CLINICAL AUDIT

Genetic testing for **haemochromatosis**

Valid to November 2020

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Audit Focus

This audit is based on a recent article in Best Tests which focused on the diagnosis and management of hereditary haemochromatosis. The objective of this audit is to ensure that unnecessary genetic testing for haemochromatosis is identified and minimised.

For further information see: "Identifying and managing hereditary haemochromatosis in adults", Best Tests (Apr, 2015).

Background

Hereditary haemochromatosis is a condition characterised by increased iron absorption which leads to elevated iron stores and iron deposition in tissues, particularly the liver, which can lead to complications such as cirrhosis, fibrosis and hepatocellular carcinoma. Most cases of hereditary haemochromatosis are due to common risk alleles; whether a patient has these alleles can be assessed by genetic testing. Hereditary haemochromatosis is an autosomal recessive disorder, which means that people who inherit only one copy of an identified risk allele will not develop clinical disease.

In addition to a clinical examination and family history, the diagnostic process for identifying patients with haemochromatosis should include measurement of ferritin levels and transferrin saturation, which provide biochemical evidence of the patient's iron metabolism. There are a number of possible causes of elevated ferritin other than haemochromatosis, including acute illness and inflammation, alcohol intake, other liver conditions such as non-alcoholic steatohepatitis, hepatitis B and C, cancer, excessive dietary intake of vitamin C or iron and iatrogenic elevations from blood or iron transfusions. Patients with elevated ferritin levels which cannot be explained by these causes or those with transferrin saturation above 45% should undergo genetic testing for haemochromatosis. Genetic Health Services New Zealand does not recommend genetic screening of asymptomatic family members unless it is performed following the recommendation of a clinician with relevant genetic experience or after discussion with a genetic counsellor.

Recommendation

This audit focuses on key practice behaviours for the diagnosis and management of hereditary haemochromatosis:

1. Patients being assessed for the possible diagnosis of haemochromatosis should have ferritin and transferrin saturation levels measured first
2. Patients with elevated ferritin levels that are not explained by other causes, or who have transferrin saturation levels greater than 45%, should undergo genetic testing for haemochromatosis
3. Genetic testing of asymptomatic family members of patients with haemochromatosis is only recommended following the advice of a genetic counsellor or clinician with genetic experience

Audit plan

The recommended steps for completing the audit are:

1. Identify patients who have had genetic testing for haemochromatosis
2. Identify what proportion of these patients had evidence of elevated ferritin or transferrin saturation levels, or genetic advice, prior to genetic testing being ordered

Standards

Consider what percentage of patients might be expected to meet the requirements above. Ideally this number will be 100%, however, 90% is suggested as a target standard.

Data for completing the audit

Identifying patients

For this audit patients who have had a haemochromatosis genetic test requested by you or another member of your practice should be identified from patient records. This should be possible using the practice's PMS system, e.g. practices that use Medtech can use the Query Builder tool to search the Inbox for terms such as "haemochromatosis", "HFE gene", "C282Y" and "H63D". Record the details of all patients who have had a genetic test for haemochromatosis

Sample size

Ideally take all patients who have had a genetic test for haemochromatosis returned from a search, because based on the estimated prevalence of hereditary haemochromatosis, the number in most practices should be relatively low. If your search returns large numbers of results, take the first 20 patients. n the previous month to allow the sample size to reach 20.

Review patient notes

Review each patient's notes to establish if there is a record of elevated ferritin or transferrin saturation levels or genetic advice obtained prior to genetic testing being ordered.

Criteria for a positive result

In order to score a positive result in this audit, patients who have had a genetic test for haemochromatosis should:

- Have had elevated ferritin or transferrin saturation prior to undergoing genetic testing if they were initially diagnosed by your practice
- Or have undergone family screening following the advice of a clinician with genetic experience or genetic counsellor

Identifying opportunities for Audit of Medical Practice

The first step to improving medical practice is to identify the criteria where gaps exist between expected and actual performance and then to decide how to change practice.

Once a set of priorities for change have been decided on, an action plan should be developed to implement any changes.

Taking action

It may be useful to consider the following points when developing a plan for action (RNZCGP 2002).

