Haemochromatosis



Part 1: Vocabulary: hereditary haemachromatosis

hereditary	knuckles
haemochromatosis	hepatomegaly
HFE gene	enlarged liver
iron overload	nausea
ferritin	restrictive cardiomyopathy
arthritis	hyperpigmentation
joint pain	alopecia areata
joint inflammation	secondary diabetes mellitus

Part 2 - Complete the definitions of the terms in the vocabulary list:

1. hereditary
2. iron overload
3. liver
4. alopecia areata
5. restrictive cardiomyopathy

Part 3: Video questions

1. in the liver
2. the liver, pancreas, heart, skin and joints
3. Between 30 and 60 years of age
4. Blood test, x-ray and genetic tests
5. regular phlebotomy

Part 4: transcript

Hereditary haemochromatosis is an inherited condition that is caused by changes in a gene known as HFE. This results in the body absorbing too much iron from the diet. When too much iron builds up in the body, this is known as iron overload.

In a healthy body, a small amount of iron is stored in the liver and used when new red blood cells are formed. However, if too much iron is stored in other organs, iron overload results. This excess iron that is stored in organs such as the liver, pancreas, heart, skin and joints causes significant damage as a consequence.

Iron overload to other organs of the body may cause the following conditions:

- hepatomegaly (enlarged liver) with the symptoms of tiredness and weakness, loss of appetite and nausea
- secondary diabetes mellitus, similar to Type 1 diabetes, often requires insulin therapy
- skin hyperpigmentation which is particularly noticeable on the face
- alopecia areata, the partial loss of body hair, is most often hair loss in the pubic region
- restrictive cardiomyopathy causes the heart muscle of the ventricles becoming stiff, so that they have difficulty filling with blood. This can eventually lead to heart failure
- arthritis, joint pain and joint inflammation, that most often affects the knuckle and joints of the first two fingers

Hereditary haemochromatosis affects both sexes equally and typically presents between 30 and 60 years of age. Women tend to present later than men, although the clinical features of the condition are more commonly seen in males.

Investigations that are performed to confirm the diagnosis of haemochromatosis include:

- blood tests to check serum iron and ferritin levels
- X-rays to identify enlargement of the heart, a symptom of cardiomyopathy
- genetic testing for mutations of the HFE gene to either confirm the diagnosis or screen at-risk individuals for the early detection of the condition

The treatment of haemochromatosis consists of the regular removal of blood, called phlebotomy. This is similar to the process of blood donation. Initially, the procedure is performed every one to four weeks until a person’s iron levels reduce to normal levels. As a guide, for every millilitre of blood removed, 500 milligrams of iron is also removed. Once normal iron levels are reached, lifelong maintenance therapy every three to four months continues, to prevent a build-up of iron levels.