Complete Summary

GUIDELINE TITLE

Screening for hereditary hemochromatosis: a clinical practice guideline from the American College of Physicians.

BIBLIOGRAPHIC SOURCE(S)

Qaseem A, Aronson M, Fitterman N, Snow V, Weiss KB, Owens DK. Screening for hereditary hemochromatosis: a clinical practice guideline from the American College of Physicians. Ann Intern Med 2005 Oct 4;143(7):517-21. [21 references] PubMed

GUIDELINE STATUS

This is the current release of the guideline.

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

Hereditary hemochromatosis

GUIDELINE CATEGORY

Screening

CLINICAL SPECIALTY

Family Practice
Gastroenterology

Internal Medicine
Medical Genetics

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

- To increase physician awareness of hereditary hemochromatosis, particularly the variable penetrance of genetic mutations; aid in case finding; and explain the role of genetic testing
- To answer the following questions:
    - What is the prevalence of hereditary hemochromatosis in the primary care setting?
    - In asymptomatic patients with hereditary hemochromatosis, what is the risk for end-organ damage or death?
    - How diagnostically useful are transferrin saturation and serum ferritin in identifying patients with hereditary hemochromatosis in the primary care setting?
    - Is phlebotomy efficacious in reducing morbidity or fatal complications in asymptomatic patients with hereditary hemochromatosis?
    - Do the benefits of screening primary care patients for hereditary hemochromatosis outweigh the risks?

TARGET POPULATION

All persons who have a probability of or susceptibility for developing hereditary hemochromatosis, including the relatives of individuals who already have the disease

INTERVENTIONS AND PRACTICES CONSIDERED

Screening for Hereditary Hemochromatosis

1. Case finding
    - Serum ferritin level
    - Transferrin saturation test
2. Genetic testing

MAJOR OUTCOMES CONSIDERED

- Prevalence of hereditary hemochromatosis
- Risk for complications of hereditary hemochromatosis in asymptomatic patients
- Sensitivity and specificity of diagnostic tests (i.e., serum ferritin level and transferring saturation)
- Effectiveness of phlebotomy
- Risks and benefits of screening

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Hand-searches of Published Literature (Primary Sources)
Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

This guideline is based on the systematic review of the evidence in the background paper (see "Companion Documents" field).

The authors of the background paper conducted a systematic review for each question in MEDLINE for papers published from 1966 through April 2004 by using PubMed Clinical Queries filters for a sensitive search of prognosis, diagnosis, etiology, or treatment depending on the question. They included only English-language studies. Two reviewers independently reviewed all abstracts. A third reviewer resolved conflicts about inclusion of an article. The authors also manually searched references from included studies. The appendix of the background paper includes details for conducting the search for each subquestion.

NUMBER OF SOURCE DOCUMENTS

Question 1: 3 studies in primary care settings and 12 studies in general population setting

Question 2: 11 studies on the relationship between transferrin saturation--serum ferritin level and hereditary hemochromatosis-related disease; 13 studies on the relationship between primary iron tissue deposition and hereditary hemochromatosis-related disease

Question 3: 3 studies

Question 4: 2 studies

Question 5: 4 studies

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Expert Consensus

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Not applicable

METHODS USED TO ANALYZE THE EVIDENCE

Systematic Review with Evidence Tables

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Expert Consensus

DESCRIPTION OF METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Not applicable

COST ANALYSIS

The authors of the systematic review (see "Availability of Companion Documents" field) reviewed published cost analyses.

METHOD OF GUIDELINE VALIDATION

External Peer Review
Internal Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

This guideline was approved by the American College of Physicians (ACP) Board of Regents on 16 July 2005.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

Recommendation 1: There is insufficient evidence to recommend for or against screening for hereditary hemochromatosis in the general population.

Recommendation 2: In case-finding for hereditary hemochromatosis, serum ferritin and transferrin saturation tests should be performed.

Recommendation 3: Physicians should discuss the risks, benefits, and limitations of genetic testing in patients with a positive family history of hereditary hemochromatosis or those with elevated serum ferritin level or transferrin saturation.

Recommendation 4: Further research is needed to establish better diagnostic, therapeutic, and prognostic criteria for hereditary hemochromatosis.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The recommendations are supported by data from cohort, cross-sectional, and case-control studies.

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

- Appropriate screening for hereditary hemochromatosis
- Serum ferritin level and transferrin saturation have been useful in identifying patients who are prone to or already have hereditary hemochromatosis.

POTENTIAL HARMS

- The value of detecting individuals who are homozygous for the mutation but do not develop iron overload is controversial. The psychological and social implications of identifying such individuals must be considered. Issues such as the impact on insurability and the anxiety of being labeled with a hereditary illness need to be considered when comparing the benefits and risks of screening.
- False reassurance in the setting of a negative genetic test result is not unreasonable.

QUALIFYING STATEMENTS

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Clinical practice guidelines are "guides" only and may not apply to all patients and all clinical situations. Thus, they are not intended to override clinicians' judgment. All American College of Physicians (ACP) clinical practice guidelines are considered automatically withdrawn or invalid 5 years after publication, or once an update has been issued.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

IMPLEMENTATION TOOLS

Patient Resources

For information about availability, see the "Availability of Companion Documents" and "Patient Resources" fields below.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Staying Healthy

IOM DOMAIN

Effectiveness
Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

Qaseem A, Aronson M, Fitterman N, Snow V, Weiss KB, Owens DK. Screening for hereditary hemochromatosis: a clinical practice guideline from the American College of Physicians. Ann Intern Med 2005 Oct 4;143(7):517-21. [21 references] PubMed

ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2005 Oct 4

GUIDELINE DEVELOPER(S)

American College of Physicians - Medical Specialty Society

SOURCE(S) OF FUNDING

American College of Physicians

GUIDELINE COMMITTEE

Clinical Efficacy Assessment Subcommittee (CEAS)

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

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FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

None disclosed

GUIDELINE STATUS

This is the current release of the guideline.

GUIDELINE AVAILABILITY

Electronic copies: Available from the Annals of Internal Medicine Web site.

Print copies: Available from the American College of Physicians (ACP), 190 N. Independence Mall West, Philadelphia PA 19106-1572.

AVAILABILITY OF COMPANION DOCUMENTS

The following is available:

- Schmitt, B, Golub, RM, Green, R. Screening primary care patients for hereditary hemochromatosis with transferrin saturation and serum ferritin level: systematic review for the American College of Physicians. Ann Intern Med. 2005 Oct 4;143(7):522-36. Electronic copies: Available from the Annals of Internal Medicine Web site.

Print copies: Available from the American College of Physicians (ACP), 190 N. Independence Mall West, Philadelphia PA 19106-1572.

PATIENT RESOURCES

The following is available:

- Summaries for patients. Screening for hereditary hemochromatosis: recommendations from the American College of Physicians. Ann Intern Med 2005 Oct 4;143(7):I-46.

Electronic copies: Available in Portable Document Format (PDF) from the Annals of Internal Medicine Web site.

Print copies: Available from the American College of Physicians (ACP), 190 N. Independence Mall West, Philadelphia PA 19106-1572.

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NGC STATUS

This NGC summary was completed by ECRI on November 23, 2005. The information was verified by the guideline developer on December 13, 2005.

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