GPSA
GENERAL PRACTICE SUPERVISORS AUSTRALIA

Hereditary Haemochromatosis

Hereditary haemochromatosis (HH) is a very common genetic disorder in Australia, affecting approximately 1 in 200 people of European origin. It can lead to iron overload and though frequently asymptomatic, especially early in the disease course, it can lead to a wide range of symptoms, organ failure and death if untreated. GP registrars need to develop an approach to the patient with possible iron overload, including understanding the role for genetic testing.

TEACHING AND LEARNING AREAS

- Pathophysiology of iron metabolism
- Genetic background of HH
- Natural history and clinical features of HH and iron overload
- Investigations, including interpreting iron studies and the role of genetic testing
- Indications for referral
- Treatment options and local pathways for venesection referral

PRE- SESSION ACTIVITIES

- Read the 2010 AFP article Hereditary Haemochromatosis – Diagnosis and Management

TEACHING TIPS AND TRAPS

- There are multiple causes of elevated ferritin apart from HH and an elevated transferrin saturation is usually required for diagnosis
- There are rarer forms of HH (non C282/H63D) which may need specialist genetics referral
- Consider HH in patients with non-specific fatigue, liver disease of unknown cause, arthritis, ED and cardiomyopathy
- Only about 30% C282Y homozygous males and 1% females will develop clinically significant iron overload in their lifetime
- Iron studies are best performed after an overnight fast
- There is no role for population screening for HH, though case finding in patients with a family history is essential
- Patients with a ferritin over 1000 are at high risk for cirrhosis and warrant urgent specialist referral
- Gene testing should occur in first degree relatives of an index case
- There is no value in a low iron diet

RESOURCES

Read

- 2012 AFP article - Elevated Serum Ferritin – What should GPs know?
- Australian Doctor How to Treat – Hereditary Haemochromatosis
- Australian Prescriber 2011 - Testing for HFE-related haemochromatosis (includes an excellent algorithm)
- 2007 GESA Haemochromatosis Guidelines

Watch

- Hemochromatosis Australia website
- Haemochromatosis Explained – clinicians and patients discussing haemochromatosis (10min)

For ACRRM registrars:

- Diagnosis and Management of Haemochromatosis (on RRMEO)

FOLLOW UP/ EXTENSION ACTIVITIES

- Registrar to review the management of 5 patients with HH, including whether family screening has occurred, and present to the supervisor
- Supervisor and registrar to complete the Clinical Reasoning Challenges together and discuss

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Clinical Reasoning Challenge

QUESTION 1. Andrew is a 45 year old accountant who returns for discussion of his haemochromatosis gene test. He was referred because his brother was recently diagnosed with haemochromatosis. Andrew's iron studies are normal. His gene test shows that he is homozygous for C282Y.

Of the following statements, which of the following is CORRECT?

- As he has normal iron studies at age 45, he does not need further monitoring of his iron studies
- He should be referred for a liver ultrasound to identify occult fibrosis
- He has a 90%+ chance of developing significant iron overload by age 70 without treatment
- He should be referred to a gastroenterologist
- His mutation is the most common cause of hereditary haemochromatosis

QUESTION 2. Amelia is a 55 year old teacher who has been found to have an elevated ferritin (654) and raised transferrin saturation (51%) on iron studies.

Of the following statements, which of the following is CORRECT?

- As a female, the risk of clinically significant iron overload leading to organ damage is negligible
- As hereditary haemochromatosis is autosomal recessive, gene testing is not indicated
- The raised serum ferritin is unlikely due to an inflammatory response
- As a female, she has the same risk of significant clinical sequelae as males
- She should be referred to a geneticist for consideration of family screening

QUESTION 3. Max is a 25 year old student who tells you that his father has just been diagnosed with C282Y homozygote hereditary haemochromatosis. He is concerned and asks you some questions about it.

Of the following statements, which of the following is INCORRECT?

- HH is autosomal recessively inherited
- Max should commence a low iron diet to prevent his risk of iron overload
- HH is common in Australians of Northern European descent
- As a first degree relative, Max should have gene testing irrespective of the results of his iron studies
- Even with C282Y homozygosity, Max's father has a less than 50% likelihood of experiencing clinically significant iron overload

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ANSWERS

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