



Complete Summary

GUIDELINE TITLE

Invasive prenatal testing for aneuploidy.

BIBLIOGRAPHIC SOURCE(S)

American College of Obstetricians and Gynecologists (ACOG). Invasive prenatal testing for aneuploidy. Washington (DC): American College of Obstetricians and Gynecologists (ACOG); 2007 Dec. 9 p. (ACOG practice bulletin; no. 88). [50 references]

GUIDELINE STATUS

This is the current release of the guideline.

COMPLETE SUMMARY CONTENT

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SCOPE

DISEASE/CONDITION(S)

Fetal chromosomal abnormalities (fetal aneuploidy)

GUIDELINE CATEGORY

Counseling
Diagnosis
Risk Assessment
Screening

CLINICAL SPECIALTY

Medical Genetics
Obstetrics and Gynecology
Pediatrics

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

- To aid practitioners in making decisions about appropriate obstetric and gynecologic care
- To provide clinical management guidelines for the prenatal diagnosis of fetal aneuploidies

TARGET POPULATION

All women, regardless of maternal age

INTERVENTIONS AND PRACTICES CONSIDERED

Screening and Diagnosis

1. Risk assessment for fetal aneuploidy
2. Invasive testing for prenatal diagnosis of fetal chromosomal abnormalities
 - Amniocentesis
 - Traditional
 - Early (considered but not recommended)
 - Chorionic villus sampling
 - Cordocentesis (percutaneous umbilical blood sampling)
3. Laboratory tests to diagnose aneuploidy
 - Metaphase analysis of cultured amniocytes or chorionic villus cells
 - Fluorescence in situ hybridization analysis
 - Comparative genomic hybridization
4. Ultrasonography

Counseling

1. Prenatal counseling concerning risks and benefits of screening versus invasive diagnostic testing
2. Counseling of women with chronic hepatitis B or C virus infections, or human immunodeficiency virus infection
3. Nondirective counseling concerning pregnancy termination

MAJOR OUTCOMES CONSIDERED

- Procedure-related pregnancy loss rate
- Procedure-related complication rate
- Risk for chromosomal abnormalities
- Positive predictive value of diagnostic/screening tests for chromosomal abnormalities

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

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

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

The MEDLINE database, the Cochrane Library, and the American College of Obstetricians and Gynecologists' own internal resources and documents were used to conduct a literature search to locate relevant articles published between January 1985 and June 2007. The search was restricted to articles published in the English language. Priority was given to articles reporting results of original research, although review articles and commentaries also were consulted. Abstracts of research presented at symposia and scientific conferences were not considered adequate for inclusion in this document. Guidelines published by organizations or institutions such as the National Institutes of Health and the American College of Obstetricians and Gynecologists were reviewed, and additional studies were located by reviewing bibliographies of identified articles.

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

Studies were reviewed and evaluated for quality according to the method outlined by the U.S. Preventive Services Task Force:

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 or case-control analytic studies, preferably from more than one center or research group.

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

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

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

Analysis of available evidence was given priority in formulating recommendations. When reliable research was not available, expert opinions from obstetrician-gynecologists were used. See also the "Rating Scheme for the Strength of Recommendations" field regarding Grade C recommendations.

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Based on the highest level of evidence found in the data, recommendations are provided and graded according to the following categories:

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

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

Practice Bulletins are validated by two internal clinical review panels composed of practicing obstetrician-gynecologists generalists and sub-specialists. The final

guidelines are also reviewed and approved by the American College of Obstetricians and Gynecologists (ACOG) Executive Board.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The grades of evidence (I-III) and levels of recommendation (A-C) are defined at the end of the "Major Recommendations" field.

The following recommendation is based on good and consistent scientific evidence (Level A):

- Early amniocentesis (at less than 15 weeks of gestation) should not be performed because of the higher risk of pregnancy loss and complications compared with traditional amniocentesis (15 weeks of gestation or later).

The following conclusions are based on limited or inconsistent scientific evidence (Level B):

- Amniocentesis at 15 weeks of gestation or later is a safe procedure. The procedure-related loss rate after midtrimester amniocentesis is less than 1 in 300 to 500.
- In experienced individuals and centers, CVS procedure-related loss rates may be the same as those for amniocentesis.

The following recommendation and conclusions are based primarily on consensus and expert opinion (Level C):

- Invasive diagnostic testing for aneuploidy should be available to all women, regardless of maternal age.
- Patients with an increased risk of fetal aneuploidy include women with a previous fetus or child with an autosomal trisomy or sex chromosome abnormality, one major or at least two minor fetal structural defects identified by ultrasonography, either parent with a chromosomal translocation or chromosomal inversion, or parental aneuploidy.
- Nondirective counseling before prenatal diagnostic testing does not require a patient to commit to pregnancy termination if the result is abnormal.

Definitions:

Grades of Evidence

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 or case-control analytic studies, preferably from more than one center or research group.

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

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

Levels of Recommendation

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

CLINICAL ALGORITHM(S)

None provided

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

Appropriate use of prenatal screening and diagnostic testing for fetal aneuploidy

POTENTIAL HARMS

Procedure-related maternal and fetal complications, including pregnancy loss, can occur with amniocentesis and chorionic villus sampling.

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

These guidelines should not be construed as dictating an exclusive course of treatment or procedure. Variations in practice may be warranted based on the

needs of the individual patient, resources, and limitations unique to the institution or type of practice.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

IMPLEMENTATION TOOLS

Audit Criteria/Indicators
Foreign Language Translations
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

Patient-centeredness
Safety
Timeliness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

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ADAPTATION

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

DATE RELEASED

2007 Dec

GUIDELINE DEVELOPER(S)

American College of Obstetricians and Gynecologists - Medical Specialty Society

SOURCE(S) OF FUNDING

American College of Obstetricians and Gynecologists (ACOG)

GUIDELINE COMMITTEE

American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins - Obstetrics

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Not stated

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline.

GUIDELINE AVAILABILITY

Electronic copies: None available

Print copies: Available for purchase from the American College of Obstetricians and Gynecologists (ACOG) Distribution Center, PO Box 4500, Kearneysville, WV 25430-4500; telephone, 800-762-2264, ext. 192; e-mail: sales@acog.org. The ACOG Bookstore is available online at the [ACOG Web site](#).

AVAILABILITY OF COMPANION DOCUMENTS

Proposed performance measures are included in the original guideline document.

PATIENT RESOURCES

The following are available:

- Diagnosing birth defects. Atlanta (GA): American College of Obstetricians and Gynecologists (ACOG); 2005. Available from the [American College of Obstetricians and Gynecologists \(ACOG\) Web site](#).
- Screening tests for birth defects. Atlanta (GA): American College of Obstetricians and Gynecologists (ACOG); 2007. Available from the [American College of Obstetricians and Gynecologists \(ACOG\) Web site](#). Copies are also available in Spanish.

Print copies: Available for purchase from the American College of Obstetricians and Gynecologists (ACOG) Distribution Center, PO Box 4500, Kearneysville, WV

25430-4500; telephone, 800-762-2264, ext. 192; e-mail: sales@acog.org. The ACOG Bookstore is available online at the [ACOG Web site](#).

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NGC STATUS

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