



Complete Summary

GUIDELINE TITLE

Carrier screening for genetic disorders in individuals of Ashkenazi Jewish descent.

BIBLIOGRAPHIC SOURCE(S)

Langlois S, Wilson RD, Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada, Prenatal Diagnosis Committee of the Canadian College of Medical Geneticists. Carrier screening for genetic disorders in individuals of Ashkenazi Jewish descent. J Obstet Gynaecol Can 2006 Apr;28(4):324-32. [44 references] [PubMed](#)

GUIDELINE STATUS

This is the current release of the guideline.

COMPLETE SUMMARY CONTENT

SCOPE

METHODOLOGY - including Rating Scheme and Cost Analysis

RECOMMENDATIONS

EVIDENCE SUPPORTING THE RECOMMENDATIONS

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

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IMPLEMENTATION OF THE GUIDELINE

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT

CATEGORIES

IDENTIFYING INFORMATION AND AVAILABILITY

DISCLAIMER

SCOPE

DISEASE/CONDITION(S)

Genetic disorders in individuals of Ashkenazi Jewish descent, including:

- Tay-Sachs disease
- Canavan disease
- Familial dysautonomia
- Bloom syndrome
- Fanconi anemia
- Gauchers disease
- Glycogen storage disease type Ia
- Mucopolipidosis type IV
- Niemann-Pick disease type 1A

- Cystic fibrosis

GUIDELINE CATEGORY

Counseling
Risk Assessment
Screening

CLINICAL SPECIALTY

Family Practice
Internal Medicine
Medical Genetics
Obstetrics and Gynecology
Pediatrics

INTENDED USERS

Advanced Practice Nurses
Physician Assistants
Physicians

GUIDELINE OBJECTIVE(S)

- To give recommendations to physicians and midwives providing pre-conception or prenatal care about carrier screening for genetic disorders in individuals of Ashkenazi Jewish descent
- To offer carrier screening for Tay-Sachs disease (TSD) only or to expand the screening to include other disorders known to occur with increased frequency in the Ashkenazi Jewish population
- To offer carrier screening to the Ashkenazi Jewish population for conditions in which the benefits to the couple outweigh the risks, which include psychological distress from screening and diagnostic interventions
- To minimize practice variation across Canada with respect to carrier screening in individuals of Ashkenazi Jewish descent

TARGET POPULATION

Individuals of Ashkenazi Jewish descent

INTERVENTIONS AND PRACTICES CONSIDERED

1. Carrier screening
2. Referral for genetic counseling before conception or prenatally when both partners are carriers
3. Prenatal diagnosis using DNA analysis done on cells obtained by chorionic villus sampling or amniocentesis

MAJOR OUTCOMES CONSIDERED

- Risk of having a child with Tay Sachs disease, Canavan's disease, familial dysautonomia or other genetic disorder
- Psychological distress due to carrier screening
- Validity of screening tests

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases
Searches of Unpublished Data

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

The MEDLINE database was searched for relevant articles published from January 1966 to December 2004 related to carrier screening and genetic disorders in individuals of Ashkenazi Jewish descent. In addition, Canadian maternal-fetal medicine specialists and medical geneticists were surveyed to determine current practices and opinions.

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Weighting According to a Rating Scheme (Scheme Given)

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly designed randomized controlled trial.

II-1: Evidence obtained from well-designed controlled trials without randomization.

II-2: Evidence obtained from well-designed cohort (prospective or retrospective) or case-control analytic studies, preferably from more than one center or research group.

II-3: Evidence obtained from comparisons between times or places with or without the intervention. Dramatic results from uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

*Adapted from The Evaluation of Evidence criteria described in the Canadian Task Force on the Periodic Health Exam..

METHODS USED TO ANALYZE THE EVIDENCE

Systematic Review

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

Classification of Recommendations*

- A. There is good evidence to support the recommendation that the condition be specifically considered in a periodic health examination
- B. There is fair evidence to support the recommendation that the condition be specifically considered in a periodic health examination.
- C. There is poor evidence regarding the inclusion or exclusion of the condition in a periodic health examination.
- D. There is fair evidence to support the recommendation that the condition not be considered in a periodic health examination.
- E. There is good evidence to support the recommendation that the condition be excluded from consideration in a periodic health examination.

*Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on Periodic Health Exam.

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Internal Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

This guideline has been prepared by the Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Prenatal Diagnosis

Committee of the Canadian College of Medical Geneticists (CCMG) and approved by the Executive Council of the SOGC and the Board of Directors of the CCMG.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The quality of evidence (**I-III**) and classification of recommendations (**A-E**) are defined at the end of the "Major Recommendations."

