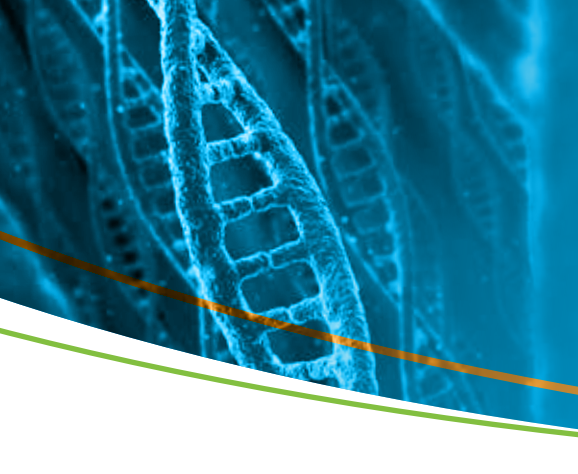

GPSA
GENERAL PRACTICE SUPERVISION AUSTRALIA

Hereditary haemochromatosis

Hereditary haemochromatosis (HH) is a very common genetic disorder in Australia, affecting approximately one 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 evidence-based 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 - a fasting value of >45% is suggestive of iron overload • There are rarer forms of HH (non C282/H63D) which may need specialist genetics referral • Consider HH in patients with non-specific fatigue, and unexplained liver disease, arthritis, ED or cardiomyopathy • Only about 30 per cent C282Y homozygous males and 1 per cent females will develop clinically significant iron overload in their lifetime • Iron studies are best performed after an overnight fast and >24hrs after taking iron tablets • 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 affected person
RESOURCES	**Read**	• 2021 GESA Clinical Update for General Practitioners and Physicians: Haemochromatosis • 2012 AFP article - Elevated Serum Ferritin – What should GPs know? • Australian Prescriber 2011 - Testing for HFE-related haemochromatosis (includes an excellent algorithm)
	Watch	• Haemochromatosis Explained – clinicians and patients discussing haemochromatosis (10min)
	Listen	• Haemochromatosis Australia podcasts
FOLLOW UP/ EXTENSION ACTIVITIES		• Registrar to review the management of five patients with HH, including whether family screening has occurred, and present to the supervisor • Supervisor and registrar to complete the MCQs together and discuss

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Hereditary haemochromatosis

Multiple choice questions

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.

Which of the following statements is CORRECT?

a As he has normal iron studies at age 45, he does not need further monitoring of his iron studies

b He should be referred for a liver ultrasound to identify occult fibrosis

c He has a greater than 90% chance of developing significant iron overload by age 70 without treatment

d He should be referred to a gastroenterologist

e 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 per cent) on iron studies.

Which of the following statementsis CORRECT?

a As a female, the risk of clinically significant iron overload leading to organ damage is negligible

b As hereditary haemochromatosis is autosomal recessive, gene testing is not indicated

c The raised serum ferritin is unlikely due to an inflammatory response

d As a female, she has the same risk of significant clinical sequelae as males

e 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.

Which of the following statementsis INCORRECT?

a HH is autosomal recessively inherited

b Max should commence a low iron diet to prevent his risk of iron overload

c HH is common in Australians of Northern European descent

d As a first degree relative, Max should have gene testing irrespective of the results of his iron studies

e Even with C282Y homozygosity, Max's father has a less than 50 per cent likelihood of experiencing clinically significant iron overload

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ANSWERS

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GPSA is supported by funding from the Australian Government under the Australian General Practice Training Program 02/07/2023