Problem solving process

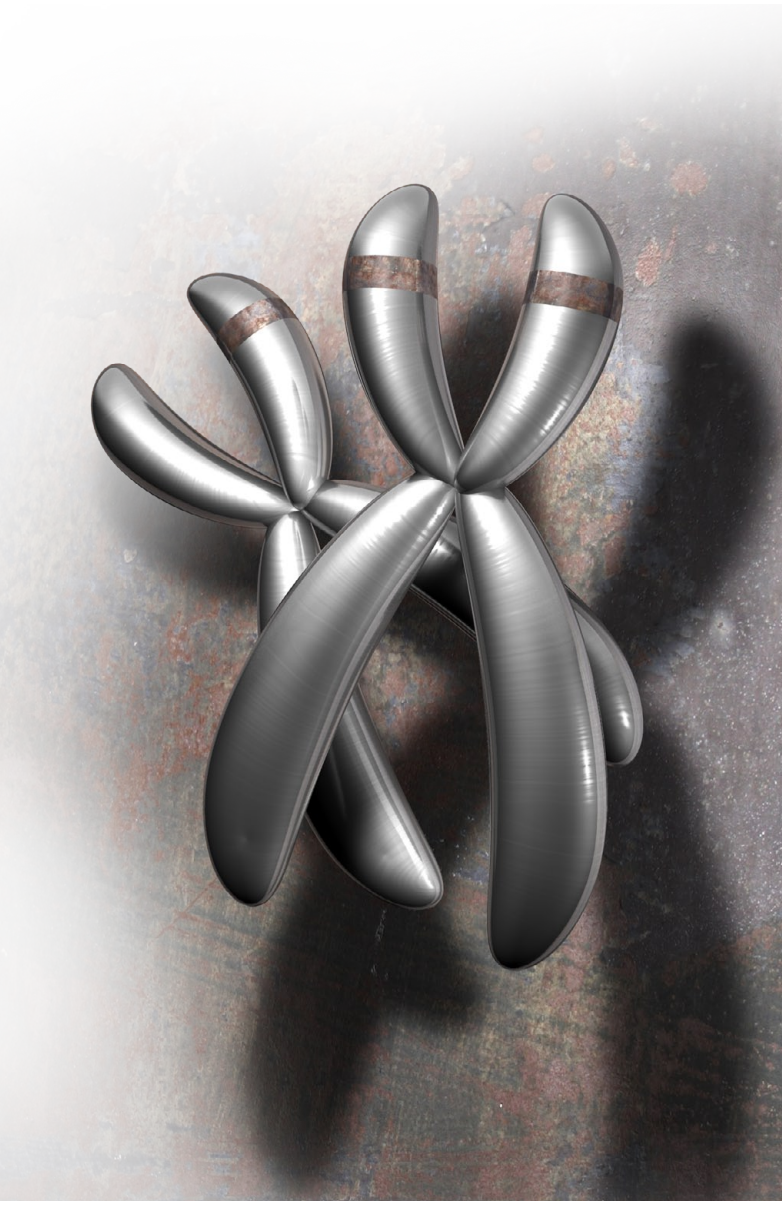
- What is the problem or underlying problem(s)?
- Change it to an aim
- What are the solutions or options?
- What are the barriers?
- How can you overcome them?

Overcoming barriers to promote change

- What is achievable – find out what the external pressures on the practice are and discuss ways of managing them
- Identify the barriers
- Develop a priority list
- Choose one or two achievable goals

Effective interventions

- No single strategy or intervention is more effective than another, and sometimes a variety of methods are needed to bring about lasting change
- Interventions should be directed at existing barriers or problems, knowledge, skills and attitudes, as well as performance and behaviour



Review

Monitoring change and progress

It is important to review the action plan at regular intervals. It may be helpful to review the following questions:

- Is the process working?
- Are the goals for improvement being achieved?
- Are the goals still appropriate?
- Do you need to develop new tools to achieve the goals you have set?

Following the completion of the first cycle, it is recommended that the doctor completes the first part of the Audit of Medical Practice summary sheet (Appendix 1).

Undertaking a second cycle

In addition to regular reviews of progress with the practice team, a second audit cycle should be completed in order to quantify progress on closing the gaps in performance.

It is recommended that the second cycle be completed within 12 months of completing the first cycle. The second cycle should begin at the data collection stage. Following the completion of the second cycle it is recommended that practices complete the remainder of the Audit of Medical Practice summary sheet.

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Claiming MOPS credits

This audit has been endorsed by the RNZCGP as an Audit of Medical Practice activity (previously known as Continuous Quality Improvement – CQI) for allocation of MOPS credits; **10 Credits** for a first cycle and **10 Credits** for a second cycle. General practitioners taking part in this audit can claim credits in accordance with the current MOPS programme.

To claim points go to the RNZCGP website:
www.rnzcgp.org.nz

Record your completion of the audit on the M**OPS Online credit summary**, under the **Audit of Medical Practice** section. From the drop down menu, select the audit from the list or select "Approved practice/ PHO audit" and record the audit name in "Notes", the audit date and 10 credits.

"MOPS online" can be completed by vocationally registered doctors or "CPD online" for general registrants.

General practitioners are encouraged to discuss the outcomes of the audit with their peer group or practice.

As the RNZCGP frequently audit claims you should retain the following documentation, in order to provide adequate evidence of participation in this audit:

1. A summary of the data collected
2. An Audit of Medical Practice (CQI activity) Summary Sheet (included as Appendix 1).

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Data sheet – cycle 1

Genetic testing for haemochromatosis

Patient	The patient's note or record includes the following information: (tick if information is present)		
	A. If genetic testing was ordered by this practice, do test results show elevated ferritin or transferrin saturation > 45% prior to genetic testing?	**B.** OR was testing performed as part of family screening following the advice of a clinician with genetic experience or genetic counsellor?	**C.** Positive result? (a tick in either column A or B)
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		Total positive results	
		% positive results	

Please retain this sheet for your records to provide evidence of participation in this audit.

Data sheet – cycle 2

Genetic testing for haemochromatosis

Patient	The patient's note or record includes the following information: (tick if information is present)		
	A. If genetic testing was ordered by this practice, do test results show elevated ferritin or transferrin saturation > 45% prior to genetic testing?	**B.** OR was testing performed as part of family screening following the advice of a clinician with genetic experience or genetic counsellor?	**C.** Positive result? (a tick in either column A or B)
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		Total positive results	
		% positive results	

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APPENDIX 1

Audit of Medical Practice (CQI activity) Summary Sheet

Topic:	**Genetic testing for haemochromatosis**
The activity was designed by (name of organisation if relevant):	**Bpac^{nz}**
Doctors Name:	

FIRST CYCLE

DATA:	Date of data collection:

CHECK:	Describe any areas targeted for improvement as a result of analysing the data collected.

ACTION:	Describe how these improvements will be implemented.

MONITOR:	Describe how well the process is working. When will you undertake a second cycle?

Please retain this sheet for your records to provide evidence of participation in this audit.

SECOND CYCLE

DATA:	Date of data collection:

CHECK:	Describe any areas targeted for improvement as a result of analysing the data collected.

ACTION:	Describe how these improvements will be implemented.

MONITOR:	Describe how well the process is working.

COMMENTS:	

Please retain this sheet for your records to provide evidence of participation in this audit.