1. Carrier screening for Tay-Sachs disease (TSD) (**II-2A**), Canavan disease, and familial dysautonomia (FD) should be offered to Ashkenazi Jewish couples. (**III-A**)
2. Carrier screening for other disorders seen with increased frequency in Ashkenazi Jewish individuals (e.g., Bloom syndrome, Fanconi anemia, Gaucher disease, glycogen storage disease type 1a, mucopolysaccharidosis type IV, Niemann-Pick disease type 1A, and cystic fibrosis [CF]) should be offered when there is a positive family history. (**III-A**)
3. When only one member of a couple is of Ashkenazi Jewish ancestry, screening should be offered for TSD only. (**II-2A**)
4. When only one member of a couple is of Ashkenazi Jewish ancestry, screening should not be offered for Canavan disease or familial dysautonomia (FD) because of a low carrier frequency and limitations of carrier screening (low detection rate in individuals of non-Ashkenazi Jewish ancestry). (**III-D**)
5. When both partners are carriers of the same autosomal recessive condition, they have a 25% risk of having an affected child. They should be referred for genetic counselling, either before conception or prenatally. Prenatal diagnosis would be offered and performed according to the patient's informed decision. Prenatal diagnosis would consist of DNA analysis done on cells obtained by chorionic villus sampling or amniocentesis. (**II-3A**)

Definitions

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly designed randomized controlled trial.

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Classification of Recommendations*

- A. There is good evidence to support the recommendation that the condition be specifically considered in a periodic health examination
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- E. There is good evidence to support the recommendation that the condition be excluded from consideration in a periodic health examination.

*The quality of evidence reported in these guidelines has been adapted from The Evaluation of Evidence criteria described in the Canadian Task Force on the Periodic Health Exam.

**Recommendations included in these guidelines have been adapted from the Classification of Recommendations criteria described in the Canadian Task Force on the Periodic Health Exam.

CLINICAL ALGORITHM(S)

A clinical algorithm "Approach to carrier screening with negative family history" is provided in the original guideline document.

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of supporting evidence is identified and graded for each recommendation (see "Major Recommendations").

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

Screening of couples of Ashkenazi Jewish descent will identify couples who have a 25% risk of having a child with a significant genetic disorder.

POTENTIAL HARMS

- The sensitivity of the tests being offered is not 100% in individuals of Ashkenazi Jewish descent and is significantly less or unknown in non-Ashkenazi Jewish individuals.
- Screening might identify couples where one member is a carrier and the other member is negative. Given that such a couple would be at low risk but not zero risk of having an affected child, screening might result in psychological distress, unnecessary prenatal diagnostic procedures, and possibly termination of normal pregnancies.

QUALIFYING STATEMENTS

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This guideline reflects emerging clinical and scientific advances on the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed. Local institutions can dictate amendments to these opinions. They should be well documented if modified at the local level. None of these contents may be reproduced in any form without prior written permission of the Society of Obstetricians and Gynaecologists of Canada (SOGC).

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

IMPLEMENTATION TOOLS

Clinical Algorithm

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)

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ADAPTATION

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

DATE RELEASED

2006 Apr

GUIDELINE DEVELOPER(S)

Canadian College of Medical Geneticists - Professional Association
Society of Obstetricians and Gynaecologists of Canada - Medical Specialty Society

SOURCE(S) OF FUNDING

Society of Obstetricians and Gynaecologists of Canada

GUIDELINE COMMITTEE

Society of Obstetricians and Gynaecologists of Canada Genetics Committee
Canadian College of Medical Geneticists Prenatal Diagnosis Committee

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Principal Authors: Sylvie Langlois, MD, FRCPC, FCCMG, Vancouver BC; R. Douglas Wilson (*Chair*), MD, FRCSC, Philadelphia PA

Genetics Committee Members: R. Douglas Wilson (*Chair*), MD, FRCSC, Philadelphia PA; Victoria M. Allen, MD, MSc, FRCSC, Halifax NS; Claire Blight, RN, Halifax NS; Valerie A. Désilets, MD, FRCSC, Montreal QC; Alain Gagnon, MD, FRCSC, Vancouver BC; Sylvie Langlois, MD, FRCPC, Vancouver BC; Gregory J. Reid, MD, FRCSC, Winnipeg MB; Anne Summers, MD, FRCPC, Toronto ON; Philip Wyatt, MD, PhD, North York ON

Prenatal Diagnosis Committee Members: Sylvie Langlois (*Chair*), MD, FRCPC, FCCMG, Vancouver BC; David Chitayat, MD, FRCPC, FCCMG, Toronto ON; Albert E. Chudley, MD, FRCPC, FCCMG, Winnipeg MB; Sandra Farrell, MD, FRCPC, FCCMG, Mississauga ON; Michael T. Geraghty, MD, FRCPC, FCCMG, Ottawa ON; Chumei Li, MD, PhD, FRCPC, FCCMG, Toronto ON; Sarah M. Nikkel, MD, FRCPC, FCCMG, Ottawa ON; Anne Summers, MD, FRCPC, FCCMG, Toronto ON; Frederique Tihy, PhD, FCCMG, Montreal QC

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline.

GUIDELINE AVAILABILITY

Electronic copies: Available in Portable Document Format (PDF) from the [Society of Obstetricians and Gynaecologists of Canada Web site](#).

Print copies: Available from the Society of Obstetricians and Gynaecologists of Canada, La société des obstétriciens et gynécologues du Canada (SOGC) 780 promenade Echo Drive Ottawa, ON K1S 5R7 (Canada); Phone: 1-800-561-2416

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This NGC summary was completed by ECRI Institute on March 9, 2009. The information was verified by the guideline developer on March 25, 2009.